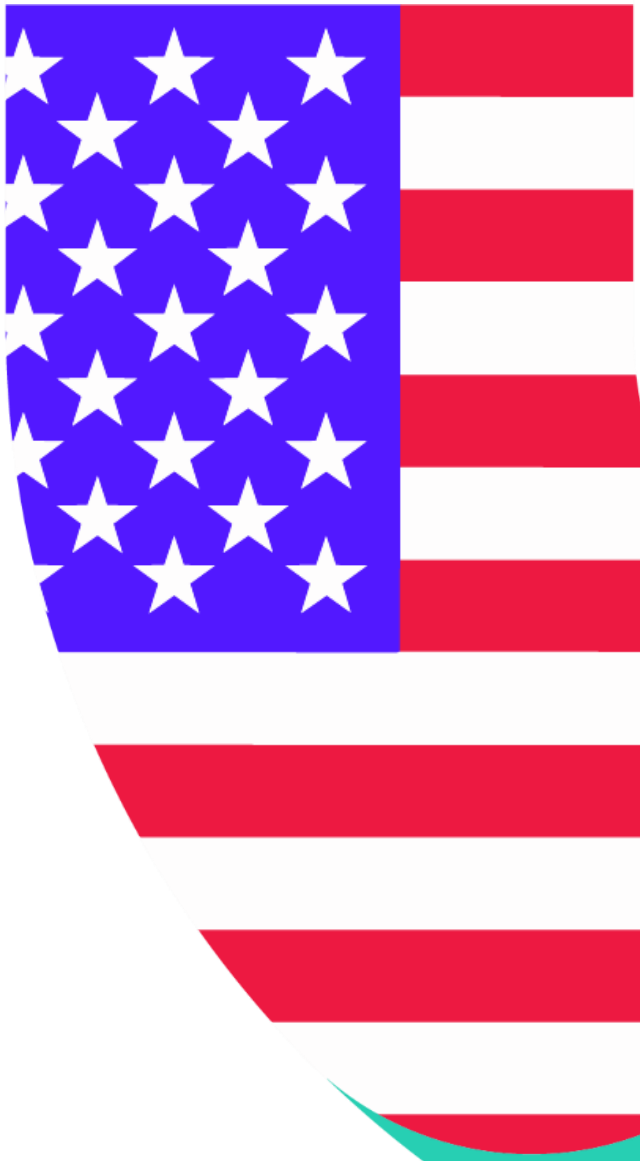


UNIQUE

UNDERSTANDING GENES & CHROMOSOMES

# After diagnosis: What happens next?

## (United States of America resident edition)

rarechromo.org

## 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.

***I was shocked and scared when the doctors gave us the news… I felt alone. They told us about it, gave us the Unique pamphlet, and that was it. I didn't know what questions to ask." - Georgia, 10 months old***

***"…When they sat us down with the results, they took us to a conference room with a social worker and really dour faces and presented it like a cancer diagnosis or something. They were really set up for us to do some kind of mourning about it. The doctors seemed kind of preoccupied with the genetics 'why' when all we wanted to do was get the baby home… there is already so much going on that it's hard to become very fixated on the subtleties of a microdeletion. My take on it has always been that a rare condition isn't some kind of injury or disease that attacked my kid, it's literally the DNA that makes her herself.” - New York, 6 years old***

***"…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” - Washington State, 25 years old***

***"The diagnosis was good to have and answered some developmental questions. However, we felt lost and still had many questions as far as what to expect going forward." - Pennsylvania, 27 years old***

***"It was devastating and really scary. Suddenly we were switching gears from financial planning for college to trying to figure out how to ensure our daughter would have funds for adequate care after we died. There was also just such a void of information on the disorder. Unique's guide was the only thing we read that gave us a sense of the range of possibilities rather than just a worst case scenario. Mourning the life you planned for someone you haven't even met yet is a surreal experience." - Vermont, 5 months old***

*"I was very scared when I first read the diagnosis… The google searches and rabbit hole I went down was SO far from the reality of what the results actually meant" - Colorado, 6*

*"We had known for some time that something wasn't quite right. The diagnosis was affirming of these feelings that had been brushed off by a lot of other people... Having a diagnosis gave us direction, but there was a lot that was still unknown" - South Carolina, 8 years old*

*"Stunned and devastated… I didn't know how to move forward. Being pregnant at the time, I sort of wished I could just stay pregnant forever, not wanting to face reality. I wish I knew that reality is different once you meet and fall in love with your child. - Michigan, 21 years old*

*"Suddenly having the diagnosis sprung on me by someone who thought I already knew was awful. 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." - California, 13 years old*

*"While the diagnosis made us concerned about his future (which it still does), we also knew that he wasn't developing typically. I think I initially really wanted there to be nothing wrong… but I think deep down, I knew that wasn't likely. I felt fairly at peace when we were first heard. It wasn't until I told our nanny that I got choked up and cried. The uncertainty of not knowing what his future will bring and his inability to tell us if something is wrong weighs greatly on me." - California, 4 years old*

*" Getting a diagnosis was in some way positive, as it acted as an explainer for some of her issues. However, it also made it seem more 'real'. I think the sheer whirlpool of emotions stands out, as our moods and emotions changed a lot every day. This has settled somewhat overtime." - New Jersey, 1 year old*

*"I felt overwhelmed, scared, and honestly a little numb at first. There had been so many appointments, tests, and unanswered questions that part of me was relieved to finally have an explanation, but another part of me was terrified of what it might mean for his future. It felt like every appointment brought new information, new specialists, and new things to worry about. At the same time, what stands out most is how strong [he] was. While I was worrying about diagnoses and milestones, he was just being himself—smiling, growing, and teaching us to celebrate every little victory." - West Virginia, 6 months old*

*"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." - Kansas, 8 years old*

# 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

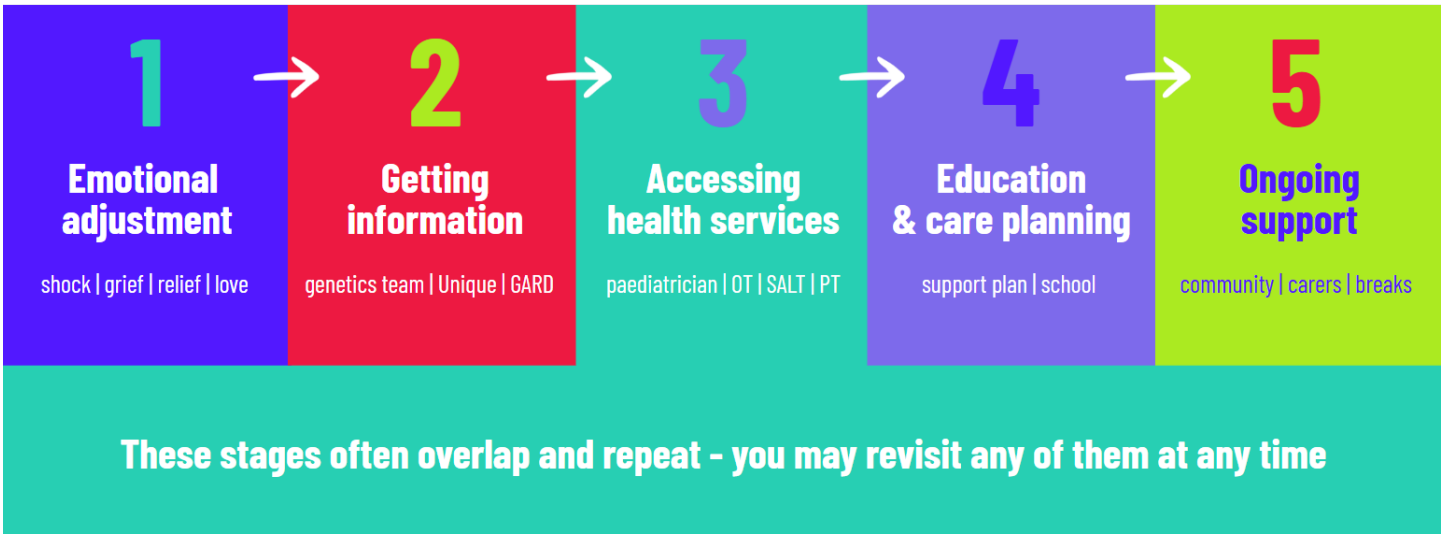
- Request copies of your child's genetic testing results
- Share the results with doctors and specialists involved in your child's care
- Schedule a visit with your genetic counselor or genetics clinic
- Contact your state's Early Intervention program if your child is under three years old
- Ask your 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 organizations or support groups
- Learn more about your insurance coverage and benefits

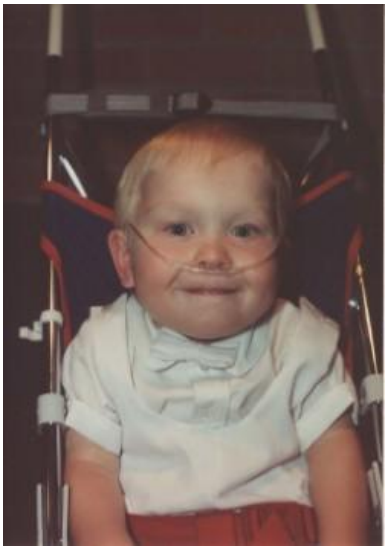
## Months 5–6

- 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




*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" – West Virginia, 6 months old*

*"In our son's life, nothing had changed because he was already getting all the therapies etc. But for me, my focus had been on connecting with other families and fundraising for our own research and drug repurposing screening." - Colorado, 6 years old*



*"There wasn't a lot that changed in the first six months. We had already begun speech therapy and at the time that's all we thought we needed. It wasn't until later that we realized we needed additional therapy, especially occupational therapy." - South Carolina, 8 years old*

*"I think the thing that has changed the most is our future planning. We have no idea what his future will look like, but it is unlikely he will be able to support himself. That is really scary, so we have set up our financial and planning future differently." - California, 4 years old*

*"Our whole routine changed we had appointments almost every day for a while. Applying for help and resources was difficult at times. The advice that I would give to parents is always ask for help, ask questions, get resources! - California, 2 years old*

## 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 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 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" - New Jersey, 1 year old*

## Seeing a clinical geneticist

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

In most cases, attending a genetics clinic requires a referral from your child's doctor or medical specialist.

Wait times for genetics appointments in the United States are often several months or longer, depending on the availability of specialists in your area. 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 intervention

**Early intervention (EI)** is a federally funded program in the United States that provides support and services to infants and toddlers from birth to age three who have developmental delays or medical conditions that may affect development. EI programs may have different names depending on the state. 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.

EI services may include speech therapy, occupational therapy, physical therapy, developmental therapy, feeding support, and other specialized services depending on a child's individual needs. In many states, a confirmed genetic diagnosis may automatically qualify a child for an evaluation, even if developmental delays are not yet apparent.

Families can usually access EI by contacting their state's EI program directly or through a referral from a pediatrician, clinical geneticist, genetic counselor, or another healthcare professional. After a referral is made, the child will typically receive a developmental evaluation to determine eligibility. If eligible, the family and care team will develop an **individualized family service plan (IFSP)**, which outlines goals and the services that will best support the child and family.

EI programs are available in every U.S. state and territory, although eligibility criteria and available services may vary somewhat by location. Starting services as early as possible can help maximize a child's developmental progress and long-term outcomes.

If you are concerned about your child's development, it is important to speak with your pediatrician 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.

> ***Early intervention was a game changer, we started at 6 months. While it is a little effort to coordinate sometimes, it is well worth it, and I encourage people to find therapists that work well for their family. I know his development would not be nearly as far without their help teaching him and us." — Missouri, 2 years old***

*"… These services have helped us better understand [his] strengths and challenges while giving us practical strategies to support his development at home. The therapists not only work with [him] but also teach us how to incorporate activities into our daily routines... My advice to other parents is to accept referrals early and not take a 'wait and see' approach if developmental concerns are present. Early Intervention does not mean something is wrong with your child—it means your child has access to additional support during a critical period of development. Ask questions, stay involved in sessions and remember that you are an important part of the therapy team. The earlier services begin, the more opportunities there are to build skills and address challenges before they become bigger obstacles." West Virginia, 6 months*

## Treatments and therapies

Your child may be referred to a specialist clinic or medical center 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:

**Developmental–behavioral pediatricians** specialize in child development, learning, and behavior. Developmental-behavioral pediatricians often coordinate care between therapists, schools, and medical specialists.

**Pediatric 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.

**Pediatric 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-language pathologists (SLP)** 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, orthopedic footwear, or spinal supports to improve alignment, posture, stability, and mobility.

**Neurologists** specialize 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.

**Endocrinologists** treat hormone-related conditions, including growth concerns, thyroid disorders, puberty differences, and metabolic conditions.

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

**Pulmonologists** specialize in lung and breathing conditions, including sleep-related breathing problems and chronic respiratory issues.

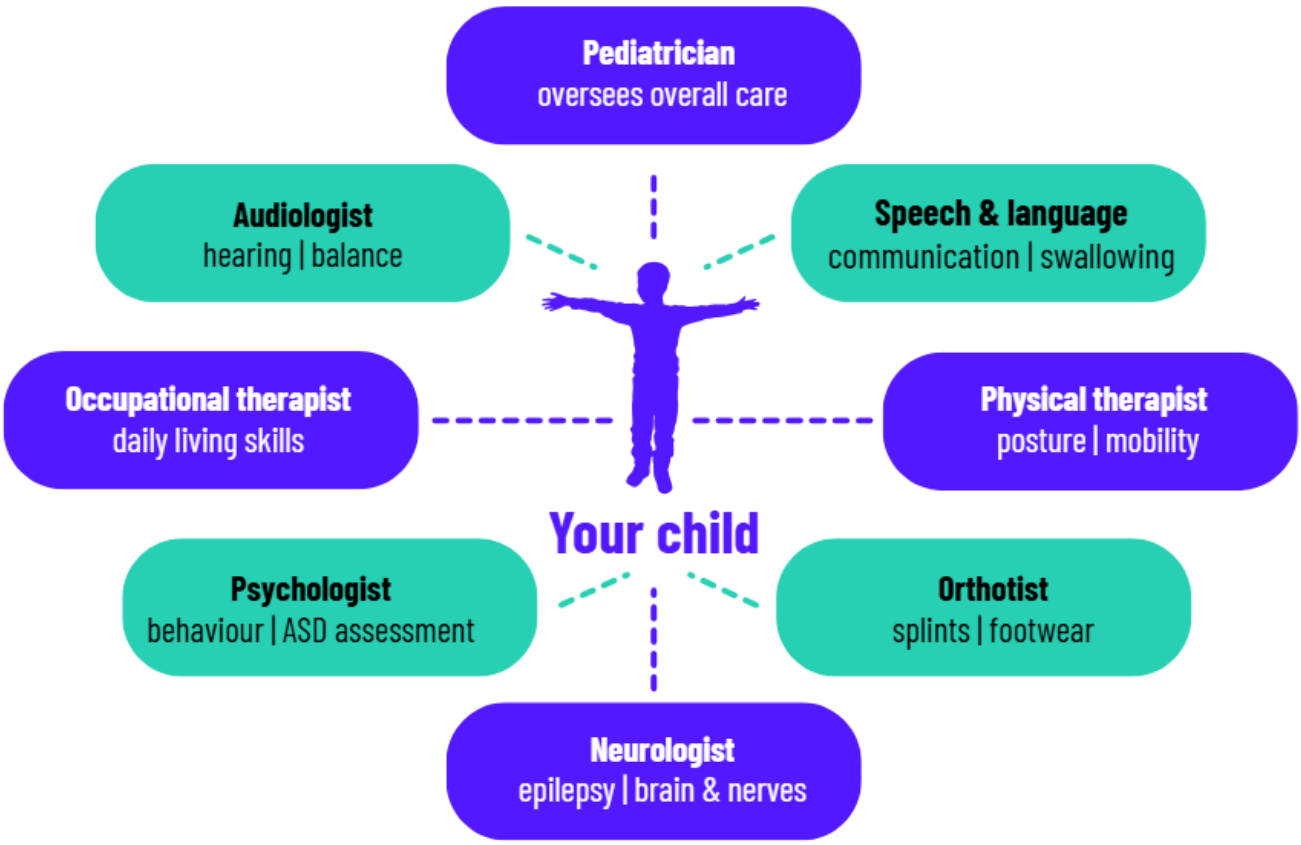
**Nephrologists** diagnose and treat kidney disorders and related medical concerns.

**Dermatologists** specialize in skin, hair, and nail conditions that may be associated with certain genetic conditions.

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

**Psychologist or psychiatrist** support children and families with behavioral, emotional, developmental, or mental health concerns.

**Care coordinator or case manager** help organize appointments, therapies, referrals, and communication between multiple healthcare providers and services.




*We saw a geneticist, GI, neurosurgery, neurology, developmental pediatrician, urology, and some others. Some of these were one or two consults and then never really saw them again... I wish we had known that it's worth a broader search for specialist clinics that are focused on kids with some of the resulting conditions, even if you have to travel once or twice a year... It's so worth the trip and we don't have to reeducate them every time, and we go to one room and get seen by urology, GI, PT, social work, everyone comes to us and has the same game plan." — New York, 6 years old*

*"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." — Vermont, 5 months old*

*"Traveling for specialist appointments is sometimes complicated and definitely time-consuming. Coordinating multiple appointments efficiently is challenging... My advice is to try to get the appointments on a similar schedule so you can spend a day here and there going to multiple providers if you have to travel out of town for them." - South Carolina, 8 years old*

*"If I could give any advice, it would be to try and stick with a single children's hospital to see all your specialists in one spot. They all use the same portal and can see what other specialists have said and communicate effectively." - Florida, 2 years old*

*"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." - California, 2 years old*

*"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." - New Jersey, 1 year old*

*"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." — Missouri, 2 years old*

*"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" – West Virginia, 6 months old*

## Care plans and follow-up appointments

After a genetic diagnosis, your child's pediatrician 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 pediatrician 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 over time.

# Medical insurance

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

There are several ways families may obtain health insurance in the United States:

- **Employer-sponsored insurance / commercial insurance** - Many people receive private health insurance for themselves and their families through their own or a family member's employer
- **Medicaid** - Medicaid is a government-funded health insurance program for individuals and families with lower incomes or significant medical needs. Eligibility rules vary by state. Some children with disabilities or complex medical conditions may qualify for Medicaid regardless of family income through special waiver programs even if the family wouldn't normally meet the standard income limits
- **Medicare via disability** - People of any age can qualify for Medicare after receiving Social Security Disability Insurance (SSDI) for 24 months, or immediately for certain conditions. Some adults with rare genetic conditions qualify for both Medicaid and Medicare (known as "dual eligibility"), which can provide more comprehensive coverage together than either program alone
- **Children's health insurance program (CHIP)** - CHIP provides low-cost health coverage for children in families who earn too much to qualify for Medicaid but cannot easily afford private insurance
- **Marketplace insurance plans** - Families can purchase insurance plans through the federal or state Health Insurance Marketplace during open enrollment periods or after certain qualifying life events. Depending on income, families may qualify for financial assistance (subsidies) to help lower monthly premiums and out-of-pocket costs
- **COBRA continuation coverage** – If a parent or carer loses or changes jobs, COBRA allows families to temporarily continue their employer-sponsored health insurance, usually at a higher cost. This can be an important bridge during transitions, especially when ongoing specialist care or therapies are involved
- **Military or government programs** – Some families receive healthcare coverage through programs such as TRICARE for military families, VA health benefits for veterans, or other government-sponsored healthcare plans
- **Private insurance** – Some people buy coverage directly from insurance companies. These plans may be an option for individuals who are self-employed, do not have access to employer-sponsored insurance, or prefer to work directly with an insurer

Health insurance plans can 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 authorization** - Approval that your insurance company may require before covering certain medications, tests, therapies, or procedures

*My daughter was given Medicaid as part of her genetic condition. The Genetic counselor helped us with this and was the one who suggested it in the first place." — South Carolina, 8 years old*

*"Health Insurance has been rough as it changed several times and care givers would change on what insurance they would take. We pay out of pocket for many of our services though we have full coverage HMO insurance." — California, 19 years old*

*"I wish we had really known about Medicaid from the beginning… I would encourage families with a new diagnosis to ask for support from a hospital advisor or advocate that can walk them through the process and connect them with the agencies they need. " — Colorado, 6 years old*

*"I could talk all day about the nightmare that we call insurance. Early on I was confused and surprised every time something was denied… Eventually I learned that children with severe disability can get a secondary insurance coverage through the state. Most of the time, if the first company denies something, the second covers it. My advice is always write down notes for yourself from every conversation. It's so confusing when there are multiple things going on, and you're put on hold, and you must retell the story." - California, 13 years old*

*"We have great health insurance and things have still been a struggle. Our providers were great too. But we've still had to call and advocate for him (compiling letters, etc.). Overall, it has been fine, we've mostly received the items necessary but still have really struggled with his care. Particularly because there is so little information. But there is such a lack of awareness that I don't think people know what is justified because they don't know his condition because it is so rare." — California, 4 years old*

*"We had no problems at all with our insurance, which I'm grateful for." — Missouri, 2 years old*

*"State insurance is the only thing we receive and it has been a huge benefit! With the number of medical appointments and therapies needed, there is no way we could afford it all. I do pay a small monthly premium for my daughter's medical care. I believe the program my kids are in is called CHIP under KanCare." — Kansas, 8 years old*

## Genetic discrimination and legal protections (GINA)

Some families worry that genetic test results could be used against them when applying for health insurance. In the United States, there are federal laws in place to help protect against this type of discrimination and to support the privacy of genetic information. **The genetic information nondiscrimination act (GINA)** is a federal law that protects individuals from discrimination based on genetic information. Under this law, health insurance companies are not allowed to use genetic test results to make decisions about coverage, eligibility, or insurance premiums.

However, it is important to understand that GINA does not cover every type of insurance. It does not apply to life insurance, disability insurance, or long-term care insurance. Your genetics team can also help explain how genetic information is protected, what may be shared in medical records, and what privacy safeguards are in place, so you can make informed decisions for your family.

# 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 behavior, 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:

- Behavior or ability to socialize
- 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 specialized 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.

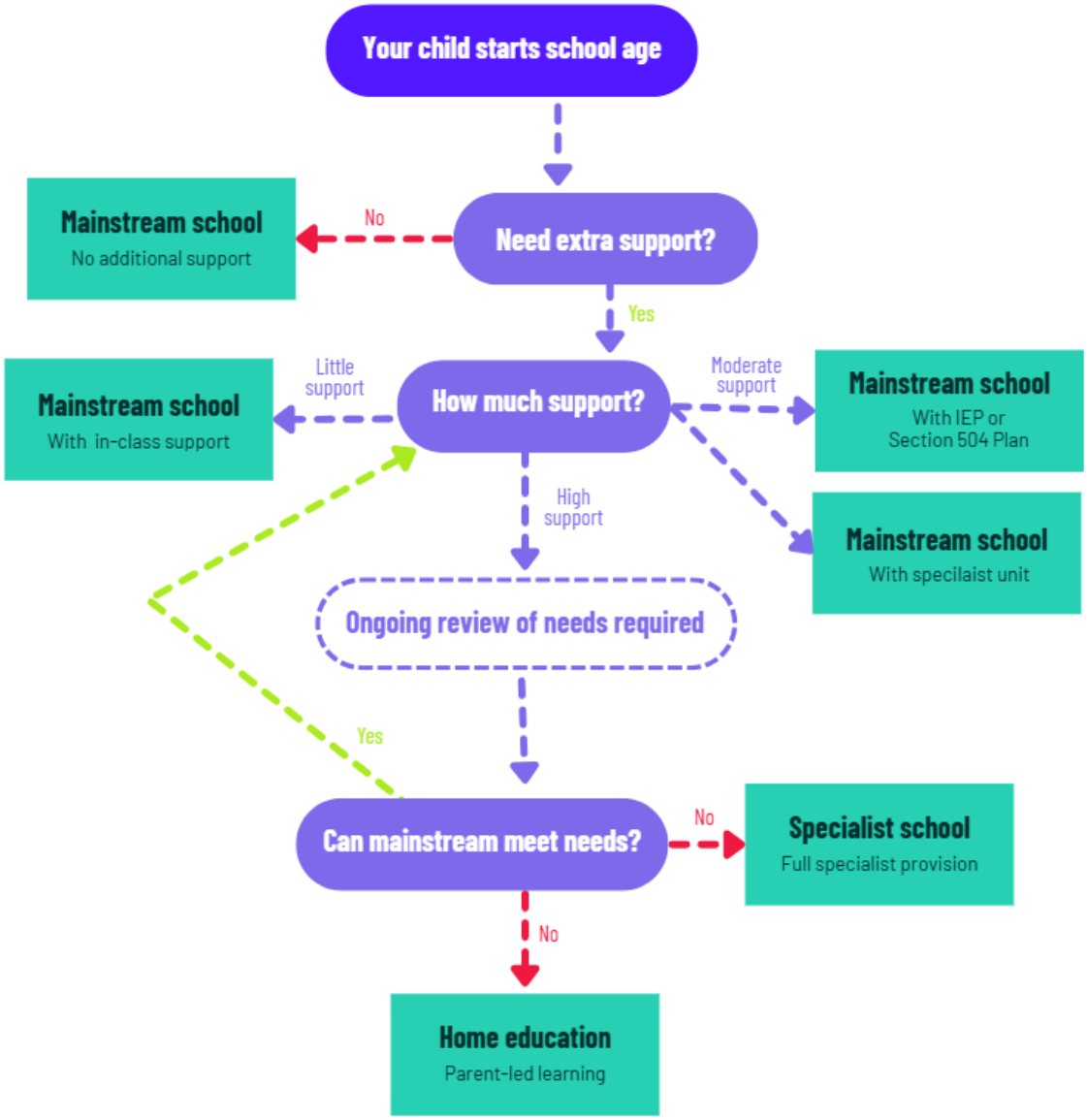
For some children, home education (homeschooling) may be a suitable alternative to attending school. Homeschooling allows learning to be tailored to a child's individual pace, interests, and abilities. For children with rare genetic conditions, this flexibility can sometimes make it easier to balance education with medical appointments, therapies, and periods of illness or fatigue. In the United States, home education requirements vary by state, so families may need to check local regulations regarding registration, curriculum expectations, and reporting.

> ***I was homeschooled and I was diagnosed at 28." — Arizona, 33 years old***

***"Originally in preschool, she was on a speech only IEP. This quickly changed in kindergarten. The conversation of repeating a grade began in 4K, but we waited until kindergarten to repeat. She started in a Montessori program for kindergarten, but it ended up not being the greatest. In the middle of her second year of kindergarten, we transferred her to a traditional school setting, and she did a lot better... Next year, we are going to try a hybrid homeschool program with the hope that the increased one to one time and small class size will be beneficial for her. We will still be able to access support services through the school district." — South Carolina, 8 years old***

***"He was included in the regular classroom from kindergarten through about fifth grade, with a 1:1 aide. In later elementary he was pulled out for a special program half days. He was in a special ed classroom in grades 6-8 with some participation in specials (music, PE) and in a functional classroom from grade 9 until he turned 22". – Indiana, 38 years old***

## Schooling options decision aid




## Individualized education program (IEP) or a section 504 plan

In the United States, children who need extra support at school may qualify for either an **individualized education program (IEP)** or a **section 504 plan**.

- An **IEP** is a written plan, unique to your child's needs. The goal of the IEP is to make sure your child gets the education they deserve, in the most typical school setting as possible. Ideally, learning will occur alongside their classmates for as much of the day as works for them, so they can build skills, keep up with their schoolwork, and prepare them for future after they finish school. For more information, you can visit the exceptional children's assistance center website

- A **504 Plan** is part of a federal civil rights law that makes it illegal to discriminate against a person because of their disability, meaning your child has the right to reasonable services and accommodations to help them in the classroom. This may include adjustments such as extra time for tests, seating changes, assistive technology, or support with organization and attention. To get a 504 plan, a healthcare provider must write a formal request describing your child's disability and need for accommodation, which you then bring to the school. The school will then create a 504 plan outlining the accommodations for your child

If you think your child may need additional support, you can request an evaluation through your local authority, or equivalent governmental or medical authority in your state. It is best to start this process early, as evaluations and eligibility decisions can take time. You can also speak with your child's teacher or the school's special needs coordinator.

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

- A specialized learning program or education plan (such as an IEP or 504 Plan)
- Extra support from a teacher or classroom aid
- Working in a smaller group or receiving targeted instruction
- Observation in the classroom or during less structured times, such as recess
- Support in participating 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

***"Advocate, advocate, advocate. Be as open and honest with the special education / IEP 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." — California, 17 years old***

***"Advocate for the maximum you can think of and then you can dial them back later, because adding things on later is harder. Use whatever paperwork you can. It can feel embarrassing at first to present your child as a medical nightmare on paper when you know they are much more than that as a person, but you really need to be comfortable turning that dial up when it will get you what you need." — New York, 6 years old***

***"The most helpful support is a 1:1 aide who can really support your child. They become integral to education success. If you don't know a lot about special education, get an advocate. You are part of an IEP team - Work together. Become friends with all members of the IEP team." — California, 17 years old***

## 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 directions your child could take after school, including college, vocational or technical school, working at a community business, volunteering, or finding another meaningful path forward. Because this transition can take time to plan well, it is a good idea to start the process when your child is around 12 to 14 years old, giving your family plenty of time to explore options and put the right support in place.

In the United States, support for students moving into further education is typically arranged through the school or college's disability services team, rather than through an IEP or 504 Plan. These teams work directly with students and their families to identify the accommodations and support needed, which might include academic assistance, communication support, accessibility arrangements, health-related accommodations, or help with organization and daily living skills.

It is also important to know that once your child turns 18, they are legally considered an adult, which means you will no longer automatically have access to their medical information or be able to make decisions on their behalf in the same way you could when they were younger. It can be helpful to plan ahead for these legal changes, as well as consider how to ensure there are no gaps in their medications, equipment, or insurance coverage during the transition.

**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 caregiver

A caregiver is someone who provides regular support and care for a child, partner, relative, friend, or neighbor 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 recognized as a caregiver (or carer). 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 caregiver can be deeply meaningful, but also physically and emotionally demanding. Many 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 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**

*I became a caregiver the day we brough my son home from the hospital. Everything changed. I grieved for a long time for the motherhood journey I had imagined. What surprised me most is how much less I hear from friends and family generally - and how irritated I get when people who know what I deal with on a daily basis tell me to 'make sure you take time for yourself. You can't pour from an empty cup'. People don't understand how pointless and impossible that can be for a caregiving parent." - Colorado, 6 years old*

*"It was honestly a relief to focus on my son, who was more important than anything to me. Over many years, I did begin to feel the loss of outside connections and lack of individual identity. I was surprised by how exhausting it is to be at home caring for my son, and how rarely I had the energy to go out even if I had a respite provider. My son and I have developed a very close bond and that has made it all worthwhile. I think it's the most meaningful thing I've ever done." — California, 13 years old*

*"Becoming [his] caregiver has changed nearly every part of my daily life. I expected motherhood to be busy, but I did not expect to become a care coordinator, therapist advocate, medical researcher, appointment scheduler, insurance navigator, and record keeper all at the same time. One of the biggest changes was realizing that being his caregiver would require me to step away from my career. That was a difficult decision because work had always been part of my identity. My identity shifted from balancing work and family to becoming my son's full-time advocate and primary support system." – West Virginia, 6 months old*

## Working parents

Balancing work and 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 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 organizations can often help you understand what options are available and what support may help your situation.

## Respite care

Regular breaks from caregiving are essential for both children and families. Short breaks give 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 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.

Depending on where you live and what insurance you have, you may be able to get financial help to cover respite care. This kind of support may be available through your private insurance, Medicaid, or local disability services, though what is covered and who qualifies will vary. If you are interested in finding out what is available to you, it is worth speaking to your doctor, social worker, or insurance provider.

## Financial support

In the United States, there are several programs and funding options that may help families who are raising a child with a disability or additional needs. These supports are designed to help with the extra costs that can come with medical care, therapies, equipment, and daily living needs. Eligibility and availability vary depending on income, state of residence, and the child's level of need.

Some possible sources of financial and medical support include:

- **Supplemental security income (SSI)** - Monthly financial assistance for children with significant disabilities and limited family income
- **Social security disability insurance (SSDI)** - Benefits for some young adults with disabilities based on a parent's work history
- **Katie Beckett / tax equity and fiscal responsibility act (TEFRA)** - an optional Medicaid eligibility pathway that allows children with significant disabilities or complex medical needs to receive care at home rather than in an institutional setting, with parents' income and assets not counted toward eligibility. Not all states offer this option, and each state has its own eligibility criteria and available services
- **Medicaid waivers / home and community-based services (HCBS)** - State programs that may help cover in-home care, respite services, personal care assistance, medical equipment, or home modifications
- **Child and dependent care credit** - If you pay someone to look after your child so that you are able to work, search for work, or attend school, you may be able to claim the Child and Dependent Care Credit on your taxes, which can help reduce the amount of tax you owe. The amount of the credit you can receive depends on your income and the percentage of care costs you paid to allow you to work or study
- **Achieving a better life experience (ABLE) accounts** – An ABLE account is a special savings account designed for people with disabilities that allows them to save and invest money for disability-related expenses without risking their eligibility for government benefits like SSI or Medicaid. The money saved in the account can grow through investments without being taxed, and as long as it is spent on qualifying disability-related expenses, it does not count as income
- **Special needs trust (SNT)** - a legal arrangement that allows money or assets to be set aside for a person with disabilities without affecting their eligibility for government benefits like Medicaid or SSI. The trust is managed by a trustee whose job is to use the funds to top up the person's existing government benefits, covering extra costs that those benefits do not pay for
- **Pooled trust -** another type of Special Needs Trust where a non-profit organization manages and invests money on behalf of multiple people with disabilities together, while keeping a separate account for each individual
- **Lifespan respite care program** - Federal money given to states specifically to help fund respite care

Some families may also be eligible for additional local or state-based assistance, such as transportation support, housing-related adaptations, or disability-related tax benefits, depending on where they live.

***"There are things our commercial insurance won't cover and some of her medications are expensive. Try to get a Medicaid waiver and it can cover some of the gaps and will be a backstop if you lose your insurance. We also max out the FSA plan (Flexible Spending Account) my wife has at work—it's empty by mid-year but it's something." — New York, 6 years old***

*"We have received an outstanding amount of financial support from pretty much all possible sources: Medicaid waiver, Medicaid Insurance, Private Insurance, A business fundraiser, private gifts from family and friends. I feel extremely ungrateful saying this, but it is not enough and it will never be enough. [Her] needs are a constant suck of financial resources." — Ohio, 9 years old*

*"He has a Medicaid waiver and has caregivers' hours to take care of him... He also gets food stamps to help buy his food, so that does help some and he gets SSI. Look into what is available to your loved one because there is financial help out there you just got to find it." — Washington, 25 years old*

*"We got my son on a Medicaid Waiver which has been a lifesaver for everything, particularly the paid caregiver program we have in Colorado. Ask other families, they are the ones that will tell you what to do and where to go. I found many local Facebook groups, and they were the most helpful." — Colorado, 6 years old*

*"Through the TEFRA program she has access to Medicaid coverage which covers what our private insurance doesn't. This is the only additional support that we. This has been a tremendous help for our family to increase our ability to provide therapy services for her that we have known has made a huge difference in her outcome. I have no doubt that it has made the biggest difference in her ability to overcome the disabilities she faces." — South Carolina, 8 years old*

## Medications Assistance

- **NeedyMeds** (Telephone: 800-503-6897, Email: info@needymeds.org) - A resource that helps people find assistance with medication and healthcare costs, including information on government programs, low-cost medical and dental clinics, prescription assistance, and financial aid programs.
- **Medicine assistance tool (MAT)** (Telephone: 571-350-8643) - A search tool created by the Pharmaceutical Research and Manufacturers of America (PhRMA) that allows patients, caregivers, and healthcare providers to quickly find financial assistance programs offered by biopharmaceutical companies to help reduce the cost of medications.
- **Medicare pharmaceutical assistance** (Telephone: 800-633-4227) - A directory of programs run by pharmaceutical companies that help cover the cost of prescription medications for people enrolled in a Medicare Drug Plan (Part D).
- **GoodRX** (Telephone: 855-268-2822, Email: support@goodrx.com) - A free tool that compares current prescription prices and discounts across pharmacies near you, helping you find the lowest possible price for your medications.
- **RxAssist** - A searchable database of patient assistance programs offered by drug manufacturers that provide free or heavily discounted medications to uninsured or underinsured individuals who cannot afford their prescriptions.
- **RxOutreach** (Telephone: 888-796-1234, Email: questions@rxoutreach.org) - A non-profit mail-order pharmacy that provides affordable medications to people who are struggling to cover the cost of their prescriptions.
- **RxHope** (Telephone: 877-267-0517, Email: compliance@triplefin.com) - A free service that acts as your personal advocate in navigating patient assistance programs, helping you access critical medications you may otherwise struggle to afford by guiding you through the process and connecting you with the right resources.

# Creating a care binder

Many families find it helpful to keep all of their child's medical, therapy, educational, and insurance information organized in one place. This is often called a medical care binder or care notebook. 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 care binder 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 IEP or 504 plans
- Insurance cards, authorization letters, and billing information
- Notes from appointments or questions for future visits

Bringing your care binder 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 programs
- Parent training and information centers
- National or condition-specific organizations
- Local disability or family support services
- Online support communities

## Online groups

Online groups can offer encouragement and shared experiences, helping families feel less alone. However, while these communities can be a valuable source of support, medical advice should always come from qualified healthcare professionals.

## Patient advocacy groups

Patient advocacy organizations 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 organizations can also provide education, connect families with resources, and help amplify the voices of patients and caregivers in the wider medical community.

## Advice for families

*"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." - West Virginia, 6 months old*

*One thing that was / is a bit difficult is how rare her genetic diagnosis is. I feel I have to do a lot of explaining to medical people any time we go to an appointment. I have gained much more knowledge from other parents I have found who have children with the same diagnosis." - Kansas, 8 years old*

*Genetic variants are not unusual and may or may not be linked with some of your child's other medical issues." - South Carolina, 8 years old*

*"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." — Washington, 25 years old*

*"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." — California, 17 years old*

*"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." — California, 13 years old*

*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." — Florida, 2 years old*

*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." — Colorado, 6 years old*

*"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." — Missouri, 16 and 19 years old*

# Useful websites and resources

This is by no means an exhaustive list. If you would like more information on support available in a particular state, 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 center** - 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 organization that provides free information guides, resources, and support programs 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, specialized medical centers, and patient support organizations 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 Center for Biotechnology Information (NCBI)

## 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, caregivers, and healthcare professionals stay informed about the latest developments, research, and resources across a wide range of rare conditions

## Autism support

- **Autism speaks** (Telephone: 888-288-4762, Email: help@autismspeaks.org) – One of the largest autism advocacy organizations in the US, providing research funding, family resources, tool kits, and a helpline to support individuals with autism and their families across all stages of life
- **American autism association** (Telephone: 877-654-4483) – A non-profit organization focused on improving the lives of those affected by autism through direct support programs, educational resources, and community advocacy initiatives
- **Autism in black** (Email: info@autisminblack.org) – An organization dedicated to raising awareness of autism within the Black community, reducing stigma, and providing culturally responsive education, advocacy and support to Black parents and families
- **The color of autism foundation** (Telephone: 313-444-9035, Email: info@thecolorofautism.org) – An organization dedicated to raising awareness of autism within the Black community, reducing stigma, and providing culturally responsive education, advocacy and support to Black parents and families
- **Autism society** (Telephone: 800-328-8476) – One of the oldest and largest grassroots autism organizations in the US, offering advocacy, education, community programming, and a national helpline to connect families with local resources and support
- **Joni and friends** (Telephone: 818-707-5664) – Provides information about disability-friendly churches and runs summer camps for individuals and families of kids with disabilities

## Blindness & visual impairment resources

- **American action fund** (Telephone: 410-659-9315, Email: outreach@actionfund.org)– An organization providing free braille books, educational materials and resources to blind children and adults
- **National braille press** (Telephone: 617-266-6160, Email: contact@nbp.org) – A non-profit braille publisher that produces and distributes braille materials for children and adults, with a mission to promote braille literacy and expand access to information
- **Seedlings braille books for children** (Telephone: 734-427-8552, Email: info@seedlings.org) – A non-profit organization dedicated to improving literacy and educational by providing high-quality braille books free of charge or at a low cost

## Deaf & hard of hearing resources

- **National association of the deaf** (Telephone: 301-328-1443, Email: nad.info@nad.org) – A civil rights organization representing Deaf and hard-of-hearing individuals, advocating for equal access in education, employment, healthcare, and technology
- **National deaf center** (Telephone: 512-471-8283) – A research and training center focused on improving postsecondary education and employment outcomes for Deaf individuals
- **American society for deaf children** (Telephone: 800-942-2732, Email: asdc@deafchildren.org) – An organization that provides information, resources and guidance on communication options and access to services

## Epilepsy support

- **Epilepsy foundation** (Telephone: 800-332-1000) – The leading national non-profit dedicated to the epilepsy community, offering education, a 24/7 helpline, seizure first aid training, advocacy, and funding for research
- **Epilepsy alliance america** (Telephone: 800-642-0500, Email: info@epilepsyallianceamerica.org) – A national organization providing community-based support, seizure first aid training, back-to-school resources, and awareness programs for individuals with epilepsy

## Financial resources

- **Patient advocate foundation** (Telephone: 800-532-5274, Email: help@patientadvocate.org) - Provides hands-on case management support for people who are uninsured or underinsured and living with serious or debilitating illnesses, as well as a range of financial grants to help cover medical costs
- **The assistance fund (TAF)** (Telephone: 855-845-3663) - Helps patients and families who are struggling with high out-of-pocket medical costs by providing financial assistance for copayments, coinsurance, deductibles, and other healthcare-related expenses
- **Accesia health** (Telephone: 800-366-7741, Email: assistance@accessiahealth.org) - Offers financial assistance to help cover the cost of prescriptions, medical treatments and insurance premiums, alongside additional support services including healthcare education, specialized legal services, and case management
- **RARE care** (Telephone: 855-567-3721, Email: help@rarediseases.org) - A collection of assistance programs designed to help patients access lifesaving or life-sustaining medications they could not otherwise afford, including help with insurance premiums and co-pays, diagnostic testing costs and travel assistance for clinical trials or specialist consultations
- **HealthWell foundation** (Telephone: 800-675-8416, Email: grants@healthwellfoundation.org) - A non-profit organization that provides financial assistance to adults and children living in the US to help cover out-of-pocket healthcare costs, including prescription drug coinsurance, copayments, deductibles, and health insurance premiums

- **PAN foundation** (Telephone: 866-512-3861) - Helps underinsured people living with life-threatening, chronic, and rare diseases afford the medications and treatments they need, while also advocating more broadly for improved access and affordability in healthcare
- **finding financial assistance for rare diseases in the US** - A helpful guide put together by the PAN Foundation that walks families through the various financial assistance options available to those living with a rare disease in the United States
- **ThinkGenetic foundation patient assistance program** (Telephone: 866-417-7348, Email: info@thinkgenetic.org) - Provides direct funding to patients and caregivers who need financial support related to a genetic condition
- **Ronald McDonald playhouse** - A non-profit charity dedicated to supporting families with sick children in their time of need, can help with transportation and overnight housing for specialist appointments

## Medical transportation assistance

- **Miracle flights** (Telephone: 800-359-1711, Email: info@miracleflights.org) - Free commercial airline flights for children needing specialized medical care far from home
- **Air care alliance** (Telephone: 888-260-9707, Email: info@aircarealliance.org) - An umbrella organization that connects patients with volunteer pilot organizations across the country that provide free private aircraft flights for medical needs
- **AirCharity network** (Telephone: 877-621-7177, Email: info@aircharitynetwork.org) - The world's largest integrated volunteer pilot network, covering all 50 states, providing free flights to medical facilities for those who are financially unable to travel
- **Lifeline pilots** (Telephone: 800-822-7972, Email: missions@lifelinepilots.org) - A volunteer pilot organization based in Illinois that coordinates free air transportation for passengers with medical or humanitarian needs
- **Mercy medical angels** (Telephone: 757-318-9174, Email: info@mercymedical.org) - The largest charitable medical transportation system in the US, offering free flights through volunteer pilots and commercial airlines, as well as ground transportation through bus tickets, train tickets, and gas cards

## Support for carers

- **Caregiver action network (CAN)** (Telephone: 1-855-227-3640; Email: info@caregiveraction.org) – The nation's leading family caregiver organization, providing free education, peer support, and resources to family caregivers nationwide, serving people who care for loved ones with chronic conditions, disabilities, disease, or the frailties of old age.
- **Family Caregiver alliance – national center on caregiving** (Telephone: 1-800-445-8106; Email: info@caregiver.org) – Works to advance high-quality, cost-effective policies and programs for caregivers in every U.S. state and serves as a central source of information on caregiving and long-term care issues.
- **National alliance for caregiving (NAC)** (Telephone: 202-918-1013) – A nonprofit that conducts research and develops policy to support family caregivers, with reports, advocacy tools, and global caregiving initiatives.
- **AARP caregiving resource center** (Telephone: 1-888-687-2277) – Provides tools, expert advice, and support for family caregivers, including guides, checklists, and access to a community of caregivers.
- **ARCH national respite network and resource center** (Telephone: 703-256-2084) – Connects caregivers with respite services and offers training, advocacy, and research tools, with a national directory to help families locate local respite care.

## 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 specific conditions. 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 I 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**

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.

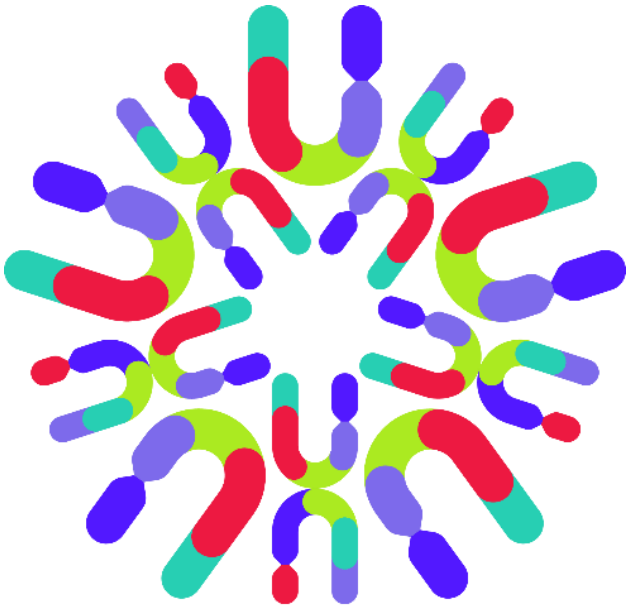
This guide was written by Olivia Morgan (MS, GC) and Unique (NM).

Version 1(NM) 2026



Rare Chromosome Disorder Support Group
Registered in England and Wales

Charity Number 1110661
Company Number 546041



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