



unique

UNDERSTANDING GENES
& CHROMOSOMES

After diagnosis: what happens next?

rarechromo.org

Unique publishes country specific 'After diagnosis: What happens next?' guides:

- United Kingdom (UK)
- United States of America (U.S.A.)

After diagnosis: what happens next?

Receiving a diagnosis of a rare chromosome or gene disorder for your child can bring a wide range of emotions. You may feel shock, sadness, anger, guilt, fear, uncertainty, isolation or even relief at finally having an explanation for why your child seems "different." Many parents describe the experience as an emotional roller coaster, filled with unexpected twists, turns, highs and lows.

Right now, the future may feel overwhelming and uncertain. But it is important to remember that your child is still the same child they were before the diagnosis. They still need love, care, encouragement and opportunities to thrive, just like any other child. Although there may be challenges ahead, there will also be moments of pride, joy, connection and celebration. Many families discover strengths and resilience they never knew they had.

Every child and every family is unique. Not all families will experience the same challenges or need the same information and support. Some children may require significant medical care or therapies, while others may need only limited support. Whatever your family's journey looks like, it is important to know that you do not have to face it alone. Reaching out for support from healthcare professionals, therapists, educators, family members, support groups and other parents can make a meaningful difference.

Although the journey may not be the one you expected, many families find hope, support and community along the way. Over time, parents often become experts in understanding and advocating for their child's needs, while also celebrating their child's individuality, achievements and unique strengths.

It's now been 12 months since the diagnosis and I've realised we've been through the grieving process since then. Initially angry at all those who'd thought us paranoid, then upset and guilty for all the things [he] had been through alone. It has taken the last 12 months for us to assimilate the diagnosis and what it means for us as a family, but I'm relieved we now have the answer."

"...Getting these results was both a relief and a concern; we had some answers to things that we knew, but now we had so many unanswered questions and unknown things for his future. It was scary and overwhelming"



"It made me feel so confused and powerless. After I started researching and learning more, I felt calmer, but still very scared and alone. It seemed like this was such an uncertain thing. I wish someone had told me it's not that rare, and that a group like Unique existed."

"I was actually thankful to have answers once we learned of her genetic disorder. It provided a road map for what to expect, what additional medical needs we needed to look into, and what we needed to check on regularly. In our case, the diagnosis explained a lot of things and gave a 'name' to what we were dealing with."

"I wish someone had told me not to be so afraid. I felt a kind of grief for something I thought I'd lost but it turns out this didn't take anything away from my son - it actually made him strong and beautiful and unique. He's not who I thought he would be, he's so much more than I could ever have imagined! I love him more than anybody on this earth and I wouldn't change a single thing about him."

A practical checklist: the first six months after diagnosis

Receiving a diagnosis can feel overwhelming. Many families find it helpful to focus on a few practical steps during the first months after diagnosis.

In the first 1–2 months

- Request copies of your child's genetic testing results
- Share the results with doctors and specialists involved in your child's care
- Schedule a visit with your genetic counsellor or genetics clinic
- Find out more about any relevant early childhood or developmental support services
- Ask your doctor about referrals to specialists or therapists if needed

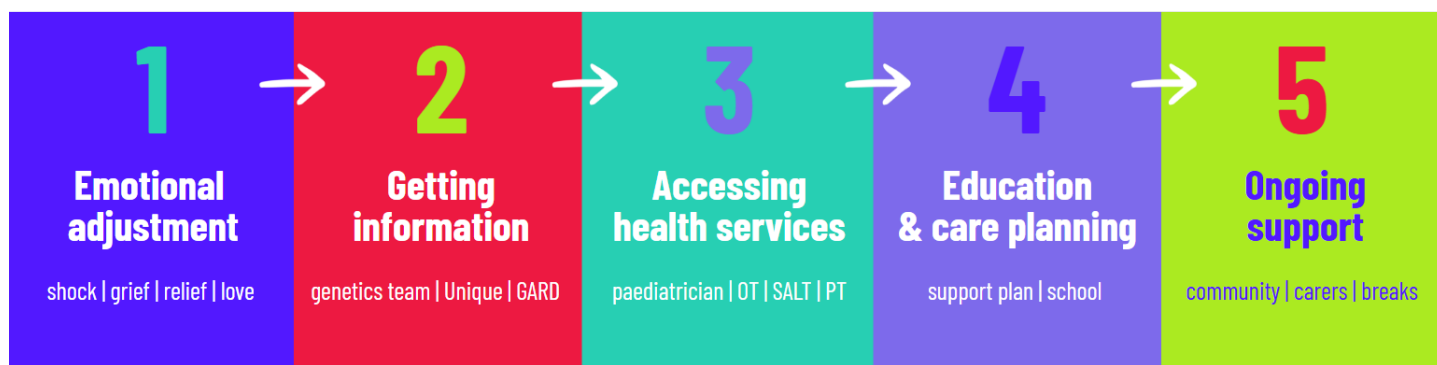


Months 3–4

- Begin recommended therapies such as speech therapy, occupational therapy or physical therapy
- Connect with condition-specific organisations or other support groups
- Learn more about your insurance coverage and benefits if relevant

Months 5–6

- Ask about financial support programs that may help your family
- Continue building connections with your child's care team and support community
- Begin thinking about long-term planning for medical and educational support



These stages often overlap and repeat - you may revisit any of them at any time

My advice to another parent in this stage would be: don't try to learn everything in the first few weeks. Rare diagnoses can feel overwhelming, especially when information is limited. Give yourself permission to process the news, ask questions and take things one step at a time. Keep records, connect with support groups if possible and remember that your child is still the same child they were before the diagnosis. The diagnosis may help explain part of their story, but it does not define who they are or all they will accomplish."

Talking about your child's rare chromosome or gene disorder

Learning that your child has a rare chromosome or gene disorder can affect many areas of family life, including relationships with relatives, friends and other people around you. Parents and carers/caregivers often find themselves having to explain complex medical information while also coping with their own emotions and adjusting to daily challenges. Deciding what to share, when to share it and how much information to give can sometimes feel difficult or overwhelming.



Family members and friends may never have heard of the condition before, and they may struggle to understand the challenges your child and family face every day. It can help to remember that you are not responsible for managing everyone else's feelings or reactions. You can choose how much information you want to share and with whom. Some parents find it easier to give simple explanations at first, while others prefer to share more detailed information, leaflets or trusted websites so relatives and friends can learn more in their own time.

You may also find that people need guidance on how to support you. Sometimes friends and family want to help but are unsure what would actually be useful. Being specific can make this easier. For example, you could ask for help with childcare for siblings, attending appointments, preparing meals or simply having someone to talk to without judgment.

Many families find comfort in connecting with others who have similar experiences. Support groups, charities, online communities and parent networks can provide understanding, practical advice and reassurance from people who truly understand the realities of living with a chromosome or gene disorder. These connections can help reduce feelings of isolation and remind families that they are not alone.

As children grow older, parents may also begin thinking about how to talk to their child about their diagnosis. Honest, age-appropriate conversations can help children develop confidence and self-understanding. Focusing on strengths as well as challenges can help build a positive sense of identity and self-esteem.

Unique publishes a separate guide to **Telling your child or their sibling about a rare genetic diagnosis**

In talking to more people about our daughter's diagnosis I feel like I have more ownership of it, and it has removed a lot of the concern I had about people reacting badly."

Seeing a clinical geneticist

When your child receives a diagnosis, you may be referred to a genetics specialist. Clinical Geneticists specialise in diagnosing and managing rare genetic conditions. They can advise on appropriate medical care, genetic testing and screening for family members. Genetic counsellors and other members of the genetics team may also provide information, emotional support and guidance for families.

Referral pathways and waiting times for clinical genetics vary between countries and, in some cases, between regions within the same country. Contact your healthcare provider to find out how to access clinical genetics services in your area and what waiting time you can expect. How often your child is seen by the genetics team can vary greatly depending on their diagnosis and medical needs. In some situations, a child may only be seen once.

Unique publishes a separate guide to **A clinical genetics appointment**

Early childhood support services

Many countries offer specialised early childhood support services which provide support for children who have developmental delays or medical conditions that may affect development. These services are designed to help young children build important skills during a critical period of brain development, including communication, movement, learning, feeding and social interaction.

Services may include speech therapy, occupational therapy, physical therapy, developmental therapy, feeding support and other specialised services depending on a child's individual needs. Access to these services varies between countries and, in some countries, between regions or local areas. Eligibility may depend on a child's developmental needs, diagnosis, age and local criteria. Referral pathways and waiting times also vary.

If you are concerned about your child's development, it is important to speak with your paediatrician or another healthcare professional. They can discuss your concerns, monitor your child's development and make referrals to appropriate specialists or community services if needed.



Treatments and therapies

Your child may be referred to a specialist clinic or medical centre where professionals with expertise in developmental disabilities, rare genetic conditions and complex medical needs work together as part of a multidisciplinary team. These teams provide assessments, ongoing monitoring, therapies and treatment plans tailored to your child's individual strengths and challenges.

The team might include:

Paediatricians specialise in child development, learning and behaviour. Paediatricians often coordinate care between therapists, schools and medical specialists.

Occupational therapists (OT) help children develop everyday life skills and increase independence. This may include support with feeding, dressing, handwriting, sensory processing, play skills and participation at home and school.

Physiotherapists / Physical therapists (PT) help children improve movement, strength, balance, posture, coordination and mobility. They may assist with walking, muscle weakness, low muscle tone or physical delays.

Speech and language therapists (SALT) help children develop communication, language, social interaction and feeding skills. They may also support children who use alternative forms of communication, such as sign language or communication devices.

Audiologists diagnose and manage hearing and balance disorders. Perform hearing evaluations to determine whether hearing loss is present and recommend appropriate treatments or supports.

Orthotists design and fit supportive devices such as braces, splints, orthopaedic footwear or spinal supports to improve alignment, posture, stability and mobility.

Neurologists specialise in conditions affecting the brain, spinal cord and nerves, including seizures, developmental delays, muscle weakness and movement disorders.

Cardiologists evaluate and treat heart conditions that may occur as part of certain genetic syndromes.

Gastroenterologists (GI doctor) manage digestive system concerns, feeding difficulties, reflux, constipation and nutritional problems.

Social workers help families access community resources, financial assistance programmes, educational services and emotional support.

Psychologists or psychiatrists support children and families with behavioural, emotional, developmental or mental health concerns.

Care coordinators or case managers organise appointments, therapies, referrals and communication between multiple healthcare providers and services.

00 *We saw a geneticist, GI, neurosurgery, neurology, developmental paediatrician, urology, and some others. Some of these were one or two consults and then never really saw them again."*

"I don't have any brilliant advice beyond trying to see everyone as quickly as possible and then weed out the ones you don't need follow up from."

"The hardest part about managing appointments was just finding the perfect time and day, especially when you have other children at home that need your time as well."

"Referrals have happened pretty quickly, but we are still waiting to see some of them due to sheer demand. Juggling appointments is hard with work, so it often feels like we have to work in evenings and weekends a lot more than before."

"Many [appointments] ended up being a couple of monitoring appointments and then we were clear. I'm so grateful for that! But the number of appointments was so overwhelming - it felt like everyone just wanted to poke at our boy and disrupt us. However, it got much better after the first year or so. Now we know who we need to see routinely and it is much more manageable."

"The hardest part of managing appointments has been the sheer number of providers involved in his care. Keeping track of appointments, recommendations, therapies, testing, paperwork, insurance issues, and communication between specialists can feel like a full-time job. My advice to other parents would be to stay organized from the beginning. Keep a notebook, binder, spreadsheet, or digital folder with appointment dates, provider names, reports, and questions you want to ask. Don't be afraid to advocate for your child or request referrals when you feel something needs further evaluation. Most importantly, remember that you do not have to become an expert overnight. Take things one step at a time, ask questions, and trust your instincts as a parent. You know your child best, and your observations are valuable to the medical team."

Care plans and follow-up appointments

After a genetic diagnosis, your child's paediatrician or specialist team will usually work with you to develop a care plan based on your child's specific medical and developmental needs. Follow-up appointments are important for monitoring your child's progress, identifying new concerns and adjusting therapies or treatments as needed. Review appointments may take place every few months, every six months, annually or more frequently depending on your child's diagnosis and health needs.

Many children with genetic conditions continue to see their paediatrician regularly throughout childhood. Some diagnoses require closer monitoring by specialists, while others may only need periodic follow-up visits. As your child grows and develops, their healthcare needs and support services may also change.



Medical insurance

In some countries, health insurance is an important part of accessing medical care as most healthcare services are paid for through private insurance plans, government-funded programmes or a combination of both. Without insurance, the cost of doctor visits, hospital stays, therapies, medications, genetic testing and specialist care can be very expensive. Having health insurance can help families afford these services and reduce out-of-pocket medical costs.

Healthcare coverage and access to services vary between countries. Families may access healthcare through government-funded or publicly-funded healthcare programmes, health insurance provided through employment, private health insurance or by paying directly for services. The options available, eligibility requirements and costs can also vary depending on where you live. Contact your local healthcare provider, health authority or insurance provider to find out how healthcare is funded and accessed in your area.



Health insurance plans can also differ greatly in what they cover. Some plans may cover therapies, specialist visits, medical equipment or genetic testing more fully than others. It is often helpful to review plan details carefully and ask questions before choosing a plan if possible. Understanding some common insurance terms may help families navigate the system more confidently.

- **Copay** - A fixed amount you pay for a medical visit, prescription or service
- **Deductible** - The amount you must pay each year before your insurance company begins covering certain healthcare costs
- **Coinsurance** - The percentage of costs you may still be responsible for after meeting your deductible
- **Out-of-Pocket Maximum** - The maximum amount you will usually have to pay for covered healthcare services during a plan year. After this limit is reached, insurance typically pays the remaining covered costs
- **Prior Authorisation** - Approval that your insurance company may require before covering certain medications, tests, therapies or procedures

Education

Children learn through everyday interactions with other people and by exploring the world around them. However, some children may experience greater difficulties than their peers in areas such as communication, understanding and learning, sensory and physical development, and behaviour or relating to other people.

When a child has ongoing difficulties that affect learning or development, they are described as having **special education needs (SEN)**. SEN can affect a child's ability to learn in different ways, including:

- Behaviour or ability to socialise
- Reading and writing skills
- Ability to communicate
- Ability to understand
- Concentration levels
- Physical ability



Schooling options

Many children with a chromosome or gene disorder are able to attend their local school, sometimes with little or no additional support. Others may attend their local school but receive additional services and classroom support to help them access the curriculum and participate fully in school life. This support might include help from a teaching assistant, accommodations in the classroom and therapy services.

Some children who need more intensive support may benefit from a specialised school setting that is designed to meet complex educational, developmental, medical or physical needs. These schools often provide smaller class sizes, a higher staff-to-student ratio and access to therapists and specialists on-site.



For some children, home education (homeschooling) may be a suitable alternative to attending school. Homeschooling allows learning to be tailored to a child's individual pace, interests and abilities. For children with rare genetic conditions, this flexibility can sometimes make it easier to balance education with medical appointments, therapies and periods of illness or fatigue.

Home education requirements vary between countries and, in some countries, between regions or local authorities. Families considering home education should check the rules that apply in their area, including whether they need to register or notify an education authority, any expectations around the curriculum or learning provided, and whether they are required to submit reports, provide evidence of progress or undergo assessments. Contact your local education authority or relevant government department for up-to-date information about the requirements in your area.

Formal education support plans

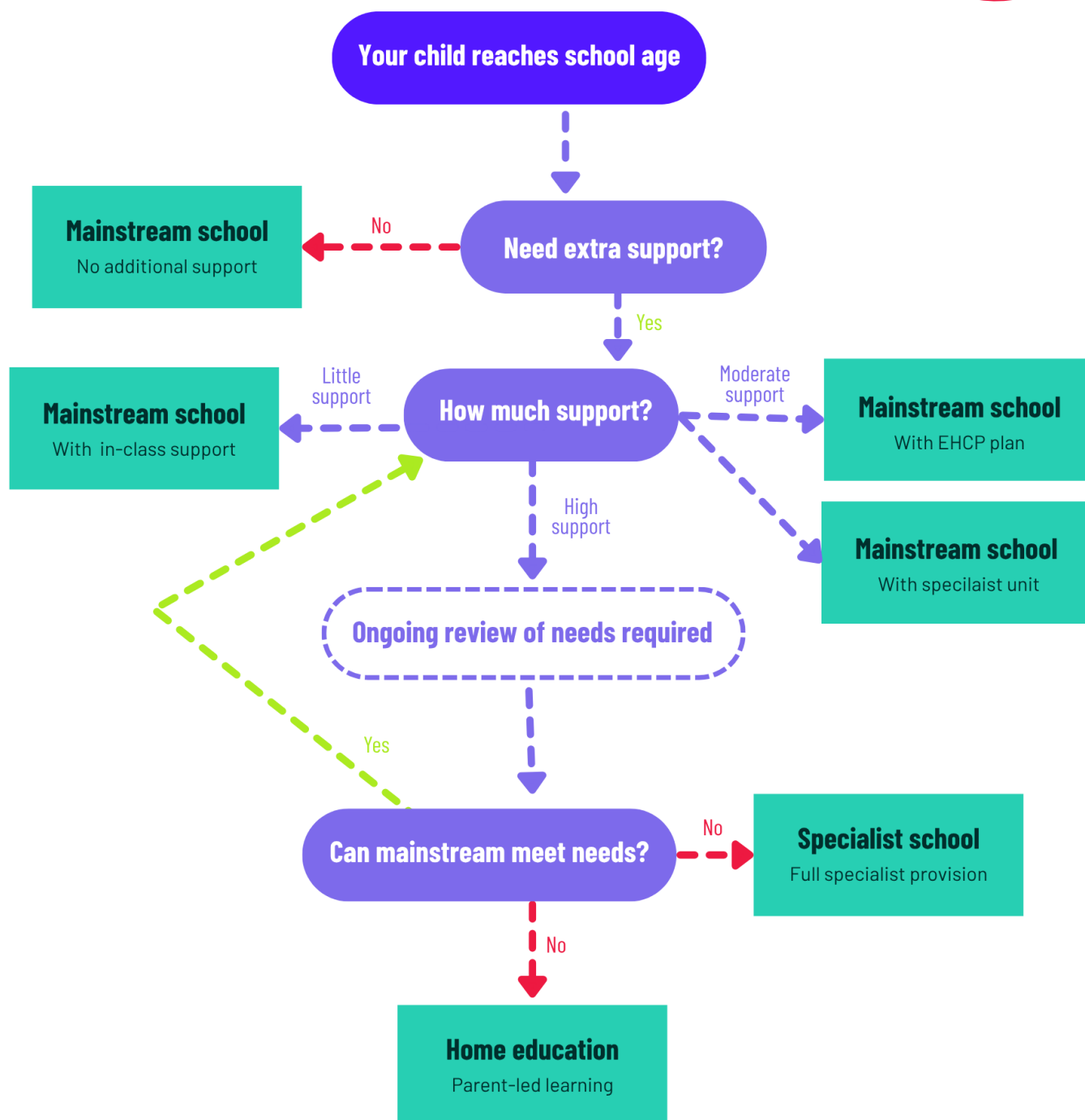
Children who need additional support at school may be eligible for a formal education support plan or similar provision, depending on the education system in their country or region. These plans are designed to identify a child's individual learning needs and set out the support, adjustments or services they may require to access education.

The name of the plan, eligibility criteria and support provided vary between countries and, in some cases, between regions or schools. If you think your child may need additional support, speak with their teacher, school or relevant education authority to find out how to request an assessment or evaluation. It is best to start this process early, as assessments and decisions about eligibility can take time.

You may want to talk with the school about any areas where your child may need extra help, such as:

- A specialised learning programme or education plan
- Extra support from a teacher or classroom aide
- Working in a smaller group or receiving targeted instruction
- Observation in the classroom or during less structured times, such as during breaks
- Support to participate in classroom activities
- Extra encouragement with learning tasks, such as asking questions or trying new or challenging activities
- Help with communication and social interaction with other children
- Support with physical or personal care needs, such as eating, mobility or using the bathroom safely

Schooling options decision aid



00 *We were relieved when we got the diagnosis because it gave proof that our kids were special. It also gave us comfort that our choice to home-school was the right choice."*

"I was homeschooled and I was diagnosed at 28."

"He was included in the regular classroom from [5 years old] through to about [11 years old], with a 1:1 aide. [From 9-11 years old] he was pulled out for a special program half days. He was in a special ed classroom [from 11-14 years old] with some participation in specials (music, PE) and in a functional classroom from [14 years old] until he turned 22."

"Advocate, advocate, advocate. Be as open and honest with the special education team as you can. The more information they have and the more willing you are to work with them the better the outcomes for your child."



Further education and beyond

As your child approaches the end of their school years, it is important to start thinking ahead about what comes next. There are many different paths they could take, including further education, vocational or technical training, employment, volunteering or other meaningful activities. Transition planning can take time, so it is helpful to start early, giving your family time to explore options, identify your child's goals and put appropriate support in place.



Support for young people moving from school into further education, employment or other adult services varies between countries and, in some cases, between regions. Support may be provided by the school, education authorities, transition services, social care or other specialist teams. These services can help young people and their families identify appropriate options and arrange support, which may include educational assistance, communication support, accessibility arrangements, health-related support, or help with organisation and daily living skills.

It is also important to plan for the transition to adulthood, as the age at which a young person is legally considered an adult varies between countries. Once they reach the relevant age, they may have greater responsibility for making decisions about their healthcare, education and other services, and parents or carers may no longer automatically have access to their information or be able to make decisions on their behalf. Planning ahead can help families understand these changes and avoid gaps in healthcare, insurance, medication, equipment or other support.

Unique publishes a separate guide to **Education, Transition and Further education**. Although these are UK based, some information might be helpful to families outside the UK



Becoming a carer/caregiver

A carer/caregiver is someone who provides regular support and care for a child, partner, relative, friend or neighbour who has a disability or additional needs. This may include physical, medical, emotional or practical support, or a combination of these.

As a parent, you naturally care for your child, but if your child has additional needs that require more support than a typically developing child of the same age, you may also be recognised as a carer/caregiver. This term can be helpful in acknowledging the extra time, energy and coordination involved in managing medical care, therapies, appointments and daily support needs.

It is important to acknowledge that being a carers/caregivers can be deeply meaningful, but also physically and emotionally demanding. Many carers/caregivers describe feeling tired, overwhelmed at times or under pressure to manage complex medical and practical needs while also trying to maintain family life, work and personal well-being. These feelings are very common, especially when support is limited, and they do not mean you are not coping - they reflect the reality of caring for a child with additional needs.

Unique publishes a separate guide to **Carers wellbeing**

Working parents

Balancing work and caring / caregiving can be especially challenging after a child receives a diagnosis or has ongoing medical or developmental needs. Many parents find they need to adjust their working patterns to attend appointments, therapies or school meetings. If you are employed, it may help to talk with your employer about flexible working arrangements, such as adjusted hours, remote work or time off for appointments. Many workplaces are able to offer reasonable adjustments to support carers / caregivers. If you are considering reducing your working hours or leaving your job, it can be helpful to explore financial assistance, employment rights and community support before making decisions. Social workers, care coordinators and family support organisations can often help you understand what options and support are available.

Respite care

Regular breaks from caring/caregiving are essential for both children and families. Short breaks give carers/caregivers time to rest and recharge, which can help prevent exhaustion and reduce stress over time. These breaks can be in the form of respite care, which is a service that provides short-term support for children with disabilities or additional needs, giving their carers/caregivers a planned break from daily caring responsibilities.

Respite care also provides children with opportunities to spend time with peers, try new activities and build independence in supportive environments. For children with additional needs, these experiences can also support social development and confidence outside of their usual routines.

Families may be able to access financial assistance to help cover the cost of respite care, depending on the country or region where they live. Support may be available through government-funded programmes, social care services, disability benefits, health or community programmes, or other local organisations. Eligibility, funding available and how to apply vary widely, so families should contact their local health or social care authority, relevant government department or disability support organisation to find out what financial assistance or respite services may be available in their area.

Financial support

Families raising a child with a disability or additional needs may be able to access financial, healthcare and practical support to help with the additional costs associated with medical care, therapies, equipment and daily living.

The types of support available, eligibility criteria and application processes vary between countries and, in some countries, between regions or local authorities. Support may include disability benefits, financial assistance with care at home, tax relief or credits, healthcare or therapy funding, disability savings schemes, trust arrangements or other government-funded programmes.

Some families may also be eligible for additional local or regional assistance, such as transportation support, housing adaptations or other disability-related benefits. Contact your local government, health or social care authority or a relevant disability organisation to find out what support may be available in your area and whether your family is eligible.



Medications assistance

Families may be able to access financial assistance or other schemes to help with the cost of medications, medical treatment and healthcare. The types of support available vary between countries and regions and may depend on factors such as income, age, disability, diagnosis or type of treatment. Support may include government-funded healthcare programmes, prescription assistance schemes, insurance coverage, charitable organisations or other financial assistance programmes. Contact your healthcare provider, local health authority, government department or relevant patient organisation to find out what support may be available in your area.

Creating a personal health record

Many families find it helpful to keep all of their child's medical, therapy, insurance and educational information organised in one place. Having important documents easily accessible can make it simpler to manage appointments, communicate with healthcare providers and keep track of your child's care over time.

A personal health record may include:

- A summary of your child's diagnosis and medical history
- Genetic testing reports and laboratory results
- Lists of medications, allergies and supplements
- Contact information for doctors, therapists and specialists
- Therapy evaluations and progress reports
- Hospital discharge summaries or emergency care plans
- School records, including formal education support plans
- Insurance cards, authorisation letters and billing information
- Notes from appointments or questions for future visits



Bringing your personal health record to medical appointments can help providers quickly understand your child's history, current needs and ongoing treatments. Many families also find it useful during emergency visits, school meetings or when seeing new specialists.

Additional support

Local support

As children grow, the level of support they need may increase, and daily care can become more complex and time-consuming. Because of this, many parents find it helpful to build connections and support networks early, rather than waiting until challenges become more difficult to manage.

Support may be available through:

- Hospitals or specialty clinics
- Early Intervention programmes
- Parent training and information centres
- National or condition-specific organisations
- Local disability or family support services
- Online support communities



Online groups

Online groups can offer encouragement and shared experiences, helping families feel less alone. Facebook is often a good place to find existing groups for families affected by a particular rare chromosome or gene disorder. If there is no group for your child's condition, you may wish to consider setting up a new group for families. Unique may also have WhatsApp groups that you can join, where available – just get in touch to find out more!

It is always important to remember that while these communities can be a valuable source of support, medical advice should always come from qualified healthcare professionals.

Patient advocacy organisations

Patient advocacy organisations can also be an important source of support and information. Many work closely with researchers, clinicians and healthcare teams to help families learn about new developments, ongoing research studies and opportunities to participate in clinical trials. These organisations can also provide education, connect families with resources and help amplify the voices of patients and carers/caregivers in the wider medical community.

Families say...

Take each day as it comes. Enjoy your child's every moment, because before you know it, they've grown up so quick and you might have missed so much that you can never get back."

"Don't waste too much time worrying about the future, it comes soon enough."

"You alone are enough, you have nothing to prove to anybody."

"I promise you it is not all doom and gloom (as it may feel like it is now), your child will surprise you at every turn and they will teach you more than you will ever know."



“Remember: diagnosis is just a word - it doesn't define who you are. Your personality and character do that.”

“Grieving the expectations you had, while still loving and celebrating your child, can both exist at the same time. Those feelings don't cancel each other out. They are both part of the journey.”

“Be patient, do research and lots of it, don't give up. Find Facebook groups and get support from other parents - it helps to know you're not alone.”

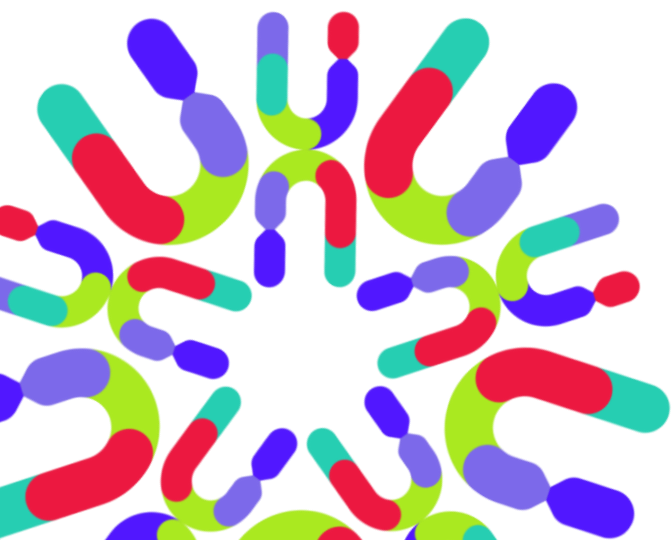
“Hold tight to your marriage and your support system. You will need them. Make absolutely sure you are investing quality time with your other children because this new normal will significantly impact them.”

“After time, I learned about Unique and eventually was connected with two other parents whose children have the same duplication. That was amazing! I would also give parents the advice that it isn't very helpful to focus on the exact genetic research and all the unknowns. The key thing to focus on is what affects your child right now, in everyday life. When therapies or medicines were suggested, it helped most to ask myself - will this make our everyday lives better and more pleasant? I learned to only say yes to things that met that criteria.”

“My advice would be don't give up, continue to advocate for your child, and educate yourself in everything you could possibly know about their condition. Write everything down- you can't possibly remember all the information thrown at you from different doctors.”

“I would tell other parents of children with rare conditions that they should be prepared to advocate hard for their kids but more importantly, find other families/parents to connect with. NO ONE else will get what you are feeling and going through like other parents will. Not even your own families.”

“I'd say don't panic. Your baby is still the baby you knew before, you just know a little bit more about them. You have answers, so use that knowledge to get them the help they need as soon as you can.”



Useful websites and resources

This is by no means an exhaustive list. If you would like more information on support available in a particular country, or think you can add to the list of resources, please contact Unique: help@rarechromo.org

Rare disease & genetic resources

- **Genetic and rare diseases (GARD) information centre** - A free, publicly accessible database run by the National Institutes of Health (NIH) that provides clear, reliable information on thousands of rare and genetic conditions, including symptoms, diagnosis, treatment and research
- **NORD** - A leading patient advocacy organisation that provides free information guides, resources and support programmes for individuals and families living with rare diseases, while also advocating for improved policies and research funding
- **Orphanet** - A comprehensive international database providing detailed information on rare diseases, orphan drugs, ongoing clinical trials, specialised medical centres and patient support organisations worldwide
- **GeneReviews** - A free, regularly updated collection of expert-written, peer-reviewed articles on hereditary genetic conditions, covering diagnosis, management, genetic testing and guidance for families, published by the National Centre for Biotechnology Information (NCBI)
- **International Genetic Alliance (IGA)** (Telephone: +31 35 603 4040) - An international alliance connecting genetic and rare disease patient and parent organisations
- **EURORDIS – Rare Diseases Europe** (Email: eurordis@eurordis.org, Telephone: +33 1 56 53 52 10) - A European alliance of rare disease patient organisations providing information, advocacy and connections across Europe



Clinical trials & research databases

- **ClinicalTrials** – A comprehensive global registry maintained by the NIH that lists thousands of ongoing and completed clinical studies, allowing patients and families to search for research trials relevant to their condition
- **CheckRare** – A rare disease information and awareness platform that helps patients, carers/caregivers and healthcare professionals stay informed about the latest developments, research and resources across a wide range of rare conditions
- **EU Clinical Trials** – A public portal for the Clinical Trials Information System (CTIS), providing searchable information on clinical trials conducted in the EU and European Economic Area, including study details, recruitment status, participating countries, and trial results where available

Australia

- **Genetic Alliance Australia** (Email: info@geneticalliance.org.au, Telephone +61 2 9295 8359) – A charity providing peer support and information for individuals and families affected by rare genetic conditions, and a contact point of support groups across Australia
- **SWAN Australia** (Email: info@swanaus.org.au, Telephone +61 404 280 441) – A charity providing information, peer support, connection and advocacy for families caring for a child with an undiagnosed or rare genetic condition

Austria

- **Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases (LBI-RUD)** (Email: office@rud.lbg.ac.at, Telephone +43 1 40160 70093) – A research institute focused on rare diseases of the immune system, haematopoiesis and nervous system, working to decode and develop personalised approaches for rare and undiagnosed diseases

Argentina

- **Fundación Red de Madres de Hijos con Discapacidad** – A group providing support and advocacy for children and adults with disabilities, including information on rights, equality and non-discrimination

Brazil

- **Instituto Vidas Raras** (Email: vidasraras@vidasraras.org.br, Telephone: +55 11 2414-3060) – Provide advocacy and support for people with rare diseases, particularly those in situations of vulnerability

Belgium

- **Rare Disorders Belgium / RaDiOrg** (Email: info@radiorg.be, Telephone +32 478 72 77 03) – The Belgian national alliance representing people and organisations affected by rare diseases

Bulgaria

- **National Alliance of People with Rare Diseases (NAPRD)** (Telephone: +359 888 323 748) – An alliance providing support and advocacy for people affected by rare diseases

Canada

- **Canadian Organization for Rare Disorders (CORD)** (Email: info@raredisorders.ca, Telephone: +1 416-969-7464 / 1-877-302-7273) – Canada's national network for organisations representing all those with rare disorders, providing information, advocacy and support
- **Canadian Rare Disease Network (CRDN)** (Email: info@canadianrdn.ca) – A pan-Canadian network connecting people, organisations, researchers and healthcare professionals to improve rare disease care, research, support and collaboration across Canada.
- **Regroupement québécois des maladies orphelines (RQMO)** (Email: administration@rqmo.org, Telephone: +1 819-943-2854 / 1-888-822-2854) – A Québec-based organisation providing information and support to people affected by rare diseases, their families and healthcare professionals, as well as raising awareness and sharing information about rare diseases

Central, North and South America

- **SPLAGen** (Email: info@splagen.org) – advancing awareness of and access to genetic counselling and maintaining a directory of genetic providers across Latin America and the US

Denmark

- **Rare Diseases Denmark (Sjældne Diagnoser)** (Email: mail@sjaldnediagnoser.dk, Telephone +45 33 14 00 10) – A national alliance of member societies advocating for people affected by rare diseases and running a free, confidential helpline

Finland

- **Terveyskirjasto** (Email: terveyskirjasto@duodecim.fi) – A Finnish public health information resource offering thousands of expert-written articles on diseases and health, including rare diseases
- **Kehitysvammaisten Tukiliitto (The Finnish Association of Societies for Persons with Intellectual Disabilities)** (Email: toimisto@tukiliitto.fi, Telephone: +358 400 968 200) – Provide information, support and advocacy for people with intellectual disabilities and their families. They also run the rare-disease unit Harvinaiskeskus Norio

France

- **Valentin APAC** (Email: contact@valentin-apac.org, Telephone: +33 1 30 37 90 97) – Provide support for children and families affected by rare chromosome disorders, connecting families and providing information
- **Maladies Rares Info Services** (Telephone: +33 800 40 40 43 (free in France) / +33 1 56 53 81 36 (from abroad)) – A national listening, information and orientation service for people affected by rare diseases
- **Alliance Maladies Rares** (Email: contact@maladiesrares.org) – A French collective bringing together organisations representing people affected by rare diseases, providing information, support and advocacy for people with rare diseases and their families.
- **AFM-Téléthon** (Telephone: 0 800 35 36 37) – A French patient and parent organisation supporting people and families affected by rare genetic diseases. It provides information and support, helps improve access to diagnosis and care, and advocates for people affected by rare diseases.
- **SOLHAND – Solidarité Handicap Autour des Maladies Rares** (Email: contact@solhand.org) – A French collective bringing together organisations and individuals connected with rare diseases and disability. It provides information, support and signposting, supports patient organisations and raises awareness of issues affecting people with rare diseases and disabilities.

Germany

- **LEONA e.V.** (Email: geschaeftsstelle@leona-ev.de, Telephone: +49 2301 1846685) – A family self-help organisation supporting and informing children and families affected by rare chromosome disorders

Greece

- **Tomellon – Association of People with Genetic Abnormalities** (Email: info@tomellon.com.gr, Telephone: +30 210 6420821) – providing psychological, social and financial support and reliable information to children and families affected by genetic conditions

Hungary

- **Hungarian Federation of People with Rare and Congenital Diseases (RIROSZ)** (Email: info@rirosz.hu, Telephone: +36 1 790 4533) – An umbrella federation providing information, support and advocacy for people affected by rare and congenital diseases, running the 'Mentőöv' information centre / helpline

Iceland

- **Einstök Börn** (Email: einstokborn@einstokborn.is, Telephone: +354 568 2661) – An advocacy and support group for children and families affected by rare disorders and syndromes

Italy

- **UNIAMO – Federazione Italiana Malattie Rare** (Email: segreteria@uniamo.org, Telephone: +39 06 4404773) – The national federation representing the rare-disease community in Italy, supporting people with rare diseases and their organisations
- **Le Malattie Rare** – An Italian online community dedicated to quality information and scientific insight on rare diseases

Kenya

- **Rare Disorders Kenya** (Email: ask@rarediseasekenya.org, Telephone: +254 701 025 753) – A patient-led organisation creating awareness and advocating for people living with rare diseases in Kenya

New Zealand

- **Rare Disorders New Zealand** (Email: enquiries@raredisorders.org.nz, Telephone: +64 4 385 1119) – The national body supporting people with a rare disorder and acts as the connector hub and collective voice for the community

Norway

- **Frambu Resource Centre for Rare Disorders** (Email: info@frambu.no, Telephone: +47 64 85 60 00) – Provide leading-edge expertise, knowledge and services for more than 120 rare disorders

Poland

- **Stowarzyszenie “Krok za krokiem” Zamość** (Email: biuro@spdn.pl, Telephone: +48 84 639 33 11) – A public-benefit organisation supporting children, young people and adults with disabilities and their families, running schools, therapy and support facilities

Portugal

- **SERaro** (Email: ser@seraro.pt, Telephone: +351 969 756 677) – An association supporting people with exceptionally rare or undiagnosed conditions and their families

Russia

- **All-Russian Society of Rare (Orphan) Diseases (BOO3)** (Email: vooz@bk.ru, Telephone: +7 800 201 06 01) – A national society providing information and support and helping people with rare diseases access treatment, diagnosis and social support
- **All-Russian Organization of Parents of Disabled Children (БОРДИ)** (Telephone: +7 800 777 234 7) – An organisation of parents and legal representatives providing support and advocacy for families of children with disabilities, including rare and genetic conditions

South Africa

- **Rare Diseases South Africa** (Email: hello@rarediseases.co.za, Telephone: +27 10 594 3844) – A registered nonprofit providing information, support and advocacy for people and families affected by rare diseases and congenital disorders

Spain

- **FEDER – Spanish Federation of Rare Diseases** (Email: sio@enfermedades-raras.org, Telephone: +34 91 533 40 08) – A federation that provides information, support, advocacy and connections for people affected by rare diseases

Sweden

- **Riksförbundet Sällsynta diagnoser (Swedish Association of Rare Disorders)** (Email: info@sallsyntadiagnoser.se, Telephone: +46 70 332 68 78) – A national organisation providing information, support and advocacy for people with rare diagnoses and their relatives

Switzerland

- **Info Maladies Rares** (Email: contact@infomaladiesrares.ch, Telephone: +41 848 314 372) – An information and guidance helpline for French-speaking Switzerland, signposting patients and professionals to specialist consultations and resources

The Netherlands

- **VSOP** (Email: zeldzameaandoening@vsop.nl, Telephone: +31 35 603 40 40) – The Dutch umbrella alliance providing information, support and advocacy
- **Platform ZON (Ziekte ONbekend)** (Email: zeldzameaandoening@vsop.nl, Telephone: +31 35 603 40 40) – A VSOP initiative connecting and supporting parents of children with an undiagnosed or ultra-rare condition

United Kingdom

Unique publishes a separate guide to **After Diagnosis, What Next: United Kingdom (UK)**

USA

Unique publishes a separate guide to **After Diagnosis, What Next: USA**

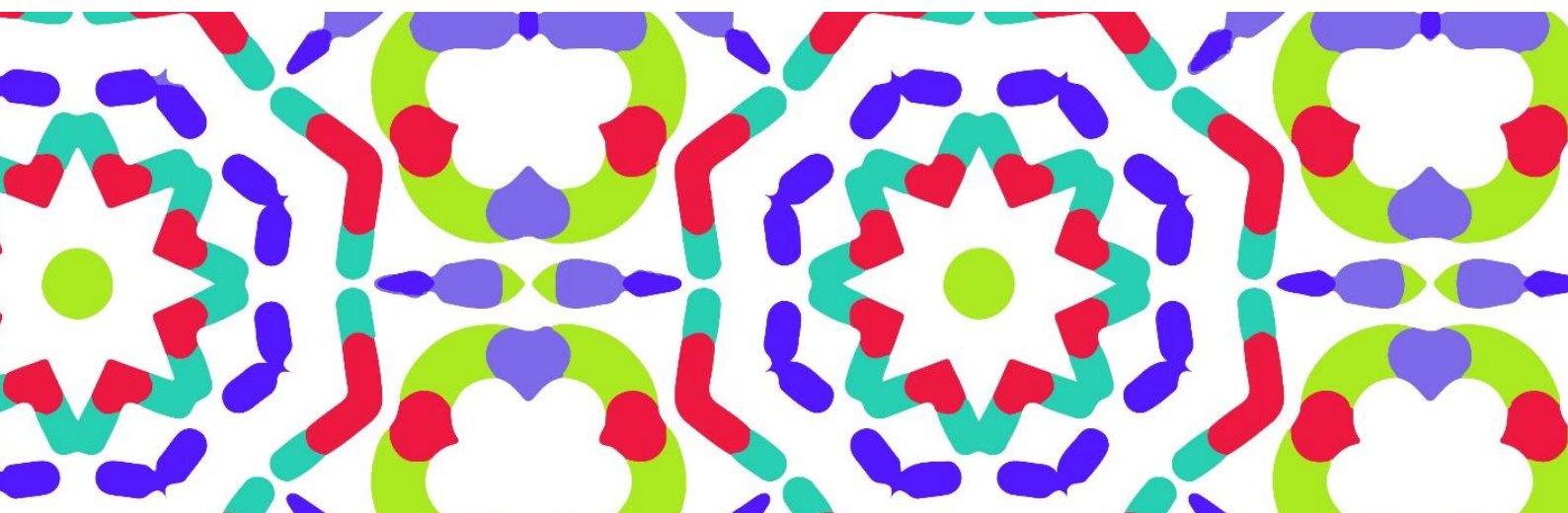
Research opportunities

Many families choose to participate in research related to their child's genetic condition. Research can help scientists better understand rare conditions, develop treatments and improve care for future patients.

There are several types of research studies families may encounter:

- **Clinical trials** - Clinical trials study new medications, therapies or medical approaches to determine whether they are safe and effective
- **Natural history studies** - These studies follow individuals with a specific condition over time in order to understand how the condition changes or progresses throughout life
- **Patient registries** - Registries collect health information from individuals with a specific condition. This information can help researchers identify patterns, improve care guidelines and connect families with research opportunities.

Participation in research is always voluntary. Families can decide whether participation feels right for them and can discuss potential opportunities with their healthcare team.



Inform Network Support



Rare Chromosome Disorder Support Group
The Stables, Station Road West, Oxted, Surrey, RH8, 9EE, UK
Tel: +44(0)1883 723356
help@rarechromo.org | rarechromo.org

Join Unique for family links, information and support:

[Become a member](#)

Please help us to help you!

Unique is a charity without government funding, existing entirely on donations and grants.
If you can, please make a donation:

[Donate via our website](#)

Unique lists external websites in order to be helpful to families looking for information and support. This does not imply that we endorse their content or have any responsibility for it.

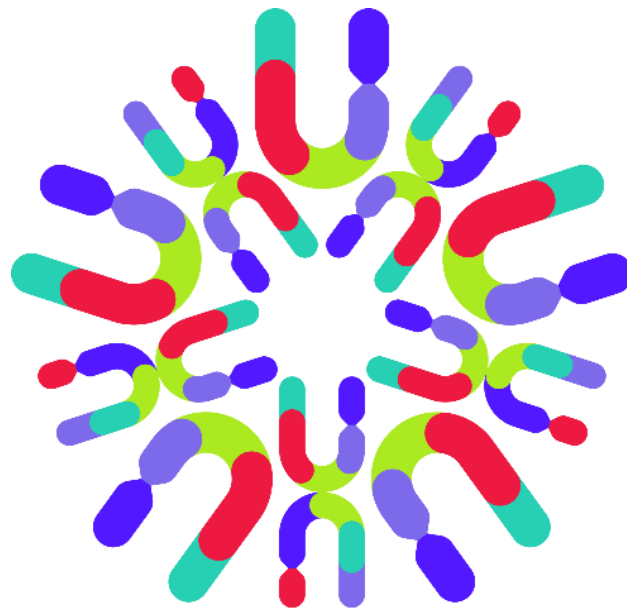
This information guide is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change.

Our thanks to all the parents who contributed to this guide.

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