Rare Ireland
Family Support Network

What comes after a rare diagnosis?

Picture: Evie-Rose, Co.Cavan (story on page 6)

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Waiting for a diagnosis

Waiting for a diagnosis for your child can be an isolating time for a family. You may have noticed your child is not reaching their developmental milestones or maybe a medical professional has raised concerns about your child's development.

It can be difficult to know where to turn next, what tests are available or what interventions will help you bring your child to their potential. Without a diagnosis this information is not always available through our health system and getting an adequate diagnosis can take some time. We can offer the support you need and advice based on our experiences at this stage of our journeys.

"Our journey with Caleb has being so hard. We saw he had problems last June when he was 9 months old. We took him to his development check and he didn't pass anything. He was perfect up until this point. We took him to a Neonatologist and got an MRI done. We were assured he was fine and his MRI clear. He still wasn't meeting milestones and Public health Nurse was very concerned. Fast forward to March this year we eventually get him to Temple Street and we are told his MRI is not clear and he has brain damage, scoliosis and development delay. The Neurologist said he thinks Caleb has a rare genetic disease so we got our bloods done and are getting a full MRI soon. The Facebook group has been a Godsend, being able to talk to other parents in my situation was so good. I really love the group and the advice I get"

Donna, Mum to Caleb who is awaiting Genetic test results

Caleb

Receiving your child's diagnosis

Receiving a diagnosis of a rare condition for your child can be an uncertain time for parents and caregivers. A diagnosis is often delivered by a General Practitioner, Paediatrician, Cardiologist, Neurologist or Medical Specialists with very little information available, this is often because the specialist is hearing the name of this condition for the first time and has no information to offer. A referral is made for the parents to be seen by a Genetic Consultant but unfortunately this involves lengthy waiting times.

We have a private parents forum which bridges the gap between diagnosis and a Genetics appointment where parents can seek advice and information from other parents perspective.

"Connecting with other rare parents is invaluable, especially at the beginning of your journey when you feel completely isolated. Through their experiences they were able to give me hope, they are amazing examples of resilience, empathy and kindness. I no longer feel alone thanks to the connections made along the way"

Lourdes-Mum to Conor

Conor is the only person in Ireland living with xq28Duplication Syndrome

“In a crowded room surrounded by people you can often feel very isolated. No one intends making you feel like this but as you gaze at all the other children play while your heart beats 90 miles per hour while you watch your rare child live in their own world afraid of what they will do or cant do your heart sinks and you can feel so alone.
However there is nothing like that feeling when you can turn to your Rare Ireland family who just get it! From a simple question to a vent there is no judgement just support. Rare is wonderful but rare can be the loneliest place ever but together we can make a difference.”

Catherine- Mum to Tom

Tom is the only person worldwide living with Trisomy 7p

After your child's diagnosis

Even if you have searched for a diagnosis for some time, receiving a diagnosis can be devastating. It can feel very final and like all hope is lost. We, as parents become experts in our children's conditions and the medical professionals learn from us because our child is often the only person with their specific condition known to their medical team.

With very little information available on how this new diagnosis will impact your child's life, the future becomes very uncertain. Rare Ireland links families together, this is invaluable to our families because they can learn and get information from other parents. It also gives parents who are new to their rare journey the opportunity to see how older children and young adults are developing. In many ways linking in with other parents who are in similar positions gives parents of younger children hope for the future.

"All I keep hearing from my son's Doctors is "its a wait and see scenario", but they drag their feet in being an advocate for my child in getting supports and services. Rare Ireland has been the support and encouragement I need as a mother to keep fighting. The only person who will put limitations on my son will be himself"

Carmel- Mum to Senan

Carmel & Senan

Evie-Rose

Evie-Rose was born by emergency section because of her erratic heart rate and no movement at 35 weeks. She weighed 1.6kg at birth and currently weighs 4.8kg at 6 months old. At birth she had no suck reflex, low muscle tone and laryngomalacia. She is delayed physically but currently has no other health complications. She is a fraternal twin, with a brother one minute older than her. she spent 35 days in special care because she couldn't feed and struggled to gain weight. Evie-Rose had Genetic testing at 8 weeks and was diagnosed with Koolen-de Vries Syndrome, which is the loss of genes from the long arm of chromosome 17. She was referred to enable Ireland after diagnosis and receives physio therapy and speech and language for feeding, she is waiting for Occupational Therapy and she is under the supervision of the Paediatric team in our local hospital.

Evie can now take a full bottle but would not be ready for weaning because of weakness in her neck and back but she is gaining weight be it slowly she is coming on.

Her personality would light up any room and thankfully we have had no major set backs in the last few months.

When we found out about Evies diagnosis, I went searching for information but also support. I came across Rare Ireland and when we reached out one of the admins very quickly got back to me, I poured my heart out in the group as a new parent of a beautiful rare baby who was worried, upset and facing something we had no idea about. The response we got was overwhelming, Laura introduced us to other parents who have children of the same diagnosis, we may all be going through different journeys with our family but we are all the looking for the same understanding, support and its really nice to know we are not alone.

Evie-Rose

Rare Ireland Family Support Network

Rare Ireland Family Support Network is a parent led charity organisation, established in 2017 by two mothers of daughters living with rare chromosome abnormalities.

Spotting the gaps in the system, the lack of support and information available to parents and witnessing the isolation associated with rare disease in Ireland we decided to create a network for families to ease this isolation and raise awareness of the challenges rare families face.

- We provide a safe space for parents within our network to engage with other rare parents.
- We put families of children with the same rare condition in contact with each other.
- We send gift boxes to rare children during hospital stays.
- We subsidise therapies for children with rare conditions.
- We subsidise counselling sessions for families post diagnosis.
- We stay linked in with other rare disease organisations and work together in an effort to improve care pathways for people living with rare conditions.
- We keep our social media platforms extremely active, raising awareness and showing the general public the faces of rare disease in Ireland.

> "Being part of Rare has not only provided me with the support from other parents in the group but also given me the opportunity to offer my help and share my experiences to others of our similar journeys on this Rare Path" -Caroline

Eric

Eric Was born 31st August 2001 with a rare illness known as TOF VACTRAL.
This affects his heart, kidneys, oesophagus, stomach and spine.
He has had 19 major operations to date and probably more to follow. He spent most of his life in Our Ladies Hospital for Sick Children, Crumlin and Great Ormond Street, London.
When Eric was born there was no Internet. We had never met another child with the same illness until the world discovered Facebook. Now we are in constant contact with parents from all over the world who's children have the same illness. A lot of their kids are very young, some only starting the TOF journey.
I can share our journey and give advice.
It's great to finally have support and also be able to give support to other parents now as back when Eric was born we had no support from anyone except our medical teams and the hospital.
It's great being a member of Rare Ireland as we can relate to each other and share experiences with other parents of children with rare illness.

"When Eric was born we had no internet. We had never met another child with the same illness until the world discovered Facebook"

Our members

We accept members from all over Ireland with any rare condition. In membership we currently have over 1.3k members, their conditions include but are not limited to:

- Chromosome abnormalities
- Autoimmune Diseases
- Neurological conditions
- Metabolic disorders
- Progressives diseases
- Childhood cancers

We are not a condition specific group, we have no age limit and membership is free of charge.

"I feel understood and validated when talking to other parents with a child with a rare syndrome. We get eachother and don't judge. This is priceless"

Natalie- Mum to Conor

Natalie & Conor

Noah

"Rare Ireland gave us support and reassurance when we needed it the most and we made lifelong friends"

Una-Mum to Noah who is living with 16p11.2 Deletion Syndrome

"Being a member of Rare Ireland means we can connect with other families with rare children and not feel so lonely or judged. Its a group we can go to for support if our days aren't going well or to celebrate our children's huge achievements. Its like being part of a family and every single person is unique to the other"

Ciara-Mum to Jayden who is living with GRIN2B Gene Disorder

Jayden

Alicia

Alicia was born 6th June 2013.When she was 2 and half years old she was diagnosed with Beckwith Wiedemann Syndrome.This is a rare overgrowth condition that can cause Wilms tumours. Alicia was monitored by having ultrasounds every 3 months since she was diagnosed. She then got a diagnosis of Hemihypertrophy. This is a rare condition causing length discrepancy in legs or arms. Alicia has it in her legs, this also has to be monitored. At 3 and half she had a tongue reduction in Temple Street hospital. We've been so lucky to get the care through Tallaght, Crumlin and Temple St Hospitals. We also came across Rare Ireland and my G.P recommended I reached out. The support we get from the other parents is amazing, its lovely having the support network. It's great knowing your not alone.

Alicia

"The support we get from the other parents is amazing"

Alanna & Ella
Rare Ireland was founded for you both and all the other children who joined the rare journey. It was founded to make life easier for you and so you all get the acknowledgement and awareness you deserve.We thank our committee who work tirelessly in the background to keep our rare group growing. We are so proud to have started this network in honour of our two amazing girls.

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