



Understanding Chromosome & Gene Disorders

After diagnosis: what happens next?



rarechromo.org



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After Diagnosis—what happens next?

Your child has just been diagnosed with a chromosome or gene disorder.

You are probably going through a whole range of different emotions right now. You may feel shock, anger, sadness, guilt, loss, a feeling of being unsure (of what's to come), isolated, or even relief that you finally have a reason why your child is 'different'. It may seem a scary place at the moment and you probably feel like you are on a rollercoaster with its many twists, turns and ups and downs. The important thing to remember is that your child is still your child and needs



love and care, just like any child. The rewards can be just as satisfying as they would be with any child. You will feel sadness at times but you will also experience incredible joy. **It is important to remember that each child is unique and not every family will need all of this information. But if parents do need help, they should reach out for support and not feel alone.**

Trying to cope with your child's condition and adjusting your own family life can be very hard. Every parent wants to do the very best they can for their child but knowing where to start, if they have been diagnosed with global developmental delay as a baby or young child, can be difficult. If the diagnosis of a rare chromosome or gene disorder comes later on in a child's life, it can be equally challenging.

The period leading up to diagnosis and getting the correct diagnosis for your child can be a very difficult time. Most parents want a diagnosis for their child; at the very least it is an explanation of why your child isn't developing at the same rate as other children of the same age. You know that something isn't quite right, and you want to know why. In some cases, the diagnosis helps you to know what problems your child could face in life and how to find help for your child. A diagnosis can also be important as it may lead to you getting the right services and support for your child, such as healthcare, therapies and education.

This guide could also be of help to families who have yet to receive a formal diagnosis for their child.

This guide contains information on services available to families with a child with additional needs, whether they are mildly, moderately or severely affected, either mentally, physically or both. The links towards the end of the guide should help you with any further information you might need.

This guide is aimed at parents living in England, there will be some differences in services for families living in Wales, Ireland and Scotland.

We also have a **Non-UK version** of this guide, which is available on our website.

Chromosome and Gene Disorders

Chromosome and gene disorders can be so rare that most people will not have heard of them. This can lead to frustration when trying to talk to



Diagnosed at
1 year

people about your child. Even once a diagnosis has been received many families continue to feel isolated and lost. This can be because their child's disorder is made up of a series of numbers and letters, which can be difficult for them to understand, let alone try to explain to other people. It can feel a bit like you are wading through mud at times.

Adjusting to a life you weren't planning for can be very stressful and your diary may seem like it is full of appointments, especially if your child is very young and not yet accessing services such as therapies. While some friends and family members may be very supportive, others might find it difficult to understand, some may not know what to say and become less close because of this.

“ 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. ”

Questions

You may have lots of questions, depending on where you are in your rare genetic condition journey. Initial questions might include:

- Will my child be OK?
- What does the genetic test result mean?
- Why did this happen?
- Is there a cure?

Information regarding these questions will be provided by your genetics team. Other questions may include:

- We haven't seen a geneticist yet, what does that involve?
- How can I get help for my child's development?
- Where will my child go to school?
- Where can I go for local support?
- I have a job. Will I have to give up my career to look after my child?
- Will I be entitled to any financial support to help support my child?

For answers relating to the questions above, please read on.....

This guide has been designed to give you basic information on where you can obtain further advice about raising a child with a genetic condition. Getting the right support will help your child and your family to live as happy and fulfilling lives as possible.

Development - how can I get help for my child?

Seeing a Clinical Geneticist

We haven't seen a geneticist yet, what does that involve?

Clinical Geneticists advise on appropriate management of rare genetic disorders, genetic testing and consequent screening of 'at risk' family members. There are regional genetic clinics all over the UK. Some follow-up appointments might be available at your local hospital. Attending a genetics centre usually requires a referral from your doctor or hospital specialist. You may have a bit of a wait for your appointment and you may only see a geneticist once in your child's life.

Unique has a separate guide on [A Clinical Genetics Appointment](#) which you might find helpful. It is freely available to download from the Families section of our website.

Early Years Support

Early Years Support is a term used in the UK for children aged five and under with disabilities or additional needs. It brings together all of the services and support available from different agencies, making it easier for families to coordinate their child's health, education and social care needs.



If you are worried about your child's development, you should contact your family doctor (GP), health visitor or other health care professional, who will be able to talk to you about your concerns. A doctor is usually able to refer your child to other services if necessary.

These services might include:

- Specialist children's community nurses (covering specific health issues such as epilepsy, tube-feeding and complex care needs)
- Specialist continence services

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



Child Development Centres - assessments, treatments and therapies

Your child may be referred to a **Child Development Centre (CDC)**, this usually houses a range of professionals who specialise in helping children who have a wide variety of symptoms and features. These professionals usually work together as a team and offer assessments and ongoing treatments and therapies as required to children with a wide variety of difficulties including physical disabilities, learning disabilities, developmental delay, speech delay and health concerns, such as epilepsy. It provides a multi-disciplinary service for children from birth to 19 years of age. In some areas, there may not be a physical building or 'centre' where all professionals work, but children will still be seen by a team of professionals in a similar way.

The team will usually include:

A **Community Paediatrician** is a children's doctor who specialises in childhood disability, developmental disorders and complex needs. Community Paediatricians regularly liaise with nurseries, schools, school nurses and the Education Authority to advise about a child's medical needs.

A **Paediatric Occupational Therapist** or 'OT' works with children to make the most of their potential in specific areas (such as self-help skills including feeding and dressing) and to increase their independence at home, school and in their recreational activities. Your child may be referred to see an OT individually, or the OT may train other professionals, for example, education staff, to help your child in these areas.

A **Paediatric Physiotherapist**. A 'PT' assesses and treats children to improve their physical skills, including their posture and mobility.

A **Paediatric Speech and Language Therapists**. A 'SALT' assesses and treats children's speech and language, including the use of manual signs and symbols. SALT's can also advise if children have chewing or swallowing difficulties.

An **Audiologist**. These are experts who can help to prevent, diagnose and treat hearing and balance disorders. Audiologists screen individuals to identify possible hearing disorders. Testing will confirm if hearing loss is present and determine the kind and degree of loss.

An **Orthotist**. These are health care professionals who are trained to prescribe, design, fit and monitor orthoses (devices that aim to help provide support or structural alignment of movable parts of the body, e.g. the limbs and spine). Orthotists can also recommend specialist footwear and insoles.

Other professionals may also be involved e.g. a **Neurologist** who treats disorders that affect the brain, spinal cord and nerves, such as epilepsy.

A **Paediatric Psychologist** may also be involved if behavioural difficulties are apparent and an assessment is required (e.g. an assessment for Autism Spectrum Disorder [ASD]).

What will happen at the first appointment at the Child Development Centre?

The first appointment with the paediatric team usually takes about an hour/ hour and a half. The paediatrician or specialist nurse, will take a detailed history, watch your child while they play and examine them. They will do a developmental assessment. Your child may be seen by another member of the team first, for example a Speech and Language therapist, who will carry out an assessment, relevant to their expertise.

Plans and reviews

The team will make a plan with you about how to help your child. They may discuss referring your child to various therapists. They may arrange a review appointment to see how things are going at an agreed date, which might be three months, six months or annually. Some children will be followed up by the Community Paediatric Service for many years, sometimes until they reach adulthood.

A paediatrician can also refer your child to the:

Children's Disability Team (social care)

What is the Children Disabilities Team?

The Children's Disabilities Team (CDT) is a specialist service that aims to provide parents with support so that they can bring up their child at home, leading as full and independent a life as possible. You can contact the CDT directly by looking on your county council website. As a parent, you have the right to ask for your child's needs to be assessed by the social services department. In Scotland this is known as the social work department and in Northern Ireland it is the health and social services trust. You also have the right to ask for an assessment for yourself. This is known as a carers assessment. An assessment could lead to services for your child and services to help you as their carer. Assessments are the first stage in sorting out what help you need from social services. A social worker will carry out this assessment and organise any care and support that is agreed. A social worker will probably visit you at home to talk about what support and services you need. They will work with you to develop a care and support plan, which should be reviewed annually. The social worker can also help you access short breaks for your child.



“ 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. ”

Children's Centres



Differing from a Child Development Centre, there are also children's centres. These centres can provide a variety of advice, information and support for parents and carers with children aged 4 and under, including parent workshops and drop-in groups. You can locate your nearest children's centre by contacting your local Family Information Service, or by visiting: <https://www.gov.uk/find-sure-start-childrens-centre>
Parents can self-refer.

“ Late diagnosis – 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.”

Education

Worries about your child's educational development

Your child learns through being with other people and exploring the world around them. However, some children may have more difficulties than other children their age with:

- Communication
- Understanding and learning
- Sensory and physical development
- Behaviour or relating to other people

Children with this type of learning disability are said to have **'special educational needs' (SEN)**.

Your child may need an education, health and care plan (EHCP), if they need more support than their school provides. An EHCP is for children and young people aged up to 25 who need more support. EHCPs identify educational, health and social needs and set out the additional support needed.

In Scotland, information can be found here:

Website: <https://www.gov.scot/policies/schools/additional-support-for-learning/>

Website: <https://enquire.org.uk/>

Wales Website: <https://www.gov.wales/special-educational-needs>

Northern Ireland Website:

<https://www.eani.org.uk/parents/special-educational-needs-sen>



Requesting an EHC assessment

You can ask your local authority to carry out an assessment if you think your child needs an EHC plan. A young person can request an assessment themselves if they are aged 16 to 25. 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. 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 doctors' assessments of your child
- A letter from you about your child's needs

The local authority will tell you within 16 weeks whether an EHC plan is going to be made for your child.

Children aged under five

SEN support for children under five includes:

- A written progress check when your child is two years old
- A child health visitor carrying out a health check for your child if they're aged two to three years old
- A written assessment in the summer term of your child's first year of primary school
- Making reasonable adjustments for disabled children, such as providing aids like tactile signs

Nurseries, playgroups and childminders registered with Ofsted follow the Early Years Foundation Stage (EYFS) framework. The framework makes sure that there is support in place for children with Special Educational Needs and/or Disabilities (SEND). Talk to a doctor or health adviser if you think your child has SEND but they don't go to a nursery, playgroup or childminder. They will tell you what support options are available.

■ National Portage Association

Website: <https://www.portage.org.uk/>

Telephone: 0121 244 1807

Email: info@portage.org.uk

A free home-visiting educational service for pre-school children aged four and under with additional support needs and their families. It is available in many parts of England. Parents can self-refer.

■ The Alliance for Inclusive Education

Website: <https://www.allfie.org.uk/>

Telephone: 0207 737 6030

Email: [via website](#)

A campaigning group promoting inclusive education for all children.

■ The British Association for Early Childhood Education

Website: <https://early-education.org.uk/>

Telephone: 01727 884925

Email: office@early-education.org.uk

Provides useful resources and downloadable publications.

■ Council for disabled children

Website: <https://councilfordisabledchildren.org.uk/resources/all-resources>
Resources for parents to navigate the Education, Health & Care planning process.

Children aged between five and fifteen

Talk to the teacher or the SEN co-ordinator (SENCO) if you think your child needs:

- A special learning programme
- Extra help from a teacher or assistant
- To work in a smaller group
- Observation in class or at break time
- Help taking part in class activities
- Extra encouragement in learning e.g. to ask questions or to try something they find difficult
- Help communicating with other children
- Support with physical or personal care difficulties e.g. eating, getting around school safely or using the toilet.



■ IPSEA

Website: <https://www.ipsea.org.uk/>

Telephone: 0300 222 5899

Email: office@ipsea.net

IPSEA offers free and independent legally-based information, advice and support to help get the right education for children and young people with all kinds of special educational needs and disabilities (SEND).

Young people aged sixteen or over in further education

Contact the school/college in good time before your child starts further education to make sure that they can meet your child's needs. The school/college will talk to you/your child about the support they need.

Unique has separate more detailed guides on [Education, Transition](#) (for children aged 13 years plus), and [Further education, work and Training](#) (for school leavers), which you might find helpful. They are available to download from our website in the Families section.

Home Education

For some children, home education can work better. Many Unique parents home-school their child with a rare genetic condition successfully.

SEND Local Offer

Also check out your SEND local offer on your county council website. The Local Offer gives children and young people with SEND and their families, information about what services are available in your local area.

Behavioural challenges

Children with a chromosome or gene disorder sometimes have what is described as 'challenging behaviour' or 'behaviour that challenges'. It can be difficult to describe it fully. It could be something sensory e.g. a reaction to doing things in a certain way, or not wanting to touch certain things. In many cases, challenging behaviour occurs because the person is frustrated at their inability to make others understand what they need or want. They could be in pain and not able to express where the pain is, or how bad it is. Some children with a chromosome or gene disorder may have challenging behaviour in their early years that improves as they get older. For others the behaviour can develop as they get older. An assessment may be recommended.

■ CAMHS (Child and Adolescent Mental Health Service)

Website: <https://www.youngminds.org.uk/young-person/blog/what-to-expect-from-a-camhs-assessment/>

Parents Helpline: 0808 802 5544 (free for mobiles and landlines)

Email: parents@youngminds.org.uk

CAMHS are specialist NHS children and young people's mental health services. If you think your child might need more help than friends, family, school and GPs can give. Or perhaps you and your child are on the CAMHS waiting list or have an appointment soon. So you know what to expect, Young Minds have put together information especially for parents and carers. YoungMinds Parents' Helpline is a telephone and online support service, they do not support families face to face, or provide mental health assessments. If this is what you are looking for, you will need to get a referral to your local CAMHS. Many CAMHS services have now trained other teams to help children and young people with mental health challenges rather than seeing all children individually. Your GP, community medical team or school will be able to advise on what is available locally.

■ Challenging Behaviour Foundation

Website: <https://www.challengingbehaviour.org.uk/>

Family Support Line: 0845 602 7885

Email: support@theCBF.org.uk

Unique has a separate guide on [Challenging Behaviour](#) issues which you might find helpful. It is freely available to download from the Families section of our website.

“ I wish someone had told me not to be so afraid. I felt a kind of grief for something I thought I'd lost, 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. ”

Being a carer

What is a carer? A carer is someone who looks after a child, relative, partner, friend or neighbour who has a disability. This could be a mental or physical disability, or a combination of both. Whilst you would naturally expect to care for your own child, if your child has additional needs that require more assistance than that of a typically-developing child of the same age, then you will be classed as a 'carer'. Being a carer may entitle you to services and help from the authorities. There may also be support available to young carers (siblings who help to care for their brother or sister). To find out if you have a carers support service nearby, visit your council website or contact one of the following:

■ Carers Trust

Website: <https://carers.org/help-and-info/introduction>

Telephone: 0300 772 9600

Email: info@carers.org

Carers Trust is a major charity for, with and about carers.

■ Carers UK

Website: <https://www.carersuk.org/>

Telephone: 020 7378 4999

Email: info@carersuk.org

Provides support for all carers and campaigns to improve the lives of carers.



Unique has separate guides on [Carers Wellbeing](#) and [Supporting Siblings of Children with a Rare Genetic Conditions](#) which you might find helpful. They are freely available to download from the Families section of our website.

Getting a Break from Caring

Short breaks increase the quality of life for children with genetic conditions and their families by giving parents a break, or providing an activity for a child with their peers separate from their parents. They provide children with additional needs with opportunities to meet new people, make new friends and experience different activities. Many families will have extended family and friends who can help to look after their child, so that they can have a break, but many will have very little support. It is important to get a break as much as possible, even if just for a few hours. You can either contact your local Child Disability Team (Social Services) or carers support service or ask your GP or paediatrician to refer you.



Local Support

Where can I go for local support?

Parents of children with chromosome or gene disorders, or any special needs are often referred to as 'carers' or 'parent carers'. Every town in England and throughout the UK should have a carers support service. It is worth registering with them as a '**Carer**' as they run groups to bring carers or parent carers together, hold workshops relevant to raising a disabled child, have access to therapists who help carers, and have subsidised treatments like Reiki, massage and aromatherapy.

Websites/social media

There are many different chromosome/gene specific websites and Facebook groups.

Working Parents

I have a job; will I have to give up my career to look after my child?

Looking after a child with a chromosome or gene disorder and working can be very difficult; it can put a huge strain on family life. You may have an arrangement with a family member to care for your child whilst you work, or you may have them looked after by a childminder or agency. It might become increasingly difficult to work as you did prior to having your child. If you are worried about your job, ask your employer if they can be flexible, to allow you to still work but also attend any appointments that your child may need. If you feel you cannot work and you care for them for at least 35 hours per week and you receive middle or high rate Disability Living Allowance (DLA) or PIP for your child, you can apply for Carer's Allowance. Unlike DLA and PIP, Carers Allowance is a means-tested benefit and how much you might get will depend on your current earnings.

■ Carers Allowance

Website: <https://www.gov.uk/carers-allowance>

Telephone: 0800 731 0297

■ Carers Credit

Website: <https://www.gov.uk/carers-credit>

You might be eligible for Carer's Credit if you're not eligible for Carer's Allowance. Carer's Credit is a National Insurance credit that helps with gaps in your National Insurance record. Your State Pension is based on your National Insurance record.

■ Working Families

Website: <https://workingfamilies.org.uk/>

Telephone: 0300 012 0312 Free helpline for parents and carers

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. This website includes a section especially for parents of disabled children.



Welfare Benefits

Will I be able to claim any benefits to help support my child?

The main benefit that you can apply for to help your child is called the **Disability Living Allowance (DLA)**. DLA for children may help with the extra costs of looking after a child who:

- Is under 16
- Has difficulties walking or needs much more looking after than a child of the same age who doesn't have a disability

Your child will need to meet all of the eligibility requirements. You can claim DLA for children if you are in or out of work as it is not means tested. DLA is made up of two components (parts): Care and Mobility.

Care component

The rate the child gets depends on the level of looking after they need, for example:

- Lowest rate - help for some of the day or night
- Middle rate - frequent help or constant supervision during the day, supervision at night or someone to help while your child is on dialysis
- Highest rate - help or supervision throughout both day and night, or if your child is terminally ill

Mobility component

The rate the child gets depends on the level of help they need getting about, for example:

- Lowest rate - your child can walk but needs help and or supervision when outdoors
- Highest rate - your child can't walk, can only walk a short distance without severe discomfort, could become very ill if they try to walk or they're blind or severely sight impaired

The mobility component can be claimed from the age of three years if eligible. Your child must need a lot more help or supervision than other children of the same age. **The Department for Work and Pensions (DWP)** can also arrange for someone to help you fill out the form if required, or you can contact your local Citizens Advice Bureau (CAB) to ask for help.

■ **Disability Living Allowance (DLA)**

Website: <https://www.gov.uk/disability-living-allowance-children>

Telephone: 0800 121 4600

■ **Cerebra guide to DLA**

Website:

<https://cerebra.org.uk/download/disability-living-allowance-dla-guide>

A step-by-step guide to claiming Disability Living Allowance for children under 16.

If your child is 16 years or over, DLA is replaced by a benefit called the **Personal Independence Payment (PIP)**. The components of PIP are slightly different to DLA, as is the application process. You'll be assessed by a health professional to work out the level of help you are entitled to. Your rate will be regularly reviewed to make sure you're getting the right support. PIP is made up of two components: Daily Living and Mobility. There are two rates: standard and enhanced.

In Scotland DLA has been replaced with the child disability payment

Website: <https://www.mygov.scot/child-disability-payment>

■ **Personal Independence Payment (PIP)**

Website: <https://www.gov.uk/pip>

DWP - PIP claims

Telephone: 0800 121 4433

If you live in Scotland, you need to apply for Adult Disability Payment (ADP) instead. Website: <https://www.mygov.scot/adult-disability-payment>

If your child is over the age of 16 they might also be able to claim:

■ **Employment and Support Allowance (ESA)**

Website: <https://www.gov.uk/employment-support-allowance>

Telephone: 0800 328 9344

There are 3 types of ESA:

- 'New style' ESA if you're entitled to claim Universal Credit (see below)
- Contributory ESA - usually you are entitled to receive this if you've paid enough National Insurance contributions (National Insurance credits can count for part of this, if you get them)
- Income-related ESA - usually you are entitled to receive this on its own or on top of contributory ESA if you have a low income

In some areas you may have to claim **Universal Credit (UC)** instead of ESA.

■ **Universal Credit**

Website: <https://www.gov.uk/universal-credit>

Telephone: 0800 328 5644

Welsh language: 0800 328 1744

Benefit Links

■ **Benefits and Work**

Website: <https://www.benefitsandwork.co.uk/>

Email: info@benefitsandwork.co.uk

Benefits and Work provide information and guides to help with benefits claims and appeals.

■ **Citizens Advice Bureau**

Website: <https://www.citizensadvice.org.uk/benefits/>

Advice line (England): 0800 144 8848

Advice link (Wales): 0800 702 2020

The CAB website provides comprehensive information on all benefits.

■ Entitled to

Website: <https://www.entitledto.co.uk/>

This is a free benefits calculator to help you estimate the financial support you are entitled to.

■ Turn2Us

Website: <https://www.turn2us.org.uk/>

Email: info@turn2us.org.uk

Turn2us is a national charity that helps people in financial hardship to gain access to welfare benefits, charitable grants and support services.

Direct Payments

Direct payments allow you to pay for and arrange help yourself, instead of receiving the help directly from social services. Direct payments and personal budgets are offered by your local authority to give you more flexibility over how your child's care and support is arranged and provided. Direct payments are given to both people with care and support needs, and also to carers. A personal budget or direct payment will be created after an assessment by social services. If the council decides that your child needs any kind of support, your child will receive a personal budget and you can choose a direct payment instead of the authorities arranging the services for you. If you aren't able to, or don't want to manage your own finances, it's possible for another person to manage the direct payments on your behalf. The council will have a local support organisation in place to offer this service. Direct payments are voluntary, and you need to request or agree to have one. You can't be forced to have direct payments. If you decide to accept direct payments, you can change your mind about this at any time. If you no longer want direct payments, you can contact your local social services and ask them to arrange services instead.

The council pays the direct payment to you so that you can decide how you want to meet your child's care and support needs. Many people choose to employ their own personal assistant(s), although there are many other ways direct payments can be used. The council should set out your obligations and responsibilities in a direct payment agreement that you may be asked to sign. This agreement could include:

- Keeping records and accounting for how the money is spent to social services
- Taking on the legal role of an employer if you're using the payment to pay for a care worker. Talk to the council about local organisations that can help manage the administration and other responsibilities of being an employer

■ Disability Rights UK

Website: <https://www.disabilityrightsuk.org/>

DRUK has specialist expertise in direct payments and other issues.



Grants and Funding

There are many different charities that can help families of children with additional needs. They each have their own criteria for how much and who they can offer support to.

■ Family Fund

Website: <https://www.familyfund.org.uk/>

Telephone: 01904 550055

Email: info@familyfund.org.uk

Family Fund is the UK's largest charity providing grants for families raising disabled or seriously ill children and young people.

Unique has a separate more detailed guide on [Grants and Funding](#) which you might find helpful. It is freely available to download from the Families section of our website.

There is also some assistance available to families called a **Warm Home Discount**. This is offered by most energy companies once a year (usually between October and March) to help with fuel bills. It isn't paid directly to you but taken off your electricity bill, or added to your pre-payment meter. For more information, see the following website:

<https://www.gov.uk/the-warm-home-discount-scheme>

Alternatively, contact your energy provider to see if you are qualify.

Council Tax reduction

■ Disabled Facilities Grants

Website: <https://www.gov.uk/disabled-facilities-grants>

These grants provide funding towards the cost of home adaptations to enable you and your family to continue living in your home. A Disabled Facilities Grant won't affect any benefits you receive.

If you have had a room built or altered for your child, you should also get a reduction in council tax. If you're on a low income you may also be entitled to Housing Benefit and Council Tax reduction from your local council.

If your child is over the age of 18 and has a **Severe Mental Impairment (SMI)** and receives certain benefits, this will also qualify you for a separate council tax reduction.

Website: <https://www.gov.uk/council-tax/discounts-for-disabled-people>



Continence and Toileting

If your child has bladder and bowel problems/issues that mean it has not been possible to achieve toilet-training by the time they reach three or four years of age, you may be eligible for free nappies/pads from your local continence service. In most areas of the UK the qualifying age is four years, but it can vary between authorities. You can get a referral to your local continence service through your GP, health visitor, paediatrician or school nurse.

■ Bladder and Bowel UK

Website: <https://www.bbuk.org.uk/>

Telephone: 0161 214 4591

Email: bbuk@disabledliving.co.uk

Provides information, publications and advice on continence products.

■ ERIC

Website: <https://eric.org.uk/>

Telephone: Freephone 0800 169 9949

Email: helpline@eric.org.uk

Provides information, publications and advice on continence products.



■ The National Key Scheme (NKS)

The NKS offers disabled people independent access to locked public toilets around the country. Toilets fitted with National Key Scheme (NKS) locks can now be found in shopping centres, pubs, cafés, department stores, bus and train stations and many other locations in most parts of the country. You can obtain a key from many local councils or you can purchase a key from Disability Rights UK shop, website: <https://shop.disabilityrightsuk.org/products/radar-key> and other recognised disability outlets. Please be aware that there are many 'fake' keys on the market that do not fit the locks, so be careful where you purchase from and do ask if it is a 'genuine' key.

■ The Changing Places Consortium

Website: <https://www.changing-places.org/>

England, Wales and Northern Ireland

Telephone: 020 7803 4814

Email: changingplaces@muscular dystrophyuk.org

Scotland

Telephone: 01382 385 154

Email: changingplaces@pamis.org.uk



For more information on [Toilet training and Continence](#), Unique has a more detailed guide which is available to download from the Families section of our website.

Vehicles and transport

If you or your child are disabled, you can apply for the following:

■ Exemption from paying vehicle tax

Website: <https://www.gov.uk/vehicle-exempt-from-vehicle-tax>

You can apply for exemption from paying vehicle tax if you get:

- The higher rate mobility component of Disability Living Allowance (DLA)
- The enhanced rate mobility component of Personal Independence Payment (PIP)

■ Parking benefits

Blue Badge Scheme Website:

<https://www.gov.uk/apply-blue-badge>

■ Disabled persons rail card

Website: <https://www.disabledpersons-railcard.co.uk/>

Telephone: 0345 605 0525

Email: [via website](#)

■ The Motability Scheme

Website: <https://www.motability.co.uk/>

Telephone: 0300 456 4566

Email: [via website](#)

Help to lease a car, in exchange for high rate mobility allowance.



“ 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. ”

Other links that may be helpful

■ Cerebra

Website: <https://cerebra.org.uk/>
Telephone: (free phone): 0800 328 1159
Email: enquiries@cerebra.org.uk
A charity helping families with children with neurological conditions. Cerebra has lots of information and useful guides on its website.

■ Child Growth Foundation

Website: <https://childgrowthfoundation.org/>
Telephone: 020 8995 0257
Email: info@childgrowthfoundation.org

■ Contact

Website: <https://contact.org.uk/>
Telephone: 0808 808 3555
Email: info@contact.org.uk
UK-wide charity providing advice, information and support to the parents of all disabled children. They are a force for change and campaign with families to remove the barriers they face every day.

■ CRY-SIS

Website: <https://www.cry-sis.org.uk/>
Telephone: 08451 228 669
This organisation provides support for families of babies who cry excessively. There is some advice for soothing the child, a helpline, and a book list of relevant publications.

■ Disability Rights Handbook Produced by Disability Rights UK

Website: <https://www.disabilityrightsuk.org/>
This handbook contains comprehensive and up-to-date information about benefits. Disability Alliance factsheets include information and advice on benefits, tax credits, social care and other disability-related issues.

■ Epilepsy Action

Website: <https://www.epilepsy.org.uk/>
Telephone: 0808 800 5050
Email: epilepsy@epilepsy.org.uk
Provides information to people of all ages with epilepsy and those living or working with them.

“ 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. ”

■ Fledglings

Website: <https://www.fledglings.org.uk/>

Telephone: 0203 319 9772

Email: Fledglings@contact.org.uk

Helping parents of a child with special needs of any kind to find simple, affordable solutions to practical problems. They offer a free product search service to locate toys, clothing, developmental aids and other items that may not be easily available, or those designed especially for children with special needs.

■ Heartline

Website: <https://www.heartline.org.uk/>

Email: intouch@heartline.org.uk

Support for children with heart disorders, and their families.

■ Home Start UK

Website: <https://www.home-start.org.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

British Forces Cyprus (ESBA): Telephone: 00357 2474 4802

British Forces Cyprus (WSBA): Telephone: 00357 2596 3748

Home-Start is one of the leading family support charities in the UK.

Home-Start volunteers help families with young children deal with the challenges they face. They support parents as they learn to cope, improve their confidence and build better lives for their children.



■ Makaton Vocabulary Development Project

Website: <https://www.makaton.org/>

Telephone: 01276 606 760

Email: info@makaton.org

Developed to help people to communicate. Makaton is now widely used with a variety of children with communication difficulties.

■ Mencap

Website: <https://www.mencap.org.uk/>

Telephone: 0808 808 1111 Monday to Friday
9am - 5pm

Email: helpline@mencap.org.uk

Works exclusively with and for people with a learning disability, their families and carers.

■ MERU

Website: <https://www.merushop.org/>

Email: info@meru.org.uk

MERU is a charity that designs and custom-makes specialist equipment for use at home, at school or college, in hospital or at play.

■ The National Autistic Society

Website: <https://www.autism.org.uk/directory>

Telephone: 0207 833 2299

Email: nas@nas.org.uk

■ National Centre for young people with epilepsy

Website: <https://www.youngpilepsy.org.uk/>

Telephone: 01342 832243

Email: info@youngpilepsy.org.uk

NCYPE is a major provider of specialised services for young people with epilepsy (school and resources & treatment centre).

■ National Network of Parent Carer Forums

Website: <https://nnpfc.org.uk/>

A network of over 150 parent carer forums from across England. Please visit their website to locate your nearest forum.

■ Netmums

Website: <https://www.netmums.com/>

A family of local websites for anyone involved in caring for young children. Netmums is a members' only site. Membership is free.

■ Newlife

Website: <https://newlifecharity.co.uk/>

Telephone: 0800 902 0095

Nursing support and equipment provider for disabled children.

■ Pyramid Educational Consultants UK Ltd

Website: <https://pecs-unitedkingdom.com/>

Telephone: 01273 609555

Email: pyramiduk@pecs.com

UK-developers of PECS, the Picture Exchange Communication System: an alternative/ augmentative communication system that teaches students to initiate spontaneous communication in a social context.

■ REMAP

Website: <https://www.remap.org.uk/>

Telephone: 01732 760209

Email: data@remap.org.uk

Makes and adapts equipment for disabled people where the exact product cannot be commercially made.



Diagnosed at the age of
20

■ Signalong

Website: <https://www.signalong.org.uk/>

Telephone: 01634 727087

Email: admin@signalong.org.uk

Signalong provides training and resources to assist those with communication difficulties.

■ Special Needs Jungle

Website: <https://www.specialneedsjungle.com/>

Special Needs Jungle creates easy to understand resources, articles and information for parents and carers of children with special needs, disabilities and health conditions to better enable them to navigate the special needs system.

■ Wellchild

Website: <https://www.wellchild.org.uk/>

Telephone: 01242 530007

Email: info@wellchild.org.uk

WellChild is a UK children's charity making it possible for children and young people with exceptional health needs to be cared for at home instead of hospital, wherever possible.



Parents say ...

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

“ We have 2 boys ages 23 and 20 and one girl 19 who were all diagnosed 2 years ago with 16p11.2 duplication. 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. My children are also all zebras, as they have Ehler's Danlos type 3. Looking back, I can see why they acted like they did socially and why we felt that the 16p11.2 duplication was actually a gift. ”

“ 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. ”

“ 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 received 12 months ago came as a complete shock. My initial thought being why had no-one thought of that earlier? Nothing has changed, but our whole world had been turned upside down. It's 12 months since the diagnosis and I didn't realise at the time, that we've been through the grieving process. Initially angry at all those who'd thought us paranoid (including family, friends and Healthcare Professionals), then upset and guilty for all the things K. 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. ”

Use this page to record your child's details if you need to.

Name:

Date of Birth:

Diagnosis:

Array/karyotype/genome result (if known):

Key phone numbers

Doctor:

Paediatrician:

Specialist nurse:

Neurologist:

Social Worker:

School:

Pharmacy:

Notes



Inform Network Support



Understanding Chromosome & Gene Disorders

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

Join Unique for family links, information and support.

Unique is a charity without government funding, existing entirely on donations and grants. If you can, please make a donation via our website at www.rarechromo.org/donate Please help us to help you!

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.

Our thanks to all of the parents that contributed towards this guide.

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This guide was compiled by Unique and reviewed by Dr Catherine Tuffrey, Consultant Paediatrician, SOLENT NHS TRUST.

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