

Factsheet – CG07

Life stories of people with CHARGE

No two people with CHARGE are the same. People can experience a wide range of physical difficulties, and these can vary in severity a great deal. Each is an individual with their own talents to express and challenges to face.

Here are some stories of individuals, and their families, who are living with CHARGE.

Ellie

“She’s got a wicked sense of humour”

Ellie, now aged 12, has a number of conditions commonly found in children with CHARGE. Her left eye is not formed, she has a coloboma of her right eye, she has nystagmus and finds bright light very difficult. This means that she has very little sight in her left eye and is very short sighted in her right eye. Despite this, “it’s amazing what she can do with the little sight she has in her right eye,” says Debbie, her mum.

She is also profoundly deaf in one ear and very hard of hearing in the other ear. She wears aids all the time although her parents are unsure how much benefit these really give her. She has also had a number of physical difficulties over the years and numerous operations. She has especially had problems with eating and it took her a long time to learn to eat.

Her parents feel that her lack of sight and hearing have undoubtedly held back her development, and finding the right education for her has been difficult. She attends a local school five days a week for children with a range of physical and learning difficulties.

“Ellie is extremely stubborn,” says Debbie. “She never gives up and is as strong as a bull. If she wants something she really goes for it, and makes us understand. She uses total communication – a mixture of signing, pictures and symbols to show us what she wants. She also has a wicked sense of humour and enjoys winding us up. She’s really funny.”

One of her favourite activities is going swimming, and she can now swim independently without armbands. “Ellie got a badge for her swimming recently which was a great achievement for her” says Mehdi, her Dad.

Looking after Ellie is very demanding for her parents. She does not need so much physical care now but they have to constantly keep an eye on her which requires a lot of patience and stamina. They receive respite care for 72 nights each year which is paid for by social services because Ellie needs one-to-one support. "It was a battle to get this," says Debbie, "but it does mean we can have a break." They have also been to a number Sense's family weekends where they have met other parents in the same position. And each year Ellie goes away for a Sense holiday which she really enjoys.

"It was very tough at first" says Debbie, "especially when she was spending so much time in hospital when she was young. But it has gradually got easier as her health has improved. Ellie has achieved a lot in her life despite what she had to go through which is wonderful."

Megan

"Megan does things in her own time, in her own way"

When Megan was born her parents didn't realise she had CHARGE at first – although she did have series of difficulties. She had a cleft palate, a blockage in her nose, and a coloboma in her right eye which was operated on when she was six months old. It was also discovered that she was partially hearing. For the first 18 months of her life she was frequently in hospital for operations and because she had a series of chest infections.

This was a very difficult time for her parents, although fortunately they found out fairly quickly about Sense who were able to offer some guidance and support.

One of Megan's biggest challenges is communication. She does wear hearing aids (but prefers to hide them in her shoe!) and can understand sign language - but doesn't sign much herself. She will make her preferences clearly known however and her mum describes her a strong character with a lot of fighting spirit. "She's a little character, always happy, always doing something," she says.

Megan started to walk when she was four, but still has some difficulties with her balance. She started at a mainstream nursery when she was two and then went on to a school for Deaf children with additional difficulties in Doncaster.

"Megan loves school and can't wait to go in and see her friends," says her mum. "There are five children in her class and she especially likes jigsaws, zooming around on her three wheel bike and going swimming".

"We never take anything for granted and always notice the progress she makes. – like, wow! she's starting to put her toys away by herself. We think 'Good for Megan.'"

Joshua

"Joshua is pretty marvellous considering the challenges he has had to overcome."

When Joshua was born he had multiple physical problems. His oesophagus was connected to his trachea and did not connect to his stomach, he had a hernia and

there was a layer of skin between his right nostril and his windpipe (choanal atresia). He also had facial palsy and acid reflux (which means that acid from his stomach came back into his throat and mouth). His parents, Hannah and David, were devastated. "We were horrified, despairing – there seemed to be no respite – one operation followed another and we virtually lived in hospital at first."

A little later they also learned that he was deaf and near blind. "The deafness and blindness were the hardest to bear really," says Hannah. "How could he communicate or interact with other people? I was absolutely horrified by that. But despite that, I always believed that Joshua would live and thrive. I always saw beyond his physical problems."

After a traumatic first year, Joshua's parents started to focus more on his communication needs. With support from Sense and other parents, they worked intensively with him and learned to communicate with him in different ways. He started to wear hearing aids and his parents learned sign language even though they were unsure whether he would be able to see and use this. In fact he was able to use around 20 signs by the time he was one and knew around 200 signs by the time he was two.

Gradually Joshua's progress continued. He started to walk at 2 years and 8 months, although he suffered a lot of cuts and bruises at first. It had been discovered that the semi-circular canals in his ears were missing and his balance was poor. Now aged 11, he loves football and will run and run, although he still needs to hold onto a banister if he is walking downstairs.

His signing also continued to develop which meant that he could express himself and he would get much less frustrated. At three he started to attend a local Deaf Bilingual School (British Sign Language with English as a second language) and at 6 he learned to read. His parents communicated with Joshua by signing and speaking and gradually he learned to speak. He has recently started at a school where he is supported to vocalise and lip-read as well as using sign to communicate.

Gradually, and with a lot of encouragement and support from his family, teachers and friends Joshua has made excellent progress. "His social skills, his language, his physical development are all delayed," says Hannah, "but he's pretty marvellous considering the challenges he has had to overcome."

James

"James has a great sense of humour and laughs at lots of things. He's very placid, and has immense patience"

When James was born, his mum could tell that "something was wrong", although it was a long time before she found out that he had CHARGE. He had a daunting range of physical problems: feeding and breathing difficulties, a hole in the heart, facial palsy, a blockage in his nose and urine reflux to his kidneys. Later it was also discovered that he had limited sight and very little hearing.

"It was horrendous," says Carol. "No sooner had we started to adjust to one problem then we found out about another. We were devastated, just trying to survive."

James had many operations, including six operations to remove the blockage from his nose and an operation to repair the hole in his heart. “After that he sat up the next day and could breathe again,” says Carol, “and he started to put on weight at last.”

“James really started to pick up when he was about three and a half”, she says. “He started crawling at four and gradually since the age of five has started walking. He can walk unaided for short distances now, although this isn’t easy given his poor balance, and that he has to tilt his head up to see.”

His parents have also gradually adjusted to this massive change in their lives. Carol is Chair of the CHARGE Family Group and receives a lot of support and advice from other parents. They have worked intensively to help James to develop his communications, and now they have another son, Adam, to look after as well. James and Adam have a lot of fun together

“It has been tough,” says Carol, “but it is true that it does gradually get easier.”

Jessica

“Her trips to the supermarket involve much signing of what is going into the trolley”

Jessica’s dad, Simon writes:

“Jessica, now five, started walking when she was three and a half – two weeks after her younger sister funnily enough – I wonder if that was sibling rivalry? Jessica has gone from scaring us with the speed of her walker frame to scaring us with her attempts to run unaided. Her style of walking is classic of children with CHARGE - widely splayed feet, stiff legged, planting feet firmly on the ground.

She has no semi-circular canals in her ears which means that her balance is poor, and the more tired she gets the worse it gets. This means that she does tend to pick up a lot of extra bruises at school and when she is playing at home. Her poor vision also contributes to this.

As she has become more confident on her feet so her communication has improved. She understands many signs and uses an increasing number herself. A lot of hard work goes into trying to improve her expressive language (which seems fine when cakes and crisps are involved!). She appears to know her fingerspelling alphabet and can associate the fingerspelling with the written letter.

Jessica really enjoys school and going out. Her trips to the supermarket involve much signing of what is going into the basket/trolley and many surprise items appear in this if we’re not looking. She eats huge quantities of food preferably with very strong flavourings but still remains thin as a stick.

Medically she is generally very healthy apart from the usual childhood illnesses. Unfortunately she has now reached double figures for general anaesthetics having had heart surgery twice, stents for choanal atresia, grommets, MRI and CT scans. We are considering a cochlear implant for her. As she is severely/profoundly deaf and has bilateral coloboma with no vision in one eye, her sensory reception is

considerably limited. Any help she could have in terms of receiving sensory information from her environment would be great.

Jessica uses her vision from her 'good' eye very well (too well sometimes - such as spotting a play area from 50 yards and having a tantrum because we are not going in that direction!).

She does have some issues with sleep. As with many children with CHARGE she often 'parties' for a couple of hours in the night. This can indicate that she is coming down with an illness but other times we just think that it is one of those things that can happen with CHARGE.

She loves books although refuses to sign more than one sign per page. Although her little sister Elly (nearly three) adores her big sister, Jessica does her best to ignore her unless it is to play a game they both enjoy – like ring-a-ring-a-roses.

Jessica has quite a lot of independence skills - she feeds herself, being very clear about which pieces of food are which and in which order she will eat them. She is very active in her self care and largely dresses herself with prompting; she is finally potty trained; she loves trampolining, riding her tricycle, running, climbing and things like that".

Where to go for help

Sense specialists have many years experience of providing assessments of babies and children affected by conditions that cause dual sensory impairment - including CHARGE. They also support families and offer consultancy services to schools. If you would like to find out more about CHARGE, or the support that is available in your area, please contact Sense.

See also Sense's website – www.sense.org.uk – which contains further information about CHARGE.

Sense also helps families by working in partnership with a number of groups for families – including the CHARGE Family Support Group. This enables parents, children and others affected by CHARGE to come together, share information and offer mutual support.

The CHARGE Family Support Group: <http://www.chargesyndrome.org.uk/>
Or telephone Simon Howard on 020 8265 3604

If you:

- would like to find out more about deafblindness or the services for deafblind people in your area
- require information in alternative formats including braille, large print, audio or disk - or would like this factsheet to be translated into your first language - please contact Sense's Information Team.

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