



Understanding Chromosome & Gene Disorders

Uniquely different



rarechromo.org

UNIQUELY DIFFERENT

The intention of this guide is to help parents to get additional support in relation to birth uniqueness and conditions that develop or progress as a child gets older. It is also to help parents who are struggling with other people's reactions to their child's look/personality/behaviours etc. Not everyone is the same, some are just **Uniquely different**. We are all individuals; no-one should be judged by how they look, walk, talk, act, etc. We are all Uniquely different in our own way, let's celebrate our children for who they are.

Quote by Albus Dumbledore from the Harry Potter books,
“ It matters not what someone is born, but what they grow to be.”
~ Credit J.K Rowling



Contents

Top Ten Tips	3
Getting Support	3
Counselling	4
Face and Head.....	5
Limbs	6
Spine.....	7
Stomach	8
Changing places facilities.....	9
Links.....	10
Welcome to Holland poem.....	12
Coping mechanisms	14
Attracting attention	15
Building confidence	15
Parent quotes.....	16

TOP TEN TIPS

1. Take each day as it comes, try not to plan your child's entire life, be fluid and roll with the waves
2. Celebrate the inch stones - they are incredible
3. Smile at people who stare, rather than getting angry; don't waste your precious energy on people who don't understand
4. Enjoy the good parts, try not to be overwhelmed by the hard parts, or you will miss the tiny miracles
5. Get involved with local support groups, like carers support services, who often run parent carer groups, or other local organisations that offer parent and child groups
6. Talk to other parents online via our private Unique network café on Facebook
7. Don't 'suffer' in silence. While it might seem like the end of the world, it really isn't. You will learn to live with your 'normal' and your child will become your most treasured gift
8. Knowledge is power. Try to find out as much as you can about your child's disorder and seek out the help that they might need
9. Don't be afraid to ask for help if you need it
10. Always be ready with a bag of spare everything's to take with you, even if you just keep it in the boot of your car, so that you can always be prepared for any eventuality.



We have a separate guide on Carers wellbeing, in the family information area of our website:
<https://www.rarechromo.org/practical-guides-for-families/>

Getting support

The most important thing is acknowledging your child's uniqueness and most importantly how you feel and being able to talk to someone about how you feel. You will go through a whole range of emotions: shock, anger, sadness, fear, denial. You might find it easy to talk to your partner, other family members or a close friend, but often that isn't easy for parents and relatives or friends might not find it easy to broach the subject either. That might be the time to seek outside support from a relevant organisation, that can link you with other parents who have experienced the same feelings.

There are organisations that can help with support; some just offer a listening ear, whilst others offer more support. Talking to other families whose child has a chromosome or gene disorder might also help. As you can imagine there are numerous types of birth differences, too many to list, so in this guide we have listed those found most often in babies with a rare chromosome or gene disorder. Dealing with other people's comments and stares is more difficult to come to terms with. Differences in our children are not of course limited to unusual facial features. For many of our children the way they might walk, talk or act can also make them Uniquely different. For many children with a rare chromosome or gene disorder, developmental delay is common, so walking and talking can arrive much later than typically functioning children of the same age. If your child is non-verbal (doesn't have any word speech at all) but makes other sounds, this can often draw the attention of others. Younger children are often curious about a child making unusual sounds and will often stare out of curiosity and comment loudly. Stares and comments from adults can be the hardest to take and once something is said, it is never forgotten by parents. Most people, though, are just curious but haven't got the courage or sensitivity to ask questions.

Counselling

Genetic counselling

You may get to see a genetic counsellor when your child is a baby, but for some parents, genetic counselling can come much later. Genetic counsellors are healthcare professionals with training and expertise in genetics/genomics and counselling skills. Their role is to provide information and explain the facts as clearly as possible, to help you understand a genetic test that is being offered and what it means for you and your family, or to explain the genetic diagnosis that you or your child has received. There are often long waiting lists to see a geneticist/genetic counsellor in the UK.

Non-genetic counselling

Having a child with a chromosome or gene disorder, or any disability, can put a huge strain on your life and relationships. Coming to terms with having a child with a disorder can be rather difficult for parents and often increased if your child has additional health issues at birth. Some professionals, like doctors, nurses, and therapists can be very helpful and supportive, but some aren't always as helpful as we'd like and can end up making you feel more stressed. You can get a referral from your GP to see a psychological counsellor, (under the NHS this is known as talking therapy), or you could refer yourself to a private counsellor (which will incur a fee). Counselling allows people to discuss their issues and any difficult feelings they encounter, in a confidential environment.

Find a counsellor near you:

◆ **COUNSELLING DIRECTORY**

<https://www.counselling-directory.org.uk/>

Medical terminology

You may hear medical professionals use the word **dysmorphic**. You have perhaps never heard of the word before. It is just the description used by doctors of a person with an unusual difference in body structure. Many parents might not even notice a difference; it could be very subtle. You may also hear the words birth defects or congenital anomalies; these are terms used to describe unusual features after the birth of a child. Sometimes children are diagnosed whilst still in the womb.

Face and head

Cleft lip and palate

Some children are born with a cleft lip and/or palate; this occurs when parts of the head and face don't join up properly during early pregnancy. They are usually fixed with surgery but some might require ongoing support from a speech and language therapist, Orthodontist, or Maxillofacial Surgeon. For more information, visit:



◆ Cleft Lip and Palate Association (CLAPA)

<https://www.clapa.com/>

020 7833 4883 Email: info@clapa.com

CLAPA has an online community forum and local area groups that meet up for support.

◆ Changing Faces

<https://www.changingfaces.org.uk/adviceandsupport/self-help/parents/you-and-your-child>

0300 012 0275 (10am-4pm Monday to Friday)

Email: support@changingfaces.org.uk

Changing Faces is a UK based charity for everyone with a scar, mark or condition on their face or body, that makes them look different. They provide advice, support and psychosocial services to children, young people and adults.

For parents whose child has been born with unusual facial features, it can be difficult, but it is no-one's fault. Parents don't need to blame themselves. For the child as they grow up, they might not like the way they look and how others perceive them. This can be especially difficult for teenagers. It is important to discuss how they feel as they grow up, as they might need help themselves and might not necessarily open up about their feelings. For some children and adults, further surgery may be an option. Psychological counselling may help and can be accessed via referral from a GP or other medical specialist.

◆ **Headlines** <https://www.headlines.org.uk/>

Helpline: 07541 106816

Email: helpline@headlines.org.uk

Headlines Craniofacial Support, is a registered charity set up by a group of parents of children with Craniosynostosis. Craniosynostosis is a birth defect in which the bones in a baby's skull join together too early.

◆ **AmeriFace (USA)** <http://www.ameriface.org>

(702) 769-9264

Toll-free: (888) 486-1209

Email: info@ameriface.org

AmeriFace helps to provide educational and emotional support to persons born with craniofacial anomalies and their families.

◆ **CCA Kids (USA) Children's Craniofacial Association**

<https://ccakids.org/syndromes.html>

800.535.3643

CCA's mission is to empower and give hope to individuals and families affected by facial differences.

Limbs

Birth Uniqueness might include webbed fingers and/or toes (Syndactyly), where the digits haven't separated properly in the womb.

Talipes (club foot)

Talipes can affect one foot or both. It is usually congenital (present at birth). It occurs when baby's foot is twisted out of shape or position. The tissues connecting the muscles to the bone (tendons) are shorter than usual.

◆ **Reach** <https://reach.org.uk>

020 3478 0100

Reach offers families the opportunity to connect and share stories with other families affected by upper limb differences. Through their website and family events.

◆ **Information on limb abnormalities (USA)**

<https://www.nicklauschildrens.org/conditions/limb-deformities>

◆ **Steps Worldwide**

<https://www.stepsworldwide.org/conditions/talipes-clubfoot/>

01925 750271

Steps is a charity offering support to those affected by childhood lower limb conditions.

Ankle Foot Orthosis (AFO) is a splint used to support weak ankles and legs.

Your child might require specialist shoes or boots to help with walking and balance and co-ordination.

Spine

Sacral dimple

Babies might be born with a 'sacral dimple', it is a small dent or tiny hole in the lower back just above the buttocks. No-one seems to know why they occur; they are not usually a cause for concern, but will be checked by a paediatrician to determine whether or not they are closed.

Scoliosis

Scoliosis is a sideways curvature of the spine that occurs most often during the growth spurt just before puberty. This can be mild but sometimes it might require wearing a brace or surgery.



◆ **The Scoliosis Association UK** <https://www.sauk.org.uk/>

Helpline: 020 8964 1166

Providing advice, support and information to people affected by scoliosis and their families.

◆ **(USA) The National Scoliosis Foundation**

<https://www.scoliosis.org/>

1-800-673-6922 Email: NSF@scoliosis.org

The National Scoliosis Foundation (NSF) is a patient-led non-profit organisation helping children, parents, and adults, to understand the complexities of spinal deformities such as scoliosis.

Spina Bifida

◆ **Shine charity**

<https://www.shinecharity.org.uk/>

01733 555988 (Monday to Friday, 9am to 5pm)

Email: firstcontact@shinecharity.org.uk

Facebook:

<https://www.facebook.com/ShineUKCharity/>

Spina bifida literally means 'split spine' and occurs during development of the spinal cord and surrounding bones (vertebrae) leaves a gap or split in the spine.

◆ **(USA) The Spina Bifida Association**

<https://www.spinabifidaassociation.org/>

(800) 621-3141

Email: sbaa@sbaa.org



Sacral Agenesis

◆ **ISACRA (International Sacral Agenesis Caudal Regression Association) (USA based)** <https://isacra.org/>

Facebook: <https://www.facebook.com/groups/ISACRA/>

email: contact@isacra.org

Sacral agenesis, also called caudal regression syndrome (SA/CRS), is a rare congenital condition in which the lower part of the spine does not fully develop, potentially also impacting the formation and function of lower parts of the body such as the legs and pelvis and organs such as the kidneys. The ways that the condition impacts individuals vary greatly.

◆ **Sacral Agenesis Contact Group (UK family)** 02380 842 661

Stomach

Reflux (Gastro-oesophageal Reflux [GOR] and Gastro-oesophageal Reflux Disease [GORD])

Reflux is quite common in children with chromosome or gene disorders.

◆ **Living with reflux** <https://www.livingwithreflux.org/>

Email: info@livingwithreflux.org

Facebook: <https://www.facebook.com/livingwithreflux/>

Gastro-oesophageal Reflux (GOR) occurs in all infants, children and adults during and immediately after meals. GOR refers to the contents of the stomach washing back up from the stomach into the oesophagus. It can go up to the throat, and even be ejected out of the mouth. Gastro-oesophageal Reflux Disease (GORD or GERD in the USA) is present when there are symptoms or signs that are troublesome, or severe, or chronic, or when complications are present.

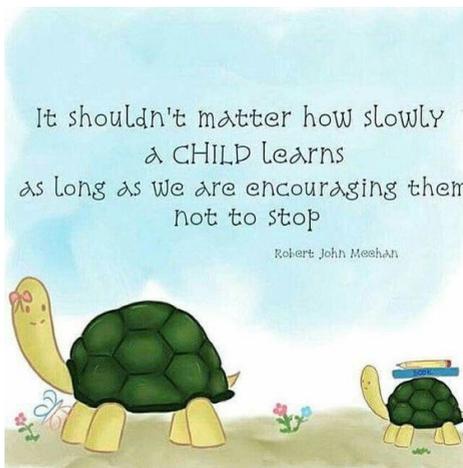
Medical equipment

Your child might not be able to feed properly or breathe without some assistance, so they will require a tube and/ or oxygen. They may be prone to choking and might require a suction machine. These are all things that could need to be adjusted/used in public, and it's possible that unusual, gurgling noises and other sounds could occur. If you are visiting a public place, for example, a restaurant, and you are bothered by other peoples stares, you might want to ask staff if you could have a quieter table. Don't let other people put you off going out and enjoying your time, wherever you want to go.



Changing Places facilities

Many of our children will be in nappies/diapers/pads for quite a few years, some never being able to be toilet-trained, while some children might also have been fitted with a stoma bag (following surgery to make a small opening on the surface of the abdomen in order to divert faeces or urine from the bowel or bladder. The waste is collected instead in a stoma bag, which is a small pouch-like waterproof bag). In the UK, people have campaigned for years to get proper changing facilities installed into public buildings. This is so that their family can go anywhere they choose without having to worry that there is nowhere to change their child (other than a standard disabled toilet and having to use a dirty floor), or possibly cutting their journey short in order to return home to change their child. No-one should have to put up with poor facilities. Changing Places facilities are becoming more common now than they ever have and legislation has been passed to include these facilities in all NEW public buildings in England. To find out more about these facilities and where they are, please visit: <http://www.changing-places.org/> Please note, not all available facilities are mentioned on this website; so if planning to visit anywhere, please contact venues direct in advance of your visit, to check whether they have a Changing Places facility.



Please check out our practical information guides on a variety of topics, on our website:
<https://www.rarechromo.org/practical-guides-for-families/>

OTHER LINKS

◆ **Birth defect Research for Children** <https://birthdefects.org/>
Facebook: <https://www.facebook.com/BDRCL/>
Birth Defect Research for Children, Inc. (BDRCL) is a USA based 501(c)(3) non-profit organization that provides parents and expectant parents with information about birth defects and support services for their children. They have an online community forum and factsheets.

◆ **Newlife** <https://newlifecharity.co.uk/>
Free Helpline phone number 0800 902 0095 (free from UK mobiles & landlines) Monday to Friday 9.30am - 5.00pm (Wed 9.30am to 7pm) answerphone facility outside normal hours
Email: nurse@newlifecharity.co.uk
Newlife's Care Services Helpline provides professional confidential care and support to families caring for disabled and terminally ill children. Experienced Nurses are readily available to give support and information. They also can help with information on equipment required.

◆ **Wellchild** <https://www.wellchild.org.uk/>
Facebook: <https://www.facebook.com/WellChildUK/>
WellChild is the national 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.

◆ **NSPCC** <https://www.nspcc.org.uk/>
Helpline 0808 800 5000

◆ **Center for Disease Control and Prevention (USA based website)**
<https://www.cdc.gov/ncbddd/disabilityandsafety/index.html>
Phone: 800-232-4636



Information and advice on bullying/cyberbullying of children with disabilities

- ◆ **Contact** https://contact.org.uk/wp-content/uploads/2021/03/dealing_with_bullying.pdf

A guide for parents of disabled children

- ◆ **Kidscape**

<https://www.kidscape.org.uk/>

020 7823 5430 Monday to Tuesday 9.30am – 2.30pm (excluding bank holidays). If the line is busy, please leave a message with your name and number and an adviser will call you back.

WhatsApp: 07496 682785

Email: parentsupport@kidscape.org.uk

Guidance and support for parents and carers on all aspects of bullying.

Autism

- ◆ **National autistic society**

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

0808 800 4104

Support and information for parents whose children have been diagnosed with an autism spectrum disorder.

- ◆ **Autism society (USA)**

<https://www.autism-society.org/>

- ◆ **Autism support (Australia)**

<https://www.autismspectrum.org.au/>

- ◆ **The Special Lioness**

www.thespeciallioness.com

Based in North East England but offering tips and support to all families.

The Special Lioness has a Facebook page

<https://www.facebook.com/groups/135449330433631/> in which they

share products, tips, info and support for one another and provide a support group and SEND specific playgroup locally (Sunderland) . They also offer a wide range of events during half term throughout the year. They would love to reach more families to offer their services, which are all free.

- ◆ **Bright Ears UK** <https://www.facebook.com/BrightEarsUK/>

Email: brightearsuk@gmail.com

Etsy shop: <http://www.etsy.com/uk/shop/BrightEars>

Handmade Inclusive Dolls and Accessories.

- ◆ **Hidden disabilities** <https://hiddendisabilitiesstore.com/blog>



Welcome To Holland

by Emily Perl Kingsley

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“ I am often asked to describe the experience of raising a child with a disability - to try to help people who have not shared that unique experience to understand it, to imagine how it would feel. It's like this.....

When you're going to have a baby, it's like planning a fabulous vacation trip - to Italy. You buy a bunch of guide books and make your wonderful plans. The Coliseum. The Michelangelo David. The gondolas in Venice. You may learn some handy phrases in Italian. It's all very exciting.

After months of eager anticipation, the day finally arrives. You pack your bags and off you go. Several hours later, the plane lands. The flight attendant comes in and says, "Welcome to Holland."

"Holland?!?" you say. "What do you mean Holland?? I signed up for Italy! I'm supposed to be in Italy. All my life I've dreamed of going to Italy."

But there's been a change in the flight plan. They've landed in Holland and there you must stay.

The important thing is that they haven't taken you to a horrible, disgusting, filthy place, full of pestilence, famine and disease. It's just a different place.

So you must go out and buy new guide books. And you must learn a whole new language. And you will meet a whole new group of people you would never have met.

It's just a different place. It's slower-paced than Italy, less flashy than Italy. But after you've been there for a while and you catch your breath, you look around... and you begin to notice that Holland has windmills....and Holland has tulips. Holland even has Rembrandts.

But everyone you know is busy coming and going from Italy... and they're all bragging about what a wonderful time they had there. And for the rest of your life, you will say "Yes, that's where I was supposed to go. That's what I had planned."

And the pain of that will never, ever, ever, ever go away... because the loss of that dream is a very very significant loss.

But... if you spend your life mourning the fact that you didn't get to Italy, you may never be free to enjoy the very special, the very lovely things ... about Holland.”

* * *



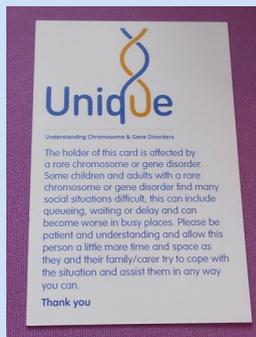
“ Believe in your child, truly believe! That is the first and fundamental thing for him to conquer all! ”

Coping mechanisms

Your child might have Autism and sensory issues that cause them to react differently. Children and adults with autism often feel overwhelmed by noise and all that is going on around them. Your child may struggle to deal with certain situations which may manifest in behaviours that may seem disruptive to others, but in fact are just your child's way of dealing with the emotions of the moment. You can explain this to onlookers if you wish but you should feel under no obligation to do so. Other people often see them as just naughty but that isn't always the case. Don't let people make you feel uncomfortable. If it is something you are comfortable with, just explain that your child has autism or sensory issues and they can find some situations difficult to deal with. There are a number of aids which can help your child cope during these situations like noise reducing earphones and fidget toys for managing anxious situations. Remember that your child will have their own coping mechanisms that work for them and help them manage their sensory/auditory/nervous system. It's all part of their wonderful neurodiversity.



We have a small awareness card (credit card size) that can help in situations like queuing etc. if you would like one of these please Email: marion@rarechromo.org



We have a separate guide on Behaviour and sensory issues in the family information area of our website: <https://www.rarechromo.org/practical-guides-for-families/>

Attracting attention

There are of course other things that are likely to attract attention; sometimes children and adults don't understand and often are too quick to judge. Don't waste your energy on negativity. Why attention might be drawn to your child.....

- ◆ your child might use a rollator to help with walking, or they might ride an adapted bicycle/tricycle.
- ◆ your child might wear a bandana or bib, or a patch behind their ear because they dribble/drool. You can buy some really trendy looking bandanas to make it look like a fashion accessory.
- ◆ Your child might have unruly hair because they cannot tolerate having their hair brushed or cut. One thing that can help with brushing is using a tangle teaser type brush, although not all children will tolerate that.
- ◆ your child might have seizures and because of that may need to wear a protective padded helmet.
- ◆ your child may wear glasses or hearing aids.
- ◆ your child might only like to dress a certain way, wear the same clothing or have the labels removed. There are several different retailers that offer bespoke items of clothing; for example, seam -free clothing. Try to dress your child in age appropriate clothing, especially as they get to teenage/adult years, so as not to make them stand out.

None of the above should be seen as an issue; they are part and parcel of your child. Everyone is Unique in their own way. People need to learn to accept **everyone** for who they are and it is up to us to keep educating them. Some people do have the best intentions, but don't always think carefully about what they are saying, before saying it; the trouble is that once something is said, it cannot be unheard and comments made can stay with you forever.

Building confidence

This is something that comes with time. Often with children, they are just that, they are the way they are and many don't feel they are any different from anyone else, until others comment. Children with disabilities can see the world in a very different way, which can lead to anxiety and frustration. Parents can build their child's confidence and self-esteem by creating for them social stories or photo books that explain their strengths and praising them. Parents can build their own confidence to help with dealing with situations that are often out of their control. Keep calm, don't jump to conclusions too quickly, think first. Talk about how you are feeling, if not to family or friends, try some counselling. Medication and mindfulness breathing techniques can also help to calm you and increase your sense of wellbeing. Focus on your own strengths and praise yourself. Smile - smiling releases endorphins that make you happy. Laughter therapy is also a good tool to help with overwhelming emotions.

Parent quotes.....

When we asked our parent members for quotes for this guide, we had a good response (sadly we had so many quotes we can't fit them all in). There are both negative and positive quotes; by sharing them, it helps other parents to deal with situations that may occur.

“ Take one day at a time. It's okay to say I'm having a bad day. It's okay to have negative thoughts, we're only human. Good friends will be with you no matter what. You will find the strength to deal with whatever is thrown your way. Go with your gut instinct, you know your child best. Those milestones, which seem to take forever to come, will fill your heart with pride and joy. Small things, like my daughter sometimes looks directly at me and gives me the biggest smile - it makes my day.”



“ One thing that sticks with me was someone that was trying to be nice - saying “well at least she's pretty, so she doesn't have that real 'disabled' look”. I have to say I didn't know what to say to that... I saw it as an ignorant comment, but felt it came from somewhere kind. She looks how she looks, because she's C. and she's beautiful.”

“ YOU are Important, the best advocate for your child, the person who knows your child best SO please look after YOU. Time out is important be it physical or mental health, we are all here for support.”

“ As my husband said to me the day our daughter was born - "every child is unique" ... 15 years later, we got a diagnosis & found Unique & finally found the support we'd been desperately missing. An early diagnosis really is a blessing - knowledge is power. I wish I'd known 15 years earlier, honestly - it's such a relief to finally know!”



“ Something positive I have found, is how educated and aware my son's journey has made me. Our Unique journey, opened up a door to a special world, a world that only those of us on this kind of journey will get to experience. Yes, some days can be so very hard...but when I think of all I have learned, from supporting my son emotionally and physically, to learning a different way of parenting that is in no means traditional, but is exactly what he needs, to the copious amounts of other parents I have met and the support system we have built... it's incredible to say the least. There is beauty in this journey and I am grateful for it, even on the hard days. ”

“ I have had numerous stares and looks from children and their parents. Most have pulled their children away but quite a few I have knelt down and introduced C. to them, saying “I can see you looking at C, I expect you're wondering why she is in a big buggy or wheelchair? It's because part of her didn't grow in the same way, as maybe you or I, and she can't use her body in the same way. Would you like to say Hi to her, she would love it if you spoke to her too”? I'd be talking to the child but also looking at their parent. The fact that these negative comments sit with us is hard, all we can do is aim to change people's perspective and educate people with knowledge. ”

“ Sadly some comments stick. When my (now adult) son was about 4 or 5, he was sitting in his Major buggy and a child of a similar age walked past with his mother and pointed at my son, saying loudly "look at that big baby in a buggy" and his mother said out loud "don't get too close, come away" as if my son had something catching! It is one comment I have never forgotten, even this many years on. I was embarrassed. I didn't react, just silently wept inside. Children don't understand, but the parents should be adult enough to know that they shouldn't respond like that, it is very hurtful . ”

“ My son uses a Kaye walker, usually the response from people is positive. The hardest thing I find is people who wouldn't usually give you a second glance will stop and speak over the child to the parent saying how wonderful it is, how sweet etc. My son is 2 but has good understanding however no verbal communication. I wonder how he will feel about being spoken about in the sense of being 'disabled' or 'different' he does sometimes scream at them as they stop him moving on. My boy also has dribble bibs as he is always dribbling and screams in communication. Both have drawn attention to him even when in a pushchair as opposed to using his walker. ”



“ Your child is still your child and still needs to be loved and cared for. Enjoy them just like any new parent would and celebrate all of the achievements they make, however small they might be. ”



“ My son was calmly sat relaxing at a bench in a local seaside town watching the people, with his forearms covering his ears, when a couple walked past and the man said “that kids autistic”. I was so shocked as there wasn't a need to say anything, he was just minding his own business enjoy his day. I didn't have chance to say anything because they moved on quickly and I was in shock. ”



“ My son has never spoken, he has always made a monotone like noise, which has drawn the attention of people of all ages. When he was very young I used to get a bit annoyed with people’s stares and sometimes comments under their breath, but then I started to think differently – why should I let other people make me feel angry and annoyed, they are the ones with the problem. So I just started to smile at them instead of saying anything, they either look down or smile back. ”

“ It is important to remember that not everything said to us is intentionally negative. Some people say things because they know someone similar either in their friends or family and are just recognising some of the signs. Most of the time people are well meaning and don’t necessarily say things in the way we like to hear them, often not thinking before speaking and sadly they are things that we don’t forget easily. ”

“ When my daughter was about a year old, I was tube feeding her in a cafe. I saw a man staring at us from several tables away, and I was getting so upset because it had been happening a lot lately. I told my husband, “That’s it. I’ve had enough. I’m tired of people staring at her, I’m saying something this time.” But before I could say anything, the man got up and hesitantly approached us. “Excuse me, I’m sorry,” he said. “But is that a g-tube? My son is in the NICU and is getting one next week, I’m so scared.” I breathed a huge sigh of relief and I was so thankful I didn’t get upset with him for staring. I made him sit down and we talked for several minutes about getting supplies, how to do feedings, doctor’s appointments, and troubleshooting. ”

“ Whenever I am out with my daughter, children will just stare at her and literally stand open mouthed. It's amazing how children will watch the weirdest and wonderful things on TV and accept characters from Star Wars, but find human beings who look/walk/talk differently weird. I always smile at the children who are staring because they (and their parents) obviously have no understanding of disabilities. If someone is looking at my daughter, she will sometimes wave to say hello. I will say to her, are you saying hello? so the other person will understand what she is doing. I would love to go into Primary schools and talk to children about their understanding of being disabled. I think you need to get to children as young as possible, so that they grow up understanding and accepting what being disabled can mean. I think the fact that a disabled child is often shown in a wheelchair gives out totally the wrong message of what being disabled means and that there are hidden disabilities, so a child can look 'normal', but may have Autism, ADHD or a chromosome condition. I think we have a long way to go in educating people about disabilities. ”



“ Ignore the world's toxic and unrealistic messages about "perfect children" and embrace your child's amazing and unique qualities. Their journey may look different than others, but you have the power to learn, educate others and be their voice to advocate for their needs. You have the power to help your child get what they need, to be the best that they can be! ”

“ My son wears hearing aids, glasses, g- tube, and braces on his feet. He has a walker and a wheelchair. He does not walk independently and he does not talk. He knows limited signs. I have had adults ask "what is wrong with him? " normally my response is " Nothing is wrong with him, but what is wrong with you?" I tend to just walk away because I can't deal with stupid. I also have a few in my family who have taken a long time understanding or admitting he has limitations and barriers and really has something going on. My mother for the longest time would say... "His hearing isn't that bad, he hears me without his hearing aids." Now however I think she gets it. ”

“ We have used a business card explaining our sons adhd/autism - I've approached people & given them a card to apologise / explain his behaviour. I've not been good at dealing with other people, I used to mostly walk away or avoid social situations in the first place. I have lost friends and drifted away from family as a result. ”



“ I recently had to take my 2-year-old son to A&E as I thought the monkey had swallowed something he shouldn't have. I already felt like a really rubbish mom, two nurses came in, they asked me why I didn't just ask him and I explained he is non-verbal with learning disabilities to which the one huffed. They treated him like he wasn't there, in the sense they made no effort to engage with him at all. I know it's not the same situation but just because he can't talk, doesn't mean he should be overlooked it was horrible and I'm not going to lie but this made me feel angry. The bloke who x-rayed my son and the doctor that came in after though were second to none and actually engaged with him. ”

“ Remind yourself that you don't have to hide and you don't have to explain. You don't owe anyone an explanation of what your child's differences or needs are. I felt in the beginning like I had to almost justify myself and actually that was completely wrong. I had to change my mind-set to be proud that my little girl was different and that I could use it as a way to promote positivity and inclusivity. That I still had a beautiful little girl who is a little miracle. ”

“ Focus on the things your child can do each day and celebrate the little steps. Share those positives with like-minded people. Surround yourself by those who'll help that way of thinking. Sometimes a longer view can help. 20 years down the line my daughter is doing things that still give me hope. ”

“ My son’s chromosome disorder causes him to be significantly taller than his peers. I remember when he was smaller, even when he was not yet 2 years old, he and I were regularly told by parents and staff at soft play areas that he needed to leave because he was "too old" to be there based on their perception because of his height; yet I knew he was often much younger than some other children playing there. I'd have to make him wear an age badge or specifically take something with me to prove his age to staff in the end, but even so, parents would frown, make comments and come over to tell me to remove him. When he'd have behavioural wobbles (as any other child his chronological or developmental age) it was even worse. I had comments such as "kids like him shouldn't be allowed in places like this", sometimes parents themselves would go up and shout at him to leave, thinking he was older, when he didn't even have the language skills to comprehend what they were saying or doing. I have a few stories of parents, staff and children being nasty to him because they thought he was older, based on his height over the years, even now he is 15, he is 6ft 5 and still growing, he is regularly on the raw end of people overestimating his abilities because of his height. Developmentally he is a decade behind his chronological age, so it's a big conflict for people to comprehend and of course you can't see how delayed he is just by looking at him. ”

“ It's ok that your child's milestones may be achieved when they are ready and not on the "standard" scale. It makes the milestone achievements that much more beautiful and worthy of celebration. ”

“ My son has a very visible eye problem. He can't coordinate his eyes and one eyelid droops. If anything his glasses mask it. No-one has ever said anything about it. Our consultant has also been sympathetic and is happy to operate to improve the cosmetic appearance, recognising the impact that it may have in later life. We have been very lucky with how people have treated him. These days I think people are much better at being sensitive and helpful with disabilities. ”

“ Don't focus so much on their differences that you forget to enjoy their babyhood and childhood. They are not defined by their differences. In our house we say that our son took the scenic route, but he got there just the same as those who took the motorway! ”

“ Notice the new things your child does. Their development may be slower but celebrate each tiny little achievement. This was the best piece of advice my genetic nurse gave to me. It changed my complete mind-set. He’s 18 now and we are still noticing and celebrating new things. ”



“ Your child will develop into the person they are meant to be, one small step at a time. Even with typical children, one crawls at 6 months and another at 9 months and mamas pull out their hair. For us, the time frames may be longer, the things we celebrate different, but our children still bring light and joy to our life. I waited 14 years for my son. I've enjoyed every day of him. Even the rough ones. ”

“ Trust yourself and your child, celebrate their achievements, however small; as they will shine and achieve in their own way daily. ”

“ Our daughter has to use reins when walking as her balance isn't always great and she still uses a pushchair for when her legs tire. She is 5. We've had several comments and looks when using either. At first they used to bother me, but now I just brush the comments off. We also have a tag on her pushchair that says treat as a wheelchair as it used to be refused in some places, when she needed it. ”

“ Our biggest struggles include:

- ◆ safety/ impulse issues (we have had some improvements with the help of ABA) and
- ◆ negative attention seeking behaviours (doing and saying things to get a rise and/or in response when not getting her way). ”



“ My granddaughter who is 11years old has drooled all her life. She had a soft trachea and choked severely on her saliva, so to this day still spits it out, even though she can swallow safely now. We were told by our PT to try wrist bands like the ones used for sports like tennis. They come in many colors and are inexpensive. She has the ability to wipe her own chin when asked, so this works out well. She also knows some sign language, so I taught her the sign for "dry" so we could use it in public and it would be more discrete especially when interacting with others. I just had to get in her line of vision and sign to her and she will dry her chin. I carry around one of those bags that can hold wet things and have 2 pockets. 1pocket had the clean wristbands and the other is where the wet ones go. It is great when out and about. ”

“ Your little person is incredibly precious and special. Being different doesn't stop that being true. Remember to celebrate all the little things that make your family your family. Reach out when you feel ready, because there is help out there and you are not alone no matter how alone you might be feeling right now. ”

“ Write down milestones and details, and then forget about them until you need them for appointments. Between appointments live in the moment, and, unlike the appointments where you are asked to dwell on the negatives, dwell strongly on the positives, enjoy the snuggles or whatever your child has to offer. And think in seasons, where you are only in THIS season for so long. ”



“ I know what I'd like to mention is not as 'serious' as all the conditions that our other lovely Unique children have to bear but - especially in this country I must add- it seems that dog owners just cannot grasp the idea that NOT everyone is a dog-lover; especially true to those children like my son aged 9, who has a meltdown immediately a dog starts barking near him unexpectedly. It happens sometimes that we either don't see the dog walkers approaching or have no time to put his ear defenders on. There has been an occasion when I turned him away from two huge German Shepherds with whom we just suddenly came across with (no leash/nozzles, of course, very rare nowadays) and sort of shielded him with my body from the sight or the 'expected loud, high pitched noise. When the owners (middle aged couple) saw us, they walked away without a word, but I noticed the man turning around after a short while looking at us - me still holding my son deciding whether further comfort is needed - as we were some kind of weirdos. I don't expect everyone to register straight away that a child with ear defenders might get a meltdown by seeing dogs, but there surely should be some platform to 'educate' the public or rather raise awareness of children with similar problems my son faces every time we go out for a walk. Such occasions can actually ruin not just a walk, but a day that has begun with lots of smiles and jumping/running around. Perhaps I don't have to mention those dog owners who despite the fact that they clearly see my son's state, they insist on persuading us that their dogs are really friendly, loves kids....etc. I used to be patient and say 'sorry, we're not keen on dogs, my son is autistic' or something similar, but now I usually just walk away with my son. ”

“ You have to remember that as humans we are entitled to all of our emotions and feelings. It's okay to feel grief, sadness, hurt, discouragement, and even anger. What's not okay is to choose to allow the emotions and feeling to swallow you whole and not allow yourself to move past them. The thing is, once you accept those feelings and move on, then you get to see the true beauty of having a unique child. Only then do you see how absolutely amazing they are and just how much you've been blessed to be their parents. ”



“ My daughter, was born with a mosaic of monosomy 18p and isochromosome 18q. This caused her spine to end at L5, created a club foot on the right, congenital vertical talus on the left and for some unknown reason, caused her tongue not to form (she has one about the size of a bird). Her spinal condition has affected her bowel/bladder control and lower leg function, so she has AFO's on both feet and a cecostomy button to deal with her bowels and keep her clean. Due to her small tongue size, she is difficult to understand. I don't have a great deal of information to share, but I find the thing that has worked for us in regard to both her AFOs and cecostomy button is to refer to them as magic. L. has been poked and prodded all her life to try and correct her feet, etc... so she has a definite aversion to appointments of any kind. When we started the process for her AFOs, it was after a long series of casts, which L. hated. The thought of another thing that might go on her feet was not looked upon favourably until we told her they were magic shoes that were going to make her walk. She already had the drive to do it, but the idea that the hated new shoes were magic helped her to lose her reluctance in wearing them. We used a similar approach to her cecostomy button. Granted, it was not an easy process and it took her a while to get used to it, but the final result definitely made her happy. In fact, once she was healed, we had the new problem of her walking through the hallway in her school, pulling up her shirt and showing everyone her “magic button”. We had to have quite the long talk and explanation about why we don't do that! ”

“ You know you are doing an amazing job as parents and you know what your child needs.”

“ Be kind to each other. It's not about who is worse off, it's about supporting each other, so that we know we aren't alone. Our children go through things as much as us adults do. Our children should have a voice too.”



When J was born I cried for weeks. A more experienced Unique mother told me that one day I would wake up in the morning no longer crying, but just wondering if I had cereals for breakfast. I thought she was mad, I just was not like that, it was too hard and I could never cope...but she was right, we are now a very happy family and J aged almost 14 speaks 3 languages fluently. Some doctors told us that it was a really bad idea and that she would never cope, but our circumstances meant that we decided to persevere and it worked!”

“ Something your doctors might not tell you - whatever health and developmental problems your child has to cope with, they may still be the one with the biggest smile on the fairground ride. The one who loves it so much, they just can't wait to go on it again and again!!! ”

“ Write down milestones and details, and then forget about them until you need them for appointments. Between appointments live in the moment, and, unlike the appointments where you are asked to dwell on the negatives, dwell strongly on the positives, enjoy the snuggles or whatever your child has to offer. And think in seasons, where you are only in THIS season for so long. ”

Inform Network Support



Understanding Chromosome & Gene Disorders

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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.

Thank you to Emily Perl Kingsley for allowing us to reprint her 'Welcome to Holland' poem.

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