

PARENTS STORIES

Experiences of Diagnoses of
Children through Parents' Eyes

Contact a Family (Northern Ireland) and the Family Information Group acknowledge the financial support from the Childhood Fund and Chuckles Playgroup for this publication.

ACKNOWLEDGEMENTS

This publication would not have been possible without the willingness of the parents to share their stories. By honestly and bravely opening up, what was for them, a very private period in their lives, they are asking us to listen to their very individual and unique experiences of diagnosis and living with a rare disorder and/or life limiting condition.

These stories are accounts, as they remember them, of times that are vivid in their memories. They are told, so that we all may learn lessons. The stories are very different, but the themes and issues are very similar. It is time now to go beyond listening, beyond the words and to act.

Families have a right to be given a diagnosis, in as supportive a way as possible, and that support to be ongoing, long after the time of diagnosis, in a way appropriate to the family. If this happens then perhaps we really have listened at last.

Contact a Family (Northern Ireland) and the Family Information Group are delighted to be associated with this project. Both organisations acknowledge their gratitude to all the parents and families involved. Also, sincere thanks are due to the Childhood Fund and the fundraising events of Chuckles Playgroup, whose combined financial assistance has put these stories into print.

'The 3D Campaign', in May 2001, was the Third Rare Disorder Awareness Week hosted by Contact a Family (NI). The aim of the week was to bring rare disorders into sharper focus, concentrating on: Doctors and Diagnosis, Daily Living and Development Treatments. In NI, a parent suggested putting a booklet together of parents' stories and sending it to the Department of Health. Thanks to the parent who suggested this.

Hopefully you, the reader, will really listen to these stories. If in any way they touch you, perhaps you will be moved to act and help change things for other parents. This is why the stories are being told. Thank you for listening.

This publication is available in large print on request from Contact a Family (NI). Information is also available on rare conditions and syndromes from Contact a Family (NI), Tel: (028) 9262 7552.

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Childhood Fund

Childhood Fund

Distributes EU funds for the Special Support Programme for Peace and Reconciliation, whose general objectives are to promote social inclusion. Specifically, the Childhood Fund targets children under twelve and their parents, who are experiencing social exclusion by reason of deprivation, disadvantage or disability. The Childhood Fund has supported the Family Information Group since its inception and co-sponsors this publication. We are very grateful for their support.



Chuckles Playgroup

Fundraising from the staff, parents and children at Chuckles Playgroup has ensured that this publication has seen the light of day. As an inclusive playgroup, which strives to enhance the whole child's development within an environment of encouragement, it is very appropriate and very welcome that everyone involved there has chosen to support this project.



Contact a Family

(Northern Ireland)

Helps families who care for children with any disability or special need. Provides information, puts families in touch with one another and with support networks, assists parents to form support groups, provides a voice to raise awareness and campaign for families and collects and provides information on rare conditions and disorders.

Address

Bridge Community Centre, 8 Railway Street, LISBURN BT28 1XP

Tel: (028) 9262 7552



Family Information Group

Presents the 'family view of disability', as experienced by families of children with physical and/or sensory disabilities. Campaigns on issues affecting families, advocating that those who use services must be involved in the planning and provision of them.

Address

c/o Holywell Trust, 10-12 Bishop Street, DERRY BT48 6PW

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Roma

Diagnosis of Morquio, a Muccopolysaccharide Disease

Roma was born on a lovely crisp Autumnal October morning. Through the windows, on my way to the operating theatre for the Caesarian Section, I could see the golden leaves flutter gently against the blue cloudless sky. It was a grand day to make an entrance into the world, and with the help of an epidural I was awake and able to welcome our first baby.

She was perfect, all 6lb 2oz of her. To us she was beautiful and still is. There was no hint or notion that anything was wrong. For the first year she completed all the necessary milestones on time, ate and slept well with frequent growth spurts. It was just as she approached her second year that her growth began to tail off. I can see all of this clearly in hindsight now, how the jigsaw of the diagnosis was beginning to be pieced together in front of our eyes, gradually and reluctantly. There were subtle clues along the way.

I can remember one evening when she was in the bath and her hair was wet thinking that her head was much bigger in relation to her little bony shoulders. However, I quickly dismissed those thoughts, telling myself that first-time mothers are supposed to be overanxious. As time passed, the clues were becoming less subtle. Her height was not rising on our height chart and the clothes she had worn from the previous year still fitted her. I poured over the growth charts with the health visitor, but was always glad when she told me that Roma's height, although small, was still 'normal' according to her centile charts.

Quite often my sister would prompt me, hinting that it would do no harm to see the GP about Roma's lack of growth. Although I was becoming increasingly aware that there was something wrong, I tried to ignore both my sister and my nagging inner conscience. I could see that she was not growing, she still could not reach the table and her little shape appeared somehow different from other children. Eventually I plucked up the courage to take her to see the GP, who assured me on several visits that she was fine, she had just inherited our shorter than average height. But as time passed, the height chart was beginning to dominate our lives.

After the third visit to a GP we were told to come back again in a few months if there was no growth spurt. When the time arrived for this visit, I knew instinctively that there was a problem. On the day of the appointment I was too frightened to go, too afraid of what I would hear. I made an excuse to stay at home with our new baby and asked my husband to take Roma to the GP. I wrote down in the back of Roma's health visiting record all the things that I was

concerned about. It felt like writing a confession, and also in some strange way a betrayal of Roma. I listed everything, from the lack of height and her bony joints, and the shape of her wrists and shoulders. I don't think I could have actually put all of this into words, I was so terrified. I told my husband to give the list to the GP and that is what he did.

After this visit we were referred to a paediatrician in the Royal Victoria Hospital in September. By then Roma was three-and-a-half years old and enjoying her first days at Nursery school. At that visit we were asked many questions, and Roma had blood and X Rays taken. As the doctor checked Roma's joints and spine I noticed that he made very little comments. The silence was ominous. By this stage I knew that the news would not be good.

Several days later the paediatrician rang me at home and asked for us all to come and see him. I implored him to tell me what was wrong. He gave me the name of this very rare condition, and when he told me that only 'bigger centres' could deal with it, like Great Ormond Street Hospital, I knew that it was very serious.

It's hard to put into words what happened from then on. It was just like a death, I can remember vividly what I was doing that day before the phone rang, what I was wearing, and the song which played on the radio in the background.

After several visits to various doctors and a geneticist, the only written piece of information we were given was a photocopy of a page, which was taken from an old medical textbook. This page

told us that Roma was suffering from a rare genetic condition, which would affect all systems of the body except for her intellect. We were to expect a progressive deterioration causing disability and a shortened life span. There was also a one-in-four chance that her siblings would be affected.

We desperately needed more information, which neither the GP nor our health visitor could offer. Both grieving and confused, we did not know who to ask or when or what to ask but we were also afraid of what answers we may receive. Around this time my sister read an article in the local press about Contact a Family (NI). She rang the local office and spoke on our behalf. This in turn led to us being put in touch with the MPS society, a support and research group for children and families like ours. It took a lot of support and courage and a few aborted attempts before I was actually able to speak by phone to a person from the MPS society.

The first contact with the MPS society marked a turning point for us. They sent us information booklets although it was several days before we were brave enough to read them. I hid the leaflets carefully away. It was difficult for us to read them, or let anyone see them because the picture of the girl on the front cover was in a wheelchair.

From then on we were catapulted into a strange new world and we had to reinvent ourselves as parents with a disabled child. A great army of health professionals came into our lives and stayed. We were the new kids on the disability block. I would sneak a look at wheelchairs when we were out and would take Roma into the

disabled toilets, this introduction eased both her and I into the world of disability. Our first contact with other families and children with the condition was at the MPS Christmas Party. This was both a frightening but positive experience. We had the opportunity to gain support from other parents, but it was also heartbreaking to see children with the same condition as Roma in their wheelchairs. This was a glimpse into Roma's future.

Throughout all this, and since then, now five years on, we have been as truthful as possible with Roma. As time passes the conversations with her change and I know that her questions will become harder to answer. Shortly after the diagnosis she had a spinal fusion operation. I was alarmed when she told me then that she thought that this would take the disease away. When she was in the bath at four-years old with her younger brother who had warts, she exclaimed, "Oh, aren't they awful, Mummy, I'm glad I have MPS and not warts!"

Today, however, she has a better understanding of her disease and the limitations, which it is starting to impose. In her prayers at night she asks God for a day off MPS, so that she can swing on monkey bars and do catapults or go on skates like her friends. She has not grown much since she was two and a half. She is only 93cm, the average height for that age, and her two younger brothers are both now taller than her. Her birthday parties and the beginning of new school years are always particularly poignant for me, as all her peers and younger cousins tower above her. She is still quite active but often wobbly and uses a buggy if we go into town. Like most eight-year-olds she loves clothes and music but is very aware that her

shape is different from that of her peers. She now knows that others perceive her as different. She asked me recently if people knew to look at her that she has MPS. I dare not think of how she will fare during the teenage years in this often-cruel body conscious world.

As time goes on the signs of the disease are becoming more apparent. It is very hard to watch this slow deterioration but we cannot escape it. Her little joints protrude and her legs are no longer straight. She seems to be very susceptible to all the various 'bugs' that circulate. We try to compensate, by endless talking, listening, reassuring and building confidence. Often this occurs when I put her to bed at night and we have a long 'girl chat' while soothing her sore legs. We try not to be overprotective but at the same time we try to equip her with an emotional suit of armour to cope with what life will throw at her.

Our lives have taken a different path from the one previously envisaged. The diagnosis affects us all in many ways. We will probably have to move into a bungalow at some stage, but that will be at the expense of Roma's social contacts and friends she has made in this area. Here, everyone knows Roma, and treats her with respect, unlike strangers who may often stare at her or refuse to believe that she is really eight-years old. We also have to consider Roma's education, knowing the problems which are faced by physically disabled children in many mainstream schools. Our days of having babies are now gone, as the throw of the genetic dice may be stacked against us, especially as Roma has two healthy younger brothers. I do not think that I could impose this condition and all its suffering on another child.

Everything has to be carefully measured. We all learn as we go along, and always wonder if we are saying and doing the right things. But we will always be forever grateful to people like my sister, Contact a Family (NI), the MPS society, and the Restricted Growth Association. After the initial diagnosis, interest from others often wanes because there are very few people who truly understand what it's like to be in our situation unless they are in it themselves. Most of the time we may feel alone with our thoughts and fears, which we feel unable to share. However, the organisations I have mentioned are always ready to listen and empathise, they know exactly where you are 'coming from.'

Some health professionals can often appear arrogant, indulging in a one-way conversation. They don't like to admit ignorance and don't always give a full picture. This can contribute to increased distress and isolation.

We are very proud of Roma; she is a bright, articulate, very positive little person. We always tell her that we are so lucky to have her. She may be short in stature and strength, but she is big in charm, heart and good humour. We all love her so very much.

Rebecca's Story

Diagnosis of Infantile Refsum Disease

Rebecca was born on 5 May 1999, in the Ulster Hospital in Northern Ireland, weighing 6lb 7oz. Rebecca spent the first nine days of her life in the special care unit where she underwent various tests.

When all the tests came back clear, Rebecca was allowed to go home. She was so small with her matchstick arms and legs. You were afraid to touch her in case she would break.

Rebecca was very slow to thrive and did not like feeding time. It took her at least 2-3 hours for each bottle and she was feeding every four hours.

By the time Rebecca was ten-weeks old, we noticed that she was not taking any interest in people or any of her toys and had not made any attempt to smile. So we took her along for another eye test, only to be told again that there was nothing to be worried about. But as the days and weeks went by, we could see that Rebecca was not thriving as she should have been.

In November 1999 Rebecca, now six-months old, was admitted to the Royal Hospital for Sick Children in Belfast for five days for a range of tests.

The results came back just before Christmas. We were told that our wee Rebecca was blind. I don't think that anything in the world could have prepared us for such a shock. To know that your own child would never know what you look like, what a bird looks like or even what the colour 'blue' is. But this was only the start of Rebecca's problems.

At nine-months old, we were told that Rebecca had 'Zellweger Syndrome'. This would affect her eyesight, hearing and her mentality.

We were told that most children would not live beyond a year old. This was incredibly hard to take in.

In April 2000, Rebecca started to have seizures. About eight a day but sometimes she would have up to twenty. Each one lasted between 1-2 minutes.

So Rebecca was admitted to the Royal Hospital again for more tests. She had to have a skin biopsy done which was then sent to Amsterdam for genetic testing.

The results of these tests took up to two months to get back, but they were better than expected. Rebecca did not have Zellweger Syndrome, but she did have Infantile Refsum Disease.

The two conditions are very similar, only Infantile Refsum is not as severe. The life span, on average, is 23 years.

There is no cure for this illness – only a special diet which excludes dairy products, red meat, lamb or fish. This doesn't have a big effect on the disease.

Rebecca is the only case that we know about in all of Ireland so it is very hard to get information about her illness.

She is a very loveable child, always laughing and smiling and just loves chatting away to herself. She is now two-years old and, as yet, cannot even sit up without support, let alone walk. She has good hearing at the minute, with the help of her hearing aids – although she would much rather chew the aids than wear them!

In a few years time, Rebecca's hearing could possibly disappear, so we are trying to make the most of it at the minute as this is her main way of learning about the world around her.

We would like to think that one day, with God's help, that there would be cures for some of these diseases but, at the moment, all we can do is to take every day as it comes and make life as enjoyable for Rebecca as we can and to think and be positive for her sake.

Eryn

Diagnosis of Incontinentia Pigmenti

My name is Michelle, I'm a nurse and I'm married to Michael. We have three children: Conor seven, Adam six and Eryn three. This is just a brief summary of our experiences during the birth of our youngest child and subsequent diagnosis of Incontinentia Pigmenti.

When we found out I was expecting our third child we were delighted as we had always felt a third child would complete our family. Adam, however, was not so sure and he asked could we not have a cat instead. Conor felt we had enough mess in the house without adding to it with a baby (profound words for a four-year old). By all accounts, like my other pregnancies, it was uneventful and just presented with the normal troubles of tiredness and looking like a womble!

When I was about six months pregnant we heard that Michael's job was taking him from Co. Down to Co. Fermanagh, a huge move to a

place where our nearest family and friends were two-hours drive away. Since we had no one to keep the boys while I was in labour we decided that I should have the baby back home and so we moved back with mum the week before Eryn was due.

Both the boys had been big babies, 9lb 5oz and 9lb 12oz so I convinced the doctor to bring me in a week early so this baby would be less likely to hit the 10lb mark! As it happened I had a very easy birth with Eryn, she was only 7lb 2oz and beautiful and of course after two boys it was a new experience with a little girl. To our great relief the boys were happy that if they couldn't have a cat that a sister was good enough.

The next two days were perfect. Eryn was a model baby, she ate and slept and really suited pink, which was just as well because after two boys the family went into pink overdrive. The only 'problem' Eryn had was a small blister on her leg, which my midwife noticed and we both agreed was probably a reaction to the starch in the sheets. I had been suffering with afterpains and on our second night the midwives gave me a sleeping tablet and took Eryn to the nursery to allow me a night's sleep. I slept really well that night and as Eryn and I were due to go home the next day I woke feeling really good.

The midwives brought Eryn back just after I woke and the first thing I noticed was that she was really red-looking about her face. I thought maybe she was a bit too warm and put it to the back of my mind. However, as the day went on I was becoming increasingly concerned as Eryn didn't waken for her feeds and when I stripped

her for a bath she was very 'floppy' and her body was covered in this strange red, blistering rash.

When I raised my concerns with the nursing staff I was reassured that this was nothing more than baby rash. The doctor checking Eryn over for discharge again dismissed this but he could see I was not happy and said that he would get his senior to check her over.

Whilst waiting on this doctor I was trying to feed Eryn as she hadn't fed well all day and she began to twitch on her left side. Thinking this was strange, I was about to call a nurse when a friend of ours who was a midwife walked in. I said to her, "Isn't this a strange thing for a baby to do?" I hardly had the words out when she shouted to get a doctor. Immediately she ran to get help and I knew there was something very wrong.

The doctor came quickly but it was all over by the time he arrived. My friend explained that she had seen Eryn take a seizure. I was shocked when I heard this but thankful that we were still in the hospital and not at home. It was decided that Eryn should stay in for another day just to observe her.

The next few hours seemed like days as we waited on this senior doctor who never came. I watched Eryn like a hawk but she seemed to settle and I sent Michael, who had come immediately after I made a frantic call to him, away saying that it was probably a one off and she'd be fine now. I tried to settle myself but just had this feeling in my heart that she wasn't going to be all right but I had already been dismissed as an overprotective mother even after the

'seizure' and the medical staff didn't seem very concerned so I sat and watched her every move just in case.

Eryn's behaviour continued to be bizarre with her squealing in her sleep like an injured kitten and her lethargy. I remember being afraid to even go to the toilet as there would be no one there if anything happened. You probably think this was major over-reaction after only a small seizure and the only explanation I have is an overwhelming sense of being on the brink of something terrible.

The events that happened next are in some parts a blur and in others still so vivid three years on that as I write them I still find it very emotional. I watched Eryn in her little plastic cot as she started to squirm and I thought she was finally waking for a feed but as she opened her eyes she began to go blue. I jumped up and pulled back the covers to look at her chest. She wasn't breathing so I called to the lady in the next bed to get help and she started to hit the call system frantically but no one came and so I ran down the corridor. I found a midwife and started to pull her up the hall, all I could say was, "She's blue, she's not breathing."

As Eryn was whisked off I remember thinking that's it, she's dead, my baby girl is dead. I couldn't breathe as the lady next to me tried to console me. I began to take a pain in my chest and I remembered Michael was going to a meeting and I knew I had to get hold of him before he left. I ran to the phone and called my father-in-law, who Michael was to leave the boys with before going out but he hadn't arrived yet. I don't know what I said but he said he would get Michael there whatever it took!

They successfully resuscitated Eryn and she was transferred to Intensive Care in the baby unit. Michael had arrived with my mum and as we watched her in the incubator the senior doctor finally arrived. Over the next few hours Eryn was extremely unstable and continued to stop breathing every time they tried to work with her until they had to sedate her.

The week that followed brought many emotions: relief, anger, guilt, to name but a few. As they tried to establish just what was wrong with her, many tests were done and a dermatology consultant was brought in to discover what was causing the blistering rash. It was also decided to send Eryn for a C.T. scan as the nurses had noticed Eryn's left arm and leg were floppy but as an ultrasound of her brain had been clear we weren't particularly worried. We asked if we could stay in while they did the scan and it was agreed. However, when the scan began, the consultant immediately said to her colleague, "There's something not right here," and we were asked to wait outside.

As we sat in the hall, every possible outcome for the scan was considered by us as we sat sobbing together. Our consultant brought us in immediately and gave us the news that Eryn had suffered a bleed at the base of the right side of her brain. It was something no one could have predicted or prevented and we would not know the extent of the damage until Eryn was older but it was thought that there was an 80% chance of a full recovery.

Michael is very different than me and likes to deal with things as and when, but I need to have every scenario covered and how to deal

with it so it was difficult as we both had different ways of coping. In the midst of all the emotion the rash that was still there became almost forgotten, after all, whatever it was, it couldn't be worse than the bleed that had almost taken our wee angel's life.

In early January Eryn was to go and see a dermatologist about the persistent rash as the tests he had carried out in S.C.B.U. had come back negative and he wanted to carry out a biopsy to reach a diagnosis. This all seemed fairly routine and I decided to go alone as Michael had only started his new job and had already taken so much time off.

This turned out to be a very bad decision as the consultant told me he thought Eryn had a rare genetic disorder called Incontinentia Pigmenti but not to worry he would talk to me about it in two weeks time when he saw me again and he left! I was in complete shock, the word 'genetic', what did it mean? I was just devastated. The nurse looked at me with a very sympathetic look, a look I knew I had used so many times in my own role as a nurse. I felt like just running out but I had to dress Eryn and I looked at her wee body covered in blisters and I wanted to make it better.

I carried Eryn back to the S.C.B.U. and the nurses sat me down as I broke down. I just didn't know what this meant. Was she going to die after surviving a bleed in her brain? And if it was genetic, which one of us had passed it on?

The nurses were very sympathetic and very compassionate but after I had composed myself I just wanted to get out of there. Why was

this happening to her? She wasn't even a month old and she had gone through more than some people do in a lifetime. I felt drained as I rang Michael and told him the news. He asked me all the questions I had asked myself but I had no answers for either of us. He, however, was not content with being left with no information for two weeks and did a dangerous thing.

He searched the internet. What he found was scary.

Incontinentia Pigmenti had a range of symptoms affecting most of the systems of the body and these could be severe or mild. I couldn't believe we had been left for two weeks with no information and I rang the dermatologist at his clinic and demanded some information to which he replied it was only a skin disorder and nothing to worry about.

It was our paediatrician who explained I.P. (Incontinentia Pigmenti) to us and has been so supportive of us during the first three years of Eryn's life. She has always listened to us and taken the view that we know Eryn best and indeed I.P. best because we are living with it.

Over the next months we settled into our normality and basically each day we would learn something new about our daughter and the I.P. We had moved back to Fermanagh and Michael had gone back to work. I found the experiences of the past months hard to shift out of my mind and in the isolation of a new home miles away from any support I relived the whole experience on a daily basis. I found by this stage I was nearly embarrassed to talk to my friends because I seemed to go over the same things again and again and I

thought they would be fed up listening to me. Michael had his own way of dealing with things and talking about it was not one of them.

It was so frustrating not being able to talk to another mother whose child had the same condition so we could share things, the way you do if your child gets measles. There is always another mum who can tell you how their child was affected and any remedies they had heard of or used. It was this isolation and the feeling that you were the only one that I found the most difficult. It was by chance I found the organisation that would be my ticket to discovering I'm not the only one and there are people whose children do not have the same conditions but do share the same experiences.

It was by chance I noticed an article in the paper about Contact a Family (NI) and I rang that day. I spoke to Nuala, the Development Officer who released me from that feeling of being the only one. She also had a child with a rare disorder. When I went home that night I felt I had found a group of people who could really understand what we were going through.

The past three years have been hard and I think the hardest thing is that, unlike measles, the condition doesn't go away and in Eryn's case it actually changes so it's a new challenge with each milestone in Eryn's life. Eryn's condition affects her in a largely 'cosmetic' way, she has no brain damage from the bleed in her brain but a mild weakness down her left side causing her to walk and run 'funny'. She has peg teeth and teeth missing, she has bald patches through her hair, her rash is presently at the pigmented stage and covers a large part of her body and she has only one nipple but because

most of this can be disguised she can function mostly without people knowing she suffers from a disorder.

This is in itself a problem because sometimes I feel she does not fit into any world. Eryn has, at three, noticed herself but just accepts her rash as normality. Eryn was diagnosed quickly and for that I am thankful but I feel disappointed that I had to search for the support I needed and that organisations such as Contact a Family (NI) were not made known to me from early in Eryn's life. I have an excellent paediatrician and health visitor who have always treated us holistically, but parents need to know what's available to them and as the health professionals are our first line of contact they need to be aware of parents' needs and also to learn through listening to parents.

We understand our children's conditions because, far from a text book, we live with them 24 hours a day.

There are not just medical needs in living with a child with a rare disorder, there are practical and emotional needs and I think sometimes our health professionals forget that and as a nurse I'm speaking as someone who has experienced both worlds.

Donovan

Diagnosis of Duchenne Muscular Dystrophy

The disbelief and numbness we felt that day in Belfast will always be remembered. Later, the isolation and loneliness were to be ever present. Fears for the future would often come to the fore. Within a few weeks our lives were turned around. The values, dreams and beliefs that we once felt we held so dear were nailed to the cross and there seemed no hope.

Belfast, in the summer of 1991, sitting in a hospital room and being told that your son definitely has a disease for which there is no cure.

It seemed so strange that in a technological age of advancement that there are still diseases for which there are no cures. However, what mattered to us was that our son had an incurable disease. It was called Muscular Dystrophy, and not just that, he had the most serious form named 'Duchenne'.

In that office, over and over, Teresa, my wife, and I felt we were being bounced off a brick wall. A wheelchair, premature death – these were alien to us. Yet these were the very things that were being said about our son. Quality of life, possibility of moving house, regular physiotherapy – on and on it went, but Teresa and I had both stopped listening. It was too unreal. Somehow, eventually, we got back to Derry that day to a changed kind of life.

Looking back now, it seems a short time between the first suspicion of something being wrong and the final confirmation of the diagnosis in Belfast. At least we were spared weeks and months of running to doctors and hospitals. Yet still, those words ‘Muscular Dystrophy’ send a chill up the spine.

The whole saga began quite undramatically. Donovan’s health visitor was in our home giving him a pre-school check up as he was coming to the end of nursery school and about to go into Primary One after the summer. We pointed out to her that Donovan seemed to be falling a lot. (We thought he might be flat footed). She asked Donovan to simply get up off the floor and she was immediately alerted by the way he performed this task. His performance of this task involved a number of movements which we now know is peculiar to Duchenne sufferers. The health visitor told us that she was concerned and referred us to Donovan’s doctor who we were to see later that week for Donovan’s pre-school injection.

All the signs of Duchenne Muscular Dystrophy were already there, but we were too blind and too ill informed to see them. Donovan

was a late walker, he had difficulty getting up and down stairs, he had a peculiar way of getting up off the ground and he fell for no reason.

Just around the same time, his teacher in nursery class brought to our attention her concerns. She was unhappy about Donovan falling so much and felt that both his larger and smaller motor skills were not developing properly. She also believed that his co-ordination and physical development were not as they ought to be. We were now becoming very concerned. Donovan’s doctor referred him to the consultant paediatrician at our local hospital. We were now worried.

About a fortnight later, we found ourselves in that consultant’s office to hear the results of a blood test carried out on Donovan a few days previously. The consultant had already told us that he suspected pelvic muscle weakness in Donovan, but we were still hoping and praying that there was nothing seriously wrong.

According to the consultant, based on his examination and the blood test, Donovan had Muscular Dystrophy – those ominous words have eaten into our lives so much since then. I didn’t know how to spell the words, never mind know what they meant. Muscular Dystrophy is a muscle wasting condition and the consultant suspected Donovan as having the most serious version named ‘Duchenne’. More tests were to follow to confirm these early tests. There are always tests, and they were to be carried out in Belfast.

So, we had discovered that Donovan had not got ‘broad feet’, but the nightmare was just beginning.

The consultant at the local hospital explained a little about the disease, how it would eventually lead to Donovan being in a wheelchair as his muscles gradually weakened. Even though there was no cure yet, he was optimistic because of current research developments.

When we arrived home, after visiting our parents, our health visitor was waiting. She was always a caring person, but there was nothing that could be said. The pain and isolation had begun. We just waited for news to come for further tests in Belfast.

Life at that time was like a constant wake. Close relatives visiting, tears and more tears. Not wanting to phone or be phoned. Not wanting to talk but wanting to talk at the same time. Life, God and everything was unfair. We resented everything going on as normal outside of our home, particularly the normal lives that our own relatives and friends were living. The isolation grew and we began to feel that no one understood and, at times, no one seemed to care. Sometimes I wanted to read about the illness, but once I came across the mention of death, I shied away.

As the summer began, we were in limbo – waiting on word from Belfast. We spent most of our time on day trips to the beach, trying to get away from reality and to just be together. The future looked bleak for Donovan, our baby girl, Ruth, and us but life had to go on.

About two weeks after our local hospital appointment, we got news to bring Donovan to Belfast for tests, including a muscle biopsy. Reality had entered our lives once again.

There was little information available at that first visit. We had to await the results of the tests. However, the cries of pain that Donovan let out, despite being under an anaesthetic, were chilling, even though he ended up going to a birthday party later that day.

It was an awful ordeal going to Belfast for the tests and meeting the consultant neurologist, but the return visit a fortnight later was much worse.

That return visit to Belfast, just about six weeks after our health visitor's suspicions, was the beginning of a different life for us.

It is a life I would not wish on anyone.

Even with the passing of time, both Teresa and myself feel that the procedure for telling us such news could have been better handled. It would not have changed anything with regard to Donovan and Muscular Dystrophy but, perhaps, it would have lessened the pain for us in those early days. We had to return to Belfast to be told the devastating news even though we were warned that we should bring someone with us. However, we were still half hoping that the news would not be all bad.

Once the consultant neurologist told us that Donovan definitely had Muscular Dystrophy and, not only that but, the most serious form – our heads were spinning and we could not take anything else in.

We believe that this information could have been passed on to either the local paediatrician or the local doctor with a follow up appointment in Belfast at a more appropriate time.

Neither of us knows how we got down the motorway from Belfast, home to Derry that day.

Donovan is a loveable boy and always was, although he can also be a little devil at times. He has large attractive eyes and gets on well with his relatives and friends. Donovan's main interest, and has been from the moment he could hold them, is playing with toy cars, but he also now likes playing with toy bricks and toy people. He creates his own world and we believe these are healthy interests for him.

Ruth, our little girl, is much the opposite. She is more boisterous, more outgoing, cheekier and more demonstrative. They complement each other.

Difficult as it might be, we had to think of Ruth as well as Donovan. In those early days our thoughts were constantly caught up with the notion that Donovan was going to die and we were constantly living in fear of this. The fear seemed to isolate us more from others and the reality of living from day to day appeared impossible at times.

It is difficult to try and explain this isolation. Again, it has something to do with everybody else living life as normal whereas, for us, life had stopped. It is so easy to sympathise with people in similar positions but much harder to empathise. Life was different now for

us, no one could come close and, furthermore, we were not going to let anyone get close.

We often did not want to talk to our relatives and friends. We did not want to be burdened with other peoples' feelings; we had enough to cope with ourselves. For fear of finding out too much, we did not want to contact the local Muscular Dystrophy Group. Contact was made instead, by letter, with the regional office in Belfast but we felt their support left a lot to be desired. A letter of sympathy, general information, bulletins, specialist holiday information, local contacts and magazines were not what we needed.

We needed one-to-one counselling, someone to reassure us, to be with us and tell us that what we were going through was natural in the circumstances. At that time there was nothing like that, we were alone and it really hurt.

We chose, and our priority was to enhance the quality of life, as best we could within our means. Thus, our immediate aim was to purchase a bungalow which had no stairs whatever.

This was a very touchy subject. We were living in a terraced house when the news of Donovan's condition broke. It did not take us long to realise that we would have to move as we really did not want adaptations and anyway, our house would not have lent itself to being extended.

However, if we had stayed, thousands of pounds would have been spent to make it more suitable. We chose to remain in the private

housing market so that we could choose the area we wanted to live in. This, therefore, excluded public housing and specialist housing.

We wanted to have a purpose-built bungalow erected, and had hoped that special grants would be available. There were no special grants, which is a crying shame, and this idea fell through when our own house sale collapsed and the price of the proposed bungalow went up.

There is no substantial financial help available if you move house for reasons such as this and when there is an extra cost involved. So, when we finally did move into a small bungalow there was no help with the additional £10,000 mortgage. Indeed, the only real help with other expenses came from family and friends as well as a government-backed charity, 'The Family Fund', who were very good even in the early days when they sent a very kind and caring worker to see us and advise us.

Our life was, and still is, ordinary. Up until that point our main concern was to find enough money at the end of the month to pay our bills. Now life had changed and so had our priorities. We had never been involved in the whole area of disability so issues such as access, special provision, medical provision, research and fund raising were all new to us. I believe this is part of the problem – your attitudes and mine. As those who have special needs are very much in the minority, those who are considered 'able bodied' are often blind to their needs. At this stage our son appeared 'normal'. However, as his parents we knew that with the passing of time his condition would deteriorate and life would become more restricted.

'Parents' choice' was emphasised by most of the professionals involved with Donovan and he was too young to fight for himself – so Teresa and I took up the challenge on his behalf.

The first main area of concern was to obtain the benefits that the professionals said that Donovan was entitled to. Of them all, Mobility Allowance was the one we believed that he would be most eligible for because of his walking restrictions. Yet, even though we went as far as an Appeals Tribunal and were represented, we lost. It was only under the new Disability Living Allowance that Donovan received the lower mobility component. By that stage Donovan had been subjected to a number of medical examinations and we were left wondering what the point of it all was.

It is unbelievable the diversity of areas that this illness has brought us into: health, housing, education, benefits, welfare... and each of them bring their own problems. At times it can be so overwhelming trying to deal with them.

This is an area where I would argue that a key social services employee would be of great benefit, to help families through the maze of bureaucracy.

At times it seemed that the various bodies who were supposed to be helping us were actually working against us. This situation appeared very unfair – as if we hadn't enough to cope with.

This pursuit for a better quality of life covers many areas, as I have said. We saw to the housing situation and took on the battle for

assistance at school for Donovan and then we turned our attention to the local Health Board.

This is an on-going affair with letters to and fro concerning the range of services locally, and the burden of having to travel to Belfast for specialist services.

We, and others, have been asking for a more comprehensive and integrated provision of services with the establishment of a multi-functional unit which would provide a wide range of services.

Time will tell if our voices will be listened to.

There is little left to be written. Life goes on. We still pester the various bodies to fulfil their various functions. We still try and be the parents that Donovan and Ruth need, despite our failings.

It has to be said that some of the professionals involved with Donovan are excellent, despite the constraints under which they work. However, they are professionals and when they leave we are still left in our own situation and it is still difficult.

The fact may be that Donovan has Muscular Dystrophy, but I can assure you that Muscular Dystrophy has not got Donovan.

Aisling

Diagnosis of Cutis Laxa Type II

My daughter Aisling was born by Caesarian Section at 37½ weeks. She weighed 5lb 10oz and my husband and I thought she was the most beautiful baby we had ever seen. She was very blue and had to be put into an oxygen tent but she recovered quickly and all was well.

I remember one midwife who used to be very particular about anybody else other than my husband or I handling her because in her opinion Aisling was a very delicate baby.

Aisling never slept well, she was feeding every three hours and when she did feed she would bring it back up again. At the time I was very worried but I put it to the back of my mind because I thought it was just a case of getting used to feeding her and being a mother.

At her check-up at the baby clinic Aisling was found to have a 'clickety' hip and was referred to an orthopaedic consultant. It

transpired that she had dislocated hips and was admitted to Altnagelvin Hospital to be put in traction. She was, at five weeks old, the youngest baby on the ward. Her legs were strapped up to a bar above her and the only part of her on the bed was her head and shoulders. We were very distressed to see Aisling like this but our spirits were high because we felt that this would help her.

Whilst in hospital, Aisling's condition deteriorated. She was very unsettled and would not feed. I was very anxious about this and voiced my concerns, but I was told not to worry, she was just 'possetting' and that because of the angle she was lying at it would be difficult for her to feed. This reassured me somewhat, but Aisling's condition got worse to the extent that she started to have a fit and a rash quickly appeared all over her body. All at once there was pandemonium. She was surrounded by medical staff and whisked off to the Intensive Care Special Baby Unit. My husband stayed with her while she got a lumbar puncture done. Aisling had got septicaemia. We found out later that Aisling had 'kidney reflux', so she probably had constant kidney infections from birth.

Eventually we got her home. She was to take antibiotics daily to prevent kidney infections. She continued to take antibiotics over the next seven years and although she had two 'sting' operations to try and rectify the problem she continued to get urinary tract infections.

When Aisling was three-months old she was put into boots and a bar to correct her dislocated hips. This is a period of our lives I shall never forget. My husband and I took it in turns to look after her. She cried almost non-stop for three days until she got used to it. She

actually lost her voice she cried so hard. It was heart breaking to watch but we knew that if we were to help her we could not take them off.

It was slowly dawning on us that all was not well. We were running to and from hospital attending orthopaedic, renal and paediatric clinics.

We were referred to the Genetic Clinic, which turned out to be a very traumatic experience. We were asked all sorts of questions about family medical histories and our own medical histories. Aisling was examined and had blood taken. She was then photographed from lots of different angles. I found this all very difficult but I was devastated by the genetic consultant's cold analysis of Aisling's features. I was told: "...her eyes were too far apart, her neck too short, there was a small bump at the top of her back, her nose was too flat, the muscle tone of her hands very poor, her skin beneath her jawbone sagged and her abdomen was protruding." The geneticist said she would look into what all these things would point to but at the time I was so hurt by her analysis of Aisling that I refused to go back to the Genetic Clinic although I changed my mind some years later.

Aisling got out of her boots and bar and by two-and-half-years old she was walking. She was very clumsy and tended to fall quite a bit. When she started nursery, the teacher pointed out that she had difficulty with her fine co-ordination skills. In Primary School she was put forward for assessment by an educational psychologist and was found to have moderate learning difficulties. She was not able to

cope at mainstream school and we made the difficult decision of sending her to a 'Special School'. Aisling was, and still is, very happy at her school. She is very confident and self-assured when socialising within a safe and loving environment, but I feel that the education system has let her and children like her down because they are isolated from other so called 'normal' children who have no learning difficulties. I think it would be much better to try to educate our children who have special needs within a mainstream system, whilst at the same time accommodating their special needs.

My family very gently advised that perhaps we should return to the Genetic Clinic and although I was fearful about returning I felt I didn't want any more 'surprises' and would rather know what to expect with regard to the future. The geneticist welcomed us back and I found that we were treated more gently second time around. After some more tests and photographs the genetic consultant said that she thought Aisling had a rare disorder known as Cutis Laxa Type II. We were told that we were very lucky that Aisling had a mild form of it, but we didn't feel at all lucky at the time. The genetic consultant gave us a few photocopied pages which I assume came out of a medical book, outlining the medical history of three children who had Cutis Laxa, but there was no long-term diagnosis because we were told that they didn't have enough information on the condition to provide us with a long-term diagnosis.

All along we have been struggling to try to get Aisling the help we need but unfortunately there is no support group and as far as we are aware there is nobody else in Ireland who has been diagnosed as having this condition. As Aisling gets older she is becoming more

aware of her limitations and is starting to ask questions as to why she is like this and why she cannot do things that her brother and sister can do. Last year we rang the Genetic Clinic because we had no one else to talk to and told them how isolated we felt and could they do anything for us. Aisling's genetic consultant very kindly told us about an organisation called Contact a Family (NI) and they have proved to be a friend indeed. To this day I shall be forever grateful to Nuala who listened to me crying down the phone and who understood implicitly how I felt because she herself had a child with a rare disorder. They helped put us in touch with a family in France who have a young girl with Cutis Laxa and we have become firm friends helping each other through our tears as well as laughter.

Later on we were told that Aisling would have been entitled to benefits and help with getting around but no one ever told us. Someone said that we didn't move in the 'correct circles' to find out about what our child is entitled to. I find it very upsetting that my daughter for years lost out on benefits solely because nobody thought to tell us, benefits that she was entitled to and which would have helped us greatly financially. I had to take a career break at one point to look after Aisling because I could not cope with going to work and getting her settled into her Special School. During this time I would have been entitled to attendance allowance but because I was unaware of this I never applied for it.

I think the biggest problem all along was that Aisling's problems were all treated by different people and there was no gathering together of information to look at the whole picture. She had an orthopaedic consultant for her hips, a renal consultant for her

kidneys and a paediatric consultant for her overall health. She had an occupational therapist and a physiotherapist, a community nurse (recently for toileting problems) and a consultant dealing in learning difficulties and an educational psychologist – I could add more to this list but I fear I would bore you!! Never at any time did these people exchange information or were even aware that she was attending other consultants, even when I informed them that she was attending other medical professionals there was no interest in exchanging information in order to help her. With such a fragmented approach it is really no surprise that she did not get diagnosed until she was almost nine-years old.

Throughout all of this I have to say that Aisling's GP was very supportive of us and did everything he could to help Aisling with the result that she has built up a warm relationship with him. Nevertheless, I do feel that other medical professionals were not interested in the whole picture, just in their little bit of the jigsaw. It was not until after Aisling was diagnosed that I realised through my own investigations that she should have had a social worker to oversee her case. As it was, we were left isolated at a time when we needed help most.

In ending, I have to say that our daughter is a very happy and outgoing child. She gives us so much joy and we love her with all our hearts. Often when the parents of a child with a rare disorder are given the diagnosis they find they have no one to turn to to get the help that is needed. This can be very frustrating and soul destroying.

I have great faith in the knowledge that my family and I will help Aisling in every way we can – after all, it is just what she deserves.

