UNIQUE
UNDERSTANDING GENES & CHROMOSOMES

# After diagnosis: What happens Next?

## (United Kingdom 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.

***“My child was 25 when he was diagnosed and we’d been searching for the answer to his health, learning and behavioural issues for 13 years. After knocking on numerous doors to seek the answer, the genetic diagnosis still came as a complete shock. Nothing has changed, but our whole world had been turned upside down. 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 (including family, friends and Healthcare Professionals), 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.”***

***“We had test after test - and actually called it playing ‘stump the Children’s Hospital’! After a few years of all testing coming back with no issue, that became the expectation. And though we still saw specialists annually, all the big, bad stuff was off the table. When genetics said they found something, I thought we were going to finally get some real answers.”***

***“The late diagnosis for us hasn't made it any easier, as it is so rare that even genetics couldn't give me any guidance on the future. The information I've been given all relates to younger children, so although it's good for them coming through, it's not for someone like us - we've passed that stage. The only comfort I've got from this diagnosis is knowing it's nothing I've done during pregnancy. So, although we have a name for it, it is still a journey that is unknown for us as a family.”***

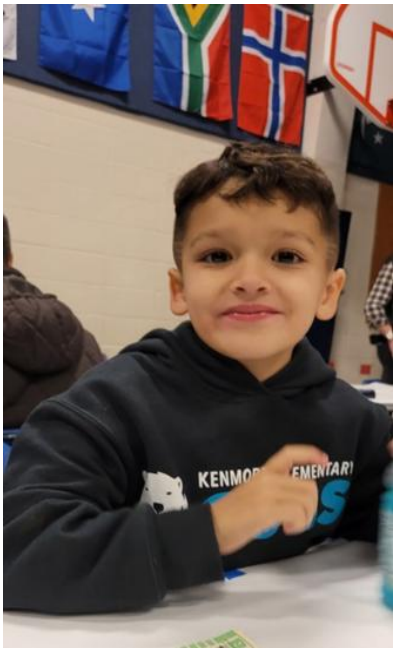
***“I wish someone had told me not to be so afraid. I felt a kind of grief for something I thought I’d lost but it turns out this didn’t take anything away from my son - it actually made him strong and beautiful and unique. He’s not who I thought he would be, he’s so much more than I could ever have 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 a genetic counsellor or genetics clinic
- Ask your doctor about early years support services
- Ask your doctor about referrals to specialists or therapists if needed

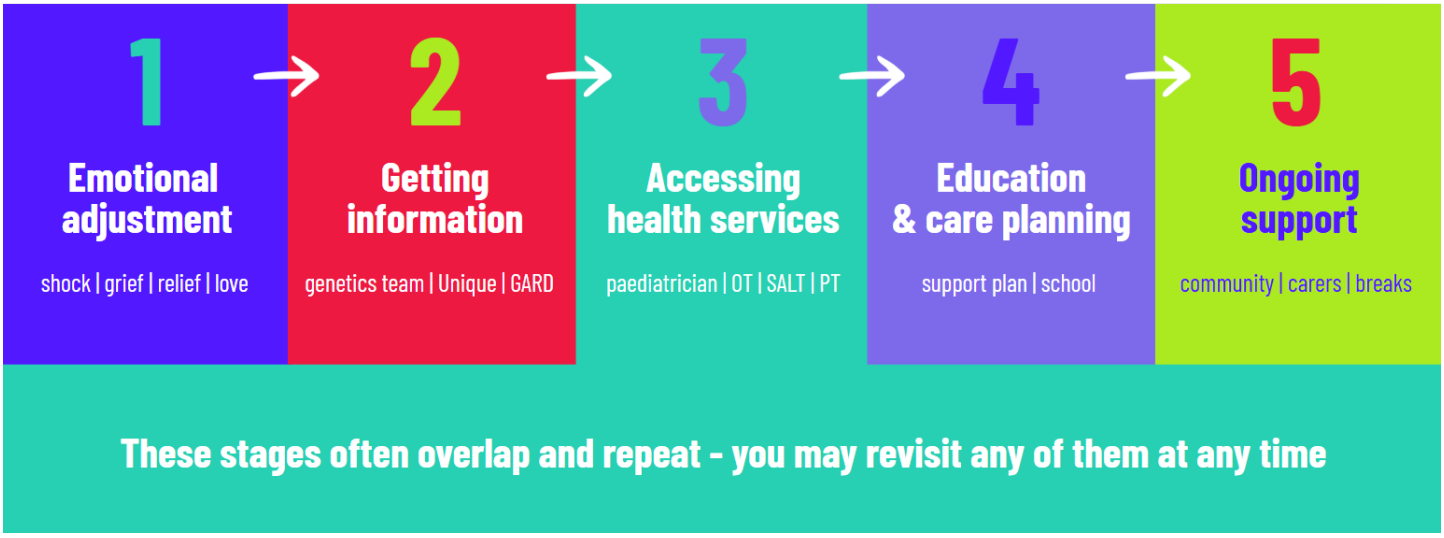


## Months 3–4

- Begin recommended therapies such as speech therapy, occupational therapy or physical therapy
- Connect with condition-specific organisations or support groups

## 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




*"No child comes with a manual. It is a steep learning curve, with life being full of ups and downs along the way. Milestones may not come very quickly but when they do, you will celebrate in a way that only you will understand and appreciate."*

*"For our family, nothing changed. We were already doing PT, OT and Speech therapy, our son was progressing slowly but surely. There was nothing left to do but love our child and watch the daily miracle of his life. We know how hard he has to work each day to be where he's at now. We get the blessing of seeing him achieve things specialists said would never be our reality. It is something special and sacred just for us."*

## 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 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.**

## 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.

A referral to clinical genetics is usually made by a GP, paediatrician or specialist nurse. The specific genetics service you will be seen by depends on your postcode, since services are organised regionally rather than by hospital. Wait times can vary but can take up to 18-weeks before you are seen. 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 years support

Early years support provides assistance and services to infants and toddlers from birth 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.

The early years support services may include speech therapy, occupational therapy, physical therapy, developmental therapy, feeding support and other specialised services depending on a child's individual needs. Early years support is delivered locally and each local authority publishes a **SEND Local Offer** on its website, setting out exactly what is available in your area.

Families can be referred for early years support by a health visitor, GP, paediatrician or hospital consultant, and many services also allow self-referral too. 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.

## Child development centres

A child development centre (CDC) is an NHS community service for children from birth to age 19, where developmental concerns, disabilities or complex health needs are assessed and supported by a team of healthcare professionals in one coordinated service. In some areas, there may not be a single physical centre where all professionals are based. However, children will still receive coordinated support from a team of specialists working together across different locations.

### The team might include:

**Community Paediatricians** specialise in child development, learning and behaviour. Community 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 (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.
**Orthotists** design and fit supportive devices such as braces, splints, orthopaedic footwear or spinal supports to improve alignment, posture, stability and mobility.
**Specialist Nurses** provide ongoing clinical support, monitoring and coordination of care for children.
**Portage Workers** deliver a home-visiting educational service for pre-school children, working with families to build play-based learning activities that support development.
**Dietitians** assess and manage nutrition, feeding and growth concerns.
**Social Workers** help families access community resources, financial assistance programs, educational services and emotional support.
**Psychologist or Psychiatrist** support children and families with behavioural, emotional, developmental or mental health concerns.

The first appointment at a CDC is usually an assessment with a community paediatrician or specialist nurse, covering medical history and current concerns. Based on this, your child may be referred for specific therapies or to other specialist teams.

## 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 over time.

## Children's disability teams

The children's disability team (CDT) is a specialist service that supports children and young people with disabilities and their families. The aim is to help children live as independently as possible and to support families in meeting their child's needs at home and in the community.

You can usually contact your local children's disability team directly through your county council or local authority website. In Scotland, this service is provided by the social work department, and in Northern Ireland by the Health and Social Care Trust. You also have the right to request a carer's assessment to consider your own needs and the support you may require in your caring role.

An assessment helps identify what support may benefit your child and family. A social worker will usually meet with you, often in your home, to discuss your child's needs, your family's circumstances and any challenges you may be experiencing. Following the assessment, they will work with you to develop a care and support plan, which should be reviewed regularly, usually at least once a year.

The children's disability team can also help families access services such as short breaks (respite care), practical support and other resources available within the local community.

## Children's centres / family hubs for SEND children

Children's centres are community hubs offering early years services, such as playgroups, health visitor clinics and parenting support typically for children under the age of five.

In many areas children's centres are being updated into family hubs, which is being rolled out nationally. These hubs extend the age range to cover children and families from birth up to 19, extending to 25 for young people with SEND. They offer SEND-specific stay-and-play sessions, quiet and sensory sessions, speech and language groups, parenting support and links into wider services. Family Hubs often include a dedicated SEND practitioner. SEND practitioners give SEND families a single point of contact to help navigate different services.

Parents and carers can usually contact or self-refer to their local children's centre or family hub. You can check your local authority's family hub or SEND local offer page for what's on offer nearby.

## The UK's learning disability register

The learning disability register is a record held by each individual GP practice in the UK, listing every patient registered at that surgery who has a learning disability. Its purpose is to let practice staff identify children, young people and adults who may need extra help or support to access healthcare.

Being on the register means your child can receive an annual health check from age 14, helping to identify and address health concerns early. It also alerts healthcare staff that your child may need reasonable adjustments, such as longer appointments or accessible communication, to ensure they receive appropriate care. Having an ongoing relationship with a GP and being on the register can also support a smoother transition into adult healthcare. In addition, people on the register are eligible for a free annual flu vaccine.

To be added to the register, ask your GP or the receptionist at your GP practice.

## 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

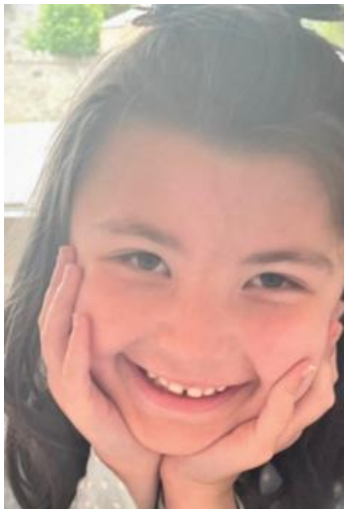
Many children with a chromosome or gene disorder are able to attend their local school, sometimes with little or no additional support. Others may attend their local school but receive additional services and classroom support to help them access the curriculum and participate fully in school life. This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

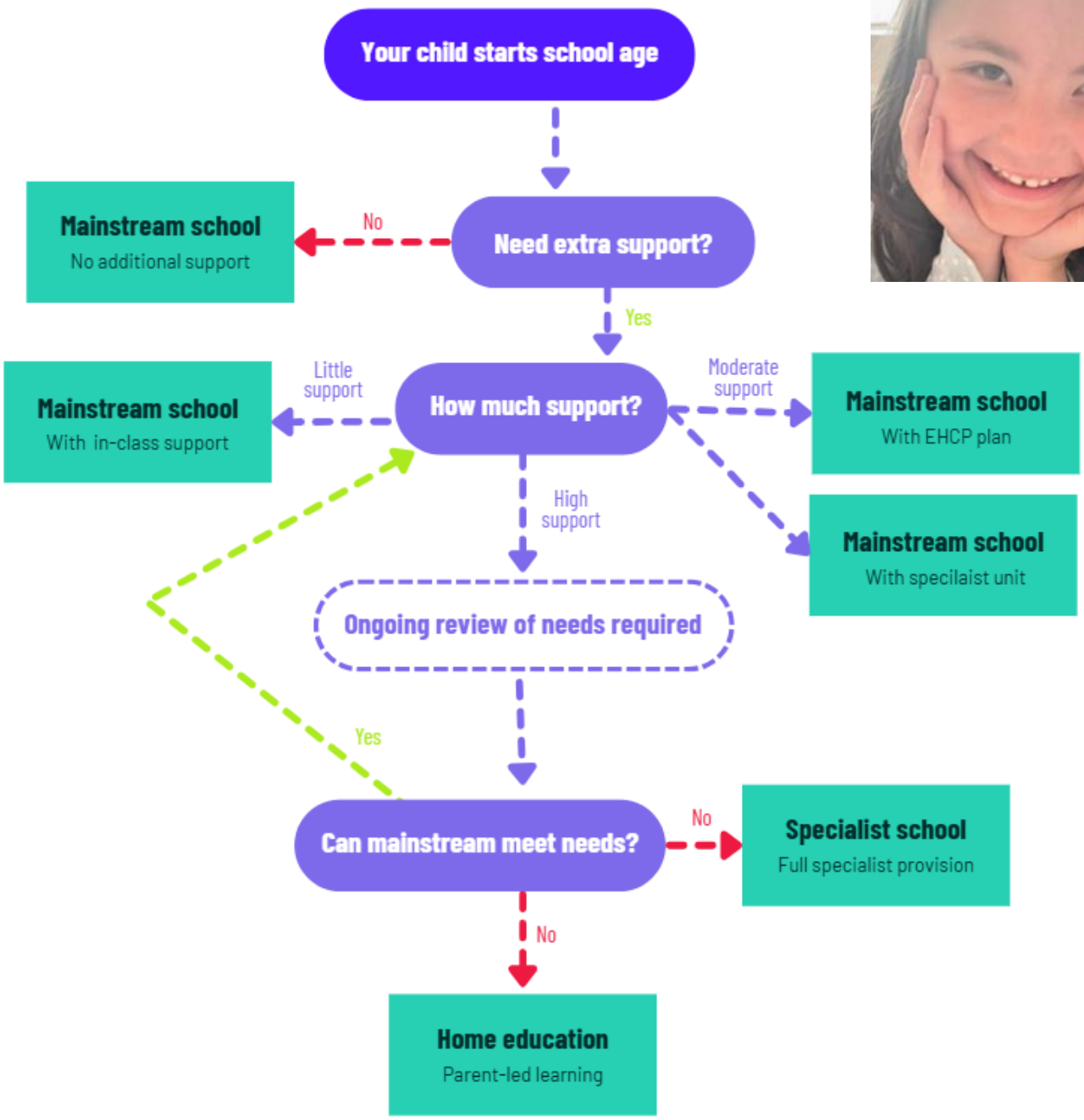
Some children who need more intensive support may benefit from a specialised school setting that is designed to meet complex educational, developmental, medical or physical needs. These schools often provide smaller class sizes, a higher staff-to-student ratio and access to therapists and specialists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

You can find out more about your responsibilities and rights using the educating your child at home portal or download the detailed elective home education: guide for parents. For ideas on different teaching styles, you can also explore the how to home educate in the UK guide.

## Schooling options decision aid






*"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."*



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## Education, health and care plan (EHCP)

In England, children who need extra support at school may qualify for an education, health and care plan (EHCP). An EHCP is for children and young people up to the age of 25 who need additional support. EHCPs identify educational, health and social needs and set out the additional support needed. Similarly, Wales uses an Individual Development Plan (IDP) under the Additional Learning Needs (ALN) system, Scotland uses a Co-ordinated Support Plan (CSP) for children with the most complex Additional Support Needs (ASN) and Northern Ireland uses a Statement of Special Educational Needs. The principles are similar across all four nations, but the names, timescales and appeals processes differ, so check your national government's website or ask your local authority which system applies to you.

If you think your child may need additional support, you can ask your local authority to carry out an assessment for an education plan. A request can also be made by anyone else who thinks an assessment may be necessary, including doctors, health visitors, teachers, parents and family friends. A young person aged 16 to 25 can request an assessment themselves.

If your local authority agrees to carry out an assessment, you may be asked for any reports from your child's school, nursery or childminder, a doctor's assessment of your child and a letter from you about your child's needs.
You may want to talk with your child's teacher or the school's SEN coordinator (SENCO) about any areas where your child may need extra help, such as:

- A specialised learning program 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

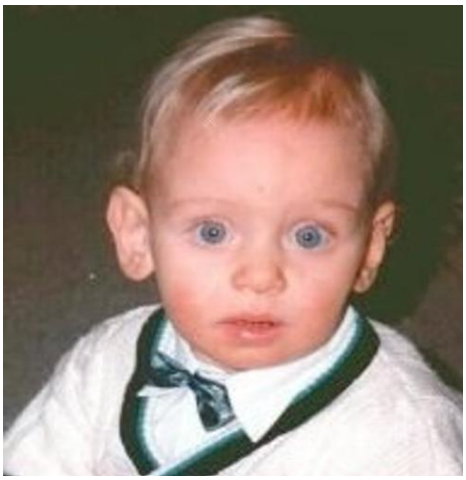
## Further education and beyond

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.

It may be helpful to contact the school or college in good time before your child starts further education, to make sure they can meet your child's needs. The school or college will talk to you and your child to identify the accommodations and support needed, which might include academic assistance, communication support, accessibility arrangements, health-related accommodations, or help with organisation 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 or equipment during the transition.

**Unique publishes a separate guide to Education, Transition and Further Education.**




## Becoming a carer

A carer 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. 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 carer can be deeply meaningful, but also physically and emotionally demanding. Many carers 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 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. 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 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 carers 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 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.

In the UK, help with the cost of respite care can come from several sources. Your local authority may offer short breaks as part of your child's support plan following a Children's Disability Team assessment, and these are often free or means-tested. Direct Payments or a Personal Budget can also be used flexibly to pay for respite, including short breaks, holiday clubs or a support worker. Charitable grants, such as those from Family Fund, can help cover the cost where statutory support is limited or unavailable. To find out what is available in your area, ask your social worker or Children's Disability Team about the local short breaks offer, which should also be listed on your local authority's SEND Local Offer page.

## Financial support

In the UK, there are several programs and funding options that may help families who are raising a child with a disability or additional needs.

Some possible sources of financial and medical support include:

- **Disability living allowance (DLA) (England, Wales, NI) / Child disability payment (Scotland)** – For children under 16 who need significantly more care or supervision than other children their age or who have mobility difficulties. DLA includes a Care component and a Mobility component. In Scotland, Child Disability Payment has replaced DLA
- **Personal independence payment (PIP) (England, Wales, NI) / Adult disability payment (Scotland)** – For people aged 16 and over. PIP provides support through Daily Living and Mobility components. In Scotland, Adult Disability Payment (ADP) has replaced this
- **Carer's allowance** – Financial support for those who spend a significant amount of time caring for a disabled person
- **Carer's credit** – National Insurance credits for carers who are not eligible for Carer's Allowance, helping to protect future State Pension entitlement
- **Employment and support allowance (ESA)** – Young people aged 16 and over who have a disability or health condition may be eligible for ESA, depending on their circumstances
- **Universal credit (UC)** – May provide additional financial support for families on a low income, including disability-related elements
- **Direct payments and personal budgets** – Funding provided by local authorities to give families greater choice and control over how care and support services are arranged
- **Housing and council tax support** – Families may be eligible for Disabled Facilities Grants to fund home adaptations, as well as Council Tax reductions, Housing Benefit, or other local authority support

For advice on eligibility and applications, families can contact the Department for Work and Pensions (DWP), their local authority, or Citizens Advice.

## Grants and funding

There are many different charities that can help families of children with additional needs. Each has its own criteria for how much support they can offer, and to whom.

- **Family fund** (Telephone: 01904 550055, Email: info@familyfund.org.uk) - the UK's largest charity providing grants for families raising disabled or seriously ill children and young people
- **Warm home discount** scheme - offered by most energy companies once a year, usually between October and March, to help with fuel bills. It is not paid directly to you but taken off your electricity bill or added to your pre-payment meter. Contact your energy provider to check if you qualify

## Vehicles and transport

- **Vehicle tax exemption** – If your child receives the higher-rate Mobility component of Disability Living Allowance (DLA) or the enhanced-rate Mobility component of Personal Independence Payment (PIP), you may be eligible for exemption from paying vehicle tax
- **Blue badge scheme** – Provides parking concessions for people with disabilities or mobility difficulties, helping them access services and facilities more easily
- **Disabled persons railcard** – Offers discounted rail travel for eligible disabled people and an accompanying adult, helping to reduce the cost of train journeys
- **Motability scheme** – Allows eligible individuals to lease a car, wheelchair-accessible vehicle, scooter, or powered wheelchair using their higher-rate Mobility component of DLA or enhanced-rate Mobility component of PIP. The scheme can help improve independence and access to everyday activities

## Creating a health record folder

Many families find it helpful to keep all of their child's medical, therapy, educational and insurance 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 health record folder 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
- Notes from appointments or questions for future visits

Bringing your health record folder 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.

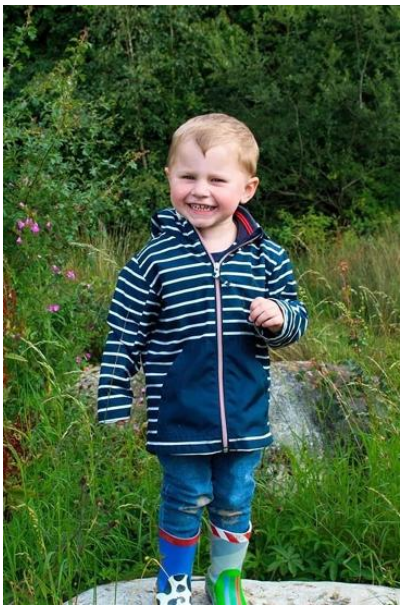
## A hospital passport

A **hospital passport** (sometimes called an "all about me" or "this is me" document) is a short, practical summary that travels with your child to any hospital admission, A&E visit or appointment with staff who don't already know them. It typically covers things like how your child communicates, what helps them feel calm or causes distress, sensory sensitivities, mobility or feeding needs, current medications and simple do's and don'ts for staff. Many hospital trusts have their own template for a hospital passport, and some charities provide free downloadable versions. It's worth updating your child's hospital passport every so often as your child grows and their needs change and keeping a copy both in your care binder and on your phone.

# 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 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. 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 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 in the wider medical community.

## Advice for families

***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."***

## 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 alliance UK** (Telephone: 0300 124 0441, Email: contactus@geneticalliance.org.uk) - An alliance of over 200 UK charities and support groups for people with genetic, rare and undiagnosed conditions, running the Rare Disease UK and SWAN UK campaigns
- **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 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, 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)

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

## Education and SEND resources

- **National portage association** (Telephone: 0121 244 1807, Email: info@portage.org.uk) - A free home-visiting educational service for pre-school children (aged 0–4) with additional support needs and their families. Portage practitioners work alongside parents to promote children's learning and development through play. Parents can often self-refer where a local service is available
- **Alliance for inclusive education (ALLFIE)** (Telephone: 020 7737 6030, Email: info@allfie.org.uk) - A national charity campaigning for an inclusive education system where disabled children and young people can learn alongside their peers in mainstream settings. It also provides information, training and policy resources
- **Early education (British association for early childhood education)** (Telephone: 01727 884925, Email: office@early-education.org.uk) - Provides evidence-based resources, professional guidance and downloadable publications to support high-quality early years education and child development
- **Council for disabled children** (Telephone: 020 7843 1900, Email: enquiries@ncb.org.uk) - Offers guidance, resources and practical information to help families understand Education, Health and Care (EHC) plans, SEND legislation and local services
- **IPSEA (independent provider of special education advice)** (Telephone: 0300 222 5899, Email: office@ipsea.org.uk) - Provides free, independent legal advice and support to families of children and young people with SEND, helping them secure the educational provision they are entitled to by law
- **SEND local offer** - Every local authority publishes a SEND Local Offer, outlining education, health, social care, leisure and support services available for children and young people with SEND in the local area
- **Pyramid educational consultants UK ltd** (Telephone: 01273 609555, Email: pyramiduk@pecs.com) - UK-developers of PECS (Picture Exchange Communication System) which is an alternative / augmentative communication system that teaches students to initiate spontaneous communication in a social context

## Behaviour and emotional wellbeing

- **Child and adolescent mental health services (CAMHS)** - Specialist NHS mental health services for children and young people experiencing significant emotional, behavioural or mental health difficulties. Referrals are usually made through a GP, school or other professional
- **YoungMinds parents helpline** (Telephone: 0808 802 5544, Email: parents@youngminds.org.uk) - Offers free advice and emotional support for parents and carers who are concerned about their child's mental health or who are waiting for CAMHS support
- **Challenging behaviour foundation** (Telephone: 0300 666 0126, Email: support@thecbf.org.uk) - Supports families and professionals caring for children and adults with severe learning disabilities whose behaviour challenges. Provides practical guidance, training and a family support service

## Carers support

- **Carers trust** (Telephone: 0300 772 9600, Email: info@carers.org) - Supports unpaid carers through local services, information, practical help and respite opportunities
- **Carers UK** (Telephone: 0808 808 7777, Email: advice@carersuk.org) - Provides expert advice on carers' rights, benefits, employment and wellbeing, while campaigning to improve support for unpaid carers
- **Working families** (Telephone: 0300 012 0312, Email: advice@workingfamilies.org.uk) - Information for working parents and carers on their employment rights; Tax Credits and in-work benefits; maternity and paternity leave; flexible working options; and maternity discrimination
- **National network of parent carer forums** (Telephone: 0207 608 8708, Email: info@nnpcf.org.uk) - A network of over 150 parent carer forums from across England
- **Netmums** (Email: help@netmums.com) - A family of local websites for anyone involved in caring for young children

## Benefits and financial support

- **Disability living allowance (DLA) for Children** – A helpful overview on the government website explaining the DLA process
- **Cerebra DLA guide** (Telephone: 0800 328 1159, Email: enquiries@cerebra.org.uk) - A practical, step-by-step guide to completing a Disability Living Allowance application for children
- **Personal Independence payment (PIP) -** A helpful overview on the government website explaining the PIP process
- **Benefits and work** (Email: info@benefitsandwork.co.uk) - Provides detailed guides and information to help people claim benefits and appeal decisions
- **Citizen's advice** (Telephone: 0800 144 8848) - Offers free, independent advice on benefits, debt, housing, employment and legal issues
- **Disability rights UK** (Telephone: 0330 995 0400, Email: enquiries@disabilityrightsuk.org) - Provides expert information on disability rights, welfare benefits, independent living and social care
- **EntitledTo** - Free benefits calculator to estimate what financial support a family may be entitled to
- **Turn2us** - Helps people access welfare benefits, charitable grants and wider financial support.
- **Benefits and work** (Email: info@benefitsandwork.co.uk) - provides information and guides to help with benefits claims and appeals

## Continence and toileting

- **Bladder and bowel UK** (Telephone: 0161 214 4591, Email: bbuk@disabledliving.co.uk) - Provides expert advice, information and educational resources on bladder and bowel health, continence and toilet training
- **ERIC (the children's bowel & bladder charity)** (Telephone: 0808 801 0343, Email: helpline@eric.org.uk) - Offers advice, resources and support and young people experiencing continence difficulties
- **National key scheme (RADAR keys)** - Enables disabled people to access thousands of locked accessible public toilets across the UK using a universal key
- **Changing places** (England, Wales and NI, Telephone: 020 7803 4814, Email: changingplaces@musculardystrophyuk.org), (Scotland, Telephone: 01382 385 154, Email: changingplaces@pamis.org.uk) - Campaigns for and maps fully accessible toilets equipped with hoists and changing benches for people with profound disabilities

## Communication and specialist equipment

- **Makaton** (Telephone: 01276 606760, Email: info@makaton.org) - A communication programme using signs, symbols and speech to support people with communication and learning difficulties
- **PECS UK (pyramid educational consultants)** (Telephone: 01273 609555, Email: pyramiduk@pecs.com) - Provides training and resources for the Picture Exchange Communication System (PECS), helping children develop functional communication skills
- **REMAP** (Telephone: 01732 760209, Email: data@remap.org.uk) - Creates bespoke equipment and adaptations where commercial products do not meet a disabled person's needs
- **Signalong** - Provides signing resources and training to support children and adults with communication difficulties
- **Fledglings** (Telephone: 0203 319 9772, Email: enquiries@fledglings.org.uk) - Helps families find specialist products, equipment, clothing, toys and practical solutions for children with additional needs
- **Newlife** (Telephone: 0800 902 0095, Email: info@newlifecharity.co.uk) - Provides specialist equipment, emergency grants and nurse-led support for disabled and terminally ill children
- **MERU** (Telephone: 01372 725 203, Email: info@meru.org.uk) - a charity that designs and custom-makes specialist equipment for use at home, at school or college, in hospital or at play
- **REMAP**(Telephone: 01732 760209, Email: data@remap.org.uk) - Makes and adapts equipment for disabled people where the exact product cannot be commercially made

## Condition-specific organisations

- **Child growth foundation** (Telephone: 020 8798 2139, Email: info@childgrowthfoundation.org) - Supports children with growth conditions and provides information for families and professionals
- **Epilepsy action** (Telephone: 0808 800 5050, Email: helpline@epilepsy.org.uk) - Offers information, advice and support for people of all ages affected by epilepsy
- **National centre for young people with epilepsy (young epilepsy)** (Telephone: 01342 832243, Email: info@youngepilepsy.org.uk) - Provides specialist education, health services and support for children and young people with epilepsy
- **Mencap** (Telephone: 0808 808 1111, Email: helpline@mencap.org.uk) - Works with people with learning disabilities and their families, providing advice, support and campaigning for equal opportunities
- **National autistic society** (Telephone: 0207 833 2299, Email: nas@nas.org.uk) - Provides information, advice, training and support for autistic people and their families
- **CRY-SIS** (Telephone: 08451 228 669, Email: info@cry-sis.org.uk) - Supports families of babies who cry excessively, offering practical advice and emotional support
- **Heartline** (Telephone: 07949 256302, Email: intouch@heartline.org.uk) - Support for children with heart disorders, and their families

## General family support and disability organisations

- **Cerebra** (Telephone: 0800 328 1159, Email: enquiries@cerebra.org.uk) - Supports families of children with neurological conditions through advice, research, practical guides and equipment services
- **Contact** (Telephone: 0808 808 3555, Email: helpline@contact.org.uk) - A national charity providing advice, emotional support and information for families of disabled children
- **Disability rights handbook** (Telephone: 0330 995 0400, Email: enquiries@disabilityrightsuk.org) - contains comprehensive information and advice on benefits, tax credits, social care and other disability-related issues
- **Home-Start UK** (England, Telephone: 0116 464 5490, Email:info@home-start.org.uk), (Northern Ireland, Telephone: 07718 912 772, Email: jmurray@home-start.org.uk), (Scotland: Telephone: 0131 281 0879, Email: scotland@home-start.org.uk), (Wales, Telephone: 029 2036 0876, Email: info@homestartcymru.org.uk) - Offers volunteer-led home support to families with young children facing a range of challenges
- **WellChild** (Telephone: 01242 530007, Email: info@wellchild.org.uk) - Supports seriously ill children and their families, helping children with complex health needs receive care at home where possible
- **Special Needs Jungle** - Provides accessible information, guides and news to help families navigate the SEND system

## Palliative care and bereavement support

- **Together for short lives** (Telephone: 0808 8088 100, Email: helpline@togetherforshortlives.org.uk) - The UK charity for children's palliative care, helping families find their local children's hospice and offering a family helpline
- **Child bereavement UK** (Telephone: 0800 02 888 40, Email: enquiries@childbereavementuk.org) - Supports children, young people, parents and families when a child grieves or when a child dies

## 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 I rarechromo.org

## Join Unique for family links, information and support:

**Become a member**

## Please help us to help you!

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If you can, please make a donation:

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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 of the parents that contributed towards this guide.

 This guide was compiled by Unique (MM) and reviewed by Dr Catherine Tuffrey, Consultant Paediatrician, SOLENT NHS TRUST, and updated by Unique (NM)

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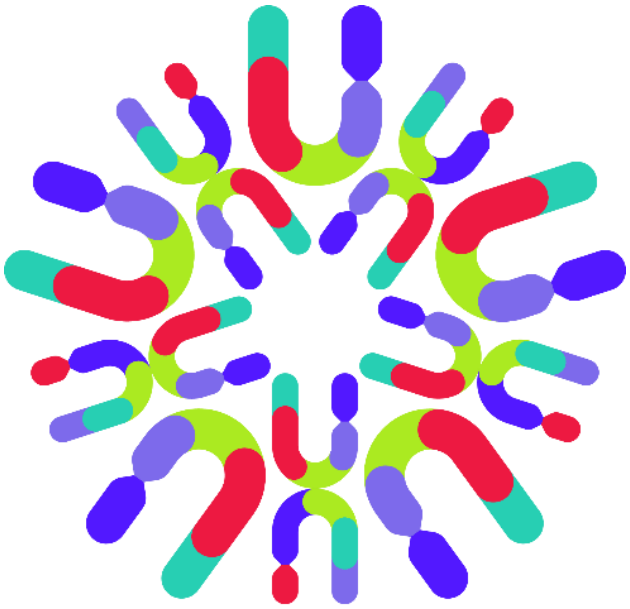

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