

Support and Information



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Please help us to help you!

There is a closed Facebook group for families affected by chromosome 7 deletions and duplications at www.facebook.com/groups/493223084038489

Unique lists external message boards and websites in order to be helpful to families looking for information and support. This does not imply that we endorse their content or have any responsibility for it.

This information guide is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change. Unique does its best to keep abreast of changing information and to review its published guides as needed. The text was compiled by Unique and reviewed by Dr Barbara Scelsa, Pediatric Neurology, V. Buzzi Children's Hospital, Milan, Italy.

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Duplications of 7q



Sources

The information in this guide is drawn partly from 29 people reported in the medical literature [Vogel 1973; Hoo 1982; Kardon 1983; Keith 1988; Bartsch 1990; Romain 1990; Humphreys 1991; Verma 1992; Stratton 1993; Lukusa & Fryns 1998; Mégarbané 2000; Ndah 2000; Courtens 2001; Kroisel 2001; Rodriguez 2002; Boccone 2004; Scelsa 2008; Nasiri 2010; Alfonsi 2011; Chen 2011; Weimer 2011; Velinov 2012; Rivera 2013]. The first-named author and publication date are given to let you look for the abstracts or original articles on the internet in PubMed (www.ncbi.nlm.nih.gov/pubmed). If you wish, you can obtain most articles from Unique. In addition, this guide draws on a survey of members of Unique conducted in 2013, referenced Unique. When this guide was compiled, Unique had 36 members aged between a few months and 56 years with a pure 7q duplication not involving any other chromosome arm. The guide also contains information from the publicly accessible Decipher database (<https://decipher.sanger.ac.uk>)

7q duplications

A 7q duplication is a rare genetic condition that occurs when there is an extra copy of part of the genetic material (DNA) in one of the 46 chromosomes – chromosome 7. This extra copy is known as a duplication, sometimes referred to as a copy number gain. People have two chromosome 7s, but the extra DNA is found in only one of them. The genetic change usually affects development, and sometimes health and behaviour as well. But how much it affects individuals, and the ways in which it affects them, can vary a lot.

In general, the right amount of genetic material is needed for correct development – not too little and not too much. How an individual develops, his/her personality, needs and achievements, are influenced by both his/her genetic material and the environment in which he or she lives.

Genes and chromosomes

Our bodies are made up of trillions of cells. Most of the cells contain a set of around 20,000 different genes; this genetic information tells the body how to develop, grow and function. Genes are carried on structures called chromosomes. Chromosomes usually come in pairs, one chromosome from each parent. Of the 46 chromosomes, two are a pair of sex chromosomes: two Xs for a girl and an X and a Y for a boy. The remaining 44 chromosomes are grouped into 22 pairs and are numbered 1 to 22, approximately from largest to smallest. Each chromosome has a short (p) arm (from petit, the French for small) and a long (q) arm (see diagram, page 3).

Looking at chromosome 7q

Chromosomes can't be seen with the naked eye, but if they are stained and magnified under a microscope, each one has a distinctive pattern of light and dark bands. Each chromosome contains millions of base pairs of DNA. Base pairs are the chemicals in DNA that form the ends of the 'rungs' of its ladder-like structure. There are millions of base pairs on every chromosome, and they are often counted in millions, where 1Mb = one million base pairs. The whole of chromosome 7 has about 159Mb (159,000,000 base pairs), and 900-1000 genes. On the right of the diagram you can see how the base pair numbers relate to the chromosome bands.

Notes

I wish I had known

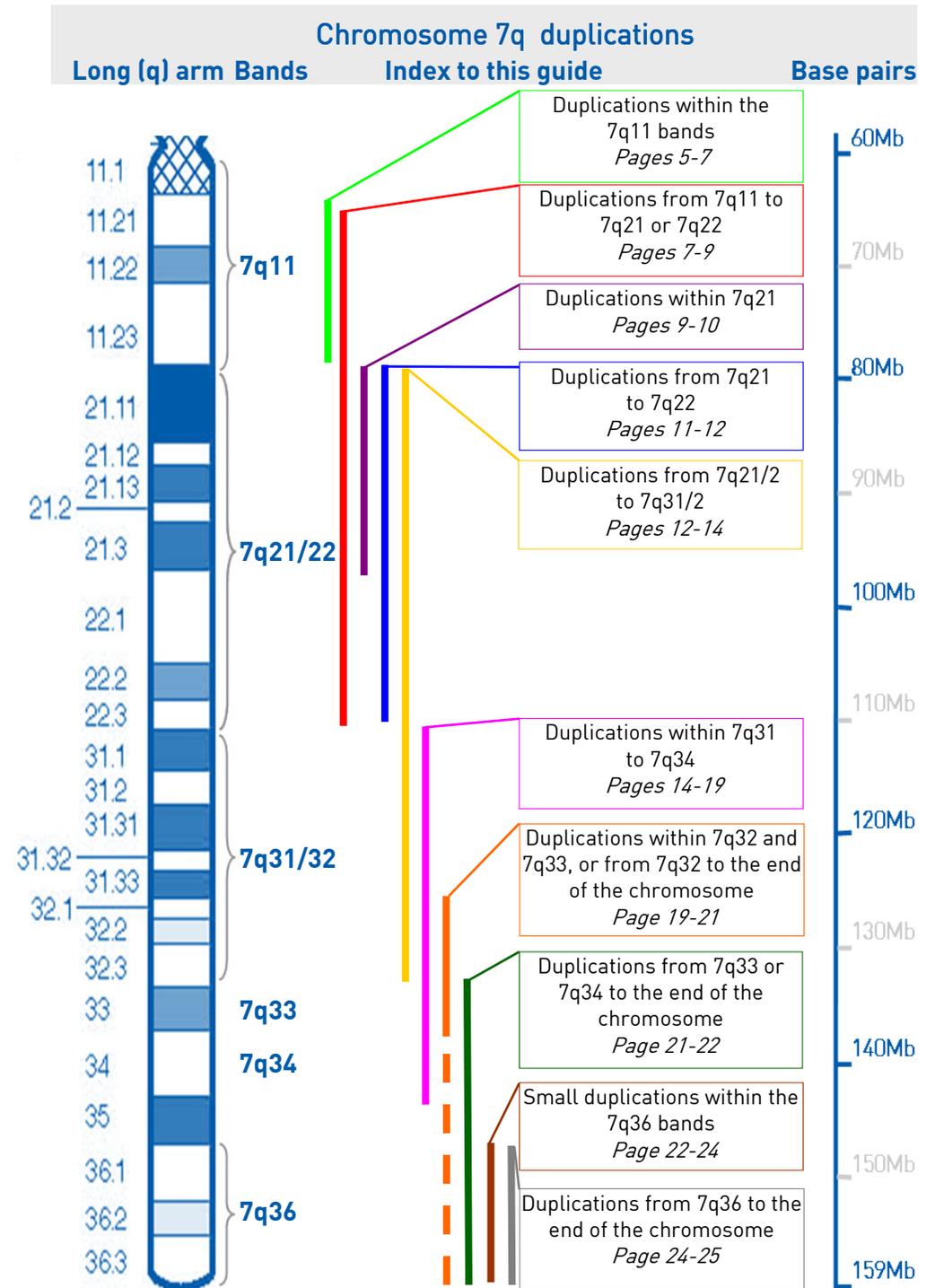
Unique asked families what they wished they had known earlier. You can read some of their replies throughout this guide. Other answers are gathered here.

One family observed significant improvements in their daughter after she started a gluten and casein-free diet. Another Unique family attributes major improvements in their child's development and behaviour to a gluten-free diet, together with a range of nutritional supplements. Unique takes a neutral position on diets, and does not recommend any particular diet for a child with developmental delay due to a 7q duplication.

"I wish I had known about the gluten free/casein free diet early on, for example, before beginning feeding therapy. I believe if we had implemented the diet very early on in therapy instead of towards the end, her eating issues may have resolved themselves very early on. I also believe her speech would have developed more normally and possibly never been considered delayed." 3 years

"I wish I had known how hard you have to fight, all the time! When we first received the diagnosis from the hospital paediatrician, we felt numb. The only solid information they could give us at first was the genetic karyotype. When we finally got to meet a geneticist and were informed it was genetic and no-one was to blame, we felt a slight relief. We were told she was unique and given printouts of similar but not the same cases – but by then we had already trawled the internet and contacted Unique. A friend had recently suggested I speak to a specialist nursery. I was horrified that she should even suggest that my daughter had any disability! Yet contacting and visiting the specialist nursery with our daughter was the best thing I ever did. We started attending weekly Mums and Babies sessions. They were supportive (I cried a lot!) and pointed me in the right direction for help. They supported our daughter's preschool statement/funding and 'shouted' on my behalf at any educational or healthcare professional I felt wasn't taking us seriously. As parents, we have truly made it up as we have gone along! Our eldest has been invaluable as a 2nd pair of hands/eyes and our youngest has provided daily challenges by developing at the expected rate, and has literally 'dragged' her big sister's development along." 6 years

"In some ways I am grateful that I did not know just how hard it would be to raise a child with our son's special needs. My husband and I took things a day at a time. The feelings of helplessness and isolation were hard to deal with, and I wish there had been greater support available." 28 years



Looking at chromosomes under a microscope, it may be possible to see the extra genetic material, if the piece is large enough. But changes smaller than 5Mb are very hard to identify, and sometimes larger changes as big as 10Mb can be hard to see. So if the extra piece is very small, the chromosomes may look normal under a microscope. Molecular DNA technology gives a more precise understanding of the size and position of the extra DNA. This is important as scientists identify genes and pinpoint their location on chromosomes.

Are there any syndromes on chromosome 7q?

Yes. People who have lost a tiny amount of a particular part of chromosome 7 in the band known as 7q11.23 develop Williams (or Williams-Beuren) syndrome. People who have an extra copy of this part have [7q11.23 duplication syndrome](#). Unique publishes a separate guide to this syndrome.

Has everyone with a 7q duplication got the same amount of extra DNA?

No. People have very different amounts of extra DNA, and different extra genes. Only two babies have been described with a duplication of the whole of 7q (the entire long arm). Some people have two breaks in the chromosome, with extra material between them: this type of duplication is called [interstitial](#) or [segmental](#). Other people have an extra copy of the DNA at the end of the chromosome: this type of duplication is called [terminal](#). In some people the extra DNA runs in the same direction as the rest of the chromosome: this is called a [direct duplication](#), or [dir dup](#). In other people, the extra material runs in the opposite direction: an [inverted duplication](#), or [inv dup](#). Generally, the amount of extra DNA is more important than its direction.

Genetic test results: some examples

A person's chromosome make-up is called their karyotype. Someone with a 7q duplication might have a karyotype that looks like one of these examples:

[46,XX,inv dup\(7\)\(q11.23q21.3\)dn](#)

This result tells you that the chromosomes were examined under a microscope. [46](#) chromosomes were seen, the correct number. The sex chromosomes were two Xs ([XX](#)), so this is a girl or woman. A piece of extra DNA was seen on chromosome 7, running in the opposite direction to the rest of the chromosome ([inv dup](#)). The start point of the extra material was in band [7q11.23](#) and the end point was in band [q21.3](#), so this is a large duplication. [dn](#) de novo (Latin for 'from the beginning') means that the chromosome change has not been inherited but has arisen 'anew'.

[46,XY,der\(3\)ins\(3;7\)\(q21;q34q32\)pat](#)

This result tells you that the chromosomes were examined under a microscope. [46](#) chromosomes were seen, the correct number. The sex chromosomes were an X and a Y ([XY](#)), so this is a boy or man. [der\(3\)](#) tells you that an abnormal chromosome 3 was found. [ins\(3;7\)](#) shows that the abnormality on chromosome 3 involves the insertion of DNA from chromosome 7. [\(q21;q32q34\)](#) tells you that

gestures (lifting her hand, giving high fives) to communicate needs and greetings. In terms of learning, one child was assessed as having an IQ of 73, while the child of almost 6 years was described as 'enjoying playing with toys that light up and make music'. This girl can also walk with support or in her gait trainer. She wears compression underwear during the day to improve her body awareness and has physical therapy (physiotherapy) to improve her low muscle tone.

"She can crawl and sit. Although it's very little improvement every day she has come a long way from lying on the floor to crawling." Almost 6 years

Information on behaviour is scarce, but while one boy of 3½ years has a short attention span and difficulties socialising, the girl of almost 6 years is happy if she has eaten and slept enough. Her family comments: 'She knows mom very well now. She comes to me when she is hungry or tired. When she was diagnosed I was worried about that.'

As for general health, the child of 10 years has daily headaches and absences, a type of seizure where the person goes momentarily blank, but other children are perfectly healthy. The 6-year-old had noisy breathing and pneumonia as a young child but has now outgrown this.

Verma 1992; Boccone 2004; Unique

She just comes to me and asks me to pick her up by lifting her hand if she is hungry or tired. She gives high five if we ask her for one.

Almost 6 years

Every year she gets better and stronger, and so do we.

In terms of behaviour, most Unique families say their child is affectionate, happy and friendly. One child needed behaviour management to control outbursts towards the end of the day when he was tired. Another child had autistic features and some obsessive behaviour, but did not merit a diagnosis of full autism, although this has been seen on Decipher.

As for general health, 2 children and an adult have seizures, but the information we have suggests that they are under control. Other conditions seen include an underactive thyroid, which can be managed by taking daily thyroxine; and low levels of some of the hormones secreted by the pituitary gland, including growth and thyroid hormones. The child born with double kidneys had frequent kidney infections; and another child had frequent bouts of pneumonia, which left him with long term lung damage, treated with a drug to increase the air flow to the lungs. As a teenager, he developed claw feet and as an adult, a progressive spinal curve. Two children had frequent ear infections, and needed aeration tubes (grommets) inserted into the ear drums to improve their hearing. Eyesight is affected in at least 3 people, and one adult has a condition known as cortical visual impairment, where the visual systems in the brain do not understand or interpret correctly what the eyes see. One child has convergence insufficiency, an anomaly of binocular vision where the eyes have a reduced ability to turn towards each other, resulting in double vision and eye strain on close tasks; he was successfully treated with vision therapy.

Duplications from 7q36 to the end of the chromosome

In this group there are 3 children of various ages, all with different-sized duplications: one from Unique, and 2 in the medical literature. These are generally large duplications, so are not found on Decipher where most have tiny microduplications (Verma 1992; Boccone 2004; Unique).

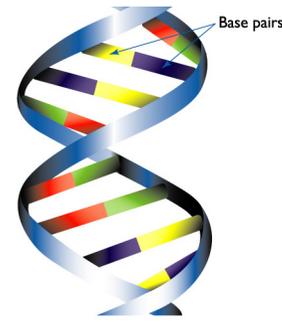
The first sign of a problem in two babies was delayed development. Babies with a known birth weight were small but within the normal range, and while one was healthy at birth, another had hepatitis (inflammation of the liver).

One child's growth is at the bottom end of the normal range, while another was an average height, and at almost 6 years was able to drink liquids (soup, smoothies) from a spoon, and was starting to cope with a fork. When given the fork, she would grab it, feed herself, and drop the fork.

In terms of appearance, one child is characterised by a large, rounded forehead and a small nose, both characteristic features of a 7q duplication.

She enjoys ... toys that light up and make music. Almost 6 years

In terms of development, all 3 have shown some delay but details are scarce. Most commonly delay has been mild, but speech and language is a significantly delayed area of development, with a child of 3½ years not yet talking clearly enough to be understood, and a child of almost 6 using



Two strands of DNA are held together in the shape of a double helix by the bonds between base pairs.

the insertion is in band **q21** of chromosome 3, and that a segment of chromosome 7 between bands **q32** and **q34** was inserted. **pat** shows that the 7q duplication has occurred because of a chromosome change in the father. Typically, the change will be balanced in the father and his development will not have been affected.

arr[hg19] 7q32.1q36.1(128065172-149301036)x3

The test was by array comparative genomic hybridization (**arr cgh**). The results follow the Human Genome build 19 [**hg19**]. **7q32.1q36.1** shows that two break points were found, the first in band 7q32.1 and the second in band 7q36.1.

(128065172-149301036)x3 shows that three copies of the material between the break points was found (**x3**). The normal number of copies is 2, so this means there is an extra copy. **128065172-149301036** are the start and end points of the extra copy, measured in base pairs. Take the first long number from the second and you find that there are 21,235,864 extra base pairs. This is about 21.2Mb.

Base pair numbers follow hg19, build 19 of the human genome. The human genome is updated as new information is found; each new version is called a 'build'. In each build the base pair numbers usually change slightly. When this guide was published hg19 was the most recent build.

Duplications of the whole of the long arm of chromosome 7

It is extremely rare to be born with an extra copy of the entire long arm of chromosome 7, and in fact just one baby has been reported in the medical literature where no other arm of any chromosome was involved (Ndah 2000). The baby girl was born prematurely at 34 weeks into the pregnancy, and had many birth anomalies, including an enlarged head, missing fingers, clubfeet and underdeveloped lungs. Sadly, despite treatment on a ventilator with oxygen, the baby died within hours of birth of respiratory failure.

Duplications within the 7q11 bands

At the top of the long arm of chromosome 7 in the diagram on page 3 is a region known as 7q11. This region is divided into four bands: 7q11.1, 7q11.21, 7q11.22, and 7q11.23. Just 10 people with a duplication in this area are known: five on the Decipher database, one in the medical literature (Hoo 1982), and five at Unique, one of whom is also on Decipher.

Duplication of genes in band 7q11.23 from around base pairs 72.7 Mb to 74.1 Mb causes a distinct syndrome. Unique publishes a separate guide to the **7q11.23 duplication syndrome**.

Apart from families where a small duplication is passed from parent to child, each of the duplications is slightly different: one at 7q11.1q21.1; six within 7q11.21; one at 7q11.21q11.23; and two at 7q11.22q11.23.

The pattern of unusual features is very variable and incomplete descriptions, particularly in the public part of the Decipher database, mean that no features are known to be consistent for everyone. Birth weight is known for 3 babies, all in the low normal range, from 2.88kg to 3.1kg (6lb 6oz to 6lb 13oz).

I wish I had known.....
... that he would eventually walk and talk. That we needed to get on waiting lists early for respite and help. That we would need to prepare for behavior issues.
11 years

An unusual head shape appears to be common: one baby had marked hydrocephalus (excessive fluid within the brain), while four others had an unusual head shape or formation: in one baby the head was pointed when viewed from on top (trigonocephaly); in two other babies, the bony plates that form the skull had joined too early; in a fourth

child the head was described as 'a little misshapen'.

In terms of development, one child is considered bright, while 4 have developmental delay. One child, considered gifted, has inherited a family microduplication in 7q11.21; other relatives with the microduplication have unaffected intelligence. This child experienced speech delay and had difficulties with concentration and hyperactivity. There is little information on mobility, but two children had low muscle tone (hypotonia), one specifically in the trunk area of the body.

Seven people, possibly including three members of the same family, have epilepsy, and in at least two of them, the seizures were initially hard to control. Three members of the same family have an immunodeficiency, but it is not known whether this is related to the family microduplication. One child was born with developmental hip dysplasia (loose/ clicky hips, routinely monitored at birth), corrected after being in plaster for 6 months, and two children had abnormal joint mobility.

One baby had a cleft (split) in the palate (roof of the mouth); another had a high palate. The baby with a cleft palate had feeding problems and put on weight slowly before the cleft was mended, but no problems afterwards. This child also had chronic constipation. Another child had too much weight gain around the waist and trunk.

One baby had significant birth anomalies, with marked hydrocephalus and only a small amount of brain tissue, a wrongly-positioned heart, a failure of the left eye, the vagina, ovaries and uterus to develop, and clubfeet. Sadly, this baby died when only 7 months old. Other features observed in only one child include a missing rib and premature growth of pubic hair.

Hoo 1982; Decipher; Unique

tiny penis with the hole on the underside; the end of the gastrointestinal tract in the wrong place; and a slight dimple at the base of the spine. One baby had breathing and swallowing difficulties at birth and had seizures, although his brain looked normal on magnetic resonance imaging (MRI) scan.

Two/5 babies had marked difficulties in feeding at first, with problems of swallowing, poor sucking and gastro-oesophageal reflux, where the feeds return up the food passage; in one of these babies, the reflux persisted into adulthood. Another baby breastfed well, but had poor weight gain. Another baby had no feeding difficulties, and ate a variety of foods with a good appetite. A milk allergy was queried in one baby, but by 5 years s/he was eating well. As for growth, we know outcomes in only 2 children, one of whom was very tall, the other growing into a short (163cm; 5'4") adult.

In terms of appearance, two families said there was nothing unusual about the way their child looked, and one described their son as a 'handsome boy'. Individual features noted by families and on Decipher show no consistency, although soft, loose skin was noted in two children. No abnormalities of the limbs, hands or feet were seen in more than one child, but one baby did have significantly short upper arms and thighs.

In terms of development, delay is to be expected: every child known to Unique has shown some aspect of delayed development. Most Unique babies had low muscle tone, making them feel floppy to hold, and were late to gain head control, to get moving, and to walk. At least 2 babies scooted or bottom-shuffled rather than crawling. The age at which children started to walk ranged from 13 months to 4 years, but some children had balance issues once walking, especially when tired: one child wore a helmet to protect his head when he fell. Despite this slow start, one child showed an early talent for sport. One child had good fine motor (hand) skills; two others had difficulties with grasping and manipulating cutlery and writing implements.

"He can dress himself and does handle his personal needs." 21 years

The most obviously affected area of development in the Unique group is speech and language: two children have a diagnosis of verbal dyspraxia (difficulty making and coordinating the movements needed to produce clear speech), while the others communicate non-verbally even in adulthood.

"He can use a communication device but has fine motor issues and is slow with its use. He can speak but is difficult to understand, and is to all intents and purposes non verbal." 21 years

Learning appears to be much more subtly affected than speech in the Unique group. Although children are generally young for their age in terms of cognitive abilities, Unique's experience is that some are quick learners, and skills like reading and numberwork are possible.

"He is slowly recognising the order of numbers and counting, though he doesn't understand time yet. He learns best in a small group, when he is interested, when he is praised for understanding, and because he is determined." 10 years

Life is challenging with our adult son. He is demanding and difficult but, thankfully, he has a good sense of humor. Seeing him laugh is one of the greatest rewards.
28 years

In terms of development, the degree of learning disability has been described as moderate to severe, with an adult performing at the level of a young child.
"He can manipulate touch screens and electronic devices, but his coordination is fair to poor, and his writing is poor. Cognitively, he functions around the level of a child of 3-7 years. He likes to be around people and can communicate well with people who are familiar with his speech problems. Strangers cannot always understand him and he

can become frustrated." 28 years

In terms of behaviour, there is information on only one adult, who has features of autism, and developed behaviour that is very hard to manage, including obsessions, aggression, anxiety, and hostility. Some medications (citalopram, quetiapine) have been helpful, but mostly his parents have learned how to manage his behaviour.

"The period between ages 14 and 16 were the most difficult. He was extremely agitated and aggressive. He has leveled off considerably since that time. Over the years, we, his parents, have discovered what works best." 28 years

As for general health, information is sparse. One baby had cysts and abnormally wide channels in the kidneys; another baby boy was born with a missing testicle, and a second testicle that was not in the scrotum (the loose bag behind the penis) at birth. Three people had an eyesight problem: a lazy eye; a squint (strabismus); and short sight. One baby had a forward spinal curve (kyphoscoliosis). One adult needed two hip operations to correct a problem at the top of the thigh bone (slipped femoral epiphyses).

Bartsch 1990; Romain 1990; Unique

Small duplications within the 7q36 bands

In this group there are 15 children of various ages with different-sized duplications within the three 7q36 bands: 5 from Unique, and 10 on Decipher. There are significant differences between different children, some explainable by smaller or larger duplications, others by limited descriptions, and others are unexplained (Decipher; Unique).

The babies for whom we have birth weights were relatively large for dates, the largest weighing 4.65kg (10lb 4oz), and two babies had a very large head – although one baby had a very small head. One baby had multiple problems at birth with swallowing, breathing and seizures. Among the abnormalities that babies faced at birth are: a heart defect; right-sided heart placement (dextrocardia); underdeveloped band of nerve fibres connecting the two sides of the brain (corpus callosum); an abnormality of the airways; a double kidney; a

Our son has future goals (to have a job, move out). He loves to clean and work – really wants to be productive all the time. He has a wicked sense of humor and loves to tease and play jokes. He is a perfectionist and wants to do the best he can at everything. He has strong metacognition, and understands what he can and can't do. He loves to cook and wants desperately to be independent in this area. He enjoys interacting with adults: sharing, playing games, just hanging out. He LOVES music and dancing. 11 years



11 years

Duplications from 7q11 to 7q21 or 7q22

In this group there are 6 people: 4 with a duplication between 7q11.23 and 7q21.11, one with a duplication from 7q11.23 and 7q22, and one with a 7q11q22 duplication. Each of the duplications is slightly different, even when the band numbers are the same (Kardon 1983; Decipher; Unique).

At birth, one baby was completely healthy although small for dates at 2.5kg (5lb 8oz), while another was unable to suck or feed properly, had constipation, passed no meconium in the first week, had a very quiet cry and slept much more than normal. The constipation persisted, but was helped by a gluten-free diet. Three babies, one with a cleft palate, are known to have had significant later feeding difficulties, and all were short or had difficulty putting on weight. One boy of 11 is in the bottom 1 per cent of the population for height.

"He had difficulty swallowing as an infant, but currently feeds himself, and is beginning to cook and prepare food on own." 11 years

Two baby boys were born with undescended testicles and one with a small penis; and two had abnormalities of the urinary draining system, with reflux back to the bladder in one, and a tiny left kidney in the other.

In terms of appearance, three babies had a pronounced, rounded forehead, and one had a large head with a soft spot on top that only closed at the age of 2; and two had slightly bulging eyes. Other facial features mentioned include a wide nose, tiny wide set eyes, a small mouth, large ears set low on the side of the head and swivelled backwards, bent earlobes, and a short neck.

As for development, all babies have shown some degree of developmental delay. One baby was not turning his head to the sound of his parents' voices by the age of 4 months, and was late to develop head control. He learned to sit early at 6 months, but never crawled and only walked at 2 years. He then wore leg supports until the age of 5, and was able to run, ride a 2-wheeler, pedal a scooter, swim, play basketball and take part in Special Olympics bowling by the time he was 11. We know that at least 2 others were late to walk, one walking at 3 years. Two children needed leg supports and a third a walker to steady him while learning to walk. Low muscle tone was diagnosed in two babies, but while

it resolved by 10 months in one, it persisted in the child who went on to compete in Special Olympics, affecting his legs which stayed 'abnormally thin'.

One child was toilet trained in the day by 8 years, and at night by 10; by 11 years he was completely independent in all aspects of personal care – toileting, washing, bathing and dressing. This may of course not be possible for all.

Speech and language development was also late, with one child of 4 years not talking yet, and another using gestures to communicate his needs and wishes. A third child on whom we have much more information has good understanding, but was diagnosed with severe apraxia (the speaker cannot say what s/he wants to clearly and consistently). At 11 years he was using complete sentences when relaxed, especially with adults rather than children of his own age, and only signing to special needs peers. He sometimes had to repeat words for clarity. He was enthusiastic about electronic communication, regularly emailing friends and family. His family believes strongly that the many nutritional supplements he takes, especially CoQ10 and carnitine, have made a 'huge difference' to his ability to communicate.

["Real aptitude for technology: digital communication has helped him tremendously with improving verbal communication." 11 years](#)

Four of the six children are known to have a learning difficulty, but details are scarce. A child of 23 months was performing at the level of a 10 month old baby, while a boy of 11 was writing legibly but slowly by hand, preferring to communicate by computer and iPad. He knew his name, address and birth date, and was reading at the level of a 6-8 year old child. He could use a calculator, and do two-digit addition and subtraction using a touch math system.

In terms of behaviour, two children have autism, although this has not been seen in the Unique cohort. One Unique child is an easy-going, happy child, while another is shy and anxious, and has OCD (obsessive compulsive disorder), and is intolerant of mess, noise and changes in routine. At 11, he is getting better at expressing his own sensory needs, and while showing little empathy, understands when others are sad or happy. He has a variety of supportive therapies, including cranial sacral therapy and heavy sensory pressure in the form of tight therapy bands, compression activities such as wall push ups and tight hugs. He also takes medications (fluoxetine, clonidine) but his family believes strongly that a gluten-free diet (see also [Duplications within 7q31 to 7q34](#)) and nutritional supplements have been extremely helpful in managing his behaviour as well as promoting his development.

["If his routine or environment are out of order he will throw tantrums until he is able to 'fix' things – lock doors, close cabinets, put mess out of sight, etc. He invades others' space in order to create order. Nutritional supplements have been major life changers for him, making a HUGE difference, especially CoQ10 and carnitine for speech development."](#)

As for general health, the child born with a cleft palate had frequent ear infections and probable hearing loss, as well as febrile convulsions as a baby.

As for general health, the two children with a large 7q32 duplication of the end of the chromosome both had seizures, one starting within 3 weeks of birth, the other starting at 16 years. In one child the epilepsy is difficult to treat: he has episodes of staring, and very abnormal EEG background activity; unfortunately he developed many side effects with most antiepileptic drugs and now tolerates only a low dosage of valproic acid with incomplete seizure control. The other child also takes valproic acid with incomplete seizure control.

One child had very poor eyesight, and when he underwent surgery to correct his strabismus (squint), he was found to have abnormal muscles holding the eye in place. Despite this, surgery to correct the squint was largely successful, but his eyesight was still poor because he had underdeveloped optic nerves. Strabismus and nystagmus (an uncontrolled jerky movement of the eyes) have been seen in other children with this duplication.

Keith 1988; Scelsa 2008; Decipher; Unique

Duplications from 7q33 or 7q34 to the end of the chromosome

In this group there are 4 people of various ages, including two adults: one from Unique, and 3 in the medical literature. These are generally large duplications, so are not found on Decipher where most have tiny microduplications (Bartsch 1990; Romain 1990; Unique).

For one of the babies the first sign of difficulties came when he was six months late in reaching most baby 'milestones', with the most pronounced delay in learning to talk. He was a good size at birth, and as a group other babies were born a healthy weight, on average 7lb 11 oz (3.485kg) at term. However they had obvious health problems at birth: poor feeding; noisy breathing caused by a floppy voicebox; and breathing difficulties, as well as dislocations of the hips and, more unusually, the shoulders.

As for later feeding and growth, we only have information on 2 people including one adult, who had no real feeding difficulties, although as an adult he needs foods such as meat cutting up, and is very tall, at 193cm (6'4"). Another child has average growth.

In terms of appearance, facial features noted by geneticists in more than one child include: a low bridge to the nose; a high, rounded forehead; tiny skinfolds across the inner corners of the eyes; ears set low on the sides of the head, sometimes with an odd shape; a small nose; a large tongue and sometimes an open mouth; a receding lower jaw and chin; and a short neck. Three babies had a single palm crease on one side; one baby was born with thumbs with an extra joint so they looked more like fingers; and 2 with incurving fifth fingers; one baby had two webbed toes.

[He enjoys ... shopping, going out to eat, going to work with his father. 28 years](#)

with a shrinking of the brain tissue, and thinning of the band of nerves that connects the two sides of the brain, known as the corpus callosum. Another had a type of brain cyst called an arachnoid cyst, where some spinal fluid has collected between two of the membranes that cover the brain.

One baby has marked difficulties in feeding, and three children are known to be short for their age, one with 'very poor weight gain', although two Unique members had normal growth, one at 22 months, and another at 17 years.

"Physically average for her age, but she looks younger. Soft skin like a baby."
17 years

In terms of appearance, any differences are often quite subtle and less obvious to families than to doctors. A Unique family found nothing unusual in their child's appearance. Doctors have reported a range of unusual features, including a very small head or one that is relatively short from front to back, tiny skinfolds across the inner corners of the eyes, large, sticking out or low and oddly formed ears, a low bridge to the nose, thin hair, long eyelashes, and a pointed chin.

One child had oddly formed teeth. Hands and feet might also be slightly unusual: short or broad fingers, deep palm creases, incurving fingers, flat feet, a wide 'sandal' gap between the first and second toes, and unusual sole creases have all been reported.

In terms of development, some delay has been reported in everyone with this duplication. The two children with a very large duplication of the end of the chromosome starting in 7q32 seem to be particularly severely affected in

terms of development. Generally speaking the delay affects all areas of development, and babies are late to learn to sit up and walk. Low muscle tone, causing floppiness, affects some, while others show ataxia (lack of control and coordination) or increased tone, so muscles are tight and stiff. Joints may also be unusually bendy. One child was walking, albeit with balance and coordination difficulties, at 22 months; another was walking by the age of 5. A

child with a large 7q32 duplication was able to roll to one side by 2 years and at 10 years can sit with support but not walk, while another has difficulty with head control as a teenager. Speech and language delay has also been found, and a child of 10 with a large duplication and a teenager with a large terminal deletion do not talk. The 10 year old vocalises and raises his voice to attract attention.

In terms of behaviour, there is almost no information available, but we do know that two children had autistic behaviour. One teenager, aged 17, with autistic traits has no behaviour problems, and spends most of the day playing with her toys, preferring toys which light up and make a noise.

She is the happiest little girl. Her cute smile and good sense of humour are our cup of tea, and give us strength to carry on.

17 years

He also has strabismus (a squint), and involuntary eye movements. A further child had a marked misalignment between the upper and lower jaw, and needed teeth extracted and braces to try to correct his bite. This treatment also aimed to expand his palate and open his airway to improve his sleep. He was to be fitted with a device to hold his tongue in place to allow him more oxygen, and hopefully enable him to sleep without waking for long periods at night.

(Kardon 1983; Decipher; Unique)

Duplications within 7q21

In this group there are 16 people with duplications of varying sizes: 4 with a duplication in 7q21.11; one in 7q21.12; 7 with a duplication between 7q21.12 and 7q21.13; 2 in 7q21.13; and 1 in 7q21.3. There are apparently great variations between individuals with duplications in the 7q21 bands, and 13/16 cases are in Decipher where very little detail is publicly accessible. Both Unique cases are families where a small duplication has been inherited through at least three generations, but only come to light in the current generation (Velinov 2012; Decipher; Unique).

At birth at term, one baby weighed 3.1kg (6lb 13oz) and had respiratory problems with fluid on the lungs. Another baby had mild breathing problems caused by softness in the cartilage supporting the structure of the voicebox. Another baby weighed 2.8kg (6lb 3oz), a low normal weight, but was very short.

One baby was born with a diaphragmatic hernia (the sheet of muscle separating the contents of the chest from the abdomen is incomplete) and a marked narrowing of the aorta, the blood vessel that leads out of the heart. Another had incorrect development of the heart, lungs, gastrointestinal system and abdominal organs caused mainly by the development of two left sides rather than a left and right, a condition known as heterotaxy. This child needed major surgery to correct the problems. Another had an open channel between the aorta and the pulmonary artery. This channel, called the ductus arteriosus, usually closes soon after birth, but when it stays open the lungs receive more blood than they should and the heart has to work too hard. The channel was surgically closed. One child with an unusually small head was found on scanning to have an underdeveloped corpus callosum, the band of nerve fibres connecting the two sides of the brain. One boy was born with undescended testicles.



He enjoys dancing, staring at trees, and he loves to cuddle. He likes bright lights and music.

4 years

One baby with a tiny duplication in the 7q21.3 band, involving only 2 genes, known as *DLX5* and *DLX6*, was born with complex deformities of the right hand and foot. Losing these two genes is known to be capable of causing a birth defect known as split-hand/split-foot, and it appears that having an extra copy of these genes can also result in this type of problem.

In terms of growth and feeding, two children are known to be very tall, and one unusually short. In terms of appearance, many unusual features have been noted, but these can be quite subtle. The most common is a broad, rounded forehead, typical of children with a 7q duplication. In one child the hairline was very high, and while in one baby the bony plates of the skull fused very late, another had a disproportionately small head. Other unusual features include: upslanting eyes; a narrow palate; a wide nasal bridge; a short nose; ears that stick out, are low set or are unusual in some other way; and large eyes. Unusual aspects of the hands or feet include broad thumbs or broad first toes; long or slender fingers and incurving fifth fingers. One child had hyperkeratosis, a thickening of the skin that can show as tough, dry patches.

In terms of development, delay is recorded in 8 children, but may well be more common. It is described as moderate in 2. Six children are known to have a learning disability, and some have mobility problems as well, with unusually bendy, lax joints and thin arms and legs. One child learned to walk at 4 years. Although speech and language delay is only noted twice, it is almost certainly more common: one child was not yet talking at 4 years.

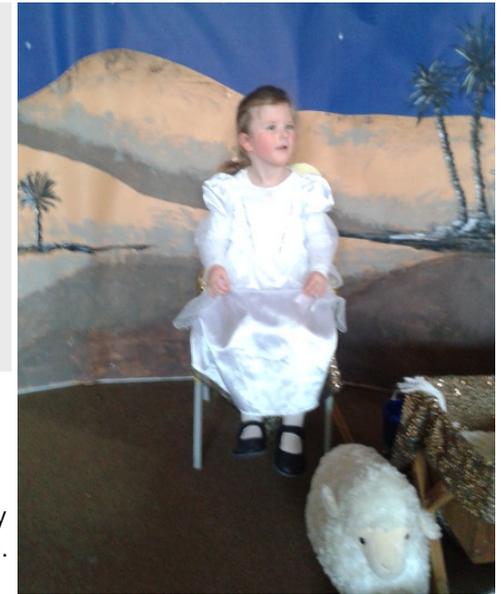
“He has problems with coordination, but this is improving, and he loves to use his iPad. He loves bright lights, music, and dancing. He has been partly feeding himself since he was one, but he is lazy, though now he loves to eat. He doesn't talk and socializes only with adults, and not with other children. Sleep is hard to manage: he doesn't sleep for long and often wakes up with a cough or bronchitis. He takes melatonin for sleep.” 4 years

In terms of behaviour, recorded problems include autism, hyperactivity, a short attention span and sleep disturbance. One Unique member is described as ‘a very active, happy child who loves to eat but screams and yells all day’.

As for general health, the baby who was born with respiratory problems continued to have coughs, colds and bronchitis into early childhood, treated with standard treatments for asthma as well as salt inhalations and tablets to soothe his persistent coughs. Two children were born with cataracts, and one had the lenses removed from both eyes and now wears glasses. One child was born with a developmental eye defect known as a coloboma. Two children had some hearing loss, one following glue ear treated with grommets (ear tubes and a hearing aid). One child had polyps (growths) removed from inside his nose. One child had a forward curve of the spine (kyphosis) by 4 years. One child experienced a delayed puberty.

Velinov 2012; Decipher; Unique

She enjoys ... cooking; ‘reading’ books; Lego (Duplo); Peppa Pig/Tweenies; night-time walks; ‘going on an adventure...’ (anywhere: zoo, train/bus ride, park, ice cream parlour, library, Legoland); playing with her sisters and visiting relatives, especially baby cousins; playing with baby dolly; giraffes and other jungle animals. 6 years



Eyesight problems can occur: one child is visually impaired and 3 have nystagmus, where the eyes move jerkily to and fro, and vision is usually affected. Three children have strabismus, a squint, where the eyes point in different directions. Children who need glasses may not wish to wear them. One child has a forwards spinal curve (kyphosis). Slight dental abnormalities are not unusual in children with chromosome disorders, and one child has a missing tooth and two fused teeth, another child has widely spaced teeth. Otherwise, problems are functional, one child grinding her teeth until they are worn down. Chen 2011; Decipher; Unique

Duplications within 7q32 and 7q33, or from 7q32 to the end of the chromosome

In this group there are 7 children of various ages, all with different-sized duplications: one from Unique, one in the medical literature, and 5 on Decipher (Keith 1988; Decipher; Unique). Two further children have a large duplication of the entire end of the chromosome, starting at 7q32 (Scelsa 2008; Unique).

One baby was born early at 35 weeks and had jaundice and difficulties with temperature regulation, as well as problems with head control; she had incurving fingers. Three babies were born with a heart problem: in one case a blocked heart valve, and in the others a cluster of different problems. Unfortunately we do not have information about the surgery to correct these. One child had a dimple at the base of the spine; another had a high, narrow roof of the mouth; and another was born with club feet.

Two babies had something unusual about their brain. One of them, identified by 23 weeks into pregnancy, had a growing accumulation of fluid inside the brain,



She is very loving. She is always hugging, and is such a good big sister to her little sister. She brings happiness to everyone she meets as she always has a huge smile on her face. 7 years

gate, and hide snacks, and hold hands while out as he will still run away. But he is very happy and loves to rub his head/ face against mine. He lights up when he sees me, although he still does not say *mama* or *daddy*." 11 years

As for general health, 3 children have had seizures, one of them 'absences' where there is a brief loss or impairment of consciousness. Two Unique families

Our daughter can be considered one of the sweetest and most empathetic children most people know. She is also very well behaved despite her issues. She reminds me every day that everyone deserves to be loved, and that she is the reason I need to be the best mom I can be. 3 years

Behaviour wise, two children in Decipher have autism, and autistic features (grouping toys) have also been seen at Unique. However, Unique children are more typically happy and very sociable, with a good sense of humour. One child is very imaginative, pretending to be a cat and meowing rather than talking. This child has had issues with sensory processing (she dislikes toothbrushing, and used to refuse certain food textures) and cannot cope with unpredictability, avoiding playing with animals or younger children.

"Very sociable and mischievous – has a great sense of humour! She is tiring! Happy. Independent. She likes to play/fight and annoy her younger sister. She enjoys all aspects of school including school transport. She can have tantrums (meltdowns). She knows what she likes (clothing choices!). Her school is aware that she is not so compliant at home as at school (eg she refuses to hold hands to cross the road)." 6 years

"Easy going, loves to play, draw, play with clay, and most of all on her iPad." 8 years

"We have to lock the fridge, doors and the two children have had repeated sinus infections and persistent nasal discharge, in one case treated with antibiotics and controlled by meticulous facial hygiene, and in the other by removing the adenoids. This child also had grommets (aeration tubes) placed in the ear drums to improve hearing. She is also more susceptible to 'stomach bugs' than her chromosomally normal sisters.

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Duplications from 7q21 to 7q22

In this group there are 6 people, all with large duplications: 1 with a 7q21q22.2 duplication; 2 with duplications from 7q21.1 to 7q22.3; one with a 7q21.2q22.1 duplication; another a 7q21.2q22.3 duplication; and one with a duplication from 7q21.3 to 7q22 (Lukusa & Fryns 1998; Alfonsi 2011; Rivera 2013; Decipher; Unique).

Two of the babies were twins, each with an unaffected twin. One set of twins was conceived using assisted reproduction (ICSI). One set of twins was born at 35 weeks, the other at 36 weeks. Another baby was born early at 32 weeks. Known birth weights are all within the low normal range for the stage of pregnancy, apart from one twin born at 35 weeks who was small for dates. The condition at birth of two babies was reported: one had low muscle tone (hypotonia), a weak cry, and sucked poorly; the other had jaundice, a sharply receding lower jaw (no chin), was malnourished and needed oxygen.

One baby was born with a small left kidney that had multiple cysts. Two babies were born with a persistent ductus arteriosus, where a blood vessel fails to close as it usually would around the time of birth, so the lungs receive too much blood and the heart has to work too hard. It can usually be closed using minimally invasive surgery. One of these babies also had a patent foramen ovale (PFO), an opening between the two upper chambers of the heart that usually closes in the first year of life, and did eventually close of its own accord. One baby had abnormally wide bones in the spine in the lower back (lumbar platyspondyly). One baby had a closed dimple at the base of the spine.

In terms of growth, two children and three young men of 18, 19 and 22 are known to be unusually short: one youth is 5'6" (168cm). Feeding difficulties are reported in 2, but were not severe: at 19 years, one preferred soft foods, needed food cutting up small, and was prone to choking. He took lactulose to manage constipation.

As for appearance, a rounded forehead and a short neck, both typical of children with a 7q duplication, are reported repeatedly. Other unusual features are less consistent, and include: low set or lop-sided ears; a very small or receding lower jaw (although one child has a 'strong jaw'); a small or large head; widely spaced or downwards slanting eyes; and an unusual mouth with a short upper lip and multiple skin webs between the cheeks. In terms of hands and feet, one youth had very bendy finger joints, broad first toes, and skin webbing between some fingers, as well as short, broad fingertips, while another has short thumbs.

In terms of development, delay is typical. Learning difficulties have been reported as moderate, although a youth of 18 was reported to have an IQ of 34. A youth of 19 works electronic devices competently, although he does not write. There is little information on speech and language, but one child of 14 had few words and a youth of 19 made a variety of repetitive sounds but no words, while

another had relatively good verbal skills. Again, there is little information on mobility, but one child was able to roll over by 12 months and to walk by 2; another child sat at 10 months and walked by 2. At 19, he has an 'uncoordinated and unsteady' walk, and has very flat feet, for which he is prescribed insoles. Others have low muscle tone (making them feel floppy). The 19-year-old is not able to care for himself or take control of his toileting needs.

As for behaviour, one youth was diagnosed with autism, while another had bouts of aggression towards other children. The youth with autism has also had episodes of aggression when he feels anxious, such as leaving his house, and is being treated with haloperidol to try to keep him calmer. Despite his diagnosis of autism, he shows great empathy when others are upset or in pain. Two Unique youngsters have disrupted sleep, and take melatonin which, one family says, 'calms him down a lot, although we still have many no-sleep nights'.

As for general health, two babies had very frequent infections, including bronchiolitis and meningitis. One has asthma and multiple allergies to pet fur, house dust, peanuts, eggs and mustard. One developed glue ear and needed repeated insertion of aeration tubes (grommets) to preserve hearing. Three children had an inward turn of one or both eyes (convergent strabismus). One child had missing teeth and poor enamel development on the teeth he had, while another tends to grind his teeth and has needed fluoride varnish to protect them.

Lukusa & Fryns 1998; Alfonsi 2011; Rivera 2013; Decipher; Unique

Duplications from 7q21/2 to 7q31/2

In this group there are 10 children of various ages, all with large duplications: 4 from Unique, and 6 in the medical literature (Romain 1990; Humphreys 1991; Mégarbané 2000; Kroisel 2001; Nasiri 2010; Weimer 2011; Unique). There is a further child with a large duplication from 7q22 to 7q34 (Stratton 1993).

Nothing unusual was noticed in the two documented pregnancies, but at birth most babies were small for dates. Among 8 babies, average birth weight was 2.667kg (5lb 14oz), although the average weight of Unique babies was noticeably higher at 3.23kg (7lb 2oz); the baby with the largest duplication from 7q22 to 7q34 was perfectly average at birth in length and weight. Two babies were born with respiratory distress, and it was severe in one baby who needed resuscitation and intubation and went on to have a collapsed lung. This baby also had renal failure, and was born with a very small penis and undescended testicles, although they had come down into the scrotum, the loose bag of skin behind the penis, by the age of 5 months. Two babies had incorrect hip development. One baby was born with cataracts, which were removed.

At least 4 babies had early feeding problems, at least one due to hypotonia (low muscle tone), and 2 were tube fed for some time. One baby had a split in the roof

"Her hand use and co-ordination is quite good. She is able to write, draws well, and loves to use an iPad." 8 years

"He can't hold a pencil or fork normally. Only scribbles, always very messy when eating." 11 years

Although speech delay is common and it may be hard to understand what a child is saying, Unique's experience is that most children are communicative and chatty. Some children benefit hugely from first learning to sign. This may not be possible for all, and one child was not talking by 11 years.

"Very verbal, but unclear with periods of clarity. Starting to use short sentences and currently focusing on sound pronunciation. Uses minimal signing at home but more at school to reinforce speech." 6 years

"Loves to chat. She is very sociable. Her speech is fairly clear." 8 years

Some level of intellectual disability is very likely, but the degree is really quite variable and some children are quick learners of certain skills. Children often struggle to focus and concentrate, and this, together with a slow processing speed, undermines their learning. Children attend both mainstream and special needs schools, and generally need support in the classroom, whether they are focusing on academic or daily living skills.

"She is a very quick learner (i.e. only needs to be shown once, possibly twice and she gets how to do certain tasks). However, any activities that require writing, coloring or using scissors, she could be thought of as delayed due to the hypotonia in her hands." 3 years

"She is slow to learn, and needs repetitive modeling. She attends a special needs school with an educational statement; and attends Beavers (Scout movement) and Rainbows (Guiding movement) sporadically if not too tired to attend." 6 years

"Even though she has learning disabilities, she still attends regular school. The other children love her and treat her like their little sister. Academically she is not able to keep up, but is still learning and socializing. She struggles with focus and maintaining concentration and also has a slow reaction time." 8 years

"He is growing in stature, but mentally he seems to stay about the same. It's like he hasn't developed past 2.3 years. He understands way more than he can verbalise." 11 years



She makes us laugh! She is very loving and has a great personality. Everyone who meets her falls in love with her. 6 years

feet – either a clubfoot on one side or both, or a ‘claw foot’, where the arch is very high and the toes point downwards. Another child had flat feet.

As for development, one child with a small duplication of around 500kb in the 7q31.33 band only showed significant delay in speech and language: her parents were more convinced of her delay than her paediatrician was. This family



Right, at 2 years, with big and little sisters

ascribes dramatic improvements in talking, mobility, muscle tone, feeding, toilet training and behaviour to a gluten-free diet, which they started when their daughter was almost 3 years old. A year later, she was discharged from all therapies, as she was achieving close to normal levels. A child with a larger duplication between 7q31.2 and 7q32 was more significantly affected.

“Two weeks after starting the gluten free diet, she was using 2 and 3 word sentences, and words that we did not know she knew! She FINALLY participated in a conversation with appropriate questions and responses within a few months of going gluten free. She also had incredible expressive language for how she was feeling and what she wanted to do.” 3 years

“Before doing or showing any new skill - standing, crawling, walking, talking - openly, she practises in secret.” 6 years

Children were a little late in learning to sit, crawl and walk, and while one child acquired these skills at the late end of normal, others were not walking independently until 2 to 4 years old. Low muscle tone (hypotonia) is very common, and once on their feet, children may still have an unsteady walk and be prone to tripping and falling. One child had a specific movement disorder (ataxia) affecting the trunk. Hypotonia also affects the hands so that despite generally good hand-eye coordination, children find it difficult to hold cutlery to feed themselves, a pen or scissors. Hand control improves with practice and maturity, but most children are happier using a touch screen or keypad than a writing implement.

“She began crawling at almost 2, used a K-walker from 3, and was walking independently at 4; today she has very good posture and enjoys walking/running/jumping/trampolining and swimming, but drags her feet and trips when tired. She was diagnosed with hypotonia (low muscle tone) and left talipes at the 24-hour check after birth, but all these activities have helped with this and now it is not so obvious. She has excellent hand-eye co-ordination, but still struggles with holding a pen, producing ‘light’ strokes, and cannot use a knife and fork (together).” 6 years

of the mouth covered by the lining of the inside of the mouth (submucous cleft palate), as well as a tongue tie, where the tongue is tethered to the bottom of the mouth by a very short piece of skin and tissue, both of which can make feeding more difficult. Babies generally found it difficult to put on weight, and some were fed with high calorie formula milk and extra fats. Despite this, growth was generally slow and both babies and small children were short and light for their age. One child of 13, by contrast, was extremely tall, in the top 3 per cent of the population for height. One child was found to be lactose-intolerant, with weight gain accelerating once her diet was changed, as well as resolution of constipation, anal prolapses and abdominal pain.

“Often very sleepy and needs prompting to stay awake for feeds. Currently spoon fed on puréed food. Very selective in what he eats.” 9 months

In terms of appearance, a rounded forehead typical of a 7q duplication is reported frequently, occasionally in conjunction with a small but more often a large head. Other common features are downslanting eyes, a small nose, a small lower jaw and chin, and large ears set low on the side of the head. Babies have occasional unusual features of the hands and feet, but these are not consistent. They include double jointed or bendy fingers and thumbs, fingers or toes fixed in a bent position, webbing between toes, and twisted feet and hands.

In terms of development, all babies and children have experienced some delay, but the range is wide and some are quite mildly affected. One family commented that their child developed in spurts rather than at an even rate. Low muscle tone (hypotonia) is very common but not universal, when it occurs affecting a baby’s ability to gain control of his body and learn to sit and walk. The joints may be exceptionally bendy: one family commented that they could fold their double-jointed daughter and ‘pack her in a bag’. Head control, rolling over, sitting and walking were generally all delayed, although one child - without hypotonia - was walking by 15 months. Another, unable to walk at 2 years, got around well by rolling and scooting. Once walking, children typically had problems with balance and coordination, and needed support.

“Appears to have delay in sitting due to poor muscle tone, but is receiving physiotherapy to strengthen the core muscles. He cannot sit up, and still has some head lag although this has improved since commencing physiotherapy. He can roll over to his front but not back again. He does not put things in his mouth yet and although he smiles, has not been heard to laugh. He struggles to pass

He fills my life with love and makes me appreciate everything that is around him. I really understand the meaning of unconditional love in a way I didn’t before. There is joy in anything new he does and acceptance of my son just the way he is, totally unique. My eldest son adores him and protects him and also accepts him and that feeling is mutual.

9 months

items from one hand to another, but bats toys and explores with his hands. He needs a sight test as he does not appear to see very well. He likes different textures to explore with his hands. He goes to baby singing and baby peep.”
9 months

Speech development is affected, but there is some evidence that effects are relatively mild. A baby of 9 months coos and babbles, and follows voices and faces. A child of 7 uses a variety of single words and understands much more; a child of 9 talks normally, even using the vocabulary of an older child; a child of 11 stammers. As for learning, the evidence is scarce, but suggests a moderate or in some cases mild level of difficulty. One child with a moderate learning difficulty is an able reader. The low muscle tone that is typical means that children struggle with hand control and writing, and cope better with light-touch methods such as touch screens.

In terms of behaviour, we have no evidence of any typical behaviour difficulties. All children with a learning difficulty can face extra frustration when they can't communicate their needs and wishes, but Unique records show no evidence of negative reactions to this. One child has Tourette syndrome, a disorder characterised by physical and speech tics.

He enjoys ... cuddles, sitting in his baby gym and playing with toys, in particular materials of different textures.

9 months

As for general health, 2 children had affected kidneys: in one, there was a solitary somewhat malformed kidney which nonetheless functioned well; the other child had reflux, in which urine flushes back from the bladder towards the kidneys with a risk of damaging them, and was treated with

protective antibiotics. One child had frequent viral infections; another had frequent ear infections; and a third had pneumonia twice before he was 3. Two children had a curved spine, caused in part by low muscle tone, and in one the curve improved as muscle tone got better. Three children have had a hearing problem, and one has abnormalities of the inner ear which mean that he is deaf in one ear; one baby has questionable eyesight. A very common problem is strabismus (a squint), most often convergent, where one or both eyes point inwards. One child had optic nerve hypoplasia, a failure of the nerves linking the eyes with the brain to develop fully, sometimes affecting vision.

Romain 1990; Humphreys 1991; Stratton 1993; Mégarbané 2000; Kroisel 2001; Nasiri 2010; Weimer 2011; Unique

Duplications within 7q31 to 7q34

In this group there are 17 children of various ages, all with different-sized duplications: 6 from Unique, one of whom is also reported in the medical literature, and 11 on Decipher (Chen 2011; Decipher; Unique).

At birth, some but not all babies were small for dates: among 5 babies, average birth weight was 2.6kg (5lb 12oz). Two babies were born with a club foot (talipes equinovarus), and one had a raised red birthmark that was removed when she was 2 years old. One baby had a narrowing of the valve between the heart and the aorta, the blood vessel that takes blood from the heart to the body. One baby born at 38 weeks had difficulty maintaining body temperature and glucose levels, and had a high bilirubin level more commonly seen in preterm babies.

In terms of growth, one child, 6, is of average height and weight, and one, 11, is tall for his age, but the rest are noticeably shorter than average and tend to be slight, with difficulty putting on weight. This is despite generally having no particular feeding problems. On the contrary, Unique families say that their children enjoy food, and often eat a greater variety than typically developing children, although they usually avoid meat because it is hard to chew. One child did have feeding difficulties and ate only puréed foods until a gluten-free diet was introduced (see also Duplications from 7q11 to 7q21 or 7q22).

“Eating is probably the best area of improvement since going on the gluten free diet. However, it definitely was not an overnight process and took a lot of work and additional interventions. In addition to variety, her volumes of food have increased: she never expressed hunger before going gluten free, however now you cannot get her to stop saying she is hungry!” 3 years

“Eating is NOT a problem - unless there is not enough food! She has a huge appetite for carbohydrates and fruit. She still gets into a mess when eating (uses her hands to push or keep food in her mouth) and needs to be washed down after every snack or meal.” 6 years

“He gorges, and puts his fingers in his mouth to sweep food to the front. With a fork and spoon he uses the opposite hand to get food to his mouth.” 11 years

In terms of appearance, many (not all) children have slightly unusual facial features, but there is little consistency and they are often so subtle that they are not obvious to families. Of 4 children with an unusual head size or shape, two have a very small head, one a very large one, and one a head that has the form of a parallelogram when viewed from on top. Four children have tiny skin folds across the inner corners of the eyes, but one of these children is of far eastern origin, where this is normal. Two children have a low bridge to the nose, 2 have a short neck, and 2 other children have an unusually short or long upper lip between the nose and the mouth. Feet may also appear unusual, and 3 babies were born with a positional deformity of the

