

Understanding
chromosome
disorders

Unique

After Diagnosis: What happens next?



rarechromo.org

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 good as they would be with any child.



You will feel sadness at times but you will also experience incredible joy.

Trying to cope with your child's disorder and adjusting your own family life can be very hard. Every parent wants to do the very best they can for their child. Knowing where to start if your child has 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 difficult. A diagnosis can be important as it may lead to you getting the right services and support for your child, such as healthcare, therapies and education. 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. Leading up to diagnosis and getting the correct diagnosis for your child can be a very difficult time. The first issue of this guide was originally aimed at parents whose children were 4 years old and under (pre-school age). In this updated version we have included information that could be of use to parents whose children were diagnosed at any age. It could also be of help to families who have yet to receive a formal diagnosis for their child.

This guide is primarily aimed at families living in the United Kingdom and 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. Contact Marion, Unique Family Support Officer, if you need any assistance with this.



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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. 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 trying to explain them to other people. It will 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 will seem like it is full of endless appointments, especially if your child is very young and not yet accessing services like therapies. Your friends and family might find it difficult to understand and not know what to say, because they don't know how to and might avoid keeping in touch because of this.

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

Questions

You will have lots of questions, you may ask yourself questions like:

- ◆ We haven't seen a geneticist yet, what does that involve?
- ◆ How can I get help for my child's development?
- ◆ Where will they 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 able to claim any benefits to help support my child?

This guide has been designed to give you basic information on where you can obtain further advice on raising a child with a chromosome or gene disorder. Getting the right support will help your child and your family to live as happy and fulfilling lives as possible. For answers relating to the questions above, please read on.....

Early Support

Early support is a term used for disabled children aged five and under. It brings together all of the services and support available from different agencies. Making it easier for families to co-ordinate their child's health, education and social care needs. The UK government website has some useful early support information here: <http://bit.ly/2ER2sD7>

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 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 genetic centre requires a referral from your GP or hospital specialist. You may have a bit of a wait for your appointment and you may only ever 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 available to download from the families section of our website.

DEVELOPMENT

How can I get help for my child?

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

These services might include:

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

They might refer you to your nearest Child Development Centre who can help advise you about these services.



Child Development Centres (CDC)

A CDC houses a range of professionals who can assess and treat children with a wide variety of problems including physical disabilities, learning difficulties, developmental delay, health problems such as epilepsy and language and communication difficulties. It provides a multi-disciplinary service for children from birth to 19 years of age. The professionals usually work together as a team and offer assessments and ongoing treatments and therapies as required.

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

The team will usually include:

A Community Paediatrician - this is a children's doctor who specialises in childhood disability, behavioural disorders and complex needs. Community Paediatricians regularly liaise with schools, school nurses and the Education Authority to help children who are struggling to access school.

Paediatric Occupational Therapists work 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.

Paediatric Physiotherapists assess and treat children to improve their physical skills, including their posture and mobility.

Paediatric Speech and Language Therapists assess and treat children's speech and language, including the use of manual signs and symbols, and can also advise where children have chewing or swallowing difficulties.

Audiologist is an expert who can help to prevent, diagnose, and treat hearing and balance disorders for people of all ages. Screening individuals to identify possible hearing disorders. Testing will confirm if a hearing loss is present and determine the kind and degree of loss.

Orthotist is trained to prescribe, design, fit and monitor orthoses, prostheses and specialist footwear and insoles.

And possibly other visiting professionals for example:

Neurologist who treats disorders that affect the brain, spinal cord, and nerves, such as: epilepsy

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

The first appointment usually takes about an hour/hour and a half. The paediatrician will take a detailed history, watch your child while they play and examine them. They may do a developmental assessment, however - if your child has already had assessments from therapists this may not be needed.

What happens then?

The paediatrician will make a plan with you about how to help your child. They may discuss referring your child to various therapists within the centre. They will 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 with additional medical needs will be followed up in the Community Paediatric Service for many years, often until they reach adulthood.

The Paediatrician can also refer your child to the Children's Disability Team (Social care) for more support.

What is the Children with Disabilities Team?

The Children with Disabilities Team (CDT) is a specialist service that aims to provide parents with support so that they can, whenever possible, bring up their child at home, leading as full and independent a life as possible. You can contact the Children with Disabilities Team directly by looking under Social and Caring Services in your phone directory or 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 disabled 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. The 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.



“ 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

Children's centres provide a variety of advice, information and support for parents and carers. You can locate your nearest children's centre by contacting your local Family Information Service, or by visiting: <http://bit.ly/2EqzE6C>

A children's centre is different from a child development centre (health). 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 difficulty or disability are said to have '**special educational needs**'.

Where will my child go to school?

Many children with a chromosome or gene disorder will be able to go to local mainstream school without needing support. For some they may still be able to attend mainstream school with support. Children that need a lot more educational support will need specialist provision that may only be provided in a specialist school setting.

Special Educational Needs and Disabilities (SEND)

Special educational needs and disabilities (SEND) can affect a child or young person's ability to learn. They can affect their:

- ◆ Behaviour or ability to socialise, for example - they struggle to make friends
- ◆ Reading and writing, for example because they have dyslexia
- ◆ Ability to understand
- ◆ Concentration levels
- ◆ Physical ability

Your child may need an Education, Health and Care (**EHC**) plan, if they need more support than their school provides. An education, health and care (EHC) plan is for children and young people aged up to 25, who need more support than is available through special educational needs support. EHC plans identify educational, health and social needs and set out the additional support to meet those needs.



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're 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 they decide to carry out an assessment you may be asked for:

- ◆ Any reports from your child's school, nursery or childminder
- ◆ 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 UNDER 5

SEN support for children under 5 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 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'll tell you what support options are available.

- ◆ **National Portage Association** www.portage.org.uk

0121 244 1807 Email: info@portage.org.uk

A free home-visiting educational service for pre-school children aged 4 and under with additional support needs and their families. It is available in many parts of England. Visit their website to find details of the nearest Portage service. Parents can self-refer.

- ◆ **The Alliance for Inclusive Education** www.allfie.org.uk/

0207 737 6030

A campaigning group promoting inclusive education for all children.

- ◆ **The British Association for Early Childhood Education**

www.early-education.org.uk

01923 438 995

Provides useful resources and downloadable publications.

◆ Council for Disabled children

<https://councilfordisabledchildren.org.uk/independent-support>

Independent support for parents & young people to navigate the Education, Health & Care planning process.

CHILDREN AGED 5 to 15

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
- ◆ Help taking part in class activities
- ◆ Extra encouragement in their 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



YOUNG PEOPLE AGED 16 OR OVER IN FURTHER EDUCATION

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

Unique has separate guides on Education, and Transition (for children aged 13 years plus), which you might find helpful, they are available to download from the families section of our website.

"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."

BEHAVIOURAL CHALLENGES

Children with a chromosome or gene disorder sometimes have what is described as 'challenging behaviour'. It can be difficult to describe it fully, it could be something sensory, for example, a reaction to doing things in a certain way, or not liking to touch certain things. In many cases, challenging behaviour occurs because the person with special needs is frustrated at his or her inability to make others understand what they need or want. Or 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 it can develop as they get older.

◆ **CAMHS** <http://bit.ly/1V4pzKQ>

Parents Helpline: 0808 802 5544 (Monday to Friday 9.30am – 4pm, 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, you can read more about CAMHS here. Or perhaps you and your child are on the CAMHS waiting list or have an appointment soon. So you know what to expect, we have put together information about CAMHS especially for parents and carers.

YoungMinds Parents' Helpline is a telephone and online support service, and we 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:

◆ **Challenging Behaviour Foundation** www.thecbf.org.uk/

Family Support Line: 0845 602 7885 email: support@thecbf.org.uk

Providing practical information for families and professionals; about understanding and supporting children and adults whose behaviour challenges.

Unique has a separate guide on Behaviour and Sensory issues which you might find helpful, it is available to download from the families section of our website.

“I wish someone 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.”

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 and have access to therapists that help carers have subsidised treatments like Reiki, massage and aromatherapy.

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 people would naturally expect to care for their own child, if that child has additional needs that requires more assistance than that of a typically developing child of the same age, then they will be classed as a 'carer' and that entitles them to carers services and an assessment of their own needs. There is also support available to Young Carers (siblings who help to care for their brother or sister with additional needs).

To find out if you have a carers support Service and a young carers service nearby, visit your council website, or check out these links:

- ♦ **Carers Trust** <https://carers.org/section/help-advice>

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

- ♦ **Carers UK** www.carersuk.org/

Support for all carers. Also campaigns to improve the lives of carers.

If your child has a secondary diagnosis of another disorder, there may well be a local support group for that disorder. For example: Autism.

For further information on local support groups, visit:

- ♦ **The National Autistic Society** www.autism.org.uk/



Unique has separate guides on **Carers Wellbeing, Young Carers and Sibling support** which you might find helpful, they are available to download from the families section of our website.

BENEFIT

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 **Disability Living Allowance (DLA)**. Disability Living Allowance (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

They 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 they're on dialysis
- ⇒ Highest rate - help or supervision throughout both day and night, or if they're terminally ill

Mobility component

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

- ⇒ Lowest rate - they can walk but need help and or supervision when outdoors
- ⇒ Highest rate - they 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, 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.

They can also arrange for someone to help you fill out the form if required.

◆ Disability Living Allowance (DLA)

www.gov.uk/disability-living-allowance-children

0800 121 4600 Monday to Friday, 8am to 6pm

◆ Cerebra guide to DLA <http://bit.ly/2GdvUCT>

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

If your child is 16 years or over, there is a new benefit that replaces DLA, it is called **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 can get.

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.

◆ **Personal Independence Payment (PIP)** www.gov.uk/pip

DWP - PIP claims

0800 917 2222 Monday to Friday, 8am to 6pm

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

Employment and Support Allowance (ESA). There are 3 types of ESA:

- ⇒ 'New style' ESA if you're entitled to claim Universal Credit
- ⇒ Contributory ESA - usually you get 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 get this on its own or on top of contributory ESA, if you're on a low income

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

www.gov.uk/employment-support-allowance

0800 328 9344 Monday to Friday, 8am to 6pm

In some areas you may have to claim

◆ **Universal Credit** <https://universalcreditinfo.net/>

Universal Credit full service helpline 0800 328 5644 Monday to Friday, 8am to 6pm

"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."

BENEFITS LINKS...

◆ **Benefits and Work** www.benefitsandwork.co.uk/

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

◆ **Citizens Advice Bureau** <http://bit.ly/2FcZp7t>

Their website provides comprehensive information on all benefits.

◆ **Entitled to** www.entitledto.co.uk/

This is a free benefits calculator to see what you can claim.

◆ **Turn2Us** www.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.

TRANSPORT

If you or your child are disabled you can apply for the following: exemption from paying vehicle tax

www.gov.uk/vehicle-exempt-from-vehicle-tax

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

- ◆ Higher rate mobility component of Disability Living Allowance (DLA)
- ◆ Enhanced rate mobility component of Personal Independence Payment (PIP)

- ◆ **Parking benefits - Blue Badge**

www.gov.uk/apply-blue-badge

- ◆ **Disabled persons bus pass or railcard**

www.disabledpersons-railcard.co.uk/

- ◆ **The Motability Scheme**

www.motability.co.uk/

Help to buy or lease a car



DIRECT PAYMENTS

If your local council has assessed your child as needing care and support services, they can get: **Direct payments**

These allow you to buy in and arrange help yourself instead of getting it 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, they will receive a personal budget and you can choose a direct payment instead of letting them arrange 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 have direct payments, you can change your mind about this at any time. If you no longer want direct payments, 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 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** www.disabilityrightsuk.org/
Has specialist expertise in direct payments.

HOUSING

♦ **Disabled Facilities Grants (DFG)**

www.gov.uk/disabled-facilities-grants

This is money towards the cost of home adaptations to enable you and your family to continue living in your home. A DFG won't affect any benefits you get. 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.

"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 miss so much; that you can never get back."

WORK

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. 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) 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** www.gov.uk/carers-allowance

0800 731 0297 Mon -Thurs, 8:30am to 5pm, Fri 8:30am to 4:30pm

♦ **Carers Credit** 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** www.workingfamilies.org.uk

Free helpline for parents and carers: 0300 012 0312 (Mon-Fri)

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 includes a section especially for parents of disabled children.

GETTING A BREAK FROM CARING

Short breaks increase the quality of life for children with chromosome and gene disorders and their families by giving a break from caring to parents, or an activity for a child with their peers. They provide children with additional needs, opportunities to meet new people, make new friends and experience different activities. Many families will have extended family and friends that can help to look after your child so that you can have a break, but many will be less fortunate. You can either contact your local Child Disability Team or Carers Support Service or ask your GP or paediatrician to refer you.



NAPPIES/INCONTINENCE PADS

If you have not been able to toilet-train your child by the time they reach three or four years you might 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. You can get a referral to your local continence service through your GP, health visitor, paediatrician or school nurse.

♦ **Bladder and Bowel UK** www.bladderandboweluk.co.uk

0161 607 8219

Email: bladderandboweluk@disabledliving.co.uk

Information, publications and advice on continence products.

♦ **ERIC** www.eric.org.uk/

0845 370 8008 Monday to Thursday 10am–2pm

Email: helpline@eric.org.uk

Information, publications and advice on continence products.

♦ **The Changing Places Consortium** www.changing-places.org/

England, Wales and Northern Ireland: 0207 803 2876

Email: changingplaces@muscardystrophyuk.org

Scotland: 01382 385 154

Email: PamisChangingPlaces@dundee.ac.uk

Changing places is a campaign on behalf of those people who cannot use standard accessible toilets.

They need Changing Places toilets with enough space and the right equipment.



For more information on Toilet training and Continence, we have a guide which is available to download from the families section of our website.

The National Key Scheme (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 from Disability Rights UK: <http://bit.ly/2G6Wwqd> and other 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.



OTHER USEFUL LINKS

◆ **Cerebra** www.cerebra.org.uk/

Helpline (freephone): 0800 328 1159

Email: info@cerebra.org.uk

A charity dedicated to helping families with children with brain conditions. Cerebra has lots of information and useful guides on its website.

◆ **Child Growth Foundation** www.childgrowthfoundation.org/

Helpline: 020 8995 0257

Email: info@childgrowthfoundation.org

◆ **Contact** <https://contact.org.uk/>

Helpline: 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** www.cry-sis.org.uk/

CRY-SIS helpline: 08451 228 669 7 days a week 9am-10pm

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**

<http://bit.ly/2E9loul>

This handbook contains comprehensive and up-to-date information about benefits. Disability Alliance Fact Sheets include information and advice on benefits, tax credits, social care and other disability-related issues.

◆ **Epilepsy Action** www.epilepsy.org.uk

0113 210 8800

Email: epilepsy@epilepsy.org.uk

Provides information to people of all ages with epilepsy and those living or working with them.

◆ **Family Fund** www.familyfund.org.uk/

01904 550055

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 guide on Grants and Funding which you might find helpful, it is available to download from the families section of our website.

◆ **Fledglings** www.fledglings.org.uk

01799 541 807

Email: enquiries@fledglings.org.uk

A not-for-profit organisation, which helps parents and carers 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** www.heartline.org.uk

Email: intouch@heartline.org.uk

Support for children with heart disorders and their families

◆ **Home Start UK** www.home-start.org.uk

***England** 0116 464 5490 Email: info@home-start.org.uk

***Northern Ireland** 07747 487938 Email: ahanna@home-start.org.uk

***Scotland** 0131 281 0871 Email: scotland@home-start.org.uk

***Wales** 0292 0491181 Email: walesoffice@home-start.org.uk

***British Forces Germany** 0049 5251 1843446

Email: homestartbfg@gmail.com

***British Forces Cyprus** (ESBA) 00357 2474 4802

***British Forces Cyprus** (WSBA) 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. We support parents as they learn to cope, improve their confidence and build better lives for their children.

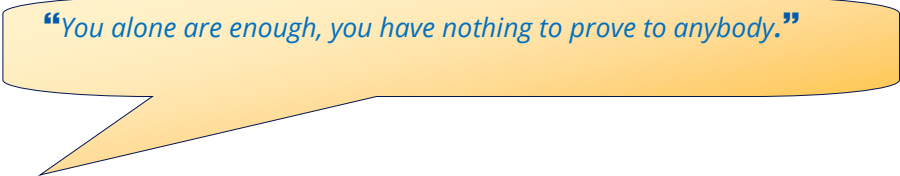
"We have 2 boys ages 23 and 20 and one girl 19 who were all diagnosed 2 years ago with 16p11.2 duplication. We were relieved when we got the diagnosis because it gave proof that our kids were special. It also gave us comfort that our choice to home-school was the right choice. My children are also all 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"

◆ **Makaton Vocabulary Development Project** www.makaton.org/
01276 606 760
Email: info@makaton.org
Developed to help people to communicate. It is now widely used with a variety of children with communication difficulties.

◆ **Mencap** www.mencap.org.uk
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** www.meru.org.uk
01372 725203
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.

◆ **National Centre for Young People with Epilepsy**
www.youngepilepsy.org.uk
01342 832243
Email: info@youngepilepsy.org.uk
major provider of specialized services for young people with epilepsy (school and resources & treatment centre)



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

◆ **National Network of Parent Carer forums** www.nnpccf.org.uk/
The National Network of Parent Carer Forums is a network of over 150 Parent Carer Forums from across England. Please visit their website to locate your nearest.

◆ **Netmums** www.netmums.com
Is a family of local websites for anyone involved in caring for young children. Netmums is a members' only site. Membership is free.

◆ **Pyramid Educational Consultants UK Ltd**
www.pecs-unitedkingdom.com/
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** www.remap.org.uk

01732 760209

Email: data@remap.org.uk

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

◆ **Signalong** www.signalong.org.uk/

01634 727087

Email: admin@signalong.org.uk

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

"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."

"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."



Diagnosed at the age of 20

**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:

NOTES

Inform Network Support



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 Please help us to help you!

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

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

Version 2 (MM)

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