I'M RARE

KNOWLEDGE
IS POWER

NOVARTIS

Dear readers,

This book is for you, to read it with your loved ones. Among its pages you will find stories of lives that have become rare, as rare as the events that define them.

You will meet mothers and fathers who have learned to smile through the tears, holding the hands of their children, and you will love the little and big warriors who have learned to value every single, precious moment of life.

Here you will meet extraordinary women and men who do their best to give these people hope and help, because they are Rare, but not alone. Never ever.

They live complicated lives, but are rich in courage and passion. We are honoured that they have decided to share their experiences, and excited at the thought of being the intermediary between them and you.

We are very proud to present you with this amazing and exciting collection of stories. This book is dedicated to all Rare Children and their families, to their determination to never give up, ever.

WE ARE RARE AND WE WILL OVERCOME

Rare Autoinflammatory Council

AIFP (*Italy*) - **AMRI onlus** (*Italy*) - **ASPANIJER (***Spain*) - **ENCA** - **FMF** (*Spain*) - **FMF & AID Global** - **INBAR** (*Israel*) - **KAISZ** (*Netherlands*) - **KOURIR** (*France*) - **Mifrakim Tze'irim** (*Israel*) - **Sunflower Foundation** (*Russia*) - **Versus Arthritis** (*UK*) -

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We don’t remember days, we remember moments.

- Cesare Pavese

Preface

If you've never heard of Rare Autoinflammatory Diseases, or if you think you know all there is to know, then this book is for you.

Let's start by defining them.

Autoinflammatory diseases are very rare chronic diseases that usually have their onset in childhood. The majority of these diseases are genetically acquired, while for some we still don't know the exact pathogenesis (i.e. why a child get the disease).

Although the clinical manifestations are variable, depending on the specific disease and even within the same disease from subject to subject, there are some common manifestations. The majority of patients suffer from recurrent episodes, that may last from days to weeks, of high spiking fever and signs and symptoms of systemic and local inflammation, such as malaise, anorexia, joint pain, abdominal pain, thoracic pain, headache, skin eruptions.

For the intensity of clinical manifestations and the recurrence of flairs, these diseases usually have a profound impact on the patients' quality of life. Autoinflammatory disease are not only very rare (some forms are described in less than one hundred people worldwide) but their identification dates to the early years of this century. That may explain why many affected individuals have wandered for decades from medical center to medical center in search for a solution for their health problem and have been seen as "clinical dilemmas" by physicians.

In these years we have even witnessed grandparents or parents that have been diagnosed in later life for a disease they have been suffering since their childhood, just because their grandchildren/children have been diagnosed with a genetically determined disease.

For these reasons, patients with autoinflammatory diseases and their family have to cope with many of the aspects people with chronic diseases and rare diseases have to deal with: a disease that arrives as a "meteorite" to destroy an entire family's life (indeed many of the people telling their stories in this book divide their life in a "before" and an "after" the disease has come), the anguish when trying to find the name of your/your child's disease, the feeling of being alone, the struggle to accomplish everyday tasks, the changes in your appearance due to the disease or the drugs prescribed, etc.

In the pages of this book you will find glimpses of real life coming from people whose existence has been changed by an autoinflammatory disease. I've always thought that being a physician means dealing with the more intimate part of a person's life, and ever tried to do my job "on tip-toes" in an attempt to respect that intimacy. The authors of these stories are giving us the most private part of their lives to feel less alone and with the hope that other people will feel less alone too because sticking together make you stronger. You will find the stories of people who simply don't ever give up: they go forward, no matter the difficulties, finding strength in themselves, in their families, in friends, in associations, and in the words of a physician or a nurse. I've had the privilege of meeting some of them in person, I've looked into their eyes and listened to their voices—I felt that strength. This is a book to read with careful consideration because it speaks about suffering, but it is also full of hope and gratitude for those great little heroes. Our task, as a society, is to make every effort possible so they won't feel alone anymore.

Marco Cattalini
Paediatrician

01. Expectations, dreams and hopes

WHEN WE BECOME INTOXICATED BY HAPPINESS

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The only thing we learn when the future becomes the past is that nothing goes according to plan.

- Daniel Pennac

KATY

County Cork, Republic of Ireland

What I like about this house of ours immersed in nothingness is just that: the colour of nothingness. Wherever you look, you're overwhelmed by a symphony of green. The Irish countryside is simply gorgeous.

We live in southern Ireland, about half an hour from the city of Cork. There are no bus lines, no shops... but there's a very close community, we can count on each other. Everybody knows everybody else. Perhaps it's because we live in the middle of nowhere that we need to be united.

I lived in London before I met my husband. I was born there, studied there and had always lived there. I used to go to art school, hang out with friends. I had fun.

London offers a lot stimulation. Just go to the market in Camden Town, with its multi-ethnic stalls full of exotic foods and clothes of all shapes and colours. Or Carnaby Street, the shopping street par excellence, or in the small places around Soho where you can drink and listen to music, be enchanted. London is fantastic if you're young and single.

My best friend from college had attended university in Manchester, and near her home lived a boy named Fergus. We started seeing each other, I liked him. He's of Irish origin and his people live in the north of Ireland and in Scotland. Fergus practically crossed all of Ireland to

go to university, and that's how we met and fell in love.

We stayed in London for the first few years we were married, even though Fergus had told me from the start about his desire to return to Ireland. We lived in the centre of the city and as long as the children were young (we have three: Tomas, 24; Niamh, almost 21; and Orla, who'll be eighteen at Christmas) we were very happy there. The schools were fantastic and there were lots of parks for the children to play in. But then we started thinking about when they would attend secondary schools, and in that respect we were no longer satisfied with London. So, we fulfilled my husband's wish and moved to Ireland.

The houses were bigger, there was more greenery, beautiful gardens and schools, and a wonderful community. There was the affection and sense of sharing that was missing in London. My husband was still working in London—he's the head of his own company and he hadn't found anything suitable for him in Ireland. Our life went on quietly: Tomas, Niamh and Orla were growing up and we were doing quite well. Although we practically lived in the middle of nowhere, surrounded by countryside, the isolation didn't seem to bother them a bit. There were few neighbours and you had to take the car into the city or to the nearest shopping centre, but there were plenty of opportunities for them to see their friends. There was a stillness but it was comforting, maternal. I was really quite happy with my life. Maybe it's not so usual these days, but I really was. Of course there were squabbles between siblings, but that's normal, as well as occasional quarrels with my husband. We fought and made up, like everyone does.

When our youngest was almost thirteen years old, I started my own business together with a partner: a small preschool for children up to five years old. I put a lot of work and money into it and it was a critical moment for the business—we had to make ourselves known. One Sunday evening I went to pick Orla up in town. She'd spent the evening at the cinema with friends. When I saw her, she seemed odd, grumpy, her face was tight, pinched. 'Oh, dear', I thought, 'either she's had a fight with someone or she's about to come down with a bug!'

When she woke up the next morning I knew she was sick, really sick.

I kept her home from school and on the second day I took her to our family doctor. He didn't waste any time and immediately sent us to the Emergency Room at the hospital where they did a series of tests. They diagnosed her with a bad infection, prescribed antibiotics, and sent us home.
My life, that of my husband, of Tomas, Niamh and Orla, and everyone around us changed course that night. We were planets in orbit following our own trajectory, but a meteorite was about to hit us.

We didn't know it yet, but nothing would ever be the same again.

ORLA

County Cork, Republic of Ireland

It all happened suddenly. I was fine and then one night—poof!—I wasn't. I wasn't the same Orla as before.

That Monday morning, I woke up feeling extremely tired. I had spent the evening before out with my friends—nothing special, I think we just went to the cinema. As soon as I opened my eyes, I realised something wasn't right. As if my body didn't belong to me anymore; every movement took immense effort. I needed to get myself ready for school, but it felt like something literally separated me from my spine and I couldn't function! I stayed home for a week with a sore throat, horrible nausea, and I hurt all over. I was sick as I'd never been before in my life.

I GOT SICKER AND SICKER

AS THE DAYS PASSED.

It seemed like a nightmare I couldn’t wake up from.

BIRGITTA

Delft, The Netherlands

I've always lived in Delft. I feel at home there and really like it's human scale. There's a very famous painting at the Rijks Museum in Amsterdam, View of Delft. The painting, by Vermeer, really shows the special kind of light we have in our city.

We are famous for ceramics and for blue Delft pottery, but the flower market five minutes from downtown, in Brabantse Turfmarkt, is definitely worth a visit.

Before I got married, I studied to be a social worker. I had always wanted to be helpful, to work on something tangible, to go out and meet people, to understand for myself their problems and the difficulties they had to face. It makes me feel good, as if I'm a part of their lives.

I'd also always wanted to get married and have children, and that's what I did.

My marriage, however, did not last long; things between us did not go well.

I was pregnant with Jasmijn when we decided to split up, it didn't change our decision. I knew it would be hard, of course, but I never imagined how hard.

I was unprepared for what would happen. I had faced difficult situations before, but only for my work!

Who knows why, but when it comes to ourselves, to our lives,

WE NEVER EXPECT OTHER PEOPLE'S MISFORTUNES TO REALLY CONCERN US.

CARMIT

Kiryat Gat, Israel

I live in Kiryat Gat in the South district of Israel. It's a small town about 56 km from Tel Aviv and about 70 km from Jerusalem.

We're only an hour's drive from the centre of Tel Aviv but the place where we live is quiet and family-friendly. Here, children have everything they could wish for to make them happy. I've always lived here, but I was born in Ashkelon, not far away, about twenty kilometres, because there's no hospital in Kiryat Gat. My parents, however, are not originally from here; they moved from Morocco.

Although I am the third of five brothers and sisters, my parents allowed me to study. My siblings and I are very close, and we're also close with our parents. They continue to be supportive and an important point of reference for all of us, despite their age and the aches and pains that inevitably come with it.

Like all girls, I did my two years of compulsory military service. Once I finished my duty, I was about twenty or twenty-one and I started to study and work.

I wanted to contribute to the development of technologies and was more passionate about scientific subjects than most girls. So I studied Industrial Engineering and went on to graduate in Social and Economic Sciences at Ben-Gurion University.

I met my husband, Dudu, at work while I was still studying. We got

married right away; I was twenty-four.

The day we got married was wonderful. There were four hundred fifty guests. But in Israel a big wedding is a cultural fact: you have lots of family and friends, and they all expect to be invited to your wedding.

Typically, the ceremony is officiated by very religious men, some of whom recite prayers in the form of songs, and then the feast begins, with lots of good food and music. When it's all over everyone is exhausted and tired! But that's how it is with us, it's part of our cultural tradition.

Life is pretty good for us and we like where we live. We're very 'westernised', as they say. The economy and education all work very well and we also have the necessary medical care, without having to look elsewhere for what we need. There's a mix of cultures here—Jews and Muslims—and you can find and see everything.

We have three children. Ofir, who's fallen ill is the oldest, and we have two boys, Mori and Arad. My husband and I both work: he runs his own delivery company and I'm a buyer for a metal and steel company.

What's the saying? Everything was fine... yes, everything was really fine and

WE CERTAINLY DIDN'T EXPECT WHAT WAS ABOUT TO HAPPEN.

AISLING

Straffan, Republic of Ireland

My name is Aisling and I live in County Kildare, Ireland. We live in a small town not far from Dublin, where the hospitals are.

I'm a flight attendant, or rather, I was until the birth of my son, Jack, who is now thirteen years old. I also have a daughter who's almost fifteen.

I used to work for Aer Lingus and I can say I've travelled the world. I liked it, but it’s clearly not a job you can easily manage if you have small children, and especially if you have small children who are seriously ill . You don't have regular schedules or the possibility to run home if you’re called from school because your son has vomited, or has a fever; in that moment you might be flying over his head or thousands of miles away.

Before I got married I had a great interest in horses, like most people around here. I loved them, and still love them, but back then I had my own horse and especially the time to take care of it. It was wonderful.

Our county emblem is a white shield with a red decusse, the cross of St Andrew. Wherever there’s space, you’ll find it: there’s the Cross of St Bridget; a horse's head, because we’re very involved in raising horses; an oak branch that recalls the meaning of the name Kildare, ‘church of the oak grove’; and a harp.

OUR MOTTO, MEANMA AGUS MISNEACH, MEANS 'SPIRIT AND COURAGE' IN GAELIC.

I always liked our motto, I think it perfectly reflects the Irish spirit.

What I couldn't have imagined was how closely it would affect my life. Spirit and courage! It really did help me a lot when our lives changed.

EMER

Kildare, Republic of Ireland

If I think back to the past, to my childhood, what I remember most happily is the carefree life we all led. I'm part of a big family—I have six brothers and one sister—and we lived in Wicklow, out in the country. Our farm was big and we certainly didn't have time to get bored!

Today things have changed a bit. It's not so common to find large families like mine or my husband Brandon's, with four sisters and two brothers.

I, myself, have three children but I would certainly not call ours a big family. Let's say we're average, which is exactly 2.5 children per family, a realistic figure for the country these days. I'd say that four children or more would be considered a large family, but today, you don't run into families like that anymore.

Even with three children, taking care of the daily stuff certainly isn't easy. You still need to know how to organise it all and, well, if you've got two who're seriously ill, it's even more complicated.

I had a job before I got pregnant. I was a manager at a biomedical company, CeramOptec, where I handled wages, recruitment, and imports and exports of optical fibres for medical purposes. I didn't have a medical background, but my work was interesting and I really

liked it.

My husband, on the other hand, is a project manager at an electrical company. He leaves early in the morning—at 5 a.m.—and gets home around 7 p.m. So, he's practically not here with us at all. He misses out on the best part of the day, to tell the truth.

Sometimes, I feel like I'm in one of the episodes of 'Father Ted'! It's a very popular TV series in Ireland about life in a small, country parish, although compared to the events in the show we're a little more... cosmopolitan! There's a scene in the first episode of the first series, I think, in which Father Dougal looks out the window and exclaims, 'Oh, what a beautiful day!' But actually, you can see that a hurricane is brewing.

WELL, SOMETIMES I FELT LIKE THAT — OUT OF THIS WORLD AND OUT OF CONTACT WITH REALITY! AND IN THE MIDDLE OF HURRICANE.

My name's Emer, and this is my story.

ANTONIO

Ludwigsburg, Germany

Thirty years ago in the province of Avellino, in what's called the Alta Irpinia part of Italy, due east of Naples, there weren't many possibilities for people who were looking for a job. I tried, but I couldn't find anything. I was sixteen years old, my relationship with my parents was rocky, we argued, and so I decided to emigrate. Lots of folks did at the time—to Switzerland or Germany, for the most part. I chose Germany. I had a sister in Stuttgart and I joined her there.

I remember leaving by train. I had very little money in my pocket, just enough to buy the ticket. Or rather, I bought the ticket, but only as far as Chiasso, on the Italian-Swiss border. It was all I could afford. So, when I got off the train in Chiasso, I waited for the ticket inspectors to work their way slowly towards the carriages at the front and slipped into one of the carriages further back. I did this at every station until Stuttgart. I felt my heart in my throat for fear they would find me and chase me away. I couldn't even return home! And if I did, it would mean even more quarrels with my parents.

It was midnight when I got to Stuttgart and I had to take the metro to my sister's house. But once I got there, I couldn't find where she lived and it was too late to ask around for help. I slept outside, and fortunately the next morning I found her. It wasn't easy to get a job in Germany either. I was still a minor, and nobody wanted minors. I started out as a dishwasher, then as time went by I worked my way up. I learned to make pizzas with a Sicilian man who, unfortunately, has passed away. I remember him affectionately because he taught me so

much.

I met my wife Anna in Stuttgart. I went to a shopping centre with a friend of mine who, as it turned out, was her uncle. As soon as I saw her I felt butterflies in my stomach, she was so beautiful. I think I fell in love with her instantly—I could hardly hear my friend's words as he introduced us.

We got married right away, I was 20 years old and my wife a little younger. It was May 14, 1990, it was a hot afternoon and it was a Friday. It's been almost thirty years but I love her as if it were that first day at the shopping centre, with butterflies in my stomach!

Right after the wedding, we left for Italy to meet our respective families. While we were in my village, my wife started not feeling well. I had a cousin who was pregnant at the time and she convinced Anna to go with her to the gynaecologist. The doctor examined her and told us she was pregnant! We were really very young but we wanted a child right away, we wanted to build our family. We were both very happy, so thrilled.

My wife named our first child. She liked the name Christopher, and so that's what we called him. Brian, on the other hand, was Christopher's choice. He listened to that American group, what are they are called?... Ah, yes, Backstreet Boys, and he was crazy about one of the members of the group, Brian.

Ten years later we were all sitting at the dinner table and my wife was pregnant again. We were eating and talking about the baby's name. We still didn't know if it would be a boy or a girl; we'd chosen Angela for a girl's name. It seemed logical to me: it was my mother-in-law's name, besides it's a beautiful name. We didn't agree on the boy's name, though. I thought of Moses but the boys thought it was hilarious and couldn't stop laughing. I suggested Patrick, but they kept laughing. Eventually they told me he'd be named after me, Anthony, and his second name, Angelo, would be after my mother-in-law. I felt the tears welling up behind my eyelids, but I held them back.

They were tears of joy,

BUT AT THAT MOMENT I NEVER IMAGINED I WOULD SHED MANY, MANY MORE. BUT NOT OF JOY.

ANNA

Ludwigsburg, Germany

My mother died when I was just seventeen. I loved her very much, we were very close. I was so upset and couldn't take it anymore. My grandmother decided I needed a change of scenery, away from Calabria, where we lived.

She sent me to Stuttgart where a married aunt lived with her family. They had a restaurant and I could help them out at work and with the children.

One day, we were at a shopping centre and I ran into my uncle, who was there with a friend of his. He introduced the friend to me—it was Antonio. I saw him more and more often because my uncle, who probably wanted to make something happen between us, always brought him along to the home of one aunt or another.

We fell in love and decided to get married. We were two kids, basically, but we loved each other and wanted our own family, both of us love children.

The wedding day was beautiful. It was hot. It was like a dream.

LENNY

Born, The Netherlands

I'm the oldest of four children. Between me and my brother, who's seven years younger than me, there are two others—a brother and a sister. I was born in Buchten, a town near my village which is too small to have its own hospital. Not that Born is much bigger... I think today it has about ten thousand inhabitants and in the next town, where my parents live, I think there are just five thousand.

I had a happy childhood, even though I had a few stumbling blocks here and there.

I've had symptoms since birth, but at the age of 10 I had the first big flare; that's when everything really began.

I STILL REMEMBER THAT DAY PERFECTLY. IT WAS A MONDAY AND I WAS AT SCHOOL.

I was about ten or eleven years old. We were in the school canteen and I wasn't feeling very well. It was autumn, I remember, and when I

got home my mother thought it was something seasonal, like a cold or a flu. She kept me in bed for a few days. By Wednesday morning my cold had almost disappeared so she said I could go back to school the next day.

When I woke up on Thursday, however, I felt awful. I couldn't walk and my whole body ached. I ended up staying home sick for eleven weeks.

I was even admitted to the hospital, where they took a blood sample. They thought I had Sjogren's syndrome and Systemic Juvenile Idiopathic Arthritis.

My grandmother exclaimed, 'So that's what it is!' Finally those red spots and widespread pain that afflicted both my grandmother and my mother had a name.

After the diagnosis of my disease, which was a rare one, they checked my brothers and sisters, but only my youngest brother had a chance of being affected as well. Almost thirty years have passed since then but he's always been well and has never suffered from anything.

When I started going to school again, my teachers came to pick me up by car for the first few days even though my house was only five hundred metres away. They didn't want me to have to walk and get tired.

It was the only time I really got sick. The following years passed peacefully enough. I had good periods and less-than-good periods, but all in all it wasn't bad. The only area where I was limited was sport, but I have to say I wasn't very interested in it.

All things considered, I lived fairly well.

It was my mother who held me back, who told me 'don't do this' or 'don't do that, it might hurt...' In fact, I think she felt guilty that she was the one who passed the disease on to me, so she tried to save me from as much pain as possible[1].

My father didn't talk about it much—he's always been a pretty quiet

guy. He had hearing problems and so he'd had a hard time at school learning to read and write, and his teachers didn't help him. I think he always suffered from an inferiority complex. When we got together with people from outside our closest family he always avoided expressing his opinion. I think he thought he wasn't smart enough.

But whenever I needed him, he was there for me. He would take me up and down the stairs or put me in the bathtub when I didn't have the strength to do it alone.

MY MOTHER LET THE DISEASE RULE HER LIFE. SHE OFTEN COMPLAINED ABOUT HER PAIN, OR THAT SHE WAS FORCED TO GIVE UP MANY THINGS.

I can't say it was a walk in the park for me, but I didn't want to follow in her footsteps because I knew they would take me nowhere. Of course I couldn't do all the things other kids did. Sometimes I had to stay at home on the sofa while my friends were out somewhere having fun without me. But I was stubborn, and I wanted to live outside my illness.

I remember that one year, when I was about seventeen or eighteen years old, there was a play that I wanted—with all my might—to participate in. It was a performance that took place every year, around the time of Carnival. It was February and it was really cold. In short, I was able to act my part, clenching my teeth through the pain. I knew I would pay dearly for the effort but at that moment I didn't care. I went on stage thanks to adrenaline, to medication... I kept my sights on my friends who were with me and on my parents who were there, applauding at the end. The following days were hard, but it was worth it. I won, not my disease.

The only thing I regret is having to give up on my dream of becoming a paediatrician. I would have liked to treat children who, like me, had major health problems but I soon realised that being ill myself, I could never guarantee that I'd be there when they needed me.

My two daughters—Tess, the oldest, who's now ten, and Kate the youngest—are also ill. Do I regret having them? No. The disease didn't beat me in that either.

My name is Lenny, and I'll tell you the story of my family.

[1] PSYCHOLOGIST'S BOX

The occurrence of a pathology, which is also not very well known, causes a profound break in the psychological balance of people. Uncertainty, fear, doubt, anger are recurrent emotions to be managed for the psychophysical well-being of the whole family.

Linda Bergamini,
psychologist, Milan, Italy

SUSANNE

Dillingen, Germany

I live in Dillingen, a small village in Bavaria in the central part of Germany. It's not as famous as Munich, and of course we couldn't organize the Oktoberfest in our town, but it’s precisely for its smallness that I like living here.

We don't have supermarkets, we don't even have a railway station. The main activity is agriculture. I really like living in the country, and we all like it very much. We're a small, very close-knit community.

My name is Susanne and I've lived here since I was born. I grew up and studied here. I’ve always liked children, so I became a kindergarten teacher. I also worked for a long time in a nursing home, then I specialised as a medical assistant and worked in a paediatrician’s surgery. After I got pregnant, I had to stop working.

I met my husband by accident, you might say. There was an outdoor concert but I didn't feel like going, I wasn’t in the mood. It was a friend of mine who insisted. She literally dragged me there, and I met Robert that evening.

We dated for five years, then decided to get married and went to Egypt on our honeymoon. It was the first time we visited that country.

When I got back, I was already pregnant and happy to be so.

We both wanted to have children right away and our wish was

coming true. We were young, in love, on our honeymoon and with a baby on the way. What more could we have wished for?

At that moment, there was no clue about what was going to happen. It seemed that our life was all set.

ROBERT

Dillingen, Germany

Susanne and I were very young when we got married. And our first child came along right after the wedding; Susanne got pregnant on our honeymoon. We didn't plan it that way, actually, but it happened and we were fine with it.

Susanne was working at the time but when she reached her sixteenth week of pregnancy, she began to have some strong contractions and had to stop.

Marco, our first son who’s now eleven, was born early, at the fortieth week.

SUSANNE

Dillingen, Germany

The day Marco was born, I felt incredibly excited and nervous. It wasn't time yet, but I knew it was about to happen. My doctor wouldn't listen to me and I spent the whole day anxiously, not feeling well. It was only in the evening, however, that the gynaecologist decided to intervene with a Caesarean section, and Marco was finally born.

That's how it started, all of a sudden.

**IT WOULD CHANGE US.
IN FACT, WE WERE
ALREADY CHANGING,
ALL OF US,
BUT WE DIDN'T
KNOW YET.**

TIZIANA

Partinico, Italy

Living in a small town in Sicily has its particularities. Compared to when the author Luigi Pirandello was alive, not that much has changed. There's a strong sense of honour and family, and it affects many aspects of social life.

In my father's house, it was 'My house, my rules.' Not that he was a despotic father—absolutely not—but he was like that, you could say he didn't have a very open mind.

One day he had a bad accident and had to stop working. I was thirteen at the time and was going to school. Little money came into the household with my father out of work so, of my own free will, I stopped going to school and found a job. I felt I had to. I wanted to help my family.

I started out working in a coffee bar, I served at the bar and worked in the kitchen too. To tell you the truth, I preferred being in the kitchen to being out front. I love to cook!

When I was sixteen I had to have an operation for appendicitis and they had to remove an ovary too because of an ovarian cyst. I was in bad shape and I quit my job.

After the bar, I got a job in an orthopaedic surgery as a secretary. I really liked it—it was quiet and I enjoyed the work.

I met Francesco, who would become my husband, in a bar when I was seventeen. He started following me around, and I liked him.

He asked my parents for permission to court me. We're not talking about a century ago, mind you, but only a handful of years in the past! But that's the way it was.

With my mother's consent, we saw each other regularly for five years, but without involving our families. But the village was small and people began to whisper. In fact, instead of whispering, they spoke quite openly, saying 'Why don't the two sets of parents know each other? Who knows what's going on…' and things like that. It was perfect material for a novella! Pirandello would have felt right at home with this kind of thing.

So, we got officially engaged, but from that moment on my already limited freedom was reduced even more. We could no longer go out on our own for an ice cream or a pizza. We were always within eyesight and there had to be someone to chaperone us—a sister, an aunt, a friend. It was quite an ordeal! Under those conditions, a couple couldn't wait to get married so they could finally be alone.

Even though I reminded my mother that if I wanted to kiss my boyfriend I would do it even with whole town in tow, she was unshakable. Alone, the two of us? Never!

It was my village and my life and, for better or worse, a person got used to it, or at least resigned to it. Anyway, I still love our traditions. I think there's too much freedom today.

So you can see how what is called a *fujitina* in Sicily is the easiest way to overcome the restrictions. You decide not to go home one evening and, at that point, there's no reason to wait any longer to get married, it's the only respectable thing to do. We were twenty-four when we did a *fujitina* too. We were tired of waiting.

We decided on it one night at dinner while we were having pizza. It was one of the few times we got away from my parents. We looked at each other and said, 'It's now or never.'

After we finished dinner we didn't go home. Since we didn't want anyone to worry, we called to reassure them, telling them what we'd done and that now we'd have to get married. After all, once we had taken that step, we already felt like we couldn't turn back. We have a saying: 'the iron's behind the door.' It means the door's been closed and can't be pushed back open. We knew we couldn't go home as if nothing had happened.

Of course, we didn't have a big wedding but we were very happy anyway.

We thought that our happy ending had finally come.

WE COULDN'T IMAGINE WHAT THE FUTURE HAD IN STORE FOR US.

ATAR

Lombardy, Italy

I was born in Israel, a beautiful country. Many of my relatives—all my grandparents, my father, uncles, cousins and stepbrothers—still live there and I visit them at least twice a year. Israel is a country that has a great deal of open-mindedness but at the same time it's full of contradictions.

Even though I only go there a few times a year, I've remained attached to my roots. You'll see a Shabbat candle lit on Fridays in my house only once in a while, but those are my roots. Although I'm not practicing, I want my daughters to know where we come from.

My grandmother is a holocaust survivor, they even wrote a book about her and Steven Spielberg's staff interviewed her for the Yad Vashem Museum in Jerusalem. Her story is a piece of the story of us all. I'm proud of her strength and I'm happy to have some of it in my blood too. Maybe that's why I didn't let myself go, despite everything that's happened to us.

I moved to Italy with my parents when I was six or seven years old, so most of my memories are linked to this country.

I graduated in economics and business in Rome, more to satisfy my parents' wishes than mine. Actually, I dreamed of being a doctor—the subjects at university were light years away from my real interests. However, once I started on the path that was set out for me, I did my best. I graduated with a maximum score and top honours.

Immediately afterwards, I participated in selections for World Foundation programmes organized by the United Nations. I was very interested in cooperation and development, and that was where I wanted to apply myself. Unfortunately, the allocation of positions follows a method of quotas for each country: if I'd been an Italian citizen, I wouldn't have had any problems being admitted but, being an Israeli citizen, I was excluded because the quotas for Israel had already been met.

My boyfriend at the time suggested I try sending my CV to Milan. 'It's *the* city for economics,' he said. 'Try it, and then you can decide.' I started receiving job offers three days later. Milan and economics had decided for me.

I was twenty-four when I met my husband and it wasn't long before we got married. We moved from Rome to Milan.

I was working for one of the largest international banks, and later I got a job in the field of high finance for Private Equity in an office near the *Duomo*. They were jobs many people my age would have sold their mother to get, but it wasn't my dream. However, I can't complain—it was stimulating and I got to deal with very interesting people and situations.

But my life was poorly suited for a mother with children. So, when my husband and I began talking about starting a family, I went to work for a large temporary employment agency. The hours weren't as long—I didn't get home at ten o'clock in the evening anymore. I was in HR, managing the 'big clients.'

That was the work I was doing when our daughter, Claire, was born in 2007. We had decided to have a child but were convinced it would take some time. Instead, we conceived Claire immediately. What an amazing emotion!

I called my grandmother, who was in America at that time with my aunt, to tell her. She was in the car with my aunt and cousins and I could hear them all laughing and shouting with joy. They told me they had to pull the car over because they were so excited, and motorists

who passed by looked at them strangely. It must have been a funny scene: an old lady laughing and screaming with other crazy people in a car!

I had just turned twenty-eight and my husband was almost thirty.

I continued working until 2009, when my husband got a job offer in South Africa, in Johannesburg—an opportunity not to be missed. That's when we decided to give Claire a little brother or sister.

Many people asked me if I was afraid of going there with a little girl and another child on the way, of giving birth and living there, but I was calm about it.

We stayed in Linksfield which, along with Bedforview, are nice areas of Joburg; we lived in a sort of 'golden cage', as we called it, because it was the only place where you could stay relatively safe. In the rest of the country, crime was almost out of control.

When we moved in 2009, work was in progress for the 2010 World Cup. In fact my husband, who's a major works engineer, was hired for that project.

I fell in love with South Africa, with the nature reserves where I could get away on weekends, with the amazing sunsets. They looked like oceans of fire burning up the horizon, with huge trees rising up in black profile against the light.

Eventually however, an episode occurred that convinced us to leave. When Michelle was 5, my husband was responsible for a construction site for a water system and he had to hire two hundred fifty people because of an agreement with the local province. When the hiring was complete, the problems began: people kept coming, demanding to be hired. Threats led to physical assaults. My husband was assaulted and wounded and we decided to leave the country.

Michelle was born prematurely and due to serious problems she was admitted to intensive care. What should have been a happy event was instead a source of anguish.

Within a month the situation improved and she was declared out of danger; they discharged her from the hospital. She just needed to gain weight, the worst was over.

One day about a week after we had returned home, I had just fed her and she collapsed.

I NEVER COULD HAVE IMAGINED, WHEN I WAS IN MY PREVIOUS LIFE, THAT THINGS COULD GET SO COMPLETELY OUT OF HAND.

LUC

Amersfoort, The Netherlands

My name is Luc, I'm thirteen years old and now I live in a village near Utrecht in the Netherlands. It's a quiet town and it's not very crowded, everybody kind of knows each other and helps each other out.

When my brother and I were little, my parents, who came from here, moved to America for work. We lived in New Jersey for about two or three years and then in Greenwich, Connecticut. It was a lot different from here. We didn't live in a place like New York City—the city that never sleeps—but there were definitely a lot more people compared to where we live now—there were more buildings and things weren't so green. I liked living in America, but I really like it here too. Anyway, I'm good at adapting. My parents used to travel a lot for work and I learned to socialise, to make new friends in every place where we moved.

Besides, having a brother helped me not to feel lonely and lost. He's sixteen, and when we were little and I was sick, he kept me company and played with me to distract me. We watched the Harry Potter films together and he bought me my favourite magazines. Now we fight sometimes, but I think that's pretty normal between brothers…

The rare disease I have started when we were still living in America, and as far as I can remember my parents were very worried about me.

I GOY SICK A LOT, EVERY MONTH.

I remember the really bad headaches, the nausea, the joint pains, and stomach pains and sore throat... I still suffer from it even now.

I don't think I noticed that I had something different from the other kids, but I saw my parents were anxious, that's for sure[2].

[2] PSYCHOLOGIST'S BOX

Parents may have concerns not only about the present but also about the future of their children and not only for their health, itself, but also for the fear that they do not have the same chances as their peers.

Linda Bergamini,
psychologist, Milan, Italy

EVA

Schoten, Belgium

My name is Eva, I'm thirty-five years old and I live in Schoten, a small town in Flanders in the northern part of Belgium. We're close to Antwerp, which is a much larger city and a tourist magnet for those who come to visit our country.

I was a popular girl during primary school, always surrounded by friends and involved in a thousand adventures. I had a trouble-free life, similar to that of many of my peers. Family, school, games, sport, friends and family. All wonderfully normal and wonderfully mine. I loved gymnastics and trained almost every day, and I also loved ballet.

One October afternoon in 1994—I remember it as if it were yesterday—my life changed. I didn't feel very well when I got home from school, so I went to bed; I had a fever, a sore throat and a stomachache. Nothing serious, a bit of laryngitis, I thought. The next day, Saturday, my grandmother came to stay with my brother and I because my parents had to go out. I still wasn't well and the thing I remember most clearly is the feeling of immobility in my legs. I couldn't move them. They felt like two tree trunks and I had to get my grandmother to help me move them.

When my parents came home, they understood immediately it wasn't a normal laryngits. My grandmother told them that I hadn't even been able to swallow the medicine because of my sore throat. The family doctor prescribed blood tests, which revealed a very high level of white blood cells. Fearing meningitis, my parents took me to the

emergency room. The bone marrow tests were negative, and my paediatrician thought it might be a rheumatic disease.

My life was already going in a different direction, but we didn't know it yet.

WENDY

Tipperary, Republic of Ireland

My name is Wendy, I live in Ireland and I have a nineteen-year-old daughter. When she was three, she was diagnosed with Juvenile Idiopathic Arthritis.

It was a shock. There were no paediatric rheumatologists at the time in Ireland and we knew nothing about the disease, except for what I could find by myself on the internet.

WE FELT ALONE, AS IF WE WERE DRIFTING IN THE UNKNOWN.

The road we had before us just couldn't be a solitary one, we wouldn't make it if it was. I feared my daughter wouldn't make it, and there was no way I could accept that.

I started contacting families who had similar problems to ours. I wanted my daughter to meet other children like her. I wasn't expecting it to be easy, but I thought it would be useful for comparing experiences, sharing information. It could help everyone to cope with the disease just a little bit.

With other parents, we set up the iCan organisation, Irish Children's

Arthritis Network. We're an association of volunteers; I'm the president and also take care of the administrative part. Five years ago we obtained status as an official charity association and our goal is to support families, promote better services, and raise public awareness of the disease.

That's how it all started. The fact that I was touched by the disease, through my daughter, made me aware of the needs of the people who have to deal with it.

I'm ready to give families all my support and understanding. I know how they feel. I know what they're facing.

NORMA

Dublin, Republic of Ireland

I had an aunt who worked as a nurse in a hospital. When I was little, I went to visit her. I liked watching her among the patients, I liked her loving way of approaching them. What I remember is her smile, she always had one for everybody.

I got my passion for the work I do from her: I'm an advanced nurse practitioner now, too.

I chose to specialize in paediatric care twenty-five years ago, and for the last eighteen years I've worked in the field of childhood pain and paediatric rheumatology.

What took me down this road was seeing how these children, even though they're cornered by a rare autoinflammatory disease, don't give up. Despite their pain, the hardships and their fears, they get up and go to school, they participate in normal social life, they do sports.

THE DISEASE HAS NOT BEATEN THEM, THEY FIGHT EVERY DAY LIKE LITTLE WARRIORS.

Sometimes I wonder if it's the illness that brings out their best

qualities, makes them so strong, and so combative. They often become ambassadors of hope and come to our meetings to talk about their experiences. They're sources of great inspiration and comfort for those who share their situation, and are an example and offer encouragement for those who don't. They're fantastic, really, and move me every time. And every time, I tell myself that no other job in the world could have given me so much.

To those who would like to enter this profession, I would say, first of all, 'Listen. Observe and listen.' You have to get in tune with the parents, the patients themselves, you have to let them vent.

Often, when they come to us, they've already been searching the internet for a long time to hunt down diagnoses, and they're scared and confused. We have to make them understand that, first of all, we are there for them and we are ready to listen to their stories. Their stories can be painful and have landed on their lives with the force of a meteorite, devastating them, but we listen. We are by their side to restore their hope.

I'm Norma, and this is my story.

MICHAL

Haifa, Israel

My name is Michal and I live in Haifa, the best city in Israel. It has long beaches and the sea is deep blue, you can surf and sunbathe, but the city's actually built on a mountain, Mount Carmel. You can go swimming and look up at the mountains. It's a fantastic sensation!

Haifa is built on three levels. It has the 'low' part with the beaches and the port; the 'central' part, where there's the commercial heart of the city; and the 'high' part, where you can take beautiful panoramic walks. Throughout the city—just like everywhere else in Israel—you can breathe in the past. Wherever you walk there are ancient ruins. In the Old Town you can still see the remains of the old constructions and you get the distinct impression of walking through history.

In Haifa there are Muslims, Christians and Jews all living side by side in harmony. When people say 'Israel' they think it's synonymous with 'war', but not here.

I was eighteen years old when I joined the army—here it's compulsory for everyone at that age, men and women. I liked it and decided to stay on, and I slowly climbed higher and higher up, eventually becoming an officer.
I was the commander of a radar station, I mentored new recruits in our field. As my last job, before my discharge as Lieutenant Commander, I was responsible for almost all the operations performed by the Israeli Navy.

I stayed in the military for almost twenty-six years and then started a new job as an administrative manager for a large company.

After five years of working there, I came down with severe inflammation, which forced me to stay at home for ten months! That's when my ordeal began, between examinations and diagnoses, before hitting on the nature of my disease.

AUTOINFLAMMATORY SYNDROMES (part 1)

Autoinflammatory syndromes are a group of rare diseases. The clinical description of the first diseases come from the second half of the last century. Although these diseases are very rare, some skilled physicians recognized their existence studying patients with recurrent episodes of high spiking fever plus signs or symptoms of systemic and organ-specific inflammation, and without an autoimmune disease or an infection justifying these episodes. For the first years, the autoinflammatory syndromes were also called “periodic fever syndromes”. The fact those diseases were genetically determined was clear from the first clinical descriptions, that showed entire families affected.

Almost 20 years ago the first genes whose mutations were causing an autoinflammatory syndrome were identified. The identification of the genes causing these recurrent fevers – that from there on will be identified also with the term “Monogenic Recurrent Fevers”- was a cornerstone in the field of autoinflammatory diseases: entire generations of people with clinical pictures that until then were considered “clinical dilemmas” finally received a definite diagnosis. The knowledge on autoinflammatory syndrome rapidly evolved.

The identification of the first genes involved pointed out that autoinflammatory syndromes are diseases caused by a perturbation in the mechanism of innate immunity that cause inflammation. Indeed, the definition we still use of autoinflammatory diseases was used for the first time when describing the genetic mutations causing TRAPS. The identification of these “new category” of diseases was very important also to distinguish them from autoimmune diseases, although we now know that some of the last identified autoinflammatory diseases may also have autoimmune manifestations. Moreover, the concept of autoinflammation was very

productive, since it helped to better understand the clinical picture and origin of other diseases, even the ones without recurrent fevers as a clinical hallmark (i.e. Blau Syndrome or Early Onset Sarcoidosis; Pyogenic Arthritis Pyoderma gangrenosum and Acne syndrome) and not monogenic but polygenic-multifactorial (that means that the disease doesn't come from a single gene mutations but is probably determined by the interaction of environmental factors with predisposing genes) such as Still's diseases including Systemic Juvenile Idiopathic Arthritis (sJIA) and Adult onset of Still's disease (AOSD). Indeed although sJIA is traditionally included in the forms of Juvenile Idiopathic Arthritis and AOSD in Rheumatoid Arthritis, autoimmune diseases, it is now clear that their clinical characteristics (and therapeutic options as further discussed) are more similar to the ones of autoinflammatory syndromes. The concept of "autoinflammation" has been very fruitful also to understand some of the clinical manifestations of other well known diseases, once considered autoimmune, such as Behçet's disease.

02. Dismay

WHEN HOPE STARTS TO TURN INTO SAND BETWEEN YOUR FINGERS

“

Apprehension, uncertainty, waiting, expectation, and fear of surprise do a patient more harm than any exertion.

- Florence Nightingale

KATY

County Cork, Republic of Ireland

When I was pregnant with Orla I was diagnosed with gestational diabetes. I was unable to carry the pregnancy to full term: the doctors had to induce the birth because there were risks for both the child and for me. They had to revive her, but she recovered immediately. Days and months went by and she grew peacefully. Of all three of my children, she was the strongest. I don't remember a single sick day from school when she was a child, unlike her siblings who suffered from tonsillitis, stomachaches, or the usual illnesses that school children get.

Orla loved sports and she was a natural gymnast. She was able to train for hours and hours without showing the slightest sign of fatigue.

When she got sick—it was a Monday—she got progressively worse, so by Wednesday we took her to the emergency room. They examined her, rehydrated her, and before discharging her told us to contact our family doctor if her condition worsened. They didn't tell us what caused her illness, and we had no idea what it was.

At home, Orla quickly worsened. The pain prevented her from moving, her fever was very high, and she threw up all the time. Both my husband and I took leave from work since we didn't want to leave her alone for a moment. We even slept next to her at night.

It was terrible. We looked on, powerless to help, not knowing what we could do to make her feel better. We didn't know what the right

thing to do was: take her back to hospital or insist with our general practitioner that he come make a house visit? The next Saturday we called him, but he didn't even examine her. He observed her from the other side of the desk and said, almost annoyed that we had disturbed him off-hours, ‘She's got a bad flu, probably swine,’ and sent us home.

But Orla was sick, and we didn't know why.

On Monday, while I was at work, they called me from the paediatric ward at the hospital. Among the blood tests they’d done, there was one that had significantly altered values: they told me to bring her to the emergency room.

They repeated the test but the result confirmed what had already emerged from the first blood sample taken. The values were off the charts!

They suspected meningitis and hospitalised her immediately, administering the initial treatment for that sort of inflammation. They also did a spinal tap to confirm the diagnosis, but the result was negative. They placed her in quarantine and adopted the protocol for similar cases, doing analyses on the whole family.

Unfortunately, after the test, Orla got worse. She had pleural effusion, pericardial effusion, and her liver started functioning less and less. Her spleen was also very swollen, as were all her lymph nodes, and her fever spiked again. In addition, she had a bad rash on her body that came and went that caused her a lot of pain.

This was an awful period that stretched on for hours and then days. We stayed at her bedside, not knowing if she would survive the next minute.

We waited and waited—we did nothing else but wait. The doctors came and went, hypotheses and retractions followed one another at a dizzying rate. Something terrible was trying to take Orla away from us and we didn't even know what name to give it. We didn't know what we needed to fight against or how.

At first, the doctors talked with us in Orla's room. I don't know how

much she picked up. I think she was so sick she had neither the ability nor the strength to pay attention to anything but her own pain.

However, soon, the doctors began ushering my husband and I out of the room to talk about her condition. This really frightened me, along with the fact that even on Sunday, at six o'clock in the evening, they did tests. In Ireland, if they run tests on a Sunday evening, you can be sure you're really sick.

After a few days, the doctors decided to consult an expert haematologist from another hospital. They suspected it was leukaemia and monitored her constantly. They decided to wait until the next day to take the bone marrow sample to be sure.

That night, Orla's condition worsened and in the morning there wasn't a single operating room free where they could take the sample.

WE FELT DESPERATE, AT THE MERCY OF THE WAVES.

The haematologist didn't delay and took the sample right there in her hospital room—there was no time to waste. They wanted to start treating her because she'd entered the macrophage-activation syndrome phase.

Her condition had become critical but they succeeded in stabilising her. There was no intensive care unit in that hospital so they moved her to the Dublin unit, which was about three hours away by ambulance.

By then, I'd entered a state of complete numbness. I didn't feel anything anymore and it seemed I was immersed in a milky bubble, everything was blurred, voices came to me as if from an infinite distance. It was as if the outside world ended right where my body began.

I embodied the border of nothingness: outside of me there was nothing but fog, a thick and oppressive curtain that concealed the

unknown. I couldn’t even remember my own name. I had no other thoughts beyond Orla. There was the risk of losing her—Orla could die—but we didn't want to know that, we couldn't look such a monstrous possibility in the eye.

We forgot, *I forgot*, about our other children. I didn’t tell Niamh’s school that Orla had been transferred to Dublin and that my husband and I were with her, and that we couldn’t accompany her to school.

Niamh somehow figured it out and only now, looking back with a clearer mind, can I imagine what it must have cost her. Fortunately, as I’ve already said, the community where we live is very close-knit and the neighbours stepped in to take Niamh back and forth to school so she could attend classes. I didn't even tell them she was home alone. Tomas was away in college at the time.

It all happened so fast but it’s true: we simply forgot about her. It was an unforgivable thing to do.

Orla's grandmother eventually showed up. Someone in that foggy limbo outside of me must have contacted her. I don't remember doing it myself, but someone did.

The first time I was able to cry was on the night Orla went into the ICU, when they thought it was meningitis.

I never let her see me in tears. I never showed her the slightest doubt—no matter how hard the disease attacked—that she would come out of it. I knew it would be an ordeal, no matter what. In fact, my sister had fallen ill with meningitis years earlier and had been in intensive care for more than twelve months. I realized I had to be strong, I couldn’t afford to allow my emotions to get the better of me.

In that period I saw so many parents fall to pieces, stressed by the delays in tests and diagnoses. I decided I couldn't do that. I learned how to live every moment for what it was, focusing on one thing at a time—the CAT scan, the clinical exams—without thinking about the next one.

I remember calling my brother one day, he’s a nurse and works in

intensive care in a hospital in London. He could understand me, and with him I was able to vent. He was the only one I let see into the dark pit of my emotions. I knew he wouldn't let me fall inside.

In my mind, I saw Orla slipping away from me. I tried to grasp her but she moved farther away, and I couldn't move, I couldn't reach her. I felt like I was being held back by an immense force.

I was losing my daughter, and I didn't know why.

BIRGITTA

Delft, The Netherlands

When my daughter was born, she and I were alone. I named her Jasmijn. I used to lead a group of scouts and there was a little girl with that name. She was sweet and kind, and I decided that if I ever had a daughter, I would give her the same name.

I was in a particularly stressful period when Jasmijn was born. I was separating from my husband and that precious little being was the only beautiful thing in my life. She really was my flower, my hymn to rebirth, and to hope.

The pregnancy had gone well and even during the birth everything proceeded as it should. Jasmijn was a quiet newborn and grew normally. She had a happy childhood and she didn’t even have the usual childhood illnesses. We were calm and relaxed.

Then, one day when she was about nine years old, her jaw became swollen. It didn't hurt her in the beginning, it was just swollen. Instead of the swelling going down, two months later it got worse and it started to hurt. I took her to our general practitioner. I wasn't particularly worried, I thought maybe she had an infection in her teeth and that they would prescribe antibiotics and it would pass. The doctor told us to see a dentist but the dentist didn’t understand what the problem was. He advised us to go to the hospital, to a specific department for oral diseases.

Two young doctors who had studied in Amsterdam examined

Jasmijn.

I WAS WORRIED, BUT NOT TOO MUCH.

Okay, so it wasn't a usual toothache and maybe Jasmijn should have given up munching on her favourite snacks a while ago, but it was just a matter of patience. Nothing serious, I really believed it.

The two doctors remembered that one of the professors they'd studied with had a patient with the same symptoms and they thought it would be appropriate to have him examine her.

So, we went to Amsterdam and the professor saw her immediately. Jasmijn's jaw continued to be very swollen, she was always very tired but had no fever. He told us the name of the disease that had afflicted my daughter. I'd never heard of it, but I didn't get scared.

At the time, I didn't realize how deeply it would affect our lives.

CARMIT

Kiryat Gat, Israel

Ofir is my only daughter; after her, we had two boys. I had no problems during my pregnancy and all the routine exams went well. Here in Israel they test for those diseases or malformations that can justify interrupting a pregnancy if there's a positive result. Nothing unusual came up, so we were unconcerned.

For the first few days after the birth, Ofir was fine. At home I breastfed her regularly, she seemed hungry and enthusiastic about being fed.

After two or three weeks, the situation changed. I realised something was wrong with her diaper. A doctor came to visit her and told us that probably something I'd eaten had passed through the milk and was the cause. But I knew that wasn't possible. I was always careful to exclude all the foods that are suggested to avoid while breast-feeding.

Ofir kept getting sicker. Almost every week we were at the doctor's to have him check her bowels—we thought the problem was there.

They didn't find anything with her intestinal tract but she wasn't growing as she should have.

Months went by and Ofir's first teeth began to appear. At the same time, the fevers started. I talked to the doctor about it but he reassured me, saying that it was common in children during teething.

Then Ofir began to walk. She crawled at first and then slowly began standing up. Sometimes in the evening, her legs hurt so much that she couldn't walk anymore. Once again, the doctors dismissed her symptoms as ‘growing pains.’

I was a young mother with no experience when it came to children. Ofir was our firstborn, and if a doctor told me not to worry, I trusted him.

It was only after the birth of my son that I realised there was something wrong with Ofir. She was about two and a half years old, and one day red spots appeared on her skin. They covered her whole body—it was shocking—and in addition she couldn't move.

We took her to the hospital and they admitted her. After a few days, they discharged her.

'SHE’S FINE, DON’T WORRY. THERE’S NOTHING WRONG WITH HER.

It was probably an allergic reaction, it’ll pass,’ they told us.

Ofir was attending kindergarten at the time, so I asked her teachers about what she’d eaten, what she’d been in contact with. I was thinking of a food that might have triggered an allergic reaction, or maybe something they’d used to paint with.

I wasn't calm anymore: I decided to take her to a private specialist.

It was a horrible time, I didn't know what to do and the same was true for my husband, for him it was so emotionally difficult that he couldn't take the child to the doctors, so I had to take over that part—contact the doctors, make all the decisions about Ofir's health, etc. He trusted me but I felt overwhelmed by the fear and responsibility.

My daughter was sick and I couldn't help her. It was dreadful.

I hoped the doctors would give us some answers. But, unfortunately, when they did come, they weren’t the ones a parent ever wanted to hear.

AISLING

Straffan, Republic of Ireland

When I was expecting Jack, we found out through routine prenatal testing that something was wrong. At first we didn't know exactly what, but it was very likely that he suffered from a heart condition and probably even Down's syndrome. Before having a diagnosis, the doctors hypothesised that he could have a rare malformation that would prevent him from surviving, such as trisomy 13 or 18.

I WAS TERRIBLY FRIGHTENED.

When they found out he had Down's syndrome, I was relieved and didn't want any more tests. I'd risked losing him and didn't want to take any more chances with invasive tests.

He was born a little prematurely, about two weeks before the due date. They realised he was very ill. He couldn't eat and they had to feed him with a naso-gastric tube. After less than a day they took him away from me. They emergency transferred him to Crumlin Children's Hospital in Dublin to test his heart.

In those first few weeks, I wanted to learn how to do CPR—with his heart problems, it could be crucial.

I felt like I was on a roller coaster, a seesaw of hopes and fears, and a

terrible anxiety hung over it all. It kept me from thinking as clearly as I would have wanted. I thought, in fact, that his situation was temporary —difficult to manage, but one I would learn to control.

I had no idea what lay ahead.

At five weeks, he came down with pneumonia. We baptized him in the hospital. There was a chance he wouldn't survive.

Jack kept getting sicker and just six weeks after he was born, he developed a rash, his body was covered with red spots. It was the first symptom of the disease, even though nobody knew what it was at the time. When he was born, he was little—he weighed only 2.3 kilos (5 lbs)—and his tiny heart was not working as it should.

Surgery was necessary, but on the morning when the operation was scheduled, Jack had a fever. They decided they would wait a few hours and see. He was scheduled to be first into the operating room that morning and they shifted him to a later time. The situation, however, got worse by the minute so they had to take the risk and operate immediately. Otherwise, he didn’t have a chance to survive.

I don't remember much about those hours. I couldn't think. I kept praying that Jack would make it. How could I have given birth to him and then have to say goodbye? He was my baby, my beautiful baby boy.

He survived the operation but the improvement was only temporary.

Four weeks later, they had to operate on him again. He was kept in the ICU the whole time. Just then, my husband—who is an accomplished journalist—accepted a new job far away from us. Jack isn’t our only child, we have another daughter whose twenty-one months older. I felt powerless on my own. My sister came to help me and to stay with the baby—she was my comfort.

Oh, God, how could such a thing actually be happening to us? It was supposed to be a time of joy, to be shared with family and friends, but instead we were there praying that he wouldn’t die. He was so small,

and his life had begun in pain. How could it be possible? It wasn't fair.

The emergency cart never left his hospital room. At just five months old, they had to insert a percutaneous endoscopic gastrostomy tube to feed him, and had to perform a Nissen-Rossetti fundoplication procedure to resolve a problem of acid reflux from his stomach into his oesophagus. The procedure had to be done again three years later because that first operation hadn't gone well.

Those first months were the beginning of an unending ordeal.

EVERY DAY A NEW DIFFICULTY AROSE AND FOR JACK THAT MEANT PAIN, AND THEN MORE PAIN.

He constantly needed oxygen, sometimes even seventeen litres a day. An absurd amount! He suffered from one bout of pneumonia after another and was always in intensive care.

Jack was about twenty months old when Dr. Cant examined him; he came from Newcastle, in the United Kingdom.

I think it was Dr. Irvine, a dermatologist, who suggested that in addition to Down's syndrome, which is fairly common, Jack could also have a rare disease, something like CAPS (Cryopirin-Associated Periodic Syndrome).

I thought I'd already seen my son endure all the pain he could, but that was just the beginning.

EMER

Kildare, Republic of Ireland

Jack was born seventeen years ago after only twenty-six weeks of pregnancy, and it was immediately apparent that he would have a rocky start in life . They had to induce birth because I had gestosis and risked having a stroke or brain haemorrhage. Among other complications, Jack also developed cerebral palsy from bleeding to the brain due to prematurity at birth.

I had to give up working because he needed constant assistance. It wasn’t a burden. He was my son and I wanted to take care of him.

We investigated whether I could develop gestoisis in a future pregnancy, but we were told it was a once-off. It just happened. He now suffers from epilepsy, is visually impaired, has severe developmental deficits, and has hemiparesis.

I remember that one day we were visiting a doctor and she looked at Jack, and told us our son would never walk. We didn't say anything, but inside of us something was triggered. I don't know exactly what. The desire to deny it, of course, but also the absolute certainty that we wouldn't give up, we wouldn't abandon our son to the shackles of his condition.

We worked hard with him, attending a special programme in England to help children like Jack recover a certain amount of joint mobility.

When he was three years old he took his first steps and it was like seeing him reborn. My husband and I locked eyes. I was crying, but my heart was bursting with happiness.

Our wish had always been to have a big family. We were learning to deal with Jack's difficulties and began thinking about giving him a little brother or sister. The doctors reassured us, even the gestosis would most likely not appear again.

I got pregnant with Amy, and everything seemed to be fine. But by the eighth week, my blood pressure suddenly rose and I realised that, unfortunately, the gestosis was returning.

I went to the hospital every week for checkups. I prayed and prayed that I would be able to pass the twenty-sixth week, knowing that if I succeeded, the baby would have a better chance of reaching a level of growth that would ensure its survival. The doctors decided to induce birth at the thirty-sixth week: my pressure was too high and the risk of harmful consequences became more and more concrete.

Amy was born and I kept her with me for three hours, but then they had to take her away because of a change in her temperature. They kept her in ICU for twenty-four hours, then discharged her and said we could go home.

Everything seemed to be going well—we were calm.

As the weeks passed, however, a shadow began to cloud our days. Amy was a newborn and like all newborns she was expected to cry, crying is a baby’s means of communication. But Amy cried *too much*. She cried when we changed her diaper; she cried if I covered her, but also if I didn't cover her; she cried when I held her in my arms and when I put her down. She cried *all the time*.

It was probably due to the experience we had with Jack that made me think my daughter's tears might be because of joint pain.

IT COULD NOT BE NORMAL

FOR SUCH DESPERATE CRYING WITH EVERY MOVEMENT, NO MATTER HOW SLIGHT.

Then, when she was about five or six months old a rash appeared. It started on her face and gradually spread to her whole body. I noticed that when I put her down for a nap, she soon started sweating profusely. I monitored her temperature. It went up as she slept, and got as high as 40° C.

I took her to our general practitioner and they thought it to be viral; this happened on several occasions. I was thinking, 'does the doctor see me as being over-apprehensive because I had a child with additional needs already?'.

I knew I wasn't crazy and I wasn't in post-partum depression. I wasn't seeing sickness when it wasn't there. My daughter was in pain and it wasn't normal. According to the doctor, it was a viral infection that would pass on its own.

The situation went on for some time. Amy was given steroids and the situation improved, and then the problems started again after a few months.

The doctor kept talking about viral infections but I knew it wasn't possible, at least not so many in such a short time. I had even taken pictures of her with the rash in progress, but I still wasn't taken seriously.

I was exasperated. I felt like I had ended up in a parallel universe, where everything was going in the opposite direction.

I asked the doctor to give me a letter of referral to the hospital emergency room. I needed to get to the bottom of this. Watching Amy be sick and not doing anything to help her was unthinkable.

More to get rid of me than out of conviction, they prepared the

letter. I asked them to detail the occasions when he had examined her and what they had found.

Amy was eleven months old when we went to Our Lady's Children's Hospital in Crumlin, in Dublin, with the letter in hand.

While we were in the emergency ward Dr. Orla Killeen, a paediatric rheumatologist who had just been hired by the hospital, came to see us. At that time, Amy had a rash and when she fell asleep, her temperature would rise.

It seemed strange to me that it was a rheumatologist examining Amy, but Dr. Killeen studied her medical record, told us that her values of Reactive Protein C, which is synthesized by the liver in response to inflammation, were extremely high which probably meant one of two possibilities: either a rare form of leukaemia or a rheumatological condition.

I didn't know which of the two hypotheses to wish for—it all seemed so frightening and absurd! There we were in the hospital again, with another child in danger of dying. It seemed like a nightmare, a perverse joke of fate.

Amy was in the hospital for two weeks, during which time she underwent all sorts of tests. Dr. Killeen proceeded by process of exclusion. Whenever a given disease was discarded, I felt both happy and frightened: happy because the diseases they had tested for and excluded were frightening; frightened because the range of possibilities was gradually diminishing and the remaining hypotheses could be more terrible than the ones eliminated.

My baby was suffering and we were fighting blindfolded, and our punches almost never hit the mark. My husband and I were exhausted and Jack needed us, too.

TIME PASSED BUT THINGS DIDN'T CHANGE—

WE WERE STUCK IN LIMBO.

ANTONIO

Ludwigsburg, Germany

Anthony was supposed to be born on the 12th or 13th of December. A week before, I took my wife, Anna, to the hospital for the last checkups. I remember I was wearing a pair shoes that were too small and they hurt my feet terribly. I couldn't wait to go home and take them off!

While they were doing the usual tests, my wife started going into labour. She probably knew at that point that the baby would be born sooner, but I didn't.

‘Listen, your baby’s about to be born!’ the doctor said, shaking me, while my wife said, ‘Run home and get the bag I prepared with things for the baby and for me. Hurry!’

I didn't understand a thing but rushed home—running like a madman—completely forgetting all about my small shoes and aching feet.

I was back at the hospital in no time. Anthony was born after I got there. I think my wife decided to wait for me!

I was happy, as if I could reach up and touch the sky. I had a wonderful wife and, at that point, three marvellous children! I’d come a long way from when I was young and penniless, hiding on the train to Germany in order to escape the ticket inspectors. All I wanted was to enjoy my family.

When Anthony started to get sick, around the age of three, my reaction was one of disbelief. I tried to calm my wife down, I told her it was normal for small children to cry.

I realise that I didn't support her enough, but I just couldn't accept—and can't accept—that there's something wrong with my son. I simply refuse to believe it.

ANNA

Ludwigsburg, Germany

In his early years, Anthony grew like all children and I didn't notice anything different from Christopher and Brian.

He began not feeling well when he started preschool. He got sick too often, more than I thought was normal. It was just... I think mothers have a sixth sense, they know when their children have something wrong.

Anthony was sick especially after physical activity. In the evening his legs hurt a lot, he complained of headaches, and his temperature rose.

WE TOOK HIM TO DOCTORS, BUT NOBODY BELIEVED ME.

It was a scary time. They told me I was too anxious, that I exaggerated

like all Italian mothers. But I knew, I had a feeling that something wasn’t right.

I also had to battle with the preschool teachers. They considered my attitude excessive and didn’t understand why I often kept him home. They thought it was because I wanted to keep him with me, that he was a mama’s boy, and that I didn't want to let him go. If I think about it, I still suffer to this day. Not only did I have to fight something that was trying to take away my son's health and life, but those around me didn't notice it.

There was only one doctor who seemed to believe me. He’d been the paediatrician for my other two sons, and perhaps he knew me well enough to know that I wasn’t the kind of mother who worries for no reason.

He sent us to the hospital, where I had to fight against the usual biases. They didn't think Anthony was that sick and treated me as if I were a crazy, overly apprehensive Italian mother.

I told them he had a stomachache. He really wanted to go to school to play with his friends but that he couldn’t because he was too sick. They didn't believe me—neither the doctors nor the teachers. It was a terrible sensation, feeling guilty even when you knew you had done absolutely nothing wrong.

It took three and a half years before we found out what was destroying our son's health and our peace.

CHRISTOPHER

Ludwigsburg, Germany

If I had to describe my family I'd say it's a bit crazy but we're very close. We like to have fun, laugh and joke together.

Anthony is the youngest, he was born when Brian and I were already big. Maybe that's why we consider him our mascot, you know, the puppy of the house. Anthony's special. He can drive you crazy until he gets you to do what he wants and sometimes he's a little quirky but he's a very determined kid.

I remember back when his problems began... not being able to understand what it was about threw us into panic. No one seemed to believe my mother's concerns. We felt powerless. I looked up symptoms on the internet, I talked about them with relatives, simple acquaintances, anyone who could help me understand. I couldn't sit on my hands and watch my brother be sick.

The umpteenth time I talked to a client about it, it turned out the woman was a doctor: I told her about my brother's fevers, his joint pains. I asked her if she'd ever encountered a case like his. She told me that in fact she had, and that she wanted my brother to come see her at the University Hospital in Tübingen, where she worked.

When I got home from work that evening, I announced, 'That's it! Time to try something new!' I told them all about Tübingen and the doctor I'd met.

That's how we got to the hospital in that city.

LENNY

Born, The Netherlands

I've played the French horn for twenty-eight years. It's an instrument I love, it's a part of me. No one knows why it’s called a French horn, actually its origins are not even French—it was used for fox hunting, and only later did it begin to appear in orchestras.

I like the instrument because it gives its best both in a solo with its melodic notes as well as when blending in with the other elements of the orchestra since it’s also very harmonic.

I met my first husband, Tess and Kate's father, thanks to the horn. The orchestra in which I played in Buchten and his orchestra, in Born, organised a concert together, and that’s how we met. We fell in love and from that moment on, I played in both orchestras.

My husband and I wanted kids. I told him about my illness, of course, but I also expressed my confidence that if they inherited it we would be able to manage it. I thought that among the diseases we see around us—such as cancer or others that undermine the chance for survival—the one I had wasn’t, all considered, a reason not to have children. I mean, in light of everything I can do despite having it, I could accept them having it too if, out of all the possible illnesses out there, they got that one.

I got pregnant and Tess, who’s now ten years old, was born. She was still a newborn when those little red spots that I knew well began to appear on her skin. Before using traditional drugs, we tried to treat

them with homeopathic therapy. Unfortunately, the remedy caused her to have a rash. We had to stop giving it to her because the rash was much more annoying for her than the inflammation she already had, which wasn't terribly bad. She also had joint pain—I could tell from her crying—that started around eight or nine months, I think.

Meanwhile, I got pregnant with Kate—there's only fifteen months' difference between one and the other. After she was born, Kate started getting sick right away, even more than Tess. All Kate did was cry. She cried non-stop, I think, for the first eleven months of her life!

One evening, while I was out, my husband called me. 'Come home right now! I'm afraid of doing something I'll regret!' He was beside himself.

We didn't know what to do and we couldn't see anything wrong with her, she didn't have any red spots like Tess did. We had no idea what was going on. The only way to calm her down a little bit was to feed her. At first I nursed her, then we switched to artificial formula so my husband could take over.

At that time, I was working in customer management for a company, while he's a computer scientist and works here in the city offices.

The first symptoms that suggested Kate might also have the same disease we have appeared around the time she was a year old. The paediatrician advised us to go to Maastricht. We submitted an application, because that's how it works here: first we had to apply for admission and then it took some time before we were summoned.

One afternoon Kate was so sick we had to run to the hospital. She wasn't eating or drinking and they had to feed her with a naso-gastric tube. She remained in treatment for a week on antibiotics and painkillers before she started to improve.

The doctor in Maastricht came to see us in the hospital; Kate's condition was such that she couldn't be transported. He examined her and told us about Dr. Frenkel, a specialist in inflammatory diseases at the Utrecht Children's Hospital.

We asked for transfer from Maastricht to Utrecht, which we got when Kate was about two years old, and she's still being treated there. It's a large university hospital and their very competent staff is taking care of us.

Kate underwent a series of tests because nobody knew what the origin of her illness was. Then when she was around four her disease finally got a name. It was the same as mine, but not the one we had thought of up until then.

SUSANNE AND ROBERT

Dillingen, Germany

Marco had just entered his ninth week of life when he came down with meningitis. He was admitted to hospital and treated for ten days, and then we brought him home.

He grew normally, like other children, and began to walk around thirteen months. It only took him a little longer than average to start talking, but we didn't worry about it. Not all children are the same.

At around eighteen months the fevers began. His temperature was very high—often as high as 40 or 41° C. The paediatrician would treat him with antibiotics and for a while he was fine, then the fever would come back. At that time he was often sick and I had to stay home from work to take care of him myself.

Sometimes he was able to go to nursery school for no more than two weeks a month and so we decided to not take him anymore. He improved at home, perhaps because he was calmer, without the stress of an unfamiliar environment.

Then I became pregnant with Jannika, who was born in October 2008. Marco wasn't so sick then and it was a pretty peaceful time.

Jannika was about six months old when her first fevers appeared. When we took her to the paediatrician, she diagnosed her with a bacterial infection and prescribed antibiotics. However, the blood tests showed that the markers for inflammation were very high. Marco was

also sick often, along with his sister, so we were regular visitors to our paediatrician's surgery.

We had a couple of relatively quiet years. Marco had improved and was able to attend school regularly. However, Jannika's illness resumed around two years of age.

WE DIDN'T HAVE ANY CLEAR IDEAS ABOUT WHAT WAS GOING ON.

We told each other things would improve, we wanted to believe it. It was hard looking at her and not knowing what to do—it was very, very difficult. We had the feeling we were powerless and even though the doctor was monitoring her, things weren't improving. And what frightened us even more was that our paediatrician seemed just as clueless.

At one point it was suggested that Jannika might have FMF, Familial Mediterranean Fever. The doctor ordered genetic tests to rule out some diseases and we did them, but nothing came of it.

After further analyses, she was diagnosed with PFAPA, a syndrome that causes periodic fevers. They explained to us that we shouldn't worry because generally it's a condition that disappears after the first few years, without leaving any negative consequences.

So, we were prepared to live with it. The important thing was to make sure Jannika suffered as little as possible when she got her fevers and wait it out. Sooner or later, it would pass.

TIZIANA

Partinico, Italy

I remember very well the moment I knew I was pregnant. We weren't married yet and had just done our *fujitina*. I was at my mother-in-law's that day and I didn't feel well. I had a fever, kidney pains... She said to me ‘Oh, honey, you could be pregnant, you know!’ I answered by laughing that I didn't think it was possible just yet. ‘Go on, do the test. What’s the harm?’ she asked.

The result of the test wasn’t very clear, so I went to the gynaecologist who examined me and confirmed that I was pregnant. We were happy, all of us—me, my husband, and our parents.

Actually, I wasn’t expecting it so soon. I thought I'd have more trouble. When I was sixteen, they’d had to remove 80% of my right ovary because of a cyst, so I was sure we would have to try for a long time before we could get pregnant.

But our serenity didn't last long, because two days later I had severe pains and at the hospital I was diagnosed with a possible miscarriage. They gave me injections and recommended I rest.

I spent the first three months in bed with very low blood pressure and terrible nausea. I couldn't even have breakfast. It wasn't until dinner that I could keep food down.

After the first three months I was a little better, and I could start planning our wedding.

At the thirty-sixth week, during one of my regular checkups, the gynaecologist decided to have me admitted to the hospital.

The birth was difficult and when the first of our twins came into the world he had trouble breathing so they had to take him to Palermo by ambulance. My husband raced after him in his car. I remember the Carabinieri stopped him because he was going too fast: he explained that his newborn wasn't well. They said they understood because they were family men too.

The second delivery was faster and Vincenzo was born healthy. They told me that Benedetto, the first twin to be born had, in a way, helped his brother during gestation. I was able to keep Vincenzo in my room because, although he was premature, he didn't need to be in an incubator and had no problems.

When we went to get Benedetto from the hospital, they told me he was a fragile, very delicate child because he'd been born prematurely and had those problems at birth. I was young and inexperienced, so I believed what the doctors said.

At home, the twins cried all the time and vomited. It was unending. My mother told me it was normal and to be patient and wait. They were *nicareddhi*, which in Sicilian dialect is an affectionate way to say little runts.

The months passed and things didn't improve so I went to the doctor. I wanted to understand why the babies spit up the milk as soon as they took it. He told me to change their position in bed, putting a small pillow under the babies' back because perhaps the valve at the top of their stomach hadn't closed yet.

We tried but it didn't work. We changed the type of milk but still it was the same. They gained very little weight and almost didn't grow.

Weaning made things a little better. They ate, though not much. They kept fussing and I felt like I was going crazy. I didn't understand what the problem was and they were too small to explain themselves.

There were times when they were better. For example in summer

because the heat helped them and they also ate more fruit, which is refreshing.

At one point, spots appeared on their bodies but my mother had always been very allergic and the doctors said they probably were allergic too. There wasn't much to do about it because as long as they were so small there weren't any tests suitable for their age.

One night we were at my mother's house for dinner. Benedetto had eaten a piece of chicken with salad. Then he took a slice of nectarine and brought it to his lips. Even before he put it in his mouth, he complained of a stomachache. I took him to the bathroom but while we were there he said his eye felt swollen. I looked at him and was scared: his left eye was very puffy. In the time it took me to call my husband, who wasn't with us, his other eye swelled up. My husband told me to give him an antihistamine, but I begged him to come home immediately because I felt this wasn't like all the other times.

In the space of a few minutes, Benedetto started to gasp. 'I can't breathe, Mama!' he was trying to say. My mind went blank. I rushed into the street and stopped the first car coming my way. We sped to the emergency room, with his eyes so swollen he could barely see.

At the hospital, they put him on treatment right away and saved him 'within an inch of his life', which were their words. The fear was too much and I almost fainted. I'd never seen my son like this before. They told me it was a good thing I'd understood what was going on. I felt like a total wreck; my son could have died.

Benedetto was about three years old and from then on whatever he ate seemed to be poison for him.

WE DIDN'T HAVE A MOMENT OF PEACE.

It was around age six that the doctor decided to have him tested for allergies. In order to run the tests, it was necessary for him to have not

taken any antihistamines recently. But as soon as we suspended them, Benedetto got very sick and so we had to start all over. ‘At this rate, he’ll be eighteen before he can do the tests!’ I sighed.

No one knew what to tell me to do. So I started searching on my own on the internet. From what I read, I found out it was necessary to put him on a very strict diet and, especially, he had to avoid all foods that cause high histamine production. He was limited to tea, plain toast, plain noodles, and turkey. For twenty days I didn't let anyone come to our house—I couldn’t run the risk of him getting sick. And so he wouldn't feel alone, we all ate the same things for the whole time. I didn't want him to have to watch us eating the food that he couldn't, he was just a child.

After twenty days, we went to the doctor for the allergy tests and the result was that he was allergic to all three milk proteins. He was treated with antihistamines, we eliminated cow's milk, and I started giving him soy milk.

According to the doctor, everything was going to be okay.

After three months of antihistamines and the change to his diet, Benedetto still didn’t improve. He had painful welts on his body: he said it felt like they were burning.

The allergist didn't believe me, even though I’d even brought her photos of his rashes. By now we weren’t sleeping anymore—Benedetto was sick and we were awake day and night. The doctor suggested we should have an immunologist check him out and I went straight to our paediatrician to ask her for the referral.

‘Of course, right away!’ she told me.

SHE WAS THE ONLY ONE WHO LISTENED TO ME, THE ONLY ONE

WHO BELIEVED ME. SHE WAS A FANTASTIC PAEDIATRICIAN.

In September of that year we went to Dr. Maggio. She welcomed us and I told her everything that had happened. She asked me for permission to do an exam and I agreed. He'd had so many without results, what harm was there in one more?

One evening, around 9:30, the phone rang and I answered it. It was her.

'Dr. Maggio? Why are you calling so late?' I was instantly worried.

'Don't be alarmed,' she said, but I replied that if she was calling at that hour—and we weren't friends or relatives—I had a right to worry!

'Ma'am, you were right. Your son is sick.'

I pointed out that I'd been saying so for a long time, but she told me, 'Don't take it personally.' But how could I not take it personally if every time I'd said so over the years, no one ever believed me?

'Listen, your son has a disease,' she continued. It felt as if the world was crashing down on me. She asked if I had any other children and I answered that Benedetto had a twin brother and I also had another son. I told her that even Vincenzo, his twin, wasn't well but every time I tried to explain it to the doctors, they replied he was probably imitating Benedetto just to get the same attention.

She decided to have all three children genetically tested.

ATAR

Lombardy, Italy

Michelle collapsed when she was about a month old after they'd discharged her from the hospital. I had given her a bottle of artificial milk and after drinking it she began to vomit, but in a very violent way, with jets two or three metres high! I didn't think such a thing was possible! We took her to the hospital in Johannesburg, where they did a series of tests. Some of them were very shocking, like pricking deeply into her heels or passing ice cubes over her body. But she didn't react.

She was in a pre-comatose state for forty-eight hours and then she woke up. They did a brain scan to check for damage but fortunately it came back negative.

For six months Michelle continued to have problems, even with the weaning. The doctors hypothesised she had gastrointestinal infections or flu. It took six months for them to figure out that food was triggering her reactions, and in the end they suggested I take her to Israel because South Africa lacked a qualified allergy department.

We went to Israel, to a clinic recommended by my family. They diagnosed her with FPIES: a food protein-induced enterocolitis syndrome, an allergy to all food proteins. They assumed it was because of the higher protein content of infant formula compared to breast milk. This meant that Michelle could only consume olive oil, breast milk, banana, and avocado.

I discovered that even in Italy, at the Meyer Children's Hospital in

Florence, there was an excellent team that dealt with the disease so we started going there every four to six months to monitor the stage of the disease. We were told the condition usually passes by the age of five. Meanwhile, they tried to desensitise her because she had developed an IgE-mediated allergy, what could cause anaphylactic shock.

Our daughter was about a year old when we left Johannesburg to move to Ho Chi Minh City in Vietnam for my husband's job.

It was a tough experience, it was not easy for me to adapt. Back in 2011, there were no adequate services for foreign families and children, and even in the American hospital, which in theory should have been excellent, there were problems. We were supposed to stay three years, but after seven months we left Vietnam to return to South Africa.

Then Michelle's joints began swelling too and she went from one fever to another. We thought the infections were being transmitted through Claire, who was going to nursery school at the time, but Michelle was constantly suffering from very high fevers.

When Michelle started talking, she complained of having a terrible bellyache. You'd see her bent in two, with her knees at her chest, unresponsive to any contact from the outside.

One afternoon she was watching TV, it was hot and she was sitting on the couch in her panties. I walked past her and when I came back through the room, not even ten minutes later, I noticed some spots on her legs. They seemed like bruises, they were purplish blue, blooming on her skin. We rushed her to the ER, where they did blood test after blood test.

THERE WAS NO DIAGNOSIS, OR RATHER THERE WERE TOO MANY OF THEM:

they talked about leukaemia, cystic fibrosis, unknown tropical

diseases contracted in Vietnam... For years we lived in complete uncertainty. It's the not knowing that throws you into the most consuming anxiety. You thrash about in a vacuum, in the grip of panic. The diagnosis, however horrible it may be, is an explanation and you can react, or at least try. Giving a name to the affliction is no small thing.

Among the many specialists, we also had her examined by a rheumatologist because rheumatoid arthritis, or juvenile idiopathic arthritis, had been considered, but the results were negative. The rheumatologist, however, insisted that this was indeed arthritis because of the presence of fluid in the joints.

She suspected that perhaps we were dealing with an unknown rare disease, and began to treat it with immunosuppressants and cortisone. In fact, our daughter improved a lot: she wasn't bleeding from her rectum anymore and she didn't have a stomachache. The immunosuppressant, however, exposed her to infections and in just nine months she contracted pneumonia seven times. We stopped the medication.

I got in touch with some Italian hospitals, and unfortunately we didn't get the answers we were hoping for. One doctor spoke to us about CINCA after reading all the symptoms and told us that we had to go immediately to Italy to be included in an experimental trial. We quickly found tickets and flew to Italy, only to then discover that we hadn't been included in the trial because there were some procedural problems. Michelle remained in the Italian hospital for three weeks but the only thing they diagnosed was arthritis of the hip, which they called a 'cold of the hip,' caused by the same virus as the one that affects the airways.

THEY SAID IT WOULD PASS SPONTANEOUSLY.

We returned to South Africa, tired and discouraged, with nothing to show for our efforts.

LUC

Amersfoort, The Netherlands

I remember the moment when I realised I had a serious illness. We were in America and my parents had managed to get a really good doctor to examine me, one who finally took my symptoms seriously and cancelled all her appointments for that day just to see me! I know that because my parents always remind me of it. After that, they took me to the **NIH**, the **National Institute of Health**—I was about eight or nine years old—and there I realised that not everyone had the same disease as me, that I was the one who was different. It didn't bother me—I also suffer from a form of autism that doesn't allow me to feel many emotions—but I remember thinking, 'Wow, cool!'

Yes, I found it strange but cool at the same time, even though I couldn't have known it would change my life.

EVA

Schoten, Belgium

I remember the months following that October afternoon as a long series of clinical tests and stays in various hospitals. I had pain in my legs, wrists, hips, elbows, ankles...all my joints were inflamed. I was in a lot of pain. The disease also threatened the functioning of my lungs and my heart. I felt terrible. I guess my parents were very worried—they had a seriously ill daughter and no one could say what was wrong with her.

I was very hungry due to the cortisone they were giving me. I ate mountains of fruit and I thought, in fact I was sure, that I would recover and would start training and dancing like before. It wasn't just a hobby, I was part of a competitive team and I wanted to resume the exercises with all my strength.

My strength, however, was slowly abandoning me. I was burning with fever because my body had to work hard to get the fever down several times a day and I'd lost a lot of weight: in those few months, I went from weighing forty-two kilos down to twenty-seven (92 lbs down to 59)! My life was in danger and my parents and doctor feared I might die at any moment if this process wasn't stopped. They fed me high-calorie drinks with baby food to try and get me back on my feet. Slowly, thanks to starting with a very low dosage of a drug for chemotherapy, I started getting better but I was very weak. There was no question of going to school and if it hadn't been for my high marks from previous years and the help of my classmates who took notes so that I could study in hospital, I wouldn't have made it.

I remember that my parents stayed with me in shifts so as to not leave me alone while I was in hospital and to also take care of my brother. My mother is a biology teacher and worked part-time, while my father worked not far from home as a technician for a pharmaceutical company.

I CAN ONLY IMAGINE THE GREAT WORRY THEY FELT AT THE TIME.

The pain, however, was like glue and it made us all much closer. That was what allowed us to overcome those terrible months. Nothing has changed since then in the sense that we're still there for each other, ready to help and support.

The disease, when it affects you as seriously as it did me, is a litmus test and the people around you react by revealing themselves for who they really are. Some of the people I thought were my friends I lost, driven away by my difficulties and my pain, while others stayed with me in those same difficulties and precisely because of that pain.

When we arrived at the Hospital of Utrecht, in the Netherlands, in July 1995, we finally discovered the name of the disease that afflicted me: **Systemic Juvenile Idiopathic Arthritis.**

We didn't know yet that event would change my life forever.

WENDY

Tipperary, Republic of Ireland

When the parents of sick children turn to patient associations, usually they haven’t seen any specialists yet. They're scared, confused, they don't know what to do, or who to go to. They're adrift, and associations like ours are their raft.

Here in Ireland, unfortunately, waiting times for an appointment with a paediatric rheumatologist can be as long as two years, and before undertaking such a choice, before standing there waiting for the call, people want to be sure they’re not wasting their time. They also ask us if there's any way to have their children examined privately. They’re so afraid—what they’ve read on internet has frightened them and they feel anger at the health system.

We’ve opened a forum on Facebook where people can compare notes and share their experiences. My advice is that people write down in a diary whatever symptoms they notice because those are things that can help the specialists formulate a diagnosis.

We hope to get a third paediatric rheumatologist with a full team in Ireland. A new paediatric hospital is being built and when it’s fully operational—hopefully within two or three years—it should have three rheumatologists and departments of physiotherapy, intensive care, and ophthalmology. All this should shorten waiting times and minimize families’ suffering.

When my daughter became ill, we had to consult many specialists

before we could understand that she was suffering from early onset JIA.

I remember we had so many appointments to go to that there didn't seem to be enough days. And then there was the care my daughter needed around the clock. I had to leave my job to take care of everything. What was missing—and I think this is something that needs work—was dialogue with the primary physician. Sometimes they don't give enough importance to the symptoms parents tell them about, which adds to the suffering and delays diagnosis.

MANY FAMILIES ARE IN THE SAME SITUATION AS ME.

I see them when they come to us with their choked voices, and there are those who hesitate even to ask for an opinion for fear of an answer they don't want to hear.

With my daughter, I felt lost, isolated, and disbelieved. I told myself that I would not allow it to happen to those who, like us, had to face the unknown of rare diseases. And so, together with other parents I founded a group. We wanted it to be like a flame of light in a dark tunnel of fear and uncertainty.

It's important to make sure families don't feel alone, to reassure them that there are people who will understand what they're going through and can give a name to their fears. And, above all, that no one will tell them that their fears are just fantasies. Listening to them and giving credit to their words means restoring their dignity, which had been taken from them by their doubt.

NORMA

Dublin, Republic of Ireland

In Ireland, the first point of contact for families when children have a problem is the general practitioner. If he or she believes the situation is outside the normal diseases that can affect children, if they think it's better to investigate, they'll make a referral to a specialist. The biggest problem is the long waiting time for an appointment. When they come to us, in the hospital, I'm there to greet them. I go with them to see the doctor and a physiotherapist, and if necessary there are psychologists for emotional support in the following days.

During the appointment, we try to figure out if they're facing a rare autoinflammatory disease. If not, the doctor will explain why that's not the case and will refer them to the most suitable specialists; but if it is, they'll explain what they're dealing with.

I'm particularly involved in this phase because such a discovery throws parents into panic. Often they've already read something on internet which has only exacerbated their fears. I try to make them feel that they're not alone, that there's a network of doctors and professionals who will assist their children in the best possible way.

They'll be given a health plan detailing the tests that need to be done, the therapies, the dosage of the drugs, and the way to administer them.

The parents have many fears, and that's understandable. They wonder how they can ensure that their children get the help they need,

especially when they're at school, or somewhere far from them. Even the management of periodic visits isn't easy. Here in Ireland there are only two centres of excellence for rheumatological paediatrics, and for many families that means having to travel for four or five hours to get to us. It can be a serious problem for work or for the children's school. Some parents have had to give up their jobs to be with their children.

It's not easy for parents—I totally understand them—and that's why, in our centre, we make sure that we schedule as many tests and appointments as possible for a single day. We give them our direct numbers so they can contact us easily; they might run into difficulty getting the drugs and in that case we contact the pharmacy and explain the situation. We try to make it easier for them to deal with the practical aspects of the disease.

The moment of diagnosis can be very difficult, but it's also often a cause for relief. Finally, the disease has a name, a face, it exists, it's not a product of their imagination. Along the way toward diagnosis, one of the greatest difficulties is precisely that: getting doctors to believe that the children are really sick, that it's not a matter of crankiness or the usual crying of newborns. It can be a very, very frustrating situation.

Being faced with doctors and professionals ready to stand by them in the management of the challenging disease of their loved ones helps them regain their credibility.

When we communicate the diagnosis of a rare autoinflammatory disease, their first reaction can be one of discouragement. They wonder why, why *their* kids. We explain that often there's no explanation, only a series of elements in the complex system that is the human body which, for some reason, aren't working as they should. Of course, it's hard to accept the shift in perspective that life has permanently changed and that they'll face challenging and painful situations, but the sooner they confront it and accept it, the sooner they'll begin to learn to live with it. That's what we try to teach parents, and we reassure them that, while these are rare diseases, we will still give them the best possible care.

MICHAL

Haifa, Israel

After the onset of the disease, more or less between 2013 and 2014, I began going over my life up to that point, to look for clues that would help me understand what was happening to me. I realised that by the age of twenty I had already begun experiencing pain in my back and joints, but each time the doctors—mostly orthopaedists—dismissed me, saying 'you should lose weight' or 'there's nothing you can do about it, you'll just have to live with it'.

With the inflammation that occurred in 2013 my situation got worse. I began to suffer from fibromyalgia, psoriatic arthritis, high blood pressure, neuropathy, and a whole series of diseases that seemed to appear from one day to the next, as if suddenly my body had gone into self-destruct mode.

AUTOINFLAMMATORY SYNDROMES (part 2)

As already described, autoinflammatory syndromes are very rare diseases of quite recent indentification. That means that only a minority of physicians are familiar with their characteristics. This happens quite often in rare diseases. For that reason, even nowadays, it may happen that a patient with a clear-cut autoinflammatory disease may experience a delayed diagnosis. Unfortunately autoinflammatory diseases, and monogenic recurrent fevers in particular, may be misinterpreted. Usually the first diagnosis is the one of recurrent infection for people with periodic fevers, “growing pains” of juvenile idiopathic arthritis in people with joint aches, “allergy” in people with recurrent skin rashes.

This is due not only to the fact that autoinflammatory diseases are still poorly known, but also to the fact that the clinical manifestations are not pathognomonic (that means distinctively characteristic of a particular disease) of these diseases. Indeed, ruling out other possible causes such as infections and tumors in a patient with the clinical picture of an autoinflammatory disease is crucial.

03. The Diagnosis

WHEN IT SEEMED THAT HAPPINESS HAD TURNED ITS BACK ON US

“

There are in fact two things: science and opinion; the former begets knowledge, the latter ignorance.

- Hippocrates

KATY

County Cork, Republic of Ireland

From the results of the bone marrow biopsy they did on Orla that terrible morning, it emerged that my daughter was suffering from Juvenile Systemic Arthritis and Macrophage Activation Syndrome.

Her condition was desperate, with all her organs compromised. She was constantly monitored by a pool of specialists. The treatment worked. She recovered quickly, and after a week she returned to the general medicine ward. I think I only started breathing normally again at that moment. We stayed another three weeks at Crumlin Hospital in Dublin.

IT WASN'T EASY TO ACCEPT THE DIAGNOSIS, BUT AT LEAST WE KNEW WHAT WE HAD IN FRONT OF US.

Orla knows all about her illness and recognizes the signals her body sends her. When an inflammation occurs, it begins with a peak of fever and an intense rash, followed by joint pain. Sometimes she needs a respirator because the disease has also affected her lungs. Orla *feels* when she's about to get inflamed. She wakes up one morning and says

‘Mom, I'm getting inflamed,’ and like clockwork it happens. She's learned to know herself really well. She knows that if she's out at the weekend, if she's tired, she'll have an inflammation, but she also knows that if she rests a bit, she might be better off in a couple of days. Sometimes she feels very tired, or under the weather, but she knows she's not inflamed. She distinguishes the various manifestations of her body and has a deep connection with her innermost self. I don’t know, but perhaps everyone who has to live with this type of disease develops a sixth sense, a kind of internal alarm that warns them when an inflammatory episode is about to occur.

The medicines used to keep the disease under control are really strong. For a long time Orla had to take steroids, as well as other drugs. The doctors had just managed to get her off the steroids when, about three years after the onset of the disease, she developed a bad lung infection and had to start them again. She stopped taking cortisone in January 2018 and since then her situation has been quite uneventful, stable. Knock on wood? Absolutely. And hope that this stage will last as long as possible!

This disease has put my baby to the test. She was the strongest of my children, the most energetic and determined. If she had something in mind, there was no doubt she would get it. She was a force of nature.

After leaving the intensive care unit, she was physically and mentally broken. Back home, she started attending the CAMHS team, our government-run mental health service for children and teenagers.

The awareness of her condition and her suffering had reduced her to a state of profound prostration. She was diagnosed with depression. But Orla refused to talk to anyone, she wanted to get through it on her own. She seemed to be succeeding, but in June 2017 she had a relapse.

Knowing the name of the disease that afflicts us makes it possible to take measure of ourselves, it’s true.

BUT LOOKING OUR DESTINY IN THE FACE

ISN’T EASY IF IT SPEAKS TO YOU OF SUFFERING.

ORLA

County Cork, Republic of Ireland

During those weeks in hospital, I was so sick that I can’t even remember my thoughts. After a bunch of painful exams they diagnosed my illness. I returned home and to my life. Well, to a new life.

There was a ‘before’ and there was an ‘after’, and that ‘after’ scared me. It was an unknown and I had no idea what the future had in store for me. This disease affects not only my body but it also got to my mind. I think it took me longer to accept the idea of the illness than accepting the physical consequences of it[3].

Going back to school after the hospital was really hard. I was on steroids and my cheeks were really puffy. It had a strong impact on me —I didn't have time to process all the changes.

I was a different Orla, physically and mentally. I had to go to class, then go home and do the injections, then go back to school and try to make up for the time I’d lost because of the illness. I lost a lot of friends, but there are others who stayed with me—they’re a great comfort. Actually, I was the one who changed so much after the hospital. Some things that had been important for me up till then

weren't so important anymore. My view of the world had changed and it no longer fit with theirs. I learned that there's no reason to keep someone in your life who doesn't accept you for who you are, or who you have become. It's just not worth it.

And besides, I had to focus on myself, on the pain, on the medications I had to take. Even if mentally I was going through a very difficult time, the mental aspect went into the background, compared to my need to feel better physically. However, the mental part—because it was being pushed to the side—ultimately builds up until it becomes unmanageable, and once it reaches that point even your physical health gets worse. I got help and it was good for me to talk about it, even if in the beginning—right after I got the diagnosis—I wasn't ready to face it. It takes time to get used to the idea that this will be your new life, that you can no longer be the athlete you were. I loved gymnastics. I was on an Irish team and trained four days a week, three and a half hours at a time, and then suddenly it was over! It had been my life, and I'd lost it overnight.

[3] PSYCHOLOGIST'S BOX

Accepting a disease, which is not a gift, is definitely complicated. Psychological support can help families to coexist peacefully with the disease. It is not possible to rejoice over an illness, but it is possible to focus attention on what is not illness and therefore on all the resources and possibilities that children have.

Linda Bergamini,
psychologist, Milan, Italy

BIRGITTA

Delft, The Netherlands

It’s thanks to the professor from Amsterdam who examined Jasmijn when her jaw was inflamed that we know what the disease is that afflicts her: she has CRMO.

At first I wasn’t worried. Okay, she’d have to take some medicines, but then it would pass and everything would go back to normal. In fact, after that first episode, she had a period without problems. During the first year after her diagnosis she seemed to have stabilized, in nine months it was as if the situation had practically gone away.

I wasn't aware of what the disease meant, so I faced it like a normal illness—a stumbling block in life that stops you for a while but then lets you get going again.

On second thought, perhaps it was better not knowing at the start what it involved. A year went by and we almost forgot about it.

Then came puberty and, from that moment on, Jasmijn's condition deteriorated rapidly. It was then that we realized our life would not be the same as before. The severe pains, the extreme fatigue, hospital admissions followed one after another, the treatments. It was all very, very stressful.

My mother really helped me a lot in that period. I worked and often had to take time off from my job. Fortunately, my employer is a nice and accommodating person and has been of great help to me, allowing

me to work flexible hours or from home, as needed.

We were also well looked after in the hospital. They explained that we weren't alone and, although my daughter's illness was rare, it didn't mean that she was the only one to suffer from it. Knowing that has helped me.

WE WEREN'T THE ONLY ONES,

there were other people in our same situation who I could ask for information on how best to deal with this difficult road.

CARMIT

Kyriat Gat, Israel

I remember very clearly the day I decided to take Ofir to a private specialist. I couldn't wait any longer, my little girl was too sick and nobody seemed to take her situation seriously.

I found a paediatric rheumatologist and contacted her. She received us at her home and after reading Ofir's medical records, without even examining her, she stated that it was most likely FMF, Familial Mediterranean Fever, and prescribed a specific test to verify it. In Israel it's not possible to perform this test without a signed recommendation from a specialist.

We got the results two weeks later: it was FMF. The test detected mutation of two genes.

The rheumatologist called us soon as she got the result, asking us to meet her that same day. She was a big help and saved us days and days of anxiety and unanswered questions. She was truly great, I will always thank her for it.

She explained what was involved with the disease, what the symptoms were, and what medications and treatment needed to be followed. As the doctor spoke, I felt a weight descending on my chest, a sense of deep oppression. It was terrible.

I CRIED, I'M NOT

ASHAMED TO ADMIT IT.

We were talking about my daughter and they were telling me that she would never be well again, that she would have to live with a disease that she would have forever and that would make her suffer. She was only three, three years old! She seemed condemned.

I have to admit, knowing the name of her illness reassured me and it comforted me to know that the doctor knew what it was.

AT LEAST WE HAD SOMETHING CONCRETE TO FIGHT AGAINST.

I felt terribly guilty, however, because I couldn’t relieve Ofir's suffering.

I was in charge of everything, the visits, the checkups, the medications. It was really, really tough. But at least I wasn't alone. The doctors understood Ofir's illness and I could share my worries with them, certain that I’d always get an answer. It doesn’t eliminate the problems, but it gives you relief and in these situations that’s already a lot.

AISLING

Straffan, Republic of Ireland

After Jack's CINCA and NOMID diagnoses, he began steroid therapy and then they gave him immunoglobulins for about five months. This was to prepare him for the specific drug. I had no idea what it meant—there were no mobile phones to search internet like today, and it wasn't easy to get information.

I had just separated from my husband and I felt lonely and scared, not even my general practitioner was supportive. Later, I found support in the doctors who handled Jack's case at Our Lady's Hospital in Crumlin, Dublin. There were so many of them because Jack has so many problems. He suffers from intracranial cerebral pressure and hypertension, has damage to his lungs from chronic disease, nocturnal apnoea, and then scoliosis and hip dysplasia. He's treated by ophthalmologists, audiologists, and I can't remember who else.

He's got a general paediatrician who treats him and coordinates all the other specialists, otherwise the situation would be impossible to manage. Sometimes, luckily, I'm able to make two or three appointments in the same day.

Fortunately, we live in Kildare, which isn't far from Dublin. Our local hospitals wouldn't be able to handle all Jack's problems.

The administration of intravenous drugs is also problematic because he has very few access points. He also came down with MRSA, septicaemia caused by the venous catheter due to a bacterial infection,

and it was necessary to surgically remove the device and look for another access route. Jack is so susceptible to infection that when I take him to the ER, I have to warn them first so they can isolate the area where he'll be hospitalized.

All the hospital staff is wonderful, they've never, ever left me alone. If it weren't for the doctors, anaesthetists, surgeons and nurses who've cared for him, Jack wouldn't be here right now.

I'll never stop thanking them.

EMER

Kildare, Republic of Ireland

Amy was almost eleven months old when Dr. Orla Killeen diagnosed **systemic JIA**, after having excluded a whole other series of diseases.

It was a hard blow when we found out that we were dealing with a rare autoinflammatory disease. But at last, it was clear to everyone that I wasn't making things up, that the disease was real and not the result of my anxieties.

When we got the diagnosis, I went back to the general practitioner. I showed him and couldn't keep myself from complaining about his past reservations. 'See?' I said. 'I wasn't making it all up, was I?'

They apologized, contrite. I asked them at that point whether it could be possible that other patients with Amy's symptoms had appeared in the past and they had underestimated their condition. Some time later, I learned that another patient of the practice who had symptoms similar to ours had also been diagnosed. The boy's mother came to thank me because the doctor had finally referred her son for the specific tests.

While you're on the difficult path toward diagnosis, it's frustrating to not be able to count on the understanding of those around you. You're forced to swim against the current and everyone asks you, 'Where are you going? Turn back, the truth isn't what you think, you're wasting your time!' You flounder and sputter, struggling to go forward, which is what I did.

I was lucky to get confirmation of the diagnosis fast enough; Jack’s difficulties probably prepared the ground for me with Amy. I already knew about joint pain and quickly recognized Amy's signs.

Amy was Dr. Killeen's first patient, and I will forever be grateful to her for identifying her disease.

ANNA

Ludwigsburg, Germany

It was thanks to my son Christopher that we arrived in Tübingen; Anthony was already seven and a half years old at that time. I would have liked to have got there sooner, because in the other hospitals they didn’t treat him properly, giving him tests and administering drugs that weren’t necessary.

I was at home with Anthony and Brian, my other son, when the paediatrician called.

I didn't understand German very well back then and asked her several times to explain it to me. She was very patient but I didn't understand. The only thing I caught was that it was a disease that would last his whole life. The paediatrician told me to come to her office so we could talk about it, and then she said goodbye. I put the phone down and began to cry and Brian punched the wall, hurting his hand. I felt the earth sink under my feet, I was desperate.

Brian called my husband and my cousin—everyone came.

I never wanted that diagnosis. I knew there were treatments and drugs that would help him, but I felt lost. How long will medicine be able to help him? Will he be able to live with this disease, I wondered.

ANTONIO

Ludwigsburg, Germany

When they called from home with the news that the paediatrician had phoned, I rushed back. I listened to what my wife told me and I felt myself turn to stone.

I believe I immediately rejected the verdict, and even today I still don’t accept it. It's simply impossible that my son is sick—he's such a healthy, determined, intelligent child... Anthony plays football, rides a bike, plays, does everything that other children do. So no, he's not sick. I know there's this thing, this FMF, but I don't accept it, can't accept it.

I don't even talk to my wife or my other kids about it. All I do is cry. But I do it alone, when no one can see me.

LENNY

Born, The Netherlands

Until Kate underwent the tests, we thought her illness was the same as what my mother and I had, and maybe even Tess. At the time, there was talk of SLE or Sjögren's syndrome, even if she didn't have all the characteristics.

One morning—I still remember it well—I was working and had left the office to go and buy some stationery. It was lunchtime and I was in my car in the car park in front of the shop when the doctor called me. I quickly and furiously tore my newly-bought notebook and pen from the packaging to write down what the doctor told me. I laughed, I felt relieved. We finally knew what it was that was making her sick. At that time, I didn't consider the implications of the disease, I was just happy it had a name. After all, we'd lived with it up till then, even though things had gone a bit differently with Kate.

With Kate's DNA test we discovered which was the 'wrong' gene; the diagnosis was Familial cold autoinflammatory syndrome, known as FCAS.

The medical staff that treated her decided to test all of us—me, my grandmother, my mother's brothers and sisters, my niece, and my brothers and sisters. It turned out that in addition to my grandmother and mother, who unfortunately are no longer with us, ten of us had it, including me and my daughters. It was a rare autoinflammatory disease, okay, but we would be able to control it, put ourselves in touch with others in our same situation, create a network, exchange valuable

information...

The girls started the therapy by having injections. They were very painful, and Tess in particular suffered from them.

It was my husband who took the responsibility of administering them. They taught him how to in the hospital. He'd come home from work at around 6 p.m. and do the injections. It was a difficult moment: for him, because he felt like the bad ogre and instead of playing with Tess and Kate he made them cry; for the girls, because the injected drug was very painful, it burned; and for me, because I couldn't do a thing.

Then my husband started having back problems and a nervous breakdown. I think it was the whole thing together: me, the sick kids, and his sense of not being able to handle it all. We hired nurses for the injections, so he was at least relieved of that thankless task.

We separated later, but it wasn't because of the disease. Or at least not just that. The situation had become too much, he just couldn't bear it anymore. I don't blame him. Maybe it's different for me because, being sick myself, I have a greater connection to the disease and its consequences. I've always lived with it and, in a way, having to deal with it with Tess and Kate didn't seem particularly distressing to me. For him it must have been different,

THE BURDEN OF ANXIETY AND PAIN OVERWHELMED HIM.

However, we're still good friends. I have another partner now who's very supportive.

SUSANNE AND ROBERT

Dillingen, Germany

When Jannika was diagnosed with suspected PFAPA, we were almost happy about it. Not happy that she was sick, of course, but they explained that it’s a disease that passes after the first years of life. So, we would only have to wait for time to take its course, taking the disease away with it.

When the fevers appeared, we treated her with cortisone and her temperature came down. After ten days, however, it came back, and we had to start with cortisone again. The situation improved in summer but in winter, perhaps because the cold lowered her immune system, Jannika's symptoms worsened.

When she was seven, the rashes began to appear. But I didn't take her to the paediatrician often—I didn't think it was necessary just for a rash. Then the joint pains appeared. We took her to a specialist, who expressed doubts about the PFAPA diagnosis.

Actually, for as much as I was almost relieved at first, I had the sensation that there was something else. I don't know, maybe we mothers have a kind of sixth sense that warns us, that alerts us when something is wrong with our children. It must be due to the fact that we carry them inside us for nine months, they grow with us. I believe an invisible thread binds mothers to their children, like a live current flowing between us, a communicative flow that needs no words.

Dr. Kümmerle-Deschner had Jannika undergo further tests, and in

the meantime I searched the internet for the symptoms my daughter complained about. I discovered that many diseases were similar to PFAPA. I came across CAPS, and many of its symptoms were Jannika's, including joint pain. So, I went to look at all her medical records and I discovered, to my surprise, that knee pain was constantly mentioned. Many of the symptoms were mine too! I was shocked, how could it be that I hadn't paid attention to it for all those years? The tests done in Tübingen confirmed a very high level of inflammation for CAPS for me as well. It was a genetic anomaly that I was carrying and that I had passed on to my children. I was devastated by the sense of guilt—they were suffering because of me!

I can say that from that moment on, our lives changed. At least now we're aware of exactly what we have to deal with every day. We know that the doctors who treat us are always ready to deal with any emergency we have and that they won't leave us all alone. We'll never be alone again, and that's nice.

IT'S LIKE FINDING FRESH WATER IN THE DESERT. IT GIVES YOU THE STRENGTH TO GO ON, DAY AFTER DAY.

TIZIANA

Partinico, Italy

After the diagnostic testing, we found out that the children suffered from CAPS, rare autoinflammatory diseases. At first I didn't understand much, I confess, and I hoped that the doctors were wrong. I took the children to therapy, without asking too many questions. But then I started to dig deeper and I wanted to know exactly what my kids had.

WHAT I DISCOVERED IS THAT UNFORTUNATELY THERE IS NO CURE,

they'll have to carry this thing with them forever. The drugs are to keep the disease under control but they don't cure it.

It wasn't easy to explain it to the kids. When they were younger and didn't ask questions it was simple, but then they started asking why they had to go to hospital so often while their friends didn't. It wasn't easy to explain to them that compared to their friends they had something else. A disease that would never go away—at least as far as we know for now—but that they would go forward with their lives anyway.

It was a relief when I heard what it was. It had taken a long time for

the doctors to agree with me there was something wrong, to take me seriously.

In Sicily it's particularly hard, and not having much education I've also had more difficulty, I think, in getting people to take me seriously. I would like doctors to realize that mothers aren't always fantasists, and that often when they say their children are sick, they're right. It would be enough to let the mothers talk, to listen to them carefully, and allow their experience as doctors to lead them to recognise when they're faced with a serious case. I wasn't given this kind of listening, and the consequences were years of suffering.

Talking, discussing with doctors, and intervening in time can save children and families a lot of pain. Isn't that a doctor's job, after all?

ATAR

Lombardy, Italy

In South Africa, the rheumatologist confirmed that we were dealing with an autoinflammatory disease. I immediately contacted the Meyer Hospital in Florence, through the allergist who had treated us for FPIES.

One of the consequences of this constant back and forth with so many doctors is that I've become friends with some of them. The allergist was amazing. She arranged for us to meet with a team of specialists who suggested starting treatment with a specific drug for this type of disease.

They told me, 'Let's try it. If there's a reaction, we'll have confirmation that we're dealing with an autoinflammatory disease even if we don't know exactly which one.'

In that period, I often visited my family in Israel. My grandfather's cousin is a head physician in one of the most important university hospitals in the country and he directed me to a friend of his: the number one doctor, worldwide, in the field of inflammatory diseases.

I was able to get an appointment without delay, he examined Michelle, and gave us his verdict. I use the word verdict because it was more a decision than a diagnosis. At least that's what it seemed like to us.

He told us that Michelle was suffering from a rare autoinflammatory

disease and Behçet's syndrome, both at the same time! He indicated the treatment and gave me a table with the progressive dosage of the drug.

We returned to Johannesburg and shortly thereafter my husband was attacked, so we decided to return to Italy.

We lived near Milan but had to go to Florence for the tests and biopsies, and it was complicated because just between going and returning it took two days. I wasn't worried for me because I wasn't working and had practically erased myself to take care of her—I no longer had a life of my own, every step and every breath of mine was as a function of my daughter. But it was tough for her because it interfered with school life. I didn't want her to feel left behind by her kindergarten classmates who were learning pre-writing and pre-reading skills. I contacted the Italian association regarding periodic fever to find out if there were any specialists in Lombardy, the region where we lived. I was directed to Dr. Cattalini in Brescia.

I wrote to him and he answered me right away, and we've been working together ever since. It's a great relief.

Rare diseases are by their very nature difficult to diagnose, which is why it's so important to gather as much information as possible.

After having diagnosed Michelle's illness, Dr. Cattalini wanted to have me undergo the same tests. It turned out that I, too, am positive for FMF, even if in a milder form than my daughter! In fact, I've always had skin problems, stomachaches, bleeding, and knee pains, but no doctor had ever put them together as parts of a single puzzle. Since then, I've been taking the same medication Michelle takes and I'm much better.

The future is an unknown, but sometimes the past has pages we can only read after a long time.

LUC

Amersfoort, The Netherlands

Even though I was a kid, I knew my parents were worried about me. They struggled to find someone who would take them seriously until they found that one doctor.

ESPECIALLY MY MOM WAS ALWAYS LOOKING FOR ANSWERS,

but no one could give her any. When we got to the **NIH** and finally they told us what I had, I saw she was relieved but still really worried because it was a rare disease and it wouldn’t be easy to deal with it.

I was seven years old and even though they told me, I didn't really understand the name of the disease or anything else. I started to understand what it meant when I was about ten or eleven.

In America, where we lived then, it was harder to live with my condition. We played a lot of sports at school and lots of the time I had to stop playing because of the pain. And it wasn’t easy to explain it to my classmates or teachers. Everything was more chaotic there, there were way more commitments. But here things are more relaxed, I live better here.

Living in a small town is important for me because we all know each other and I don’t have to always tell people about my problems. My friends and teachers, or the people close by, know and they help me, but I didn't have to tell everyone. What I do, now that I'm in middle school, is tell my new classmates every year but, maybe also because of my autism, I don't feel like it’s a problem. Then I see that it’s not such a big deal for them, in the sense that they know about it and try to help me as much as they can. I don't think they really understand what it’s about, but sometimes not even doctors understand it very well, so…

My life didn’t change all that much after the diagnosis because, well, I'm always the same. I think it changed for my parents because now they know what I have and I think it helps them so they understand how to act, but I just keep doing things like before. I go to fencing practice or do physiotherapy just like before, but if I'm too sick I just stay at home. I try to stay in touch with my friends, including Tyler, my best friend from when we were in America. We Skype a lot.

EVA

Schoten, Belgium

After my diagnosis of **Juvenile Idiopathic Arthritis**, the paediatrician told me I couldn’t do gymnastics for a whole month. It seemed like an eternity and I didn't understand why an illness should keep me away for so long from the thing I loved most in life! But I wasn't worried, I thought my parents would fix everything, and that I would recover and start living again as before. I was sure it was just a stumble, a little detour.

I was only eleven years old, in elementary school, and the furthest future I could imagine was the end of those thirty days. I understand only now, however, that it was a good thing I didn’t know, at the time, what awaited me.

I had met suffering on that day in October, and I continued to perceive it as an annoying and bothersome stranger, a guest who arrived uninvited at the wrong time. It was only much later that I realized it would be my partner forever, and that I would have to learn to live with it.

I don't have only painful memories from that time, though. In the summer of 1995 we were all in Utrecht, waiting for the newly started medicine to improve my condition. My parents and brother were staying in a facility that accommodates the families of children admitted to hospital. Despite the fever burning in me and the pain in my joints, it was almost like being on holiday because we were together and it was an almost playful environment.

In a way, I think finding out what I was suffering from was a relief for my parents. At least they knew what I was up against. I began to be treated with medication, and I alternated between periods when I felt better and others when my condition worsened. I realized I couldn't devote myself to gymnastics and ballet anymore, and it was a painful recognition. I needed to rethink my life and focused on possibilities instead of impediments.

WENDY

Tipperary, Republic of Ireland

Giving a name to the diseases that afflict children means crossing the line that separates uncertainty and fear—which magnify everything—from awareness. When you're faced with rare diseases that put a strain on parents and patients, it's important to know what you're fighting so you can come up with a specific strategy.

The greatest difficulty that people encounter, before and even after receiving diagnosis, is communicating it to others[4].

Saying that your child won't always be well, that they won't always be able to do what they had done up to the day before, but that they might be able to do it again, is incomprehensible to most people, whether they're friends, teachers, schoolmates or others.

We suggest talking about what your children are suffering from with all the people around you so they can get involved and can understand and help the children when they're in trouble.

Each child is different, each lives with the disease in their own way. There are those who want to tell their friends or teachers, but there are those who prefer to keep it to themselves and don't want to do that. My daughter, for example, couldn't talk about it. She was ashamed of a disease that she herself thought should only affect older people, and she was convinced that no one would understand that arthritis could affect even a little girl.

[4] PSYCHOLOGIST'S BOX

The difficulty lies both in making people understand adequately the situation of the child, their needs and requirements, and in the fear that this will make them different in the eyes of others. It is important to support the families to avoid any feeling of shame. There is nothing to be ashamed of.

Linda Bergamini,
psychologist, Milan, Italy

We talk a lot to the kids, and it's surprising to realize how they live with their condition. I've noticed that they often see it in a completely different way than we'd expect. And it's important for them to talk to someone, even someone who's not in their household.

Often I go to schools to talk about rare autoinflammatory diseases. Sometimes sick children want to participate, help me prepare the presentation, stay with me while I explain the characteristics of the disease, while others want me to come to their school to explain to their classmates and teachers everything that their pathology involves but they prefer to stay in the shadows, not be named. The latter group are children who still feel a strong sense of insecurity, who have not yet metabolized the disease. You have to give them time, let them learn to trust those around them before they open up.

NORMA

Dublin, Republic of Ireland

The moment when parents and children receive the diagnosis of a rare disease is one of the most difficult on the journey that began with discovery of the first symptoms.

Children, in particular, may be disoriented. We always try to give them the information in the simplest possible way. It makes no sense to tell them more than necessary, it would just confuse them.

We encourage children and young people to ask questions, and if they don't feel comfortable talking to us—people who in their eyes are still strangers—we ask parents to get them to talk to them. It's important that they speak, that they express their thoughts, as this will help them deal with what they're experiencing.

I feel so much tenderness for these children, some of whom are frightened when they come to us. A lot depends on how the parents respond to the diagnosis.

I use a book of fairytales that tell the story of rare diseases, to show how people can still do a lot of things, study, have fun with friends, do sports, and so on. It helps young children understand that they can have a life like everyone else and, above all, that they're not alone.

Older children can also be very worried at this stage about the impact the disease will have on their physical appearance. In some cases, they will have already begun to take medications, although not

specific ones, and these may cause changes in their bodies. For example some drugs make you gain weight and in the delicate phase of growth in which they find themselves, their physical self is a fundamental aspect of their lives and it can greatly affect their social relationships.

We always encourage them to talk to one of the team if they have concerns or worries. Kids may find it easier to ask us questions they wouldn't dare ask their parents, either because they wouldn't want them to worry or because they're embarrassed about them. When it comes to questions about physical relationships, for example, very often they wouldn't be able to find the words to ask their parents but, I've noticed, with us they feel free to talk about them.

Sometimes they're angry, and wonder why the disease affected them. We explain that there is no reason, the disease didn't choose them, it simply happened the way it did for various reasons related to DNA anomalies.

Younger children are worried about when they'll be able to play again and when the pain will end.

Children are at different stages of development and therefore they can have very different needs and to which we try to give the best possible answers. Hopefully we reduce their worries and anxieties when they come visit us.

MICHAL

Haifa, Israel

After seeing various specialists in an attempt to figure out exactly what I was suffering from, I was examined by one of Israel's leading experts who diagnosed me with ankylosing spondylitis**.** It wasn't a good thing to hear, but at least all those crazy pieces of the puzzle began to fit together. After almost twenty-eight years, I finally met the enemy that was taking possession of my body, I could give a name and a face to the thing I would have to fight. And believe me, knowing your opponent is critical in any context[5].

So, I started getting busy since I was used to working hard. I studied up on the disease and joined the Israeli Association for RMDs Patients "Mifrakim Tz'eirim", an association that works to support families affected by diseases like mine.

Helping others has always been my passion and when I was offered the role of Project Manager in the association, I enthusiastically accepted it. Now I'm also the Managing Director and I do everything I can to alleviate the suffering of others.

I KNOW HOW THEY FEEL BECAUSE I EXPERIENCE IT MYSELF.

For me, my desire to help others and to be close to my fellow human beings is in my DNA. My parents survived the Holocaust and, ever since I was a child, I've experienced a sort of solidarity in my family and a love for the whole of humanity. Listening to others, trying to understand their needs, doing everything to satisfy them, that's my goal.

I'm honoured to be a member of the association and our aim is to be close to patients and their families, right from the moment the disease begins to manifest itself. Unfortunately, rare diseases, by their very nature, are neither easy to diagnose nor easy to identify, so it happens that those who get sick feel alone and lost. It's vital that the work we do is known by as many people as possible because no one should feel like they are abandoned at the mercy of the unknown. People need to know that there are associations like the Israeli Association for RMDs Patients "Mifrakim Tz'eirim", ready to provide all the help they need along the difficult path of the disease.

[5] PSYCHOLOGIST'S BOX

Diagnosing a rare disease is unfortunately often long process. When patients finally arrive at a diagnosis, they feel relief, finding confirmation in what they have experienced for years and they regain confidence in themselves and feel more able to cope with what awaits them.

Linda Bergamini,
psychologist, Milan, Italy

AUTOINFLAMMATORY SYNDROMES (part 3)

One of the most striking characteristics autoinflammatory syndromes, from the mild to the more severe, is the recurrence of a “stereotyped” clinical picture. It is often the patient themselves or their family whose attention is attracted by the recurrence of episodes that are identical. One very simple instrument that can help when there is the suspect of an autoinflammatory disease is a clinical diary. It’s not unusual when we first see a child with “recurrent fevers” and we cannot come to a definitive diagnosis, that we ask the parents to come back in a matter of months (depending on the severity of clinical manifestations) with the fever diary completed. In this diary it is important to note:

• the exact date of each (fever) episode
• the exact duration of each (fever) episode
• the clinical manifestations that repeat themselves at each episode. In the case of cutaneous manifestations (i.e. skin rashes) taking a photograph can be very useful
• if there are symptoms also between the episodes
• if therapies were prescribed, what was the response
• if laboratory or other tests were prescribed, and what were the results

It is also very important that the family doctor visits the patient every time they have an attack, to help fill in the diary. Since autoinflammatory syndromes are characterized by systemic inflammation, it is very useful to have bloodwork done during an episode, to check for inflammatory markers and also to help rule out infections. It is also very important to check inflammatory markers between the episodes to verify they come back to normal. When the picture is clear, it’s not mandatory to do bloodwork at every episode,

but that should be decided on an individual basis with the physician. If the clinical diary shows recurrent episodes of systemic inflammation (i.e. fever, elevated inflammatory markers and signs/symptoms of organ inflammation detected by your doctor) without evidence of bacterial origin, probably the most useful thing to do is consult a physician with expertise on periodic fever syndromes for further work up. Since centres with expertise on autoinflammatory syndromes are not widespread, the waiting times for such a referral may be long. In such a case, continuing to keep track of the episodes may be very useful, and ease the job of the referred physician when the time of the visit finally arrives. As centres specialized on autoinflammatory syndromes are not always easily accessible, a few strategies may help optimize the care of every child with autoinflammatory disease.

When the final diagnosis comes, it could be useful to ask the medical team of the centre to provide the family with a detailed medical letter that includes the following:

a. the main clinical characteristics of the disease and the therapy needed (not only medical treatment, but also other needs, such as physical therapy, etc.) with explanation of the most common side effects and how they should be managed;
b. how frequently the child has to be seen by a physician, and what tests should be run. This “follow-up plan” maybe individualized, depending on how difficult it is to reach the specialized centre. In many cases it is possible to agree on visits to the centres just once or twice a year, and to refer to local hospital/primary physicians for more regular follow-up;
c. whether the disease or related treatment contraindicate everyday activities (i.e. school, sports, etc.) or universal medical procedures (i.e. vaccination);
d. what to do in case of common childhood diseases (i.e. upper respiratory tract infections, gastroenteritis) and, if relevant, what drugs should be avoided; e. when it is crucial to contact the medical team at the referral centre.

All this information should be shared with primary care physicians and patients themselves, and their parents could use it to inform teachers, coworkers or other reference figures from other activities

(such as sports teams). Usually patients feel much more comfortable if they know that people from the places they frequent (school, gym) are well aware of their diseases and the needs that come from them. Of course this information is very personal, and some patients prefer not to share all of it.

Once a diagnosis of autoinflammatory disease is made, it is also very important to look for a patient association. Usually the team from referral centre knows the associations very well and can also suggest websites that provide verified information on periodic fever syndromes. As in all rare diseases, the parents of children with autoinflammatory diseases usually feel isolated and helpless, even after the diagnosis. Patient associations may be of invaluable support by maintaining contact and helping the community grow stronger.

Dr. Marco Cattalini

04. Beginning again

WHEN, LIKE A RAINBOW, HAPPINESS DREW AN ARC ON THE HORIZON

Every day is a new opportunity to begin again. Every day is your birthday.

- Dalai lama

KATY

County Cork, Republic of Ireland

It's not easy for me to tell you what Orla went through in 2017.

She was sick and I, as her mother, couldn't help her. She had locked herself into a bubble that removed her from everything, a bubble of fear and pain.

None of us knew what to do to make her feel better but we realized that nothing and nobody, from the outside, could help her. We couldn't be with her all the time; she would have had to find the strength to deal with her illness within herself.

I don't know how it happened, but one day Orla decided to start attending the meetings with the CAMHS psychologists again, the ones of our mental health service for children and adolescents. And her schoolmates were fantastic, they stood by her and supported her in everything. From that moment on, Orla seemed to recover some serenity. She really committed herself to it and worked hard with the specialists.

I wondered what it was that depressed her. Well, of course, the disease itself and the pain, because Orla had suffered a lot physically, but that wasn’t all. She was shocked to see herself so transformed after the hospital. The drugs she had to take had made her gain a lot of weight and for her—a beautiful, agile and slender gymnast—it must have been a real trauma to look at herself in the mirror and not recognise herself.

She came close to dying because of the disease, and those days in the intensive care unit had frightened her. She was afraid it might happen again, and that next time she wouldn't make it.

Since she's started back at CAMHS, she seems to be getting used to the idea of the disease. She knows that her life has changed, but she's beginning to realize that 'changed' doesn't mean 'over'. She also agreed to go to an event organized by the **iCan association**, organized by Wendy Costello, as a mentor to tell about her experience. Before, she would never, ever have done that. We understood that she was really learning to deal with her disease and that she realized she wasn't the only one with such a problem.

When we returned home it hit me, as if for the first time, how wonderful all those people in our community were. They were there for us, from friends to neighbours to teachers. We had all made them aware of Orla's disease and they understood and supported it.

During that first terrible period when my husband and I were in hospital with her, I was shut off from everything else, we both were. I had neglected Tomas and Niamh, I hadn't kept them informed about what was happening to their sister, and I found them confused and frightened. Fergus' and my work was also affected and we both had to ask other people to step in for us when we weren't able to keep up.

Our life as a couple was also affected, I think it was inevitable. For almost a year and a half Fergus had to sleep in the guest room because Orla was sleeping in bed with me. I wanted to be close to her after the hospital so I could monitor her continuously.

He and I sometimes fought—we had different opinions on many things—but all in all, no more than before. What saved us was not letting the disagreements between us stew for a long time. We always found a way to say, 'Okay, that's how you see it. I don't, but now that's enough, we have to go on together.' And that's what we did.

After the first days when Tomas and Niamh were angry with us, mostly with me, for having literally forgotten them, they became even closer to Orla. What I really appreciated was the fact that they never

pitied her. When they saw her down, they encouraged her to go out for a walk; they were close to her, always. They never made her feel she was *different*. And they're the same way today. We still take our holidays together. They're wonderful, and I'm very lucky. We're slowly returning to our lives.

Fergus and I also went on a weekend alone, without any of them. We needed to regain our space as a couple because up till then we'd only been Orla's parents. But never, not even for a moment, was I afraid of losing Fergus.

ORLA

County Cork, Republic of Ireland

I remember the euphoric feeling I had after I worked out, that physical fatigue that's so rewarding. It was all, 'Oh, it was exhausting but, boy, am I happy!' Sometimes I felt like I needed to be carried home on a stretcher but there was nothing better I could have wished for.

When you stand on the balance beam and think, *Oh God, I'm not going make it* and it seems your feet don't belong to you and you can't make them take even a small step and then, as if by magic, the movement flows and you land on your feet and not on your head. You did it, and it's fantastic! It drove me crazy, all of it. I tried training again but I had to stop, it just wasn't right for me anymore.

But, I continue to exercise. I understand it's important for my physical and mental well-being, it keeps my morale up. It's not healthy

to focus on what you can't do anymore. And I'm still good friends with all my old teammates, we care a lot about each other.

As I already said, it hasn't been easy to accept all this. It took time. If I look back at the Orla I was then, I see that I'm different from her.

I FEEL LIKE I'VE GROWN A LOT, ESPECIALLY IN THE LAST YEAR. AND I'M NOT AFRAID TO SAY THAT I'M PROUD OF MYSELF BECAUSE I'VE MANAGED TO OVERCOME MANY THINGS,

to accept them as part of my new life. I know now that I should have made room for more people, accepted help from wherever it came, but even if I wish I'd understood it before, I can say that now I've achieved a good mental balance. I'm in a delicate phase because I'm changing drugs, but I'm not worried, I know I can deal with it.

I'm in my last year of secondary school and it's a rather intense period, but I try not to get overwhelmed. School's important, of course, but I'm also giving myself more time to be with friends, to take walks. I want to do things that make me happy, it's a gift I give to myself. I don't want to feel pressured anymore. I love the silence of tranquillity.

We live in the countryside and we're surrounded by fields as far as you can see. I like it a lot and take advantage of where we live to wander through the woods. It's something that makes me feel good and that I can do. But other things, like drinking, which for the Irish is almost as natural as breathing, I can't. I quickly learned what's good for my body and what's not. I've found my balance.

Of course, I didn’t just have to turn a page, I really had to change the whole book! I had to accept the fact that there will always be good days and bad days, when you feel tired as soon as you open your eyes and you think you can’t get out of bed. I like going to school but I understand that it’s better, when my body slows down, to miss a day instead of trying to force myself to go to school anyway and then miss three.

My brother Tomas, who’s twenty-four, and my sister Niamh, who’s twenty-one, help me a lot. They're my best friends, even if they make me angry sometimes. But it's always been like that between us and I don't want it to change. They don't treat me differently just because I have this thing. They know how to be there for me in the right way and they’re really helpful. I know they’ll always be there for me.

BIRGITTA

Delft, The Netherlands

Jasmijn was going through that delicate moment of transition from childhood to puberty when the disease reappeared.

She had to return to the hospital for the administration of intravenous drugs and had to endure enormous physical pain. At the same time, she went from primary to secondary school.

Jasmijn had to deal with three things all together: adolescence, new school, and illness. One of them would have been enough to upset her but she had to handle all three of them together. She was often sick, and couldn’t go to school regularly or go out with her friends. It wasn't easy at all.

I informed the school and she informed her classmates about what was going on, to avoid misunderstandings. Jasmijn might need more time for her classwork or might have to miss breaks from time to time, and her classmates needed to know why.

Only then did I realize how terrible the situation was and how much it would change her life and mine. But somehow we had to face it. It was the two of us, and we had to move on.

My mother was our lifeline. She dashed to our house whenever Jasmijn was sick, and still, along with my father, continues to help us.

I also started to get information. I'm part of a group on Facebook

that brings together families with the same problems, where you can ask questions, compare notes, discuss the therapies proposed by one doctor or another. It's a great help knowing there are others in the same situation as us.

At first, when families are told, 'Your child suffers from a rare disease,' they think, 'Oh, my God, how are we going to face this? If it's rare, we will be alone, they will not know what to do...'

Having the help of doctors and nurses and seeing that there are others in the world like you helps a lot. It's the sense of loneliness that kills much more than the disease does, the fear of not being understood, of not having a chance.

That's how it is with life, and you have to face it as it is. One step at a time, one day after the next, never outguessing what's next.

It happens sometimes that Jasmijn can't stay at school all day or that she has to be absent, but we deal with it when it does.

JASMIJN

Delft, The Netherlands

There is a moment when you're surfing—when you're standing on a board riding a wave or when you chase a wave and then see it break—when everything else disappears. I love that feeling. In the sea, I feel like everybody else, there's no disease, there's no pain, there is nothing else. And even when my friends are with me, when we are together in Scheveningen, it's the same because in the water, on the wave, it's each

person alone. We're together, yes, but everyone has their own wave, their own moment. And I can forget about the disease.

I love to dance too, ballet and modern dance. It's exhausting, but it gives me amazing energy and a whiplash of good humour. It makes me feel... powerful, that's what I feel, and free.

I remember that when it all started—I was about nine years old—I was very scared of the pain but I didn't realize I was sick. I thought that it would go away quickly and I could go back to playing with my friends, like before.

But in reality everything changed. In a second,

MY LIFE CHANGED FOREVER.

I was all swollen with medication, I think, and I was always really tired and sore. My classmates asked me what was wrong but I just wanted to distract myself, I didn't want to talk about it.

Fortunately, I have a few good friends who've stayed with me. Not many, but the ones I have are irreplaceable. And when you've got friends like them, there's no need for more.

We go out together, we watch movies, we chat, but I know that if I'm sick, they'll help me. I don't have to explain anything because they already know everything, and that's a relief.

I've been thinking about what's happened to me for a long time. Sometimes it's frustrating to know there's no cure, or that we don't have all the answers. But I've found my way of dealing with all this because I've understood that no one's life can be called 'normal.' For me, now, it's normal to live with pain, with drugs; for someone else it's normal not to. I think normality is a relative concept.

This is my life now and I want to enjoy it as much as I can. I take every day as it comes but I know that for every good day, there'll be a bad one, and vice versa.

I won’t let my disease take over, I'm not the disease. I'm Jasmijn who faces an illness. It's just me.

CARMIT

Kiryat Gat, Israel

You know Gal Gadot, the actress who plays Wonder Woman? Well, sometimes I feel like her: I have the world against me and I have to change into a super-woman and face it. I have to go to the doctors, take Ofir for her checkups, take care of her medicines, contacts with the school, everything.

I remember one day, in particular, Ofir had an appointment at the hospital and my husband drove us in his car. We stayed there almost five hours, between one thing and the other, but he stayed in the car waiting for us. He preferred to stay there alone rather than come with us. He can't cope with his daughter's illness. He's just not capable of it. Yet, it would be a big help for me to be able to talk with him about it, decide together what's best, let off steam, get support from him, but it's just not possible. I tell him about everything, of course, and he relies on me completely but he doesn't participate. I've known him for twenty years and he won't change. He loves Ofir with all his heart but can't bear to see her suffer.

I'm supposed to be Wonder Woman but I'm lonely. Sometimes I cry when no one sees me, and then I pull myself up and go on.

We have an unwritten agreement, Ofir and I, a pact between us: when we finish up at the hospital, we always give ourselves the rest of the day together. We go shopping, have lunch, relax. It's the little things that help get life back on track. Then, the next day she goes back to school and I go to work.

I gave up my job in the beginning because of her illness, but then I went back. I told my employer, ‘There's something you need to know,’ and explained to him about my daughter and her illness. He was very understanding, and I'm grateful.

There is an exact moment that marks the turning point between the Before and the After. For us it was in the rheumatologist’s office when she told us that Ofir has FMF. In a flash, our life—the one we knew—slipped away and the new one that came to meet us was so dark and difficult. I truly had doubts we would make it.

Everything was complicated. Even simply giving Ofir, who was three years old, the drug was a feat. It’s only available in tablets and I had to crumble them into her food and make sure she ate all of it, otherwise there would be problems. And this is just one example, every day there were obstacles and more obstacles.

Now that Ofir's grown up, things have changed a bit. She lives her life, goes to school, goes out. Her friends and teachers all know about the FMF and what it means for Ofir and how to help her.

My husband and I work near our home so it’s possible for us to check on things, make sure everything’s okay. I can be absent from work if I have to take her to the hospital for checkups or analyses.

Ofir has become strong. She loved to dance but after training she was too sick and had to stop—making the decision was hard for her. She enjoyed it, it made her feel free.

One day, at home, we were watching some old family films from when she was younger. In one she had a hairbrush in her hand and was singing at the top of her lungs. We laughed to tears, it was really funny. And that was when she decided to start singing. Now she goes to lessons, takes part in shows. She's got a beautiful voice and she's really good.

If I'm Wonder Woman, Ofir is Wonder Girl!

OFIR

Kiryat Gat, Israel

My mom says I'm very strong, but I don't know. I try to live my life in a normal way, without letting the disease take up too much space.

I KNOW I CAN'T DO EVERYTHING. I HAVE LIMITS

but I try not to think about what I can't achieve and instead focus on what I can do well. I had to give up dance, which I loved very much—it's true—but I felt too badly after training. I started singing, though. And I really like it. My teacher even made a short film and I look like a star! It made me happy.

I think this is the way to react: never give the microphone to the disease, never let it speak for you. We're the stars, not her.

AISLING

Straffan, Republic of Ireland

Since Jack was born thirteen years ago, I haven't been the same. I see things from a different angle than before, I pay attention to moments, individual moments.

My day is set by the times I have to give Jack his medications, or his food, or another one of his thousand little needs. He's thirteen years old but he's the size of a child of four or five, he wears a diaper, he feeds through a tube, he's in a wheelchair, and sometimes he has to resort to oxygen. He takes so many drugs that I've lost count of them!

I remember when doctors proposed treating him with a new medicine, I spent days in terror. I'd heard about a child in Japan who died because of that particular inhibitor. Before that, my son was treated with another drug, the one that meant he'd been able to return home from the hospital. It made me feel safe, I could tell it wasn't bad for him.

Actually, changing the medicine has done him a lot of good. I see that he's happier.

Jack doesn't talk, but he can make himself understood. He goes to school, a special one for children like him; there are six of them in the class. They paint, they do puzzles, but they also teach the children sign language. They do something they call 'circle time.' That's when they tell stories. Jack loves stories—his expression changes, literally. There's also a pool full of coloured balls and a sensory room, and often

they take the children outdoors. I'm really happy for Jack, it's a very positive experience for him.

Because he needs constant assistance, there are several nurses who help me. Currently I've got three; one of them went on maternity leave. I also have night care four times a month. It's not much, but it helps.

Those are the only nights I can really rest well. The other nights are always tense because I can't let myself go so far as to truly sleep. I'm always afraid Jack will have a problem and I won't notice in time.

I don't think I could do it all without the friends of the Extra Special Kids group. They're people like me who have children with problems similar to Jack's, who need assistance around the clock. We often talk with each other, not only about our children and their needs, but also just for a chat.

I ALMOST DON'T HAVE A SOCIAL LIFE ANYMORE.

With Jack, I can't go out with my friends like I did before, or go horseback riding.

Towards the beginning, I got in touch with another group in America, the NOMID group, run by Kate Barton and her husband, and Karen Durrant. They were of great help to me and I asked them all my questions in that hellish period. One day, I went to Dublin airport to meet Karen in person. She had a layover before flying to Rome, where she was to attend a conference, and we decided to meet for lunch. It was the only time we saw each other, but I still remember those two hours with joy. She was a friend by then, not just an email contact overseas. She was a person, in the flesh, who could look at me and understand what I felt, without even needing words. She explained so many things to me about Jack's illness, including the fact that he could suffer from high intracranial pressure and that it was necessary to check it. All things I was able to report to the doctors here in Ireland.

Information is essential for everything, of course, but in the case of rare diseases it can make a difference. And in the case of children like Jack, who can't talk and can't communicate if they feel pain somewhere in their body, it can be vital.

Even if only one in fifty children with Down syndrome gets arthritis, it can happen. I'm trying, together with **Down Syndrome Ireland**, the reference group for people affected by Down syndrome, to get regular meetings with rheumatologists. Many complications could be avoided if there were also visits of this kind because children who cannot explain themselves may have to wait a long time before receiving a diagnosis of bone damage, often when it's too late to intervene.

Every two years my son is seen by the entire team, although in the meantime he's followed by several specialists, one for each disease. The visit with the team serves to *put the pieces of the puzzle together* and to bring attention to the overall picture, so that the results of each individual checkup dialogue with each other. The overall view is always the starting point.

Jack was eight years old when I started to accept the idea that I needed a break. I had a new partner in my life and began treating myself to a couple of nights a month just the two of us. It's an oasis of serenity to find the carefree Aisling of many years ago and, even if my thoughts often go to Jack, I enjoy every single moment of those hours.

I also realized I had to spend more time with Molly, who's a couple of years older than Jack.

Since her father and I separated, she often spends time with him and he can devote himself completely to her. I think it's been good for her to be brought back to the forefront. Until her brother was born, she was the baby in the house and had our exclusive attention.

Then things changed, and my husband and I split up. I can't say it was only because of our son's health problems, probably it would have happened anyway, but they also contributed their weight.

Now Molly's fifteen, a teenager, and she also needs love and

attention from her mother. A while ago we went to Wicklow for two nights, just the two of us, and we also manage to set aside evenings sometimes for us to be together. Molly is very empathetic and loves Jack. When we organize parties for him and his friends, even if she doesn't like to participate too much, sometimes she comes and is very affectionate. But she also needs my undivided attention, and the occasions when we're together, just mother and daughter, are precious for me too. I see her growing up, starting to become a woman, and I want her to be serene and independent.

It's different with Jack. Everything's up to me and I think he'll always be my baby. If I see other kids his age, I never think, 'Oh, Jack could be like that now,' or for example, 'Jack could be swimming now.' Jack is Jack, and that's it. It's a miracle that he survived and I take every day as a gift.

Jack's intelligent—I think he has a kind of photographic memory because he recognizes objects and places. And he has a dog he adores, a female from the **Irish Dogs for Disabled.** She's great with him, like a caring mama. Jack loves her.

Sometimes I take him to the movies, which he likes a lot.

Looking at his medical records, the doctors can't believe that he really can do everything I tell them, they think it's incredible he's made so much progress. But it's not just the medicine or the therapies that explain it. It's love.

Maybe it's true that daily life with him is not exactly a cakewalk, but I feel proud of the results he's achieved and I'm happy to share the journey with him.

MOLLY

Straffan, Republic of Ireland

Sometimes I look at Jack and wonder what his life would be like without the disease. We would argue, probably. I see what happens with my friends and their siblings, squabbling and grabbing, but then they make peace. I'll never have this peace-making, we won't ever argue, I won't be able to tell him that I was angry when, as a child, I saw my mother disappear for days and days because she had to take care of him while I was alone at home with my aunt. I can't tell him that I cried—for Mom, for me, for him—and that I still cry sometimes. Jack doesn't know that, no. Instead, I feel that he senses my love. I read it in his big eyes, and even if I can't embrace him and tumble with him as I would like because in his condition he doesn't like to be touched,

I KNOW HE'S HAPPY WHEN I'M NEAR HIM.

It's not easy, even with Mom, but we've reached a balance. We take little vacations, just the two of us, like when we went to Wicklow. It was great! I missed Jack, he's my little brother, I love him and I like to be with him, or go with him to his friends' parties, but having her all to myself was wonderful! I love our mom-daughter nights when we tell each other our secrets. Don't try asking me what they are, because I won't tell you!

EMER

Kildare, Republic of Ireland

When Dr. Killeen diagnosed Amy with systemic JIA, the world literally fell on top of us. After the difficulties with Jack, I hoped that at least Amy wouldn't have to fight her whole life with an invalidating condition.

At the same time, however, we were ready to face it. If we made it through with him, then we wouldn't give up on Amy either.

From the beginning I wanted to know everything there was to know about the disease. Before becoming acquainted with **iCan**, organised by Wendy Costello, I connected on Facebook with groups of parents with similar problems and it was useful to exchange information, also because specialists often took part. What I didn’t do, however, was to search for information through Google. I had started but then stopped —it’s terrifying and useless because unless you have specific expertise, you’re not able to give the right weight to each piece of information. I did find many medical pages on American sites that instead gave me lots of useful explanations.

But it's iCan that's really supportive. Often, the time that doctors dedicate to each patient during examinations is limited and you don’t have the opportunity to ask them all the questions you’d like. At the events organized by iCan, on the other hand, parents meet and talk to each other, exchange information, young people meet each other and compare notes. They have experiences in common and can give advice. Friendships are born that also extend outside of the group and we

continue to have contact in other contexts.

Social networks are important, especially for people who share the same living conditions because they understand what you're going through.

Now Amy's twelve and we're able to control her arthritis with almost no medication. These days, she suffers mostly from fatigue, pain in her knee or foot, but we intervene with painkillers alone, when needed. In recent months she's only had a high fever three or four times, but we've treated her successfully with antipyretics.

Life can be a difficult game sometimes and it can throw you a curve-ball or two, but you have to be ready to hit the ball and run so you can get to home base before it does.

My Amy is almost a teenager and is about to enter one of the most delicate stages of growth for anyone. She has a good personality, however, and is much more mature than her actual age, as is Ryan, our youngest. I think she's been affected not only by her disease, but also by having to deal with the disabilities that Jack, the eldest, has. Both Amy and Ryan are close and compared to other families, where siblings maybe spend half of their time fighting, they don't. At least not for long, or not long enough that I have to step in!

I don't want the disease to define who Amy is, she has to know that her condition is just a glitch, like having to wear glasses, for example. It's not her. It's something that's happened to her.

When you're sick, especially with a disease that affects your daily life and you know that it will do so forever, it can happen that you identify with it, it becomes a physical effusion. *I'm an idiopathic arthritic and I react this way or that*, or things like that. One tends to reason like the disease itself, wearing its characteristics, but the risk is of it becoming a second skin that's hard to peel off.

I think I've managed to prevent Amy from feeling like her illness and to help her see it as something separate from herself. I've never fed her self-pity and have always pushed her to move and do things, maybe stopping to rest when she was tired or in pain, but to then start

again. I don't think it's healthy for anyone to sit on their own troubles.

AMY

Kildare, Republic of Ireland

In the game *camogie*, the female version of hurling, you play on a rectangular field, with fifteen players per team. It's our national sport, and I've been playing it a long time. With a hurl made of ash wood, called a *caman,* you have to hit the ball (*sliotar*), and puck it over the goal stick, which looks like a big H. If the ball goes under the bar, it's worth three points, and one if it goes over. The team that scores the most points wins. You run a lot and you have to be skilled because you can't take more than five steps once you have the ball, before throwing it to a teammate or trying to score.

I can't deny that it's hard in my condition, but I can't give it up. It makes me feel alive and strong. Even if I have to recover at the end of the game, and I'll have my pains, it's worth it. Exercise makes me feel better, more confident, and when I'm on the pitch there's no illness: it's only me, my legs and my desire to score goals and take my team to victory. Yes, I love the sport.

I dedicate most of my afternoons to it, training with my team after getting out of school and doing my homework. Sometimes I can't practice because I have too much pain, but I take it in stride and don't let myself be discouraged. I'll be there next time, that's what I say.

I don't like to talk about my illness with my classmates. Sometimes they ask questions I don't want to answer. But we've told my teachers

so they can understand if sometimes I need to take a break in class. I also wear a bracelet that has my illness written on it so that in case something happens to me, the rescuers know and can consult my medical file which is kept at school.

If I can't get outside, I like to relax on the couch and listen to music. I close my eyes and don't think. I just listen, and slowly the pain disappears, melting into the notes.

ANNA

Ludwigsburg, Germany

When we found out Anthony was suffering from FMF, we thought we were going crazy. It wasn't possible for my son really to be sick all his life!

At first I couldn't even talk about it. I closed in on myself and waited for the days to melt into each other, without doing anything to react.

Things didn't change, though. It was just me who felt badly and I realized that I lacked the energy to give the best of myself to Anthony. I couldn't let off steam with my husband—he still rejects the idea of his son's illness and doesn't want to talk about it.

Gabi Erbis, one of the pedagogists at the hospital in Tübingen, who also became my friend, helped me understand, however, that by keeping everything inside I was in danger of getting sick myself. So I began talking to her about it, and I still do. When I feel particularly down, because I see my son suffering, I call her and she reassures me. Or I call Christopher, he understands me and manages to calm me down. I rarely talk to Brian about it—he's more like his father, he's sensitive and I see that he suffers too much when I talk with him about the subject.

It would be nice to be able to count on the comfort of my husband, but I know it's not possible. He loves our children, literally they are his whole world, but he just can't handle the fact that one of them has health problems. I think it's his defence mechanism: if he doesn't talk

about it, it doesn't exist. At least that's what Christopher told me, and he's also studied pedagogy.

In first grade, when Anthony was diagnosed, we asked the doctors in Tübingen to go to his school to tell the teachers and children what it meant to suffer from FMF. Gabi Erbis was the one to explain what it means to have an autoinflammatory disease.

I was so hopeful that the explanations would make everyone understand that Anthony only needed a little more support, and especially that his illness was not contagious.

I remember that many people, afterwards, expressed their solidarity and most of the children stopped avoiding him.

One of the most painful things, however, was to see that he wasn't invited to birthday parties because some of the mothers thought he was contagious. Ignorance leads to fear, and fear doesn't allow a clear view of things.

I think talking about it and informing people about what rare autoinflammatory diseases, like FMF, are can help the people involved feel not so alone and understand how special these children can be.

Teachers have an important role to play in integrating the child in the classroom and in informing classmates and their parents, which is why we wanted the school to know. We've had, and still have now a lot of cooperation from them.

My son is my ray of sunshine. He's always cheerful, lively, a whirlwind. The thing he loves most is music and he listens to it for hours. He plays the piano and has a passion for drums, to my neighbours great joy! If I had to define him, I would say without hesitation that my son *is* the music.

MY SWEET ANTHONY IS A NOTE ON A VIOLIN.

ANTHONY

Ludwigsburg, Germany

My mom says I'm a note on a violin, but I prefer the piano and I also like the drums. Maybe she doesn't, though, and maybe not even our neighbours do!

I really like playing football, even if my feet hurt after a while and I have to stop. But I don't care, I don't want to be a footballer like Maradona, who my dad liked so much. I just want to have fun with my friends. They know I have FMF, and they're not surprised if I suddenly stop playing. I also like Thai boxing too and my teacher, Timo Noack, says that it’s a sport that teaches how to work as a team, even if you fight alone. Timo says that Thai boxing is a discipline that teaches us to trust each other and ourselves, and to be brave, and I think he's right.

I feel good when I'm with the guys at the gym, and so far I haven't had to stop for the pain while I'm training, so I'm just like everyone else.

I DON’T LIKE BEING CONSIDERED DIFFERENT

and I don't want my illness to stop me from doing the things I like. You just have to try, and knock it out with a good punch!

CHRISTOPHER

Ludwigsburg, Germany

My little brother is my sun, my treasure, my... everything! I really don't know what I'd do without Anthony. I like to help him with his schoolwork, or go out for a walk or spend our holidays together. Even just standing there watching him be mischievous fills me with joy. To see him happy and serene is a wonderful gift for me.

I also think that it's important for a family to share its fears, to express them out loud, to confront each other. It's no good burying your head in the sand, like Wile E. Coyote, the disease won't magically fade away. You have to listen to the advice of the doctors and build a relationship of trust with them. Afterwards, everything will be easier and you won't feel alone and lost, like we did at the beginning. If I had to recommend something to those like us who have to cross this raging river that is Anthony's disease, I'd say to talk to each other and listen. And stay united, because strength comes from cohesion, from the love that only a close family can give.

WE'VE DONE IT AND WE'RE STILL HERE, STRONGER THAN ANY DISEASE.

LENNY

Born, The Netherlands

I used to ride my bike to school when I was a kid. It was no more than five or six kilometres away from home and I usually managed to go the whole way without getting too much pain.

One day, however, the pain was worse and I preferred to take the bus. I got on and sat down, and soon the bus filled up. I could feel the eyes of elderly people staring at me. I knew they expected me to stand up and give them my seat, which would be the polite thing to do, but I just couldn’t. My cheeks burned and I couldn't wait to get to school. I could have explained it to them and they would have understood, but in that moment I couldn't do it.

I have first-hand experience with what it means to have FCAS. It’s one of the rare autoinflammatory diseases that is part of CAPS.

I understand my daughters when they have difficulty explaining what they suffer from to people they don’t know well yet. It's not a pleasant thing, but in some ways it's necessary. The school needs to know this, because the girls may not feel well in class or during other school activities, they may have to take breaks, and their classmates need to know that Kate and Tess can't do exactly what the others do, at least not in the same way.

Last year, at their school, they organised a Rare Disease Day and both Tess and Kate talked about FCAS. I remember Kate still had the Insuflon, the cannula for delivering drugs subcutaneously. Both the

teacher and our nurse explained what it was for, which was important for Kate because she was ashamed for kids to see it when she was wearing shorts in the gym.

It wasn't easy for either of them to be in front of the whole school talking about something so delicate, but then at home they told me they felt better. Now they wouldn't have to give so many explanations, they could simply be Tess and Kate.

Finally, they've both stopped taking the medicine they were taking before and have moved on to another one. Kate had serious problems with the Insuflon fixed to her leg. It became infected twice, perhaps in the pool, and had the disadvantage of being visible in summer when she wore short clothes. People kept staring at her on the street and even though she didn't let on it bothered her, I know it did.

She and Tess are different, especially in terms of personality. Often people ask me if they're twins, and in fact they look alike and are only fifteen months apart. Tess is more like her father. She's very intelligent and does very well at school, but she has some difficulties in interpersonal relationships. It's not easy for her to make new friends, new situations disturb her. She's afraid of flying, or rather *thinks* she is, because actually she's never done it before! She's stubborn, and if she says she doesn't want to do something, she doesn't, and that's the end of it.

Kate, on the other hand, has more difficulty with her school work, especially mathematics, she doesn't like calculations very much. She reads a lot and she's very sociable, she likes to make friends. She's getting closer to puberty and I'm already trembling at the idea of when they'll both be teenagers!

They're very close to one another and help each other too. Tess is less able to control the pain, she's like my mother who was all 'ouch, ouch, ouch!', while Kate doesn't complain much. She may not be her usual exuberant self, but she suffers in silence.

I can say I've struck a balance with the disease. It's not the one who runs our day, we're the ones who adapt it to our needs.

SUSANNE

Dillingen, Germany

With Marco and Jannika's condition, it’s important to know there will be trained doctors ready to give them all the assistance they may need.

For now, they are two serene kids, all things considered. Marco is quiet and he prefers to play alone, he doesn't like to be disturbed. I remember that he spent hours and hours assembling and reassembling Lego pieces of Star Wars characters, he’s always been a Star Wars fan. He also plays a lot outdoors, especially with his best friend. He rides a bike or a go-kart, and sometimes he takes his sister. When she does go with him, Jannika is overjoyed because it's not often that her brother takes her. She loves going with him in the go-kart.

Jannika is different. She’s lively and very exuberant, she’s never still. I tell her she's got pepper in her rear end and she laughs and runs away! She's an adorable little imp. She loves being the centre of attention and is always coming up with something. She's engaging and always manages to convince others to follow her.

She and her brother are very close and if one is not well the other is ready with water or a blanket. They're very thoughtful.

AFTER LIVING WITH THE DISEASE FOR A LONG TIME,

I wouldn't have expected such misunderstanding, though, especially at school. The children were often absent but the teachers were not cooperative. When a teacher says, 'You're always sick. I always have to explain everything again,' it hurts a lot.

We even had to change Marco's school, we gave up trying to get collaboration out of his teachers. In the new one, things have gone differently: he fits in well and his health has also benefited.

One aspect that should not be overlooked, in these diseases, is the psychological one. To be surrounded by affection and to be encouraged improves a person's mood and gives the strength to go on. And in growing children, it's important for their self-esteem[6].

[6] PSYCHOLOGIST'S BOX

Children have a pathology, they're not the pathology. It is important to make them feel safe, to support their growth by helping them understand the different aspects of themselves, to allow them to have rewarding experiences, and to bolster them in their journey as people. If they are guided in the discovery of who they are regardless of the disease, they may be able to cope with the various situations they encounter.

Linda Bergamini,
psychologist, Milan, Italy

Over the years, we've lost a lot of friends along the way. Or rather, people we thought were friends turned out to be superficial. They didn't understand the seriousness of the situation and said they thought the disease was a consequence of going to see doctors too often. Unbelievable! We've learned to do without them and now our

friends—the real ones—are fewer but we can count on them.

The relationship between Robert and I has also strengthened: having to face this has united us even more. At least one evening a week we all do something together, all four of us, but we also take time just for ourselves.

Robert and I really like diving, it's a hobby that we share and continue to practice together. It's an opportunity to be alone, to be together as a couple, and as parents. I think it's important. It gives us balance and it's good for the kids too when their parents are happy.

This disease hasn't changed our habits, it's only modified them a bit. We continue to travel—we've been to Egypt twice, and the last time I took the kids alone. The warm climate is very good for us and all I did was make sure that there was a refrigerator where we were staying to store medicines.

We're not going to stop travelling, ever.

MARCO

Dillingen, Germany

I have a bunny named Evoli. He's only eighteen weeks old, a baby. He likes to eat and I like watching him munch on the grass I give him. Mama says it's good for him. I want Evoli to be well all the time. It's not nice having a disease like mine.

I have CAPS. Mama explained it to me, and she and Jannika have it too. When I don't feel well, I have headaches or my joints hurt and I

can't concentrate.

In the school where I was before, my teachers didn't understand and my parents made me change school. Now it's better. My classmates, and even the teachers, know that sometimes I need more time for my work, or that I have to be absent because I'm not well, but they don't blame me.

I told my friends that I have CAPS, and sometimes I can't play with them all the time and it's okay. I like swimming and riding my bike. Often I go out to play with Danny and Hannes—they're my best friends—and if I'm not well, they help me.

I also really like going around in my go-kart, and sometimes I even take Jannika with me. But only when she doesn't make me angry!

JANNIKA

Dillingen, Germany

My brother Marco says I'm naughty, but I'm not! He's just saying that because he doesn't want to take me in his go-kart. But he's not mean and sometimes he even takes me. I really like riding in the go-kart.

I have a kitten, his name is Ticki and he sleeps with me at night. He likes to cuddle, and when I hug him I pretend he's my stuffed animal. He's really sweet. Sometimes the disease gives me headaches, joint pains, stomachaches, sore throats or rashes on my skin. It's not very fun and I always have to take medicine for it, but at least then I feel a

little better.

And then I dance and sing, and I like to go swimming.

Since I'm in a new school I don't have many friends and for now almost nobody knows about my illness, only my teachers and the headmaster. I haven’t had any problems so far, so nobody’s noticed.

I really like diving and I'm sure I'll get better at it than Mama and Daddy! Then Marco will have to take me in the go-kart more often as a reward!

TIZIANA

Partinico, Italy

After we got the CAPS diagnosis, we went through a period of disbelief. We couldn't cope with the fact that the twins would be sick forever, and with a rare disease! My husband hasn't been able to accept it yet, and I think he never will. He has trouble even saying the name of the disease, but he kept telling me things would eventually get better. In a way, this gave me the strength to face the situation.

Right now the situation is relatively under control. The twins have their periodic exams and take their medications. I know the drugs can't cure it, because for now a cure doesn't exist, but they serve to keep the disease under control, to prevent it from attacking organs, or causing other damage.

When they were little, it was easier to take them to the hospital—they didn't ask questions. But then, as they grew, I told them the truth: they have a rare disease that we're keeping under control and that we won't allow it to affect their lives. They understand and, indeed, are happy now when the time comes to do the treatment because they know they'll be better.

We have our own ritual every time we go to Palermo. Once we finish at the hospital, we go for a walk, we go play, or do some shopping. We've turned it into a playful moment. My husband can't always drive us by car, so sometimes we take the bus. The boys like it and have fun.

We've had some difficulties at school. I had to explain that the boys

didn’t have learning disabilities, as the teachers thought, they just needed more time to do their work, even in class. Especially when it’s almost time start back with the medication because they’re not in the best shape, they have headaches, joint pain, and can’t concentrate. It takes a lot of patience, but I'm tough and I'm not giving up. I’ll go to the end of the world, if necessary, for my children, and I’ll do it on foot if I have to!

Since we found out what disease they have, we haven’t been able to go on vacation, my husband works and his vacation time is used for coming to the hospital. But the situation doesn’t weigh on us. We’re a relaxed couple and giving up travel isn’t a sacrifice. We would do it, and even more, for them. Besides, we live in a place with an ideal climate.

I remember years ago, before the diagnosis, we went to Germany to visit my uncle. The first few days were sunny and then it started to rain. The boys got sick and we had to stay at home for three weeks, the three of us! But it’s okay, we’re moving forward.

ATAR

Lombardy, Italy

Some of the few friends we have left ask us how we did it, how I moved forward. I've wondered too, but the only possible answer is that you have to go forward, your daughter depends on you and the choices you make because those choices can mean that she lives or dies. It's simple.

TOSTOPORTOFALL INTODEPRESSIONARE NOTVIABLEOPTIONS.

Of course, you don't come out unscathed. I made up for it with food, and gained twenty-three kilos. It was my outlet valve, as were tears in the shower, which get mixed up with the rest of the water and nobody sees you. I never cried in front of the girls.

I've learned to move on somehow. The stress level is very high. At first, my husband reacted by denying it was true, saying the doctors were wrong and that soon Michelle would be fine. Then he slowly learned to deal with it, to live with it.

Now he's abroad for work for most of the year, so I'm the one who has to take care of everything. I have to be strong for both, because the girls only have me with them every day.

I try to plan everything, to be ready for any eventuality. I wish I had a crystal ball and could read the future in it, but obviously that's not possible. And so I set out for myself as many scenarios as possible and the best way to deal with each of them.

I also want to always be up-to-date and so I started collaborating with associations that deal with rare diseases, both internationally and in Italy. I help the ones here keep in contact with groups in other countries. It's a way to get a preview of what's new in therapies, drugs, and so on.

Michelle is eight years old now and she's starting to understand, so I explained some things about her illness to her. One day I overheard her talking with some friends. She was explaining in her own words that she has two rare diseases, and she even pronounced their names perfectly. It was so touching I had tears.

She's a very strong child and copes with her condition. When we go for her blood tests, I hear her reciting in a low voice 'I can do it, I can do it, I can do it,' like a mantra. No one explained to her how to do it, it just comes from inside. She's like me—just like me.

We have our consoling rituals after the hospital: we go to lunch together and then go shopping.

When she was younger I took her to a psychologist because I was afraid what she had to endure would undermine her, that all that pain would mark her forever. After a few appointments the psychologist reassured me: Michelle had accepted her condition and was a well-balanced child.

She's very attached to Claire, who's very protective of her. But Claire's suffered greatly from my being away from home to stay with her younger sister in the hospital.

I'll never forget one day in particular. Michelle and I had come home after three weeks of hospitalization, it was during the Christmas period. Claire had stayed home with her daddy, and it had really upset her. She saw her friends with their families having a good time all together, while she hadn't seen her mother and sister for a long time

and didn't really understand why. All she knew was that during the Christmas holidays she didn't have her mom with her. When she saw Michelle again, she treated her badly. I heard her say, 'Mommy's mine too. You're not the only one, you and your hospitals. What about me?' Those words full of suffering tore my heart apart. I probably cried more that time than any other, of course without letting anybody see me as usual.

I REALIZED I COULDN'T COPE WITH CLAIRE'S PAIN ON MY OWN

and took her to see special therapists who helped her. They told me she's divided between a very strong love for her little sister and her fear of losing her, and anger toward me because I take attention away from her. It's not easy, not at all.

Moreover, through the years I've lost a lot of friends, including the ones I thought were closest to me. They didn't always want to hear me talk about Michelle and her problems, it annoyed them. I've let them go without regrets—I want to devote my limited energy to my family and my true friends.

LUC

Amersfoort, The Netherlands

Compared to when we lived in America, it's easier here in the Netherlands. I had a lot of trouble managing my illness at school, instead here we have different solutions that let me do what I want without creating too many problems.

My life's pretty quiet, I can work things out for myself pretty well. School's okay—my favourite subjects are science and technology—and I'm working on computer programming. I also have my own website and can program using languages—that's what they're called in computer science—like Python, C, JavaScript and SQL. At school, we're also doing a project in English. We have to prepare a CV with an application letter, it'll help us when we start looking for work. Once a week I go to fencing, but no more than that because the next day my legs hurt from the exertion. Pretty normal, all in all, at least for someone who found out he was different when he was ten!

EVA

Schoten, Belgium

I always loved school, and I loved it even more in those years. The more the disease knocked me down, the more I got up, aching and bruised but determined not to let it win. It wasn't an easy time. I started secondary school in a wheelchair with my wrists supported by rigid splints. In primary school I had been Miss Popularity but at my new school I soon became the outcast. Classmates who were initially around to help me with my books and schoolbag began to disappear. I was no longer a novelty—however unusual—and they got tired of following me around. I wasn't fun, I couldn't run and play with them. They ignored me. I simply ceased to exist in their eyes. Some of them were even jealous of the extra attention the teachers gave me. They didn't know how willingly I would have done without it! But I longed to succeed and didn't let myself get discouraged; if they didn't want me, I just had to come to terms with it. And I no longer looked in the eyes of people who saw in me a disease, but concentrated my gaze on those who saw me as Eva. I never confused my identity with my illness, and nor did the people who saw the person in me, not the problem I was carrying.

Fate hasn’t been generous with me. When I was sixteen I had hip surgery, and then again when I was twenty-one.

However, when I was eighteen I enrolled in university. I was happy, the disease was more or less under control and I felt like a blank page, waiting to be written on.

I didn't hide my condition from my new friends, and I still don't. I understood that talking about it simplified relationships and avoided those embarrassed and elusive looks that can hurt deep down. I was the way I was—the whole package—and if they loved me, great; otherwise, they could go their own way. Making things clear from the start let them feel free to ask questions, and allowed me to give them answers so they could understand me. There’s nothing simpler or more liberating than truth.

Despite my condition, I always managed to do what I wanted, and my parents encouraged and supported me. I also studied abroad, in Spain, through the Erasmus program and once my mother took a plane to be with me because I had a stomach flu, I wasn't well and I didn't have the strength to go shopping or cook. She made me soup and stayed with me until I got back on my feet a bit.

It wasn't easy, but I had something inside that kept pushing me forward. Maybe everyone who has to live with a disease like mine has it, because otherwise you can't even consider moving on.

Since there are episodes when you risk having your days end in a second, you have to be ready to fight. Three times my immune system turned against me. When it came in contact with a virus, it attacked my blood, liver, and so on. The doctors don't know what virus it was that caused this anomalous reaction, but three times I risked dying. The condition is called **MAS, Macrophage Activation Syndrome,** and it's a really bad thing, and there's no guarantee it won't happen again. But I’ve come out the winner and I refuse to live in fear.

The first time it happened, I was twenty-seven and I had just met a boy. We were seeing each other and I was beginning to think that maybe our special friendship could turn into something more. I told him about my condition right away because I didn't want any misunderstandings. He could have run away when he realized how fragile my life is. But he didn't, and he's still by my side.

WENDY

Tipperary, Republic of Ireland

With our association we organize a Family Day, bringing together several groups of people with the same problem when they're at the beginning of the journey. Speakers explain the various diseases and then we involve the children and kids in various activities, such as cookery workshops, water games, or climbing. We let them be free and have fun together. There are special days for older children too where they can do yoga, psychotherapy, and art therapy.

These days are very useful, for children and adults. You can discover a lot more about what children feel by listening to what they say when they're playing than you can by talking to them directly. Often kids don't want to open up in formal contexts, but when they're free they have no problems talking to other people of the same age about what they feel. They feel at ease, understood, they don't have to justify themselves or explain, they don't have someone in front of them who looks at them with curiosity. They don't feel they're 'on the other side,' instead they're equals.

THESE MEETINGS OFFER THEM OPPORTUNITIES FOR GREAT LIBERATION.

Not all families have the same needs. Many don't want to participate

in the events but prefer only to know other people who share their reality. And there are others who participate in all the events, while still others just want to talk to us over the phone. However, I find that those who participate in the meetings derive great benefit from them. The ones who are shy and quiet at the beginning learn to open up with time, and that makes them more self-confident. For parents like me who've had to leave work to devote themselves full time to their children, finding themselves with others who've made the same choice is a precious moment of sharing and mutual support. And believe me, both are really necessary!

When you're facing a road that's unknown, it's important to find someone who can show you the way.

NORMA

Dublin, Republic of Ireland

A rare autoinflammatory disease is a big challenge. It upsets all the mechanisms, undermines the body, makes the mind waver, creates a sense of temporariness like never before. Certainties are few and you risk drowning in a sea of apprehension.

One of the consequences can be overprotection on the part of parents. Most of them are confused. Perhaps they're well informed about the medical aspects of the disease but are worried about the relational, practical aspects. They wonder if their children will be able, when they're not with them, to take care of themselves; if they'll follow the instructions; if they'll be able to avoid hurting themselves. It's perhaps what frightens parents the most: losing control and not being able to intervene in case of need. It's one of their worst nightmares. On the other hand, there are the children who are pressing for greater autonomy and their need becomes all the more urgent as they grow up and approach puberty.

Very often I'm the link between the parents' anxiety and the children's desire for freedom. Generally, I help them work out small concessions on both sides. The need for confrontation, to put oneself to the test, is fundamental in the process of growth and acceptance of the disease and it would be an enormous damage to prevent children from expressing themselves freely outside the protected context of the family. You can't keep them under a glass dome, they'd suffocate.

It's also a challenge for the teachers, some of whom tend to exclude

children from the activities that could—in their opinion—be risky, especially during the hours of physical education. Yet, we always advise children to move and be active, keeping in mind the necessary precautions. Sometimes, on the contrary, as most of the problems are 'invisible' disabilities, some teachers don't take the special needs of these pupils into account and don't understand their difficulties.

Parents know they can always ask for our intervention at school to provide information about the children's illness. Sometimes it's the teachers themselves who contact us and ask for advice. It depends a lot on the headmaster's sensitivity and the school's ethics. School teachers and headmasters often report that it is difficult for them to be familiar with the numerous diseases and conditions that children can have.

It can be frustrating for parents and children to not have the support of the school or institutions in general. I know there is, in this regard, a group that's working with the government to get attention and visibility for rare autoinflammatory diseases because the main focus is currently on diseases such as cancer or cystic fibrosis, which have the attention of the media.

Here in Ireland, when kids are sixteen years old they're transferred from a children's hospital to an adult service. It's a very delicate phase because the adult medical service is structured differently and the children may not feel comfortable with new people who don't know them. In our hospital we try to keep the kids with us for as long as possible—until they go to college or university if possible—unless the young adults decide to move on. It's happened that after shifting to the adult system some of them have come back to tell us they're not feeling well, and we've found them an alternative. In any case, whether they stay or go elsewhere, they always have our phone numbers. We never abandon them.

We're always ready to help at any time. It might be in the form of a letter to school or university explaining that the kids need extra time for exams, or requests for government grants for parents. Not everyone in Ireland is entitled to free health care and families who are not covered by the system

MAY HAVE TO PAY AS MUCH AS € 100-150 PER MONTH FOR MEDICINES,

beyond the cost of the medical card, which is not a small amount. Sometimes they ask us to help them obtain a free medical card so they don't have to pay for the assistance of a family doctor. Today it's a little easier than it was in the past: the government is trying to meet parents' needs, but up until now that hasn't always been the case. We also help those who may have to make modifications to their homes for their children's needs so they can get subsidies.

We try to be close to families in every aspect of their daily lives. The team I work with, although it's not very large, is able to provide medical, nursing and psychological assistance. Our team consists of Dr. Lowry who's a Consultant Paediatric Rheumatologist, myself and a paediatric physiotherapist, along with assistance from occupational therapy and psychology departments.

While many of these teenagers adapt very well with this condition, others really struggle with the impact it has on their lives. I remember a girl a while ago who told me she was really struggling with life. She said she wanted to die. Her parents were waiting outside when she opened up to me. Thank goodness. I managed to convince her to let my colleagues in psychiatry chat with her. I'll always remember her face, it was so endearing.

Just like other children, some may also have reported gender identity issues. They're no different from other kids and this can happen, but it's a very delicate issue that is part of an already very complicated picture. It's essential that we give them the right support to face these challenges.

In addition to material and practical help, we provide families with contacts to organisations active in the field of rare autoinflammatory diseases. For example, Wendy's iCan and Arthritis Ireland. I leave it up to the parents to choose whether or not to get in touch with these

organisations. Families need time to understand the diagnosis and not everyone wants to talk or share with others about what's happening to them.

When you have to live with diseases like these, life changes forever and daily routines get distorted. Children may need tests and investigations, medical visits, and checkups that require a great deal of time. Not everyone lives in the vicinity of the hospital where they're being cared for with their children, and sometimes they have to travel for hours. For those parents who work, it can be very complicated to find the time to attend hospital appointments and I know that some parents decide to put their careers on hold for a while, especially with very young children. Frequently, it's the mothers who make this choice as they're the ones who usually bear the main responsibility for care. And unfortunately, it also happens that some fathers might not participate at all. I feel a great deal of compassion for these parents—life really puts them to the test.

In all this, there's also the delicate management of the relationships with other children in the family. They may feel neglected because their parents devote most of their attention to their sibling who's unwell and, while they love him or her, they may feel jealousy. It's not easy to find a balance, but I see that most families do their best.

Sometimes we've had to deal with parents who seem very relaxed about their son or daughter's illness, as if it's not anything too serious. We advise that they be vigilant regarding their child's symptoms and report any problems to us, and that they don't delay appointments because in the meantime their child could get worse. Or at least that they should go to their family doctor.

Unfortunately, unless there are urgent situations, waiting times for appointments here in Ireland can be long. Our challenge at the moment is to be able to shorten them. For this reason it's important that patient groups such as the one being set up involve the media in order to obtain visibility. There aren't many patients with rare autoinflammatory diseases and at the moment it's difficult for the government to devote attention to them as there are other competing support groups and organisations. Therefore we need an awareness

campaign because research can still do a great deal in this field to improve our understanding of this condition.

MICHAL

Haifa, Israel

Every association like the **Israeli Association for RMDs Patients "Mifrakim Tz'eirim"** needs to have internal support groups that provide information and help patients to better cope with the disease and to not fall victim to depression. We have an excellent website where specialists write articles about diseases, their symptoms, the drugs to take and their side effects, and the best lifestyle to minimise discomfort and suffering. This information is invaluable for those who find themselves having to fight these kinds of morbid events.

We're also very committed to trying to obtain from the government a more vigorous policy in the field of social protection. Currently, requests for recognition of invalidity must be made to a special institutional committee composed of doctors who are tasked with examining patients and judging whether they're actually entitled to recognition or not. It's important for everyone to know that it's not a label that says 'crippled' but a proper assessment of a condition that is, objectively, disabling and that has serious repercussions in everyday life. We're fighting hard to obtain this type of recognition, which entails a small financial contribution useful for those who are forced to work at 50% capacity or for those who are no longer able to work at all.

The risk of losing one's job is the thing that most frightens people affected by this type of serious illness. One of the first questions that people who come to us ask is precisely how to be granted a percentage of invalidity. People also want to know what social security has to offer. They get information about drugs and the characteristics of

biological drugs. Nowadays, most of the people who turn to us have already consulted 'Dr. Google' and are still in shock from the amount of terrifying information on the web. It's really scary to read that you're taking pills used to treat cancer! That's why it's so important to turn to a support group like ours, because you can have contact with experts who will dispel your doubts, reassure you, and give you the correct information on a case-by-case basis.

At the **Israeli Association for RMDs Patients "Mifrakim Tz'eirim"** we often organise meetings and conferences. We've launched a new model, called **A Doctor and Coffee**: we invite a doctor to a coffee bar to help about twenty people at a time, so they can take the opportunity to ask him or her any questions they want. For example, they can ask about the therapeutic effects of cannabis. Most of them are people who, like me, spent years trying to get a diagnosis. For this reason, we're doing our utmost to raise the general awareness of these diseases and to spread as much knowledge as possible. We're happy that more than three hundred people turned to our association in 2018. Everyone has been thrilled to have found us and curious to know how we could help them.

As we explain,

THE MOST IMPORTANT THING IS THAT THEY'RE NOT ALONE ANYMORE

and that our support groups will be with them for as long as it takes. Everybody's got a difficult story to tell, just like me.

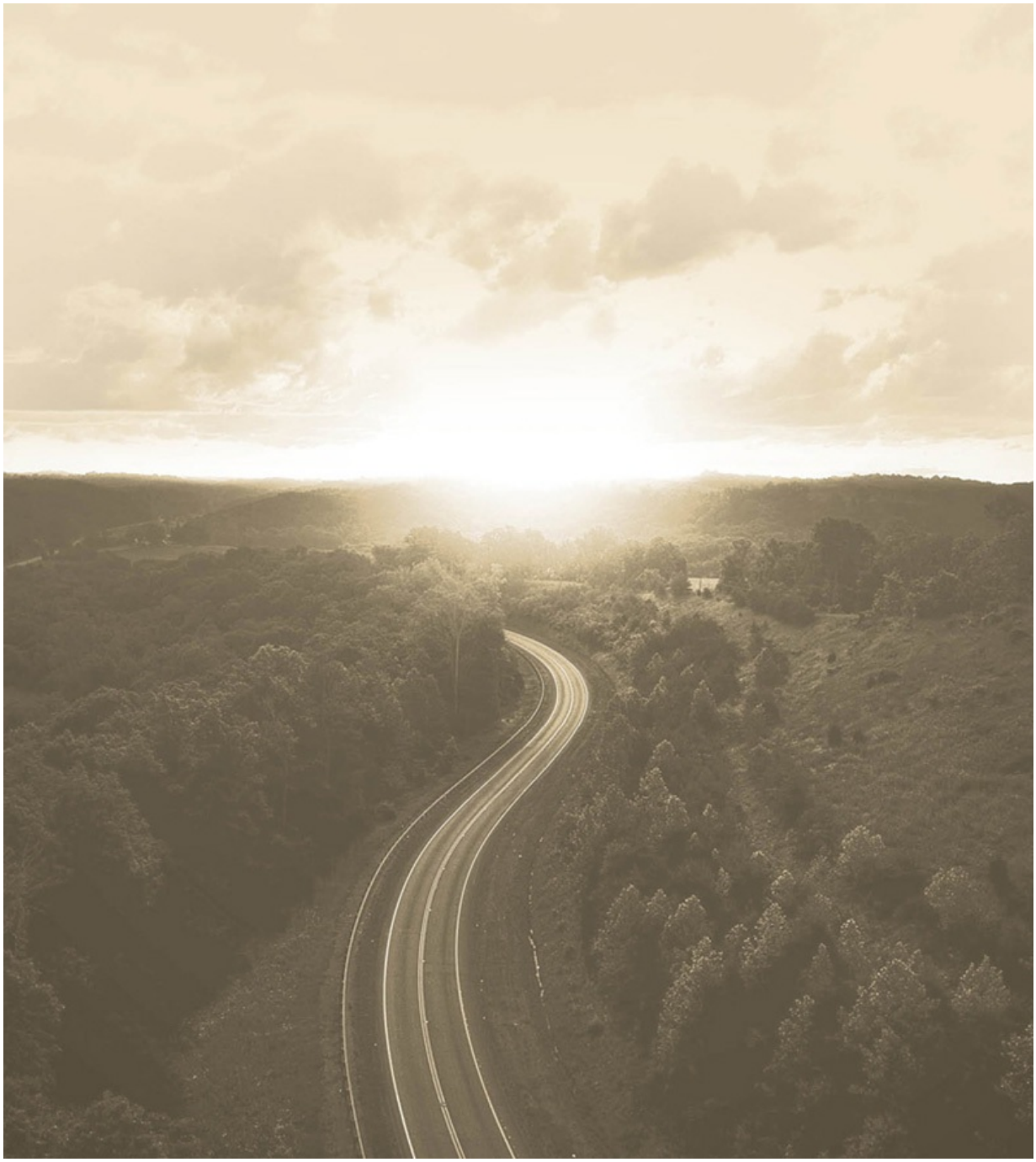
I want to tell you about a special person I've met. She's a mother of six who's sick with **ankylosing spondylitis**. She's so sick that she's in and out of the hospital almost every week. But the beauty of this woman is her large family. Her eldest daughter is sixteen years old and she's practically the lady of the house, she takes care of her siblings and everything that revolves around family life—from laundry to shopping—basically everything. The woman's husband works to

support the family—she can no longer work herself—and oversees the children’s school life. It's wonderful to see how everyone revolves around the mother, how she continues to be the centre of the family, and everyone is with her and for her, helping and supporting each other. It's a wonderful example of family love. I really don't know how a woman who’s as petite as she is could have had six children! She's really great and has a big heart, and thinking about her magically erases the pain from my face, making way for a smile.

AUTOINFLAMMATORY SYNDROMES (part 4)

All the progress in undestanding the origin of autoinflammatory syndrome also leads to the identification of possible therapies. It is now clear that the hyperproduction of IL-1β, a potent mediator of inflammation, is causing the clinical manifestations of many of the autoinflammatory diseases. This leads to the application of drugs able to block IL-1 in patients with autoinflammatory syndromes, and in particular monogenic periodic fevers (CAPS, TRAPS, FMF, MKD). Although these treatments should not be seen as a definite cure, they are able to control with great efficacy the signs and symptoms in the majority of patients, also abating the risk of possible long term complications. IL-1 blocking agents have radically changed the life of patients with many autoinflammatory diseases, permitting them to live almost normally.

Dr. Marco Cattalini



05. The Future

WHEN WE REALISED THAT HAPPINESS WOULD NOT DISSOLVE

“

Life can only be understood backwards; but it must be lived forwards.

- Soren Kierkegaard

KATY

County Cork, Republic of Ireland

What scares us when we realise what it means to have a rare autoinflammatory disease is the future. At first, it appears cloudy, and much more uncertain than it had been up to that point. But then, life goes on and we realise that the future is what we build for ourselves day by day.

Life can be short, for a million reasons that have nothing to do with the disease, and it makes no sense to sit and wait for it to pass. Life should be lived, enjoying every single moment.

Now that Orla's grown and she transferred to the adult health service, she seems more poised, more responsible. She's beginning to understand that she can do many things, that she can have a life just like everyone else can. Of course, she'll need to make a few small 'adjustments', but nothing can stop her from fighting for her dreams.

She's going to a festival next summer and all she has to do is make sure there's a fridge where she can store her medicine. Aside from that, she'll do exactly what the other kids do. It won't be her illness that's going to stop her. Sure, it will always be there, but it's not going to stop her from living.

I don't fear the future and have great faith in research and have high hopes for it. New drugs, new therapies—I'm confident. I want to be.

And when Orla confides in me her doubts about becoming a nurse

one day—with her illness—I say, ‘Orla, you can't know if you don't try it. So go out there and try it, it's always better than living with regret.’

The future is exciting and we must never stop believing it.

ORLA

County Cork, Republic of Ireland

I don't know what my future will be... I don't think about it very much. I'm finishing school now and am focused on what I want to do afterwards. I'm very determined, that's for sure, and usually if I want to do something, I do it! When I was younger I said I wanted to join the army and now I know that’s something I can't do. Some of my medicines need to be refrigerated so I know I’ll have to face a series of restrictions, but it doesn't matter. I'd like to study to be a paramedic, a nurse, or a midwife, which seems more realistic to me. But I don't really care. Things might not work out but the important thing is to have the right people around me and to be happy.

Whatever has to happen, I'll deal with it when the time comes.

NOW I JUST WANT TO LIVE.

BIRGITTA

Delft, The Netherlands

Imagining the future is a challenge for those like us who live with a rare autoinflammatory disease, because life is played out in the moment you live it. It's made up of many small steps, one after another. Some are easy, others more challenging. The disease will always be with you, and you can't ignore it. It follows your footsteps, but you can leave it behind as much as possible. Lots of things can change—new medical discoveries, new drugs—it makes no sense to imagine the future using the face of the present.

When she grows up, Jasmijn wants to study for a profession which will let her help other people; for example a job in the military to help soldiers recover when they're wounded on a mission. Maybe she could study kinesiology.

JASMIJN

Delft, The Netherlands

Yes, I would like to do a job that would allow me to help people. It's wonderful to look at the happiness that lights up people's eyes when, after having suffered, they feel better. The light that radiates from their faces will guide me, I feel that this is my path. A piece of advice I'd give pediatric doctors is: always tell people the truth because it's scary to not know what's happening, even if you're very little. And when you don't have a clear answer, just say 'I don't know,' because that's better than giving wrong answers, which make people even more anxious. I know because I've experienced it first-hand. My future? I have no doubts. I'm going to ride the wave. Always.

CARMIT

Kiryat Gat, Israel

There was a moment, when they told us that Ofir was suffering from FMF, when I wasn't thinking and I let myself fall into tears. Then I realised that I didn't want my future and my daughter's future to be soaked in tears, and I snapped out of it.

I know that the disease can be kept under control;

WE'RE COUNTING HEAVILY ON THE HELP THAT CAN COME FROM RESEARCH.

In the meantime, I do everything I can to ensure that she has the best treatment possible. She has her life and will have her work, like everyone else. She's learned to know her body, to manage the signals it sends her. She won't let the disease control her; she'll be the one who sets the rules. My Ofir is a fighter.

AISLING

Straffan, Republic of Ireland

I ADMIT TO BEING AFRAID OF THE FUTURE.

Jack, deprived of autonomy and with a rare disease, will never be able to take care of himself; he'll always need someone to assist him. My greatest fear is that something will happen to me, and he'll end up in an institution. There's my sister, she's a nurse and takes care of him when my partner and I leave for a while, but she hasn't got children and staying with Jack is a full-time job. I don't know if she could handle it. There's my ex-husband, but I don't know how he could do it either.

I'm also worried about when he'll have to shift from the paediatrician to the adult service because they don't know much about Down syndrome, which at the moment is more critical than his autoinflammatory disease, and in a way it dictates the rules.

The early days were so hard that I didn't think about the future. Jack could have died at any moment and I was living only for the here and now.

Maybe that's what I should keep doing. Life is unpredictable, it's better to take it in small doses.

EMER

Kildare, Republic of Ireland

My daughter Amy is a ball of energy; I don't know any other way to describe her. She's pretty, fun, extroverted.

I've never treated her like a sick person; I've never pitied her. I taught her that it's not important *what* you do, but *how* you do it. There's no sense in standing around feeling sorry for yourself and giving up on doing the things you love. When a bump occurs, you stop and resolve it, and then you go on.

I think that Amy's internalised it, because I see that she doesn't give up.

If I were to give a word of advice to those in a similar situation, I'd tell them not to panic. And then to continue to look at their children as they did before the diagnosis, because nothing's changed. They are and will always be their children. They should be scolded and praised just like any others. Self-pity would hamper their future, clip their wings. It would be unforgivable.

AMY

Kildare, Republic of Ireland

My mother is a strong person, she has to be with Jack, and me, and all the rest. She's taught me to fight and that there are no obstacles, only changes.

Life is like a match, you can win or lose—in the end it doesn't matter. What does matter is how you hit the ball on the pitch.

ANTONIO

Ludwigsburg, Germany

I have no doubt that my son will be able to do what he wants to do in life. He's determined, he can convince anyone to do what he wants them to do. He's a rascal who can steal your heart.

ANNA

Ludwigsburg, Germany

The future? I don't know, I don't stop to think about the future that much. When I do think about it, I would like to stop time because I'm afraid of the moment when Anthony grows up and I won't be able to keep him under my wing. Sometimes I wish he'd stay the way he is forever, the child he is now. But then I look in his eyes and all I want is for him to grow up happy, making the choices that will make him happy, and that's all I want. All children are special to parents, but children like Anthony are something different. They manage to give so much just through their presence. They're like spheres of light that

illuminate the horizon.

AND LIGHT WILL ALWAYS CONQUER DARKNESS, ALWAYS.

CHRISTOPHER

Ludwigsburg, Germany

My brother is a stubborn kid. He's very determined. When he sets his mind on something, you can be sure he's going to get it. I don't see any difference between him and the other kids in terms of opportunities for personal development. He can do what he wants, he's smart and he'll make it. I really think he's going to be successful. In any case, he's got his Plan B in case things don't go according to his expectations! He's a really resourceful kid with a gift for adapting. For sure!

ANTHONY

Ludwigsburg, Germany

When I grow up, I want to work in a bank like my brother. But I'd be happy being a football player too, if the bank thing doesn't work out.

LIKE THE GUY MY DAD LIKES, WHAT'S HIS NAME? OH YEAH... MARADONA!

CHRISTOPHER

Ludwigsburg, Germany

See? The kid's willing to settle!

LENNY

Born, The Netherlands

I know that Tess and Kate are going to be alright. I, myself, suffer from the same disease, but it hasn't prevented me from working, marrying and having children. Today things have improved compared to when I was a little girl, and they'll improve even more, I'm sure. I know that the research is continuing, also thanks to the groups devoted to rare autoinflammatory diseases that are helping to make them known. It's important that people are informed: knowledge leads to improvement. I wish Tess and Kate all the best in life. God gave me the two of them and for that I'm grateful every single day.

SUSANNE

Dillingen, Germany

The future? To be honest, we've kind of backed off from even thinking about it!

Before, we tended to plan everything, to imagine what we would do in five or ten years' time. Since our children got sick, we don't think that way anymore, it's useless. We live day by day, and all that counts is what we were able to achieve by the end of the day and how we did it. Tomorrow is another day and there's no crystal ball that can reveal what will happen.

THE FUTURE? YES, WE'LL THINK ABOUT IT, WHEN IT'S PAST.

MARCO

Dillingen, Germany

When I grow up I'm going to be a rabbit farmer. I think I'll be good at it. I like rabbits. *Be a god boy, Evoli. Hey, where are you running to? Come here...*

JANNIKA

Dillingen, Germany

I think I'll be a doctor when I grow up, like Dr. Kummerle, the lady who's so nice to us.

I WANT TO HELP OTHER PEOPLE WHO ARE SICK LIKE ME AND MARCO,

and tell them that there are lots of drugs that can help them feel better. *Hey Ticki, leave Evoli alone*!

TIZIANA

Partinico, Italy

‘I want to get my license for motorbikes when I’m fourteen, Mama. Can I get it?’

‘Can I get my driving licence when I’m eighteen?’

‘Mama, can I go to music class?’

‘Maybe I’ll be a music teacher, Mama. What do you think about it?’

‘Of course, darlings, you can do it all!’ This is the answer—my only answer—to my children's questions. And even if the future scares me sometimes, I know that’s the only answer I can give them. The only one I *want* to give them.

You know, it would be nice if people could read this book. Making people aware of the problems of families like ours can be of so much help.

BECAUSE IT SHOWS THAT WE’RE NOT ALONE.

ATAR

Lombardy, Italy

I don't think about the future. Actually, it scares me so I avoid thinking about it.

MORETHANANYTHINGI HAVEHOPES,THOSEIHAVE.

Hopes that medical science will make advances and that safer drugs will be discovered; hopes that public assistance will make things easier for those like us who are struggling with rare diseases. Just because they're rare doesn't mean people can simply forget they exist.

Along with the Italian and international associations I fight a lot to help families who have difficulties, especially economic ones. The drugs are really very expensive and if public health doesn't step in and help, making it through to the end of the month is hard for many.

So I move forward with small steps, but I hope that research, on the other hand, will make great strides. I hope I'm ready when the future comes.

LUC

Amersfoort, The Netherlands

Now that I'm a little older I sometimes wonder what my future will be like. Things like ‘Can I have a family? What are the risks?’ or ‘What will I do when I grow up?’ I'm not afraid, though, because I don't think I won’t be able to do what I want.

I love programming and I want to become a programmer for a big tech company or start my own. My father has an important job as a computer scientist for a large Dutch financial group, and that could help me.

When I think about the future I sometimes imagine that things might not always go how I want, that I won’t get the job I hope for. But at the same time I try to stay confident.

I remember a quote, it says, “Always look at the bright side. Even when it’s dark, there’s always the moon.” So, I keep looking at darkness but I see the moon in my future - there’s always a positive side, even when you think there isn’t.

EVA

Schoten, Belgium

If I look back, I don't see the difficulties that have tried their best to bring me down, I see what I've managed to achieve. My studies, my experience abroad alone, my wonderful family. I also have a car that's modified especially for me so I don't have to strain my wrists. I have a wonderful job and a wonderful boyfriend.

I can already imagine him at the altar, waiting for me in a few months, and I'll be a cloud of white. We're not going to have children—it wouldn't be possible for me—but we have his sister's children, fantastic nieces and nephews, and it will be great to be able to simply spoil them.

The future? I see it by looking at the days that have passed, and it doesn't scare me.

WENDY

Tipperary, Republic of Ireland

We’re constantly working to build the future. Each small step we take brings us closer to a greater awareness of rare autoinflammatory diseases.

When I joined the European Arthritis Children's Network (ENCA) five years ago, there were almost no relations with other European associations, and children's voices were hard to hear.

In the last two years the scenario has changed, there’s more attention to rare diseases. What’s needed is a greater involvement of the media, but there are organisations, such as the one in York, that are working hard to involve them.

Associations like the one I belong to offer the possibility to share information, experiences, and spread the word about best practices. It can be really useful for specialists who often don’t know what’s happening beyond their own reality. Greater knowledge can solve many problems for their patients.

When I founded iCan with a group of parents, I had no idea if there were organisations like ours around the world. I found so many of them that I had to choose which of them to follow. We're all volunteers, and you don't always have the time to take care of everything. Every year I attend a conference on rare diseases, and it's a big event. It gives me the opportunity to talk about what’s happening in Ireland and how we can work together to make things better.

Most of the work consists of trying to get better services in different countries and raising public awareness. Activities that are very, very time-consuming.

Almost all the families we've assisted have learned to face life without making too many plans. They live every single day to the fullest, but not because they have no hope, but because they have understood that no one can tell them what the future will hold. They face each challenge as it comes, and after overcoming it what they have left is the satisfaction and the awareness of having once again made it.

When we organise events in which families participate, the future is there on stage speaking to us. It might have the face of a twenty-year-old boy who talks about his experience, his studies, his life, the sport he follows. And that's what keeps everyone going, what makes them say, 'If he's made it, so can my child.'

We build the future every day.

NORMA

Dublin, Republic of Ireland

It’s not easy to deal with children who are constantly struggling with all the consequences of a rare autoinflammatory disease; I’ve been doing it for almost twenty years. The pain can be really devastating, and the children are so little! What gives me strength and inspiration is seeing them, every day, despite their problems, get up and go to school, go out to do physical activities, engage in their hobbies such as music, dance and sports, and realise that they often get even better results than their peers who have no health problems. It's not uncommon for these children to get the Best Student of the Year awards, and they give me hope that they can achieve great results in the future, becoming leaders in whatever field they’d like to pursue. Life may have robbed them of their health, but they have no intention of surrendering.

THEY FIGHT AND FIGHT,

and every day they fight back from this disease is a day that writes their future.

Many of these children become involved in a variety of co-curricular activities such as becoming ambassadors against bullying in the classroom; they’re extraordinary examples of young children who can thrive, flourish and reach their potential. Maybe it's because of something they have developed precisely because of the disease that

pushes them to get up and go forward and take on the challenges of life.

The future? It's them. They're going to make it, and we're going to be right there by their side.

MICHAL

Haifa, Israel

In the future I hope to see more and more people turn to us at the **Israeli Association for RMDs Patients "Mifrakim Tz'eirim".** I wish that everyone who suffers from rare diseases, whether they're autoimmune or inflammatory, can find us because I know we can offer them our experience. It's possible to register with us on our website, and it's completely free. We can make the difference between a life of unknowns and a life full of awareness. Our task is to **help, inform, and support** patients and their families, and our support groups are ready to stand side by side with them through all the difficult times they have to face. The path may be difficult, but we always reach out a hand to help.

Never alone anymore.

This book was made possible by healthcare professionals and patients who commit to studying and sharing knowledge about rare auto inflammatory diseases.

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Norma | ***Dublin, Republic of Ireland***

Michal | *Haifa, Israel*

Glossary

This section is for information purposes only and does not represent medical documentation.

Ankylosing spondylitis

is a type of arthritis in which there is a long-term inflammation of the joints of the spine.

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Behçet's syndrome

is a rare inflammatory condition that causes a number of symptoms, including: mouth sores, eye inflammation, skin rashes and lesions, and genital sores.

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CAMHS - Child and adolescent Mental Health Services

is the name for NHS-provided services in the United Kingdom for children, generally until school-leaving age, who are having difficulties with their emotional or behavioural well-being.

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CAPS - Cryopirin-Associated Periodic Syndrome

also called cryopyrin-associated autoinflammatory syndrome, consists of three types (Neonatal Onset Multisystem Inflammatory Disease - NOMID, Muckle-Wells syndrome and Familial Cold Autoinflammatory Syndrome - FCAS), related to a defect in the same gene. The differences in these diseases types are in their severity and the organs involved.

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CPR - Cardiopulmonary resuscitation

is an emergency procedure that combines chest compressions often with artificial ventilation in an effort to manually preserve intact brain function until further measures are taken to restore spontaneous blood circulation and breathing in a person who is in cardiac arrest.

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CRMO - Chronic Recurrent Multifocal Osteomyelitis

is an inflammatory bone disease occurring primarily in children and adolescents. The consistent feature of CRMO is the insidious onset of pain with swelling and tenderness localised over the affected bones.

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ENCA - European Network of Children with Arthritis

is organised as an informal network for national associations working with children and young people with paediatric rheumatic diseases and their families.

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FCAS - Familial Cold Autoinflammatory Syndrome

is a condition that causes episodes of fever, skin rash, and joint pain after exposure to cold temperatures. These episodes usually begin in infancy and can occur throughout life.

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Fibromyalgia

is a medical condition characterised by chronic widespread pain and a heightened pain response to pressure. Other symptoms include tiredness to a degree that normal activities are affected, sleep

problems and troubles with memory.

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FMF - Familial Mediterranean Fever

is a periodic fever syndrome characterised by recurrent attacks of fever with accompanying pain. FMF flares can last from a few hours to 3-4 days and can involve the following symptoms: fever, abdominal and/or chest pain, inflammation of a joint, knee, ankle, wrist, painful and severe skin redness in nearly 50% of patients, often on the feet and/or lower legs; spleen enlargement, swelling of the ends of fingers and/or toes.

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FPIES - Food Protein-Induced Enterocolitis Syndrome

is an uncommon and potentially severe form of non-IgE-mediated food allergy, commonly characterized by profuse vomiting and diarrhea. Poor growth may occur with continual ingestion. Upon removing the problem food(s), all FPIES symptoms subside. The most common FPIES triggers are cow's milk (dairy) and soy. However, any food can cause an FPIES reaction, even those not commonly considered allergens, such as rice, oat and barley.

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Histamine

is an organic nitrogenous compound involved in local immune responses, as well as regulating physiological function in the gut and acting as a neurotransmitter for the brain, spinal cord, and uterus. Histamine is involved in the inflammatory response and has a central role as a mediator of itch.

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iCan - Irish Children's Arthritis Network

it is a volunteer charity, providing support & information and advocating for best care for those affected by Juvenile Idiopathic Arthritis.

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Immunoglobulins

an antibody (Ab), also known as an immunoglobulin (Ig). It's a large, Y-shaped protein produced mainly by plasma cells that is used by the immune system to neutralize pathogens such as pathogenic bacteria and viruses.

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Immunosuppressants

drugs that inhibit or prevent activity of the immune system.

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JIA - Juvenile Idiopathic Arthritis

also known as juvenile rheumatoid arthritis (JRA), is the most common form of arthritis in children and adolescents. JIA is an autoimmune, noninfective, inflammatory joint disease of more than 6 weeks duration in children less than 16 years of age.

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Macrophage-activation syndrome

is a severe, potentially life-threatening, complication of several chronic rheumatic diseases of childhood. It occurs most commonly with systemic-onset juvenile idiopathic arthritis (SJIA).

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Mifrakim Tz'eirim

Israeli Association for RMD's (Rhythmic Movement Disorder) Patients.

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MRSA - Methicillin-Resistant Staphylococcus Aureus

refers to a group of gram-positive bacteria that are genetically distinct from other strains of Staphylococcus aureus. MRSA is responsible for several difficult-to-treat infections.

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Nissen-Rossetti fundoplication

is a surgical procedure to treat gastroesophageal reflux disease (GERD) and hiatal hernia. In GERD, it is usually performed when medical therapy has failed; but, with a Type II (paraesophageal) hiatus hernia, it is the first-line procedure

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Percutaneous endoscopic gastrostomy – PEG

is an endoscopic medical procedure in which a tube (PEG tube) is passed into a patient's stomach through the abdominal wall, most commonly to provide a means of feeding when oral intake is not adequate.

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Pericardial effusion

is an abnormal accumulation of fluid in the pericardial cavity. Because of the limited amount of space in the pericardial cavity, fluid accumulation leads to an increased intrapericardial pressure which can negatively affect heart function.

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PFAPA - Periodic Fever, Aphtous stomatitis, Pharyngitis and cervical Adenopathies

is a medical condition, typically starting in young children, in which high fever occurs periodically at intervals of about 3–5 weeks, frequently accompanied by aphthous-like ulcers, pharyngitis and/or cervical adenitis (cervical lymphadenopathy).

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Pleural effusion

excess fluid that accumulates in the pleural cavity, the fluid-filled space that surrounds the lungs. This excess fluid can impair breathing by limiting the expansion of the lungs.

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Psoriatic arthritis

is a long-term inflammatory arthritis that occurs in people affected by the autoimmune disease psoriasis. The classic feature of psoriatic arthritis is swelling of entire fingers and toes with a sausage-like appearance.

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Rheumatoid arthritis

is a long-term autoimmune disorder that primarily affects joints. It typically results in warm, swollen, and painful joints.

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SJIA – Systemic Juvenile Idiopathic Arthritis

is a rare inflammatory disorder and the rarest form of juvenile idiopathic arthritis that affects children. The most common symptoms are recurrent attacks of fever with high daily temperature spikes, skin rash, and painful, stiff joints.

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Sjögren' syndrome (SjS, SS)

is a disorder of the immune system that affects the body's moisture-producing glands, resulting in decreased tears and saliva.

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