



The Newsletter of SWAN UK

Life without a Diagnosis

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SWAN UK is a project run by Genetic Alliance UK offering support and information to families of children with undiagnosed genetic conditions. Registered charity No. 1114195

If you are interested in becoming a member of SWAN UK contact Lauren on 0207 704 3141,
SWAN@geneticalliance.org.uk
 Facebook www.facebook.com/SWANchildrenUK
 Twitter @SWAN_UK



Genetic Alliance UK
 Supporting. Campaigning. Uniting.



About SWAN UK

SWAN UK is a support project run by Genetic Alliance UK offering support and information to families of children with undiagnosed genetic conditions.

Genetic Alliance UK is the national charity of patient organisations with a membership of over 140 charities supporting all those affected by genetic disorders.

Our aim is to improve the lives of people affected by genetic conditions by ensuring that high quality services and information are available to all who need them.

www.geneticalliance.org.uk

The support group Syndromes Without A Name (SWAN) was originally formed in 1999 by the grandmother of a child with severe developmental delays but no diagnosis for her condition.

Over the following decade SWAN provided support and information to numerous families with over 1300 members. Close links were formed with a number of professionals in the UK and with similar support groups abroad.

In 2009 the group's founder was advised to give up her role as chair of SWAN because of health problems.

Most SWAN members were parents of children with severe disabilities, there was no-one able to take over. With minimal resources the group had to close and has not been operating for the last 2 years.

In 2010 Genetic Alliance UK secured funding from the National Lottery through the Big Lottery Fund to employ a full time coordinator for 5 years to re-establish support for families of children with undiagnosed genetic conditions.

Based on the aims identified in the consultation exercise the project will:

1. Develop a community of families with undiagnosed genetic conditions for mutual support and information sharing
2. Develop a network of health and social care professionals with expertise in undiagnosed conditions
3. Increase awareness and understanding of undiagnosed genetic conditions

In May 2011 the new SWAN project coordinator Lauren Roberts started in post. The project has an advisory committee made up of parents of undiagnosed children and professionals working in the field of genetics, paediatrics and the voluntary sector.

Membership of SWAN UK is free to parents of undiagnosed disabled children.

If you are interested in becoming a member of SWAN UK please send your contact details to SWAN@geneticalliance.org.uk or call 0207 704 3141 ext 110.

THE SYGNET is the name of the original SWAN newsletter. You can read these old articles online at : <http://www.geneticalliance.org.uk/projects/swan.htm>

SWAN UK is funded by the National Lottery through the Big Lottery Fund.



Please note that this newsletter is for information purposes only. All information is correct to the best of our knowledge at the time of going to print. Any organisations or products included here does not imply that they are recommended by or endorsed by Genetic Alliance UK or SWAN UK.

SWAN Dorset

Do you live in the Dorset area?

Would like the chance to meet up with other families of undiagnosed children near you?

If you would like to meet other families for friendship, information sharing and mutual support then please get in touch with Claire on clairechissell@hotmail.com

#SpecialSaturday



"It's [#specialsaturday](https://twitter.com/specialsat) today my specialneeds child has taught me understanding and love and NEVER TO GIVE UP!"

Special Saturday is a group of people who want to raise awareness about children and adults with special needs.

We aim to do this by devoting every Saturday to informing and spreading the news to as many people globally as we can about both the difficulties we face raising our children or by having special needs AND how amazing our special children are, including their achievements.

To find out more about how you can get involved or to read what has already been submitted over the previous weeks please visit:

www.specialsaturday.org
www.facebook.com/specialsaturday
www.twitter.com/@specialsat

What do you want to see in future editions of the newsletter?

How do you think it can be improved?

Would you like to contribute an **article or tell your child's story?**

Do you have information you would like to share with SWAN families?

If so please get in touch, SWAN@geneticalliance.org.uk

Deciphering Developmental Disorders Study (DDD)



Over the next 5 years a study based at the Wellcome Trust Sanger Institute in Cambridge called **'Deciphering Developmental Disorders (DDD)** is aiming to use the latest genetic testing methods to help understand why some children get developmental disorders.

Advances in testing chromosomes means that scientists may now be able to detect changes to **chromosomes that they couldn't see** before so they may be able to make new diagnoses. If your child has previously had a microarray test done but nothing was found it is still worth being on DDD as they are using the latest technology that can pick up much smaller things than is detected in NHS labs.

The study aims to collect information from 12,000 undiagnosed children and their families. It will be done by doctors from the 23 NHS Regional Genetics Centres and scientists at the Wellcome Trust Sanger Institute.

Families who take part in the study will need to provide DNA samples from their child and where possible, from both biological parents. This will be done by providing a sample of saliva. Ideally they would like samples from both parents. If this is not possible, the study may in some circumstances be able to enrol a child where a sample from only one parent is available. However because parental samples are so important in sifting and interpreting

the huge amount of genetic data generated by the study for each family the chances of finding a diagnosis will be much reduced.

The study will not accept samples where they are known to be from an incestuous relationship. They will be able to tell this from the samples. If it is discovered they have a duty to report it.

There will also be some questions to answer about your family history. The samples will go to the Wellcome Sanger Institute to be stored and analysed. The results will be added to the DECIPHER database using a code to protect the privacy of families who take part.

The DECIPHER database is an international database used by doctors and researchers around the world to help diagnose developmental disorders. Because the changes that happen to chromosomes can be very rare, sharing information across the world should help doctors and scientists learn more quickly so they understand more about rare conditions.

Most children on the DECIPHER database have a diagnosis (i.e., microdeletion/duplication) that is thought to account for their problems. There are only a few children who are on the DECIPHER database who do not yet have a diagnosis. Such children can be enrolled in DDD but have to be specifically recruited - they will not automatically be recruited.

There is strict eligibility criteria to take part in the study and you must be referred through UK NHS Regional Genetics Centres. Only certain developmental disorders will be appropriate – if you are not already under a Regional Genetics Centre you will need to get your GP to refer to one before you can then see if you are eligible for the study. If you are not sure how to go about this please get in touch with us here at SWAN UK so we can support you.

Anything found by the research study will be fed back to your regional clinic via the DECIPHER database for your clinical geneticist to talk to you about. They may need to do another test to validate / confirm what was found by the DDD study team. Ultimately it is up to your clinical geneticist to make the decision if a diagnosis is appropriate and to let you know about it. They will be emailed when/ if the DDD team find something about your child and enter it onto DECIPHER. The DDD team aim **to feedback even if they don't find anything** so that you will know one way or another if anything was found.

The DDD study are not accepting :

- children over 16. If your child is approaching their 16th birthday and you think they may be eligible then time is of the essence so get in touch with your regional genetics centre to see if you can be on the study. You will need to give your consent and provide your samples before your child turns 16:
- they are not accepting samples from terminations or still births.
- samples from adopted children
- samples from children of known incestuous relationships

You can find out more about DDD at www.ddduk.org

If you would like more information about DDD please contact SWAN@geneticalliance.org.uk

A Diagnosis

We're pinning our hopes on Deciphering Developmental Delay To provide some answers come what may!

If one more doctor says a diagnosis doesn't matter,
I think I might go mad as a hatter!
We know it won't change our special boy,
Making him better isn't our ploy.
You don't understand why we've made it our mission.
That's because you're not in our position!

Thanks to Claire Smyth for sharing

Living Without A Diagnosis : Information for Parents, Carers and Families



"If you have a diagnosis you're on some sort of track. Without a diagnosis it feels like you're lost in a swamp."

How common is it not to have a diagnosis?

Even though most people do not realise it, having an **'undiagnosed condition'** is actually quite common. For example, it is thought that about half (50%) of children with learning difficulties have no definite diagnosis.

Even without a diagnosis, children are still entitled to receive healthcare, education, benefits and services that are appropriate to their particular needs. Not having a diagnosis should not stop a child from reaching their full potential.

Why are some conditions difficult to diagnose?

There are a number of reasons why making a diagnosis is not easy:

Sometimes children have a number of different problems that do not all fit into one specific recognised condition

Many conditions have similar features and health problems which makes it difficult to be accurate about a diagnosis

Certain conditions are so rare that it can be difficult for doctors to identify the condition. The same condition may affect children in different ways which can add to the problem of

Some features of the condition may **not appear until later on in the child's** life. This may cause a delay in getting a diagnosis, or in some cases a change of diagnosis

(Taken from Contact A Family, *About Diagnosis*)

Doctors are now able to diagnose more conditions than ever before due to advances in our knowledge, and improvements in testing techniques and medical equipment.

Some of these conditions will be genetic, (caused by mistakes in the **"blueprint" of information we get** from our parents), others will be the result of problems at birth or infection during pregnancy or in the first few years of life.

However, because of improvements in science, there may be an expectation that doctors can always find a diagnosis. If they can't, it can be extremely disappointing and confusing for parents.

It is worth remembering that, even if your child does not have a diagnosis now, advances in medical knowledge and testing techniques means that in the future there may be an **explanation for your child's** problems.

The emotional impact

Finding out a child is not developing as expected can be a very worrying and anxious time for some parents. Parents described feeling alone, angry and sad, particularly because **their child was not the 'problem-free'** child for whom they had hoped.

"I think parents need to know that it is OK to feel resentful, angry and negative at times"

Waiting for the results of tests was also stressful. It was disappointing and frustrating when the results did not give any definite answers. Some women said that when they first found out there was a problem they were worried that it may have been their fault, or something they had done while they were pregnant.

However, the reality is that these things are rarely anyone's fault. In most cases these things just happen, and there is no way that the parents could have changed the outcome.

Some parents said that not having a diagnosis made them feel out of control of the situation. They were unsure whether they would ever get **a diagnosis and they didn't know what the future would hold for either them or their child. Without a 'name' for their child's problems, they were** unable to search for information. This was very frustrating.

"I don't know what is going to happen to him in the future and that is the worst thing I think, the 'not knowing'"

Parents often used the term **'emotional rollercoaster'** to describe the day-to-day experience of looking after a child with a disability.

At times it can be isolating, difficult and frustrating, but there was also an overwhelming sense of love and admiration that parents had for their child.

"She's so lovable. She makes me smile, she makes me glow. I just love her to bits."

Advice from parents

Try to enjoy your child. It is easy to focus on all their problems and forget to enjoy watching them grow up.

Once you accept your child for who they are, your life becomes easier.

Talk to other parents in a similar position, such as members of the SWAN UK support group, as they are likely to understand what you are going through.

Don't compare your child to other children of the same age, or how your other children were at that age, as this can be upsetting. Every child is different.

Try not to focus on the milestones your child has not reached. Focus on those that they have. Once you accept your child for who they are, your life becomes easier.

Try not to worry too much about the future. Try to focus instead on dealing with each day at a time.

"People kept saying to me 'just enjoy him because they grow up so quickly', and I do feel that we didn't enjoy him as much because of all the anxieties that we had about what was going to happen. If it was to happen over again I'd definitely say try and put that to the back of your mind and just enjoy who he is."

Raising a child with a disability can be hard work, both physically and **emotionally. Don't be afraid to ask** for help, and take help when it is offered.



"As much as you think you are coping, take a step back and say, well, perhaps I need help with this or that. Don't be afraid to ask for help. It doesn't make you any less of a parent."

Having someone you can really talk to and who is not emotionally involved (such as a counsellor or health visitor) can be a great help, **even if they don't have all the** answers.

For peace of mind, try to be as organised as you can. You might want to put certain procedures in place, for example organising your will.

It's easy to forget to look after yourself when you are looking after a child with a disability. Take time out every once in a while; go out with friends; look after your health.

Doctors and specialists

Going to lots of hospital appointments and seeing different specialists was a frustrating and time-consuming experience for some parents. Often, a number of different specialists were involved in their child's care, and children were often referred

from one specialist to another as the search for a diagnosis continued.

One issue that parents found particularly frustrating was the frequency with which they have to **repeat their child's 'life story'**, including a history of all the symptoms, tests and the specialists they had seen. It can be even more frustrating when you don't have a diagnosis because you do not have a **'label' that summarises all your child's** problems.

Whilst repeating this information every time you see a new specialist is understandably frustrating, in most cases it is necessary because there will be very specific things the specialist is interested in finding out that relate to his or her particular speciality.

"If you are able to go in and say, 'He's got so and so disease' then they go 'Okay that is fine', but you can't. You have to go back from birth when you were first told, and explain the whole thing."



This information is taken from the EuroGenTest leaflet 'Living Without A Diagnosis'

Every Penny Counts!

Raising a disabled child can be an expensive business, if you have something specific you need money for these organisations / websites might be able to help:

www.turn2us.org.uk
www.familyfund.org.uk
www.caudwellchildren.com
www.disability-grants.org

If you aren't already getting it you should also see if you can get Disability Living Allowance (DLA). This form is very long and it is a good idea to get advice before you complete it. Try and remember to use **key phrases like 'needs ongoing and frequent support with . .' or 'requires constant adult supervision.'** Make sure you photocopy the form before sending it in so you can refer to when you need to do future renewals. For more info see www.direct.gov.uk

If you or someone in your house has a recognised condition that means you need to use more water than other households (such as frequently washing sheets) you may be able to get a discount on your water rates. Check your with your local water company to see if they offer this and you are eligible.

You may also be entitled to a reduction on your council tax if:

Your house has an additional bathroom or kitchen to meet your **child's needs**

There is an additional room (not bathroom / kitchen) to meet their needs.

Extra space is needed for a wheelchair

If your child is getting middle or high rate Disability Living Allowance (DLA) and you can provide a doctors certificate stating that they are **'severely mentally impaired'** you may also be able to get a reduction. If your home is eligible, your bill will be reduced to that of a property in the next Council Tax band down.

Education Information



IPSEA is a national charity providing free legally based advice to families who have children with special educational needs. They have specially trained volunteers who can offer information and advice to families of children with special educational needs.

They also have lots of useful information and resources available on their website including lots of template letters to help you request support for your child from your Local Education Authority.

It is always a good idea to get information and advice before you start trying to get more support in school, especially if you are going to be applying for a statement of special educational need. www.ipsea.org.uk

Nate's Story

I find the best way to describe our life is that it's a roller coaster. Though every time I say this out loud, or even in my head I face an incredible urge to punch Ronan Keating in the face. But I digress...



In the last stages of my pregnancy I felt something was wrong. My baby moved less and my bump seemed to have stopped growing. I was concerned, the professionals were not. I went into labour at term, my son was undiagnosed breech (he must like that word) until the pushing stage (oops midwife dropped a right clanger there methinks) and was born 6lb 15oz by emergency c section.

He didn't cry, move or breathe needing a little oxygen but then started on his own. As I lay in recovery with my son I could hear other babies crying. Mine didn't. He didn't even open his eyes. He wouldn't feed. The midwife took him to scbu "back soon" she said. They were concerned with his "tone". I thought they meant his funny purple colour, little did I know.

My husband returned and I said the baby has gone to scbu but not to worry. I was pushed along to scbu and the dry gave us the news. A potentially very serious genetic condition she said. Did you notice his unusual features she said. Then gave a long list. Lovely.

Thus began 4 weeks in scbu where my gorgeous boy developed a need for oxygen, needed to be tube fed, had ultrasounds, x rays, bloods, an MRI, all normal. We had to wait 4 weeks for the micro array chromosome test to come back and

were told the geneticist was confident of a diagnosis- Zellweger's- 6 months to live.

We concentrated on getting used to tube feeding and encouraging the bottle. And he came home. His chromosomes were normal. He was still undiagnosed. One of the many consultants he saw was concerned about his breathing at night so after we were home for 6 weeks we went onto hospital for a sleep study. Got home 8 weeks later with a ventilator, sats monitor, and after several episodes of apnoea.

The next "confident diagnosis" was of Schinzel-Gideon disease. We waited 13 weeks for that life limiting diagnosis to be disproven. He has been in and out of hospital with several respiratory viruses over the winter. Its been rough. Things have been hairy at times. These are all the downs on our rollercoaster.

But the ups, oh the ups are so good. The half rolling, saying mama, gaining strength, playing, smiling, laughing, eating. These things are made so much better when you don't expect them. He doesn't have a "path". We don't know how he will develop. But, well, I've always liked surprises.



And we've changed, my husband and I. We are his advocates. We don't agree with Doctors automatically, we ask probing questions, we push at times, and get professionals to take a step back at others. We battle for equipment and resources, and have to explain constantly that we "just don't know". We access support and help where we can.

Having an undiagnosed child is hard. The testing, waiting, and frequent appointments are draining. But when faced with some diagnoses offered I happily bury my head in the sand. Ignorance is bliss... Some days!

I hope that doesn't sound too depressing. We have a lovely family life. A new version of normal if you like.

Tommy's Story



Tommy was born 6 weeks early .He was very weak and cold. When he was born it took him a few weeks to begin to feed properly. At 4 days old it was discovered he had no response in his right ear and that it was also deformed. We had many concerns about Tommy but just kept getting told it was because he was premature - but to us things were not right.

Tommy seemed so unaware of his 4 brothers and sisters running around him. He was so floppy it was like holding a rag doll. He never gave eye contact and was so slow to feed. At 4 weeks old it was discovered Tommy had a under active thyroid so he was placed on thyroxin for this.

2 days later he began to be very sick and it was discovered he had pyloric stenosis (a narrowing of the outlet from the stomach to the small intestine) and had to have an operation.

As the months passed Tommy didn't seem to get any stronger and as the winter months drew in Tommy was forever chesty and would spend the nights going blue due to the build-up of phlegm that he could not clear. He also had very painful constipation.

Finally, in the February they gave Tommy some patches to help dry up the phlegm and he finally smiled for the 1st time. He still could not roll and had no head control. It was then discovered Tommy had Nystagmus.

We spent months going back and forth to different appointments which gave no answers. Tommy finally rolled a week before his 1st birthday. By now it was very noticeable that he had problems and so the physio and OT appointments began and so did many tests.

In Sept '10 it was discovered Tommy had a heart condition called Hypertrophic cardiomyopathy which meant yet another lot of meds for him to take every day. By November '10 Tommy finally sat on his own for the 1st time ever. As the winter months began so did the chest infections in March 2011 Tommy was very ill with pneumonia. He soon recovered but then it was also discovered he had lost hearing in his good ear. It's now May 2011 and Tommy has had many tests which have all come back clear.

The reports from all Tommy's doctors state as follows:

"Thomas is a cheerful young boy. He has development delay; he has hyper mobility and muscle weakness. Tommy's has nystagmus which affects his vision. He had a slightly large forehead, a deformed ear and foetal toe pads. He has a high palate. Thomas doesn't not produce any tears and has a funny high pitched cry"



Tommy is now weight bearing with support and bum shuffles around the room. **He doesn't say any words as yet.** Tommy has days where he is very weak and will just flop. On these days it seems as if his whole body closes down.

Tommy is a very happy little boy full of lots of smiles and love for everyone. We hope one day the **cause of Tommy's problems will be found** but for now he is our little swan.

You can read more stories from families with undiagnosed children at www.swanuk.wordpress.com

If you want to share your **undiagnosed child's story please** email SWAN@geneticalliance.org.uk

New Lifestyle Magazine



My Child & Me is a new lifestyle magazine for parents of young children with additional needs. It is bi-monthly and includes realistic, down to earth and friendly advice about parenting when your child has additional needs as well as providing support for you. Each edition will offer guidance on a range of topics from general parenting to the more specialist:

The latest issues includes:

Solving sleep issues
Your rights as a carer
Grandparents

My Child & Me's Guide to Choosing a School
Makaton

Step by step: Autism
Eating and drinking products reviews

You can subscribe for only £14.95.
www.mychildandmemagazine.co.uk

We always want to hear from you so please send your thoughts, comments, stories, experiences or article ideas to editor@mychildandmemagazine.co.uk or via twitter

Breaking Down Barriers

The 'Every Disabled Child Matters' (EDCM) campaign has expressed "concern about the impact of the Government's proposal to reduce disability additions for low income and out of work families with disabled children, on making work pay for disabled children.

In their recently published report "Breaking Down Barriers" which was undertaken jointly with The Family Fund, EDCM suggest that "Reducing disability additions will make it much more difficult for these families to afford work related costs that they pay a premium for such as childcare. For those families who are unable to work because of their caring responsibilities this will make the move to employment much harder as they will be unable to afford costs associated with becoming work ready. This policy is therefore likely to result in many more families with **disabled children living in poverty.**"

For many families parental leave is crucial for balancing their working and caring responsibilities. It is vital that parents of disabled children applying for parental leave feel supported both from their employer and colleagues, during difficult times of their life

They make 3 recommendations to the government to help families of disabled children make work pay:

1. Help families afford high quality accessible childcare
2. Provide financial assistance to families of disabled children
3. Promote family friendly working



Every Disabled Child Matters (EDCM) is a campaign to get rights and justice for every disabled child www.edcm.org.uk.



The Family Fund is the UK's largest provider of grants to low-income families raising disabled and seriously ill children and young people.
www.familyfund.org.uk/

Special Siblings

"I am the proud mother of a disabled child. He is a dangerously cute, disarmingly intelligent eccentric who wouldn't be the amazing little boy that he is today without all the added extras that he was born with. I wouldn't change any part of him, because he never would have become the child he is without everything else, and he's perfect, in a beautifully human and imperfect way.

But I am also the proud mother of two able-bodied children (or whatever generally accepted pc definition you wish to categorise them as). One is serious and smart and capable of an extraordinary ability to love and an even more extraordinary ability to survive on very little sleep. The other skips and pirouettes through life managing to juggle frivolity with a deep and sensitive understanding of other people's emotional complexities, and just happens to have the funniest bottom of anyone I have ever met. These two are the unsung heroes, the forgotten siblings in the special needs world.

They are the ones who have sacrificed the most but are rarely applauded. They are the ones who learnt to dress themselves and comfort themselves before nature was ready for them to grow up, the ones who willingly put their needs to one side so a crisis could be managed, the ones who remember to cry quietly at night so they don't worry their exhausted parents. They are the ones who have to say goodbye to their mummy when she has to stay in hospital and return to a lonely house not knowing when they will get to enjoy the comfort of a complete family again. They are the ones who still manage to be thrilled for their sibling when a charity brings a smile to their faces, without lingering on, or giving a voice to their own sadness inside.

They are the ones who stand up to the children who poke fun at those don't understand, and quietly listen

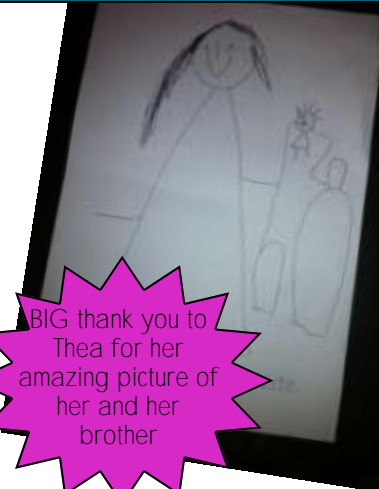
to their friends talk about days out and holidays that they don't enjoy. They are the ones who clap and cheer the loudest as their sibling comes last in the sports day race and won't give up trying to find a hidden smile in an unwell child. They are the ones who learn lessons in grief and injustice alongside their times tables and who start in counselling at the same time they start school. They are the ones who reassure the adults that they are fine, knowing that the burden their parents are bearing is heavy enough at times.

It is these children that are special, these children that should be celebrated and applauded just as much as their disabled siblings. It is my family, my three precious individuals who *all* overcome the challenges that disability can throw our way each and every day, who are *all* remarkable, who are **ALL heroes."**

You can comment or read more at www.justbringthechocolate.com

"Jack is the older brother of my little Swan Joe. At just two and a half years old he has had to grow up such a lot in the last year and I worry so much that our family circumstances have changed him. I imagine to some extent they have, but I hope **that he doesn't miss out on a care free childhood as a result.**

He is by nature a boy's boy; lively and boisterous, he loves trains, cars, Thomas the Tank Engine, Fireman Sam, wrestling and running around **like a lunatic.** He'd spend all day outside come rain or shine and, thanks to Peppa Pig, has developed a fondness for jumping in muddy puddles. Yet he is kind and caring in a way that you would never expect from someone so young. He is so gentle with his little brother and it is obvious he loves him dearly. I **watched them 'playing' together one day.** Play between the two brothers is not and never will be the rough and tumble play of boys just 14 **months apart. Joe's vision is very**



BIG thank you to Thea for her amazing picture of her and her brother

poor, he doesn't always respond to sound, he has low muscle tone and can't sit. Jack lay on the floor beside Joe and took a soft, tactile ball. He **held it up to Joe's face and stroked it gently along his cheek.** I genuinely nearly cried watching them. **It's not** how I expected my two boys to play together but Jack intuitively knew how Joe needs to play. Long before our visiting teacher suggested on-body signing – a form of basic communication for visually impaired children with additional needs, Jack **would hold and shake Joe's hand to say hello to him.** Coincidentally this is **the sign for hello.** I've no idea why Jack started doing this – **it's certainly not something he's seen me do.** He **strokes Joe's face gently as he talks to him** and even as a very young child himself, perhaps just one and a **half, he would move to stay in Joe's line of vision while he spoke to him.**

Other children at two and a half would get jealous when their mothers spend time with other children and not them. Not Jack, he gets jealous **on Joe's behalf.** **"Talk to my Joe", he'll say if he feels that attention is being directed elsewhere, particularly if it's to another baby.**

Jack is an amazing little boy. It's easy to forget that he is really only a baby himself. Having a child with special needs affects the whole family and special siblings like Jack should be celebrated too. Joe really **couldn't have chosen a better big brother than Jack"**

Sibs

for brothers and sisters
of disabled children & adults

Sibs is the UK charity for people who grow up with a disabled brother or sister. Siblings who grow up with a disabled brother or sister often have more worries, more responsibilities and less attention from parents than **those who don't. They have a** lifelong need for information and often have to cope with difficult situations. Many feel isolated and that others do not understand how they feel. They also want to have positive relationships with their disabled brothers and sisters and to be able to choose the role they play in current and future care.

What Sibs does

We run a helpline and website for siblings, deliver parenting phone sessions, run workshops for siblings, train professionals and volunteers to run sibling groups for children, and run a support project for adult siblings. We also influence service provision for siblings across the UK.

Help for parents on supporting siblings

Many parents come to Sibs to ask for help with sibling issues, such as:

How do you explain to a six year old that his brother's condition may be life limiting?

I need ideas for giving my daughter attention as she is feeling very left out.

My son seems withdrawn since his sister has come out of hospital, how can I help him?

My daughter is being teased at school about her disabled sister but she won't talk to us about it.

My son is frustrated with his brother's behaviour – how can I help them play together?

Parents can talk to a Parenting Siblings Advisor in a one hour phone session on how to deal with specific sibling issues. The advisor helps parents feel confident about supporting their sibling child provides

them with activities that are easy to use in a family where time and resources are limited.

Any parent who would like to arrange a free parenting phone session can call Sibs on 01535 645453 or email info@sibs.org.uk

Visit our website www.sibs.org.uk for tips for parents and for more information about the needs and experiences of siblings

UNIQUE Support

Each year many hundreds of babies are born with a rare chromosome disorder. Many are very sick and disabled, some unable to walk or talk, whilst others are relatively unaffected and only find out about their chromosome disorder when they come to have children themselves.

Chromosome disorders involve missing, added or rearranged genetic material. They can happen to absolutely anyone and yet support, information and advice for families are patchy and can be difficult to **access. It's often the case that professionals simply don't have the** knowledge of specific rare chromosome disorders to be able to **help. That's where Unique comes in!**

Unique fills a crucial gap in service provision, providing specialist information, support and guidance and helping to reduce the isolation felt by the parents and carers of those affected. Individual chromosome disorders can be very rare, some, as our name suggests, are quite literally Unique!

Collectively though, they affect approximately 1 in 200 live-born babies. With the roll out of more sophisticated genetic techniques such as array CGH analysis, many more families are joining us whose children were previously undiagnosed but have recently been shown to have a chromosomal microdeletion or microduplication.

A registered charity, Unique's services to families and the professionals working with them include a Listening Ear, telephone,

postal and email helpline service which is often the first point of contact for parents of a recently diagnosed child. Our Information Project has so far produced over 130 family-friendly chromosome disorder-specific information guides, using information from our specialised database and the peer-reviewed medical literature.

We publish a regular magazine containing articles from families and lots of special needs resources and have a network of more than 160 local volunteer contacts, raising awareness of us in their area, supporting local families and signposting them to services they need. We are also pleased that our CEO, Dr. Beverly Searle, is a **member of SWAN's new advisory board.**

Above all, Unique means that those living with rare and poorly understood chromosome disorders are able to find a home, an identity and others in the same boat. This is particularly important when many rare chromosome disorders have only a technical label rather than being a syndrome with a name.

Some exciting news is that we're currently planning our next family conference weekend, taking place in **April 2012. It will be Unique's 13th** conference and will bring together families and professionals, including expert geneticists, therapists and others to learn more about chromosome disorders and talk to families about topics of their choosing. The event will be held in a disability-accessible hotel where families will get the chance to relax and socialise with others in the same boat and there will be a crèche for children with special needs and their siblings.

For more info please visit our website at www.rarechromo.org



Do you feel isolated?

contact a family
for families with disabled children

Families sometimes report that they feel isolated and alone when they find out their child has a disability. But with the growth of the internet, social networks and the establishment of an increasing number of parent support groups and forums, there are more and more ways of connecting with others and making links and friendships. But is this enough?

Contact a Family wants to find out what the emotional situation is for you and your family in 2011 and if isolation impacts on your family life.

The survey will take just five minutes to complete and the findings will help us highlight this issue through a media story. The survey will be closed on 23rd September. You can find it online at www.surveymonkey.com/s/ContactaFamilyIsolation2011

SWAN UK has an active online community, come join us at www.facebook.com/SWANchildrenUK

Car Tax Exemption

If your child/young person is in receipt of the Higher Rate Mobility Component of Disability Living Allowance you may be entitled to apply for Vehicle (Car) Tax Exemption (cars on the Motability lease scheme are automatically taxed).

You should receive a form with the award, but if you have mislaid it and the certificate, please contact the DWP who will supply you with another.

Disability Living Allowance Unit,
Warbreck House, Warbreck Hill,
Blackpool, FY20YE
Phone: 0845 7 12 34 56
Textphone: 0845 7 22 44 33 (for hearing or speech difficulties).
Lines are open Monday to Friday 7.30am till 6.30pm.
You can find out more online www.direct.gov.uk

Lifting a teenager will damage your back!

You see you can lie to yourself for years. Tell yourself that you're doing fine.

Other people will say she's not that heavy - I mean she only weighs about 7 stone. And a few years ago when she was lighter I could be picking her up 20 times a day. That's what does the damage - it's the cumulative effect, you make things a little bit worse each time.

I got my first fright about 3 years ago, when I damaged my back very badly from lifting Smiley in and out of the car. So I went out and bought the Smileymobile, this great big enormous 'car' that I can push her wheelchair onto, and it also takes her walker, hoist, ramps, 3 passengers and me! It's about as exciting and sexy as a Transit Van, but means that Smiley can once again come out and about with me everywhere - which she loves - even a trip to Tesco is an opportunity for fun, learning and meeting new people.

Since then, I have had more back problems, but I keep getting patched up and carrying on. Last Spring I spent a lovely few days in Wexford and damaged my back again. On the Friday morning I stretched everything thoroughly and it seemed okay.

But on Saturday it was worse, and by Sunday morning I could hardly get myself down the stairs. Once again my friends came to the rescue, opened up their clinic and gave me an emergency treatment. We had a long talk about what was wrong and what I should do about it. Basically I was warned that I was sustaining crush injuries through all the carrying, and that it had to stop or my spine would be permanently damaged. That could mean that I would no longer be able to care for my daughter.

So Smiley's bed was moved downstairs so it was bye bye dining room until I moved house. Now she has a room downstairs, but I can't get the hoist into the bathroom so I'm still

doing 'level transfers' from her wheelchair onto the toilet. This may have to continue until I'm in a position to get a ceiling hoist put in, or I put my back out again. More treatment was needed this week and my knees are now giving me trouble as well.

I should have acted much sooner. But I didn't know any better. So please, if you are reading this and a carer who has to regularly lift a child or adult, take care. Don't just soldier on hoping for the best, listen to your body and if you start to feel under strain, get help and make changes.....before it's too late.

Read more at www.looking4bluesky.blogspot.com

Epilepsy Resources

epilepsy society

The Epilepsy Society have helpful downloadable resources about epilepsy medication and children.

www.epilepsysociety.org.uk

You can also call them on 01494 601 400 Monday to Friday 10am till 4pm if you want to talk to someone.

NCYPE
The National Centre
for Young People
with Epilepsy

NCYPE also have free handbooks for parents and great diaries designed for children—which come with free stickers! They have recently also launched an online seizure diary for teenagers.

Both are available from their website www.nycpe.org.uk

They also have a confidential enquiry line you can call with any **questions you may have. It's open** Monday to Friday 9am till 1pm 01342 831342,



Q : “Billy’s got a palliative care nurse who made a very poor attempt at explaining why he had one when I’d only ever heard them used in terminal patients before. He assured me that Billy isn’t terminal, but couldn’t explain why he was involved”

When I heard this, it reinforced my belief that one of the major barriers to people being able to find the appropriate care is that families **don’t always know about the support** available to them, because some of the words and terms used by professionals are not part of our day to day language. I was disappointed that **Billy’s family didn’t receive an explanation** from the nurse about why Billy had been referred to him – and what help he could offer, beyond his job title.

Palliative care for children and young people is defined by ACT, the **UK Children’s Palliative Care** organisation as:

“...an active and total approach to care, from the point of diagnosis or recognition, throughout the child’s life, death and beyond. It embraces physical, emotional, social and spiritual elements and focuses on the enhancement of quality of life for the child/young person and support for the family. It includes the management of distressing symptoms, provision of short breaks and care through death and bereavement”.

What this aims to describe is the care children and young people who

Piecing together the jigsaw : Your questions answered

have a health condition for which there is no reasonable hope of cure and from which they will probably die in childhood, or early adulthood.

ACT believes that children’s palliative care is not something that comes in at the ‘terminal’ or ‘end of life’ stage, but something that should happen right from a child’s diagnosis, or recognition of a life-limiting or life-threatening condition, throughout their life, no matter how long or short.

Receiving palliative care (and having a palliative care nurse) right from diagnosis can ensure that any symptoms can be managed and emotional, spiritual and social support is made available to enable the child and their family to enjoy the best quality of life possible. This care and support should be available **throughout the child’s life, not just at the end.**

Sometimes people do talk about **“terminal conditions” but this is not used commonly in children’s care; I am not sure why we have moved away from this, perhaps because many children and young people live for many years, or even decades with “life-limiting” conditions, and the word “terminal” is associated with end of life, whereas in children’s care, living matters!** Parents may find the ACT Family Companion useful to help answer **questions about children’s palliative care.** You can download it free from www.act.org.uk/families or call 0845 108 2201 to order your copy, or to ask the team any questions you might **have about your child’s care.**

Thanks to ACT for this answer.



ACT is the only organisation working across the UK to achieve the best possible quality of life and care for every life-limited or life-threatening child or young person and their family.

Q : What does it mean when the hospital refer to my child’s tone?

When doctors or nurses talk about **your child’s ‘tone’ this does not mean their colouring.** They are referring to their muscle tone. It means the amount of tension or resistance to movement in a muscle. If your child has low muscle tone this is called **‘Hypotonia’.** **If they have muscles which are abnormally tight and there is a reduced ability stretch out the muscles this is called Hypertonia.**

Q : What is a ‘Child in Need’ assessment?

A child in need assessment is undertaken by a social worker to see if your child has extra support needs and how these can be met.

The assessment happens in 2 stages: the initial assessment and then the core assessment.

As a parent you have the right to ask **for your child’s need to be assessed.** You would need to contact your local **children’s social care team.** **Under section 17 of the 1989 Children’s Act** all disabled children are classed as **‘Children in Need’.** **This does not mean they will automatically be entitled to services but they should receive an initial assessment.**

There is a strict timeframe set for the assessment process:

24 hours from receiving the referral to decide whether to do an initial assessment

7 working days to complete the initial assessment and decide if a core assessment is needed.

35 working days to complete the core assessment

Your social worker should talk to you and your child as part of the assessment.

You can find out more about your rights and the law relating to disabled children here: www.ncb.org.uk/cdc/resources/legal_handbook.aspx

Glossary of Terms : Remember to keep this page as it will be different each edition

Amniocentesis: This is a test that is used to take a sample to test an **unborn baby's genes/chromosomes**. The baby is surrounded by fluid in the womb which contains a few of **the baby's skin cells**. A small sample of the fluid is taken with a thin needle through the skin of the mother's tummy. The fluid is then sent to a lab for testing.

Autosomal : Something that involves the **autosomes**.

Autosomal dominant genetic conditions: These are conditions where a person only needs to inherit one changed copy of a (**mutation**) of the **gene** in order to be affected by the condition. The changed **gene** is dominant over the normal **gene**.

Autosomal recessive genetic conditions: These are conditions where a person has to inherit two changed copies (**mutations**) of the **gene** (a changed copy from each parent) to be affected by the condition. A person who has only one copy of the changed gene will be an unaffected **carrier**.

Autosomes : We have 23 pairs of **chromosomes**. Pairs 1-22 are called autosomes and are the same in men and women. Pair 23 are different in men and women and called the **sex chromosomes**.

Balanced Translocation : A **Translocation** when no **chromosome** material is lost or gained but is rearranged. A person with a balanced translocation is not usually affected by it.

Carrier: someone who carries one faulty copy of a gene but is not affected with the condition (at that moment)

Cell: The human body is made up of millions of cells which act like building blocks. Cells in different parts of the body look different and do different things. Every cell (except for **eggs** in women and **sperm** in men) contains two copies of each gene.

Chromosomes : These are thread-like structures which can be seen under a microscope and contains **genes**. The

The usual number of chromosomes in humans is 46. One set of 23 chromosomes comes from our mother and one set of 23 chromosomes comes from our father.

Deletion: This means that part of a person's genetic material is missing. It can mean either a missing area of a **gene** or a **chromosome**.

De Novo: This is from the Latin **phrase meaning 'new' and is used to describe a situation where the affected person's parents both have normal genes or chromosomes**.

DNA: Is a chemical substance that makes up the genes and contains the information needed for the body to work.

Duplication: This is the abnormal repetition of a sequence of genetic material in a **gene** or **chromosome**.

EEG : (Electrocardiogram) is a test that records the rhythm and electrical activity of your heart. It can tell if you are having a heart attack or have had one in the past. Sometimes it can be used to tell if your heart is enlarged or thickened.

Gene: This is the information needed for the body to work. It is stored in chemical form (**DNA**) on **chromosomes**.

Genetic Counselor: This is a specialist who gives information and support to people who are concerned about a condition which may have a genetic basis.

Insertion : This means there is additional genetic material in the **gene** or **chromosome**.

Karyotype: This is a description of the **chromosome** structure of an individual including the number of chromosomes and the type of **sex chromosomes** they have (XX or XY) and any variation in their **chromosomes** from the normal pattern.

Mutation : A change in a **gene**. Sometimes when a **gene** is changed its information is altered so it does not work properly. This may cause a **genetic condition**.

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Ring Chromosome: This is the term used when the ends of a **chromosome** have joined together in a ring shape.

Reciprocal translocation : This is when two fragments from different **chromosomes** break off and swap places.

Robertsonian translocation : This is where one **chromosome** becomes attached to another one.

Sex chromosomes: The **X chromosome** and the **Y chromosome**. The sex person is male or female. Females have two X chromosomes and Males have one X and one Y chromosome.

Tone: When doctors use this term they are usually referring to muscle tone.

Translocation : This is where there is a rearrangement of **chromosome** material. It happens when a piece of **chromosome** is broken off and attaches to another one.

Unbalanced translocation : This is a **translocation** where the **chromosome** rearrangement has extra chromosome material, some missing material or both

X Chromosome: This is one of the **sex chromosomes**. Females usually have two X chromosomes. Men usually have one X and one **Y chromosome**.

X-Linked conditions : A genetic condition caused by a **mutation** (change) in a **gene** on the **X chromosome**.

Y Chromosome : This is one of the **sex chromosomes**. Males usually have one Y chromosome and one X chromosome. Females usually have two **X chromosomes**.

Information here is taken from the EuroGenTest leaflets which you can access at www.geneticalliance.org.uk/publications_patients.htm

