

## Support and Information



**Rare Chromosome Disorder Support Group,**  
GI, The Stables, Station Road West, Oxted, Surrey RH8 9EE, UK  
Tel/Fax: +44(0)1883 723356  
info@rarechromo.org | www.rarechromo.org

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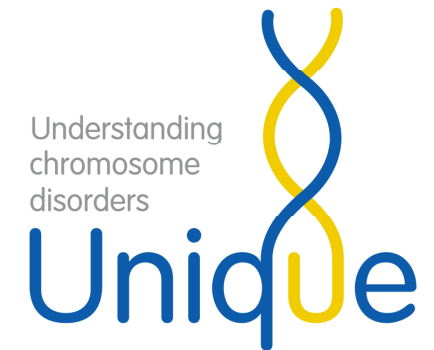
The publication of this guide was made possible through the generous support of Cornerstone OnDemand



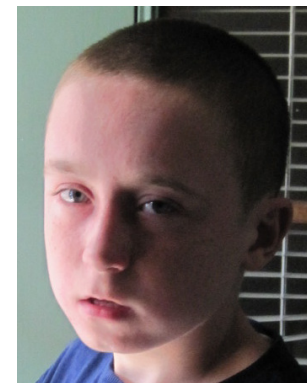
This guide is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be 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. This guide was compiled by Unique and reviewed by Dr Katherine Schon, Specialist Registrar in Genetics, Addenbrooke's Hospital, Cambridge, UK.

2014 Version 1 (PM)

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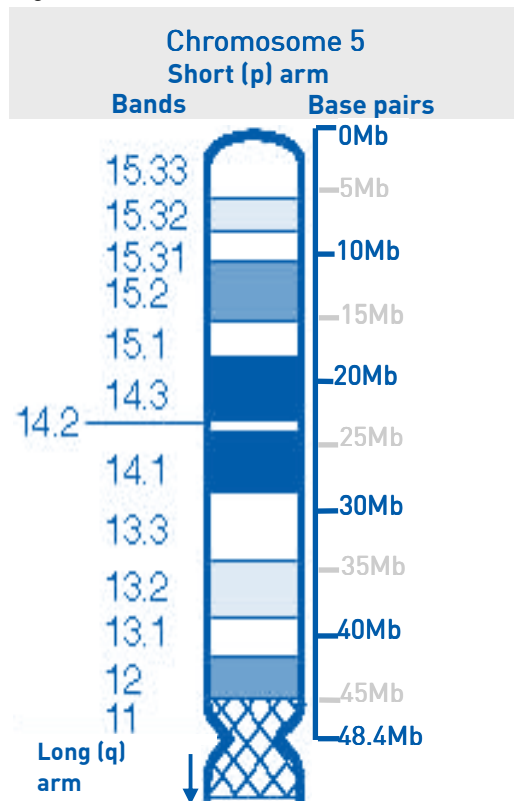
# Trisomy 5p: Inverted duplication and deletion of 5p



## Inverted duplication with deletion of 5p

Inverted duplication with deletion of 5p, known as inv dup del 5p, is a very rare genetic condition in which there is an extra copy of part of the genetic material (DNA) that makes up the body's 46 chromosomes, and a missing copy of another part. Like most other chromosome disorders, this usually affects development, and sometimes health and behaviour as well. It is likely that both the extra and missing parts of chromosome 5p have an effect, but a lot depends on their position and size. The precise effects of gaining material from a chromosome vary depending on how large the duplication is, how many genes it contains and what those genes do. The same applies to deletions. The effects may not be limited to the genes within the duplicated or deleted piece of chromosome because these genes may interact with other genes on the same chromosome or other chromosomes.

Chromosomes usually come in pairs, and we inherit 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. The diagram below shows the short arm.



## Can it happen again?

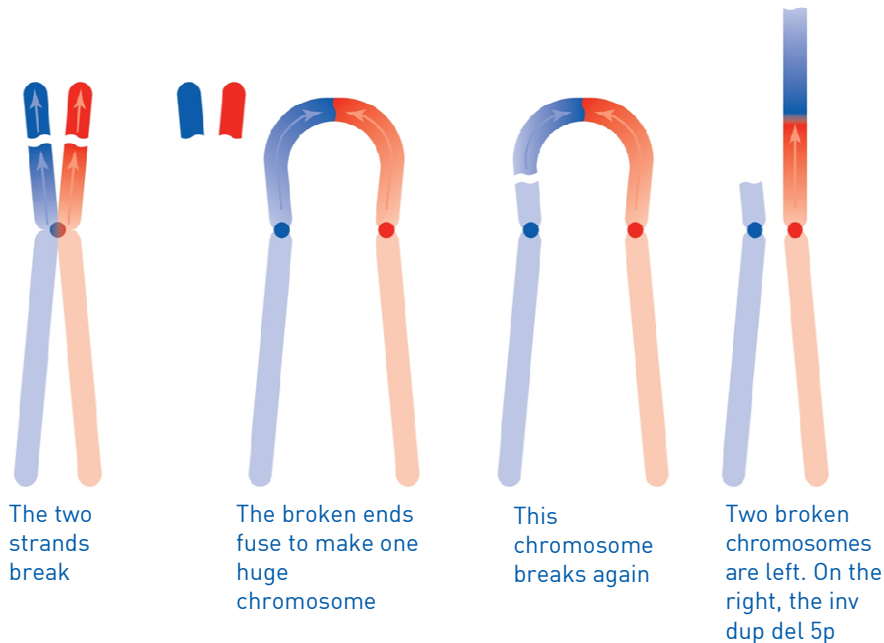
If a child has a  $n$  inv dup del 5p, the parents' chromosomes may also be checked with a blood test. This most often shows that the parents both have perfectly normal chromosomes. The inv dup del 5p has then just happened as a mistake in the immensely complex process of DNA copying and assembly that happens in human chromosomes when the parents' sperm or egg cells were formed or in the very earliest days after fertilisation. The duplication has therefore occurred for the first time in this child, an event that geneticists call *de novo*, shortened to *dn*. As a parent there is nothing you could have done to change or prevent this, just as there are no known environmental, dietary or lifestyle causes of these types of chromosome disorder.

Where both parents have normal chromosomes, it is unlikely that another child will be born with an inv dup del 5p or any other chromosome disorder. Very rarely, both parents have normal chromosomes by a blood test, but a few of their egg or sperm cells carry the same chromosome change as in their child. Geneticists call this *germline mosaicism* and it means that a parent whose chromosomes appeared normal when their blood was tested could have more than one child with the 5p chromosome change. But this is only theoretical: it has never been reported with inv dup del 5p, but it is a theoretical risk.

If they wish, parents should have the opportunity to meet a geneticist or genetic counsellor to discuss the specific recurrence risks and options for prenatal testing and preimplantation genetic diagnosis (PGD). PGD is a technique which uses in vitro fertilisation and embryo biopsy, and only embryos without the chromosome disorder are transferred to the mother's uterus. If the parents choose to conceive naturally, prenatal diagnosis options include chorionic villus sampling (CVS) and amniocentesis to test the baby's chromosomes. Testing is generally very accurate, although not all of these tests are available in all parts of the world.

## How did the inv dup del 5p happen?

Changes to the structure of chromosomes such as inv dup del 5p are believed to occur most often during the cell divisions that lead to the creation of eggs in the mother or the sperm in the father. It is thought that each arm of each of the 46 chromosomes first splits lengthwise into two strands that are held together at the centromere. At this point, something unusual happens. The two strands break and the broken ends fuse, to create one continuous strand, in effect, a huge chromosome with two centromeres. Next, this continuous strand breaks again, leaving two broken chromosomes, one missing a large segment of one arm, and the other the inv dup del chromosome. Look at the diagram below to see how this works.



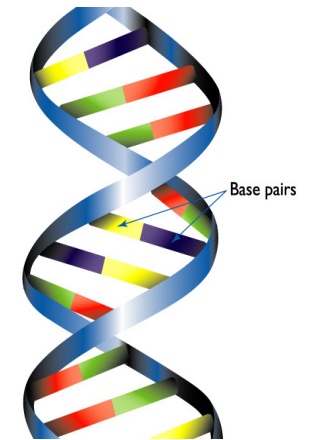
That tells you how the inv dup del 5p probably happened, but it doesn't tell you why. The first step in finding this out is to check the parents' chromosomes with a blood test. With an inv dup del 5p, this is most likely to show that the parents both have perfectly normal chromosomes. The inv dup del 5p has then just happened as a mistake in the immensely complex process of DNA copying and assembly that happens in human chromosomes when the parents' sperm or egg cells were formed. It is a mistake that has occurred for the first time in this child, an event that geneticists call *de novo*, shortened to *dn*. As a parent there is nothing you could have done to change or prevent this, just as there are no known environmental, dietary or lifestyle causes of these types of chromosome disorder.

People have 2 copies of chromosome 5 in most of their body cells. However only one of the copies will have the inv dup del 5p. Part of the short arm is duplicated (dup for short). Straight after the end of the duplication, the tip of the short arm of this chromosome 5 is missing. This is termed deletion (del for short). The extra duplicated part runs in the opposite direction to normal and so is termed inverted (inv for short). You can see a picture of this on page 14.

## Looking at chromosome 5p

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. You can see these bands in the diagram of chromosome 5 on page 2.

Each band of 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. The whole of chromosome 5 has about 181 million base pairs, shortened to 181Mb; the short p arm has rather more than 48Mb. The position of each of the 900 or so genes on chromosome 5 is measured in base pairs. On the right of the diagram you can see how the base pair numbers relate to the chromosome bands.



Looking at chromosomes under a microscope, it is usually possible to see a large piece of extra or missing genetic material, but changes smaller than 5Mb or even 10Mb can be hard to identify. So if the extra pieces are very small, the chromosomes may look normal under a microscope. New techniques, particularly one known as array CGH, are better for finding small changes, and are now often used to find the size and position of the extra and missing DNA, helping to identify genes and pinpoint their location on chromosomes.

## Sources

The information in this guide is drawn partly from 2 reports in the medical literature of children born with a 'pure' inv dup del 5p, not involving any other arm of any chromosome (Sreekantaiah 1999; Vetro 2008; Wang 2008; Mosca 2011; Izzo 2012). Additional sources for the information on the way an inv dup del 5p is formed are Rowe 2009 and Yu 2010. There is a further report of an inv dup del 5p at Vera-Carbonell 2009, but since an extra duplication of 5p12 was involved, this is not a simple inv dup del 5p, and is therefore omitted. 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](http://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 2014, referenced Unique. When this guide was compiled, Unique had 6 members with inverted duplication and deletion of 5p.

## Genetic test results: two examples

A person's chromosome make-up is called their karyotype. Someone with inv dup del 5p might have a karyotype that looks like one of these examples:

### 46,XX,der(5)(p15.33)(5ptel48-)(inv dup(5)(p15.33p14)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. An abnormal (derivative) chromosome 5 was found, with a break in the band known as p15.33. This band is near the tip of the chromosome. 5ptel48- shows that a marker normally found near the tip of 5p, in the p15.33 band, was missing (-), so there is DNA deleted from the tip of the chromosome. inv dup(5) shows that in addition, there was extra material on chromosome 5, running in the opposite direction (inv) to the rest of the chromosome. The start point of the extra material was in band p15.33 and the end point was in band p14, so this is a large duplication. dn de novo (Latin for 'from the beginning') means that this chromosome change has not been inherited from either of the child's parents but has occurred for the first time in this child.

### arr[hg19] 5p15.33(51948-743521)x1,5p15.33p13.2(867903-32630865)x3

The test was by array comparative genomic hybridization (arr). The results follow the Human Genome build 19 [hg19], which is a recent version of a kind of atlas of human chromosomes. 5p15.33 shows that a break was found in the 5p13.33 band, closest to the tip of the chromosome. (51948-743521)x1 shows that one copy (x1) of the material at the break in 5p13.33 was found, instead of the normal 2 copies, so this means material is missing. 51948-743521 are the start and end points of the missing copy, measured in base pairs. Take the first long number from the second and you find that there are 691,573 missing base pairs, which is about 691kb. 5p15.33p13.2(867903-32630865)x3 tells you about the extra (x3) material. It starts in band 5p15.33 and extends as far as 5p13.2. 867903-32630865 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 31,762,962 extra base pairs, which is about 31.7Mb.

“We have been struggling to get a diagnosis, and delayed the genetic tests. If we had known, we would have pushed for this investigation much earlier. We also delayed treatment in a special needs toddler group, but exactly this is what helps us and her the most!” 3½ years

## Pregnancy, birth and the newborn baby

Out of 4 babies, we know that 1 was born early, at 32 weeks of pregnancy, and 3 were born around or after term. Two pregnancies were essentially normal, although in 1 the mother had a high level of alpha-fetoprotein (AFP) when she was tested at 15-20 weeks. The AFP test screens for common anomalies including spina bifida and Down's syndrome. In this pregnancy the baby also had enlarged kidneys (hydronephrosis (see Kidneys, page 11).

In general, babies were on the small side but within the normal range for weight at birth (see Normal growth, page 9), and usually had initial difficulties with breastfeeding. Apart from these common features, individual babies were quite different at birth. One was born near the due date with jaundice (hyperbilirubinemia); another had enlarged kidneys at birth, and then caught pneumonia at just a month old; another had repeated episodes of what looked like breath holding after birth, and was taken to special care (see Seizures, reflex anoxic seizures, page 8) (Sreekantaiah 1999; Wang 2008; Unique).

## General wellbeing

Two families told us how their child with an inv dup del 5p is doing. This is a little girl of 3½ years: 'Apart from her seizures, she was a healthy baby. She ate and drank well and grew within the boundaries, even though she was small.

She appears to be healthy, energetic and shows a lot of age average behavior. She is happy, caring and loving. She appears to feel safe and can be a menace, as she is supposed to at her age!' In just the same way, a boy of 12½ years is 'a healthy child.'

The diagram on page 5 is reproduced with the permission of Wiley-Liss, Inc., a subsidiary of John Wiley & Sons, Inc. from Kjaer, I. and Niebuhr, J. Studies of the cranial base in 23 patients with cri-du-chat syndrome suggest a cranial developmental field involved in the condition. *Am. J. Med. Genet.* 82, 6-14. © 1999 Am.J.Med.Genet.

### ■ Slightly unusual facial features

Your child may look like the rest of your family, but he or she may also have some facial and other features that are common in people with trisomy 5p or in those with cri du chat syndrome. No particular features appear to be characteristic, but those noted here have been seen in one or two babies or children. In order of frequency they are: ears positioned low on the side of the head, perhaps with an odd appearance; somewhat bulging eyes; a small lower jaw; an asymmetric or flat face; a rounded or sloping forehead; tiny tags of skin in front of the ears; eyes set too far apart or too close; an upturned nose; a long groove between the nose and the upper lip; a thin upper lip; a small or large mouth; and a protruding tongue (Sreekantaiah 1999; Vetro 2008; Wang 2008; Mosca 2011; Izzo 2012; Unique)

“Low muscle tension around her mouth: her tongue used to hang out, but is rapidly improving.” 3½ years

### ■ Squint

Any problems with eyesight have only been reported among Unique members. One baby was born with strabismus, where the eyes cross, or squint. Another has a ‘small nick’ in the eyelid, which is monitored, and another had mild but unspecified eyesight difficulties.

“She had strabismus from birth and surgery at the age of 2. The esthetic correction is wonderful. She still has regular investigations, but no abnormalities in strength. The limited view of depth that comes with strabismus appears to be present.” 3½ years

### ■ Kidneys

Kidney and urinary tract problems are not uncommon among babies with chromosome disorders, and a variety of kidney problems are seen in babies with cri du chat syndrome. Kidney anomalies seen in 2 babies with an inv dup del 5p are pyelectasis, where the part of the kidney where urine collects is dilated, and generally enlarged kidneys, which were identified on antenatal ultrasound and led to surgical correction at the age of 5 months (Izzo 2012; Unique).

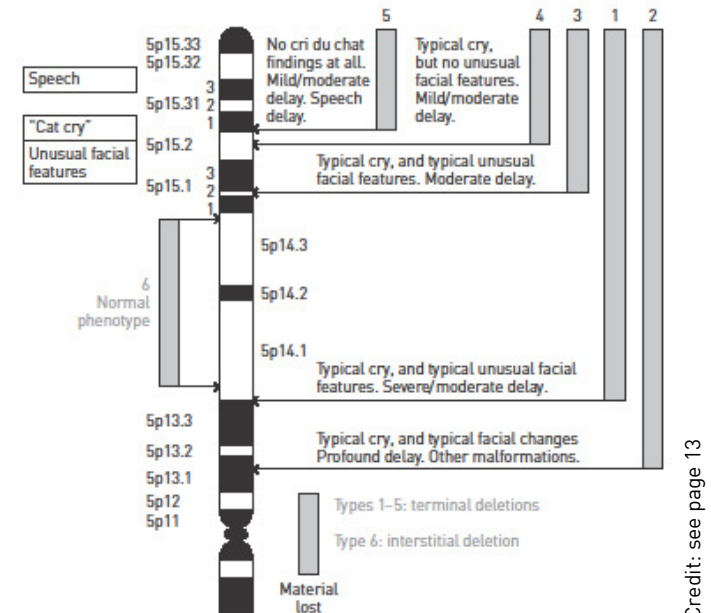
### ■ Hands

Slight anomalies of the hands are quite common among babies and children with a chromosome disorder, but in this group with an inv dup del 5p, the anomalies are scarcely noticeable. One baby was born with broad thumbs, and another with square-shaped finger tips (Wang 2008; Unique).

## What are the effects of an inv dup del 5p?

The effects of an inv dup del 5p are a combination of the effects of the deletion and of the duplication. The full effect is not a simple additive one, with the effects of the deletion added on top of the effects of the duplication, but in interpreting how a child with an inv dup del 5p is, and is likely to develop, that is the starting point.

## Effects of a 5p deletion



People with inv dup del 5p have very different sizes of deletion. Some have a tiny deletion from near the very tip of the short arm in the 5p15.33 band, while others have deletions so large that they should be visible under a microscope. There do not appear to be any places where the chromosome is particularly liable to break.

Deletions of the end of chromosome 5p cause a condition known as cri du chat syndrome, so the overall effects are quite well known, and are shown in the diagram above. Overall, a larger deletion tends to have a more serious impact.

## Effects of a 5p duplication

The size of the duplication, like the deletion size, varies widely between individuals. While most have a duplication that starts from the end of the deletion in the 5p15.33 band, the duplications end anywhere between the 5p15.31 band (a small duplication) and the 5p12 band (a duplication of almost the entire short arm). The largest duplications are more than 20 times as big as the smallest. There do not appear to be any points where the chromosome is particularly likely to break.

### Effects of small duplications within 5p15

The most common features of a small duplication within the 5p15 band are, running from most to least common: speech delay; behaviour difficulties and autistic features; developmental delay; feeding difficulties; seizures; small size; learning difficulties; unusual joints and limbs; low muscle tone; heart problems; eye and vision problems; slightly unusual hands; slightly unusual facial features.

For more detailed information about duplications within 5p15, see Unique's guide to [Trisomy 5p Duplications of 5p15](#).

### Effects of medium-sized duplications extending to 5p14 or 5p13

The most common features of a duplication extending to 5p13 or 5p14 are, running from most to least common: developmental delay; learning disabilities; small size; feeding difficulties; low muscle tone; speech delay; autistic features; a large head; anomalies within the brain; unusually positioned feet or clubfoot; seizures; heart problems; eye and vision problems; slightly unusual facial features slightly unusual hands.

For more detailed information about medium-sized duplications extending to 5p14 or 5p13, see Unique's guide to [Trisomy 5p Duplications of 5p13 and 5p14](#).

### Effects of large duplications extending as far as 5p12

The most common features of a large duplication extending as far as 5p12 are, running from most to least common: developmental delay; low muscle tone; normal size at birth, but slow growth after birth; a large head; seizures; tracheobronchial/ respiratory involvement; club foot and other foot anomalies including a short first toe; and unusual facial features including small upslanting eyes, tiny skinfolds across the inner corner of the eyes, a low nasal bridge and low set ears.

For more detailed information about large duplications extending as far as 5p12, see Unique's guide to [Trisomy 5p Duplications of the whole 5p arm](#).



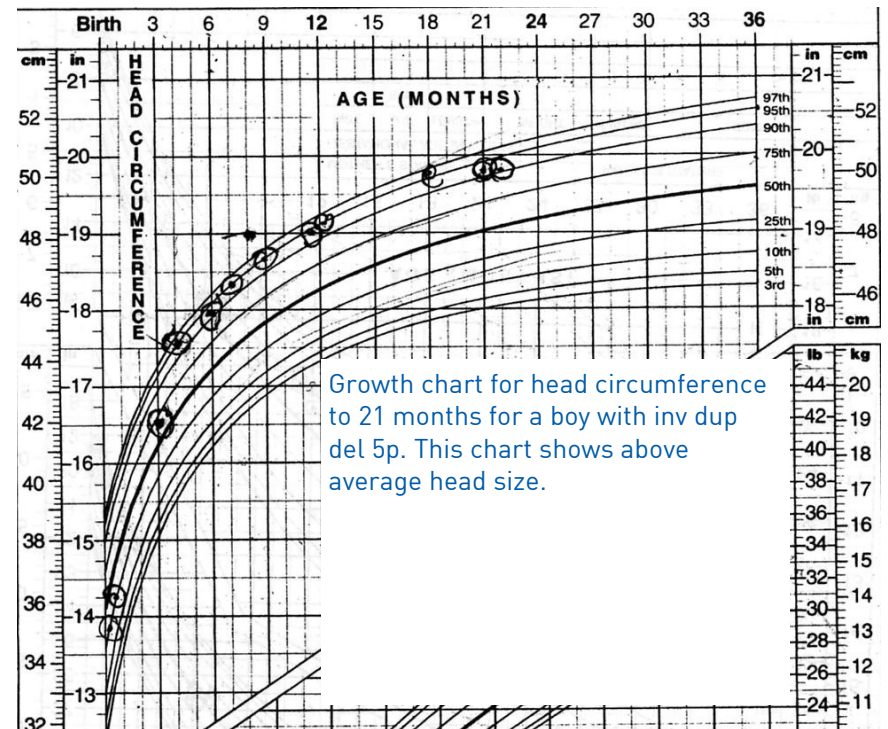
### ■ Head and brain

Babies and children with a duplication of 5p most typically have a large head, but this was only found in 1 child with a relatively small duplication within the 5p15 band, out of 7 for whom we have information. Three children have completely normal head shapes and sizes, while 3 others have a slight abnormality: in one case a square-shaped head; and in another, a relatively small front part of the skull and face compared with the back.

Among 5 cases where the brain was imaged or examined, 2 were completely normal. Both these children are living and well, compared with the 3 who were either foetuses where the pregnancy was terminated or babies who had sadly died. These different outcomes mean that this group almost certainly presents a distorted picture: if more was known about the brains of healthy children and adults with an inv dup del 5p, more of them would probably have quite normal brains.

The anomalies identified varied from case to case: one had a cyst between the two halves of the brain; another had an underdeveloped band of nerve fibres (corpus callosum) linking the two halves of the brain; and a child who died had a small, shrunken brain (atrophy).

(Sreekantaiah 1999; Vetro 2008; Wang 2008; Mosca 2011; Unique)



## ■ Unusual feet

5/11 babies with inv dup del 5p are known to have had some kind of foot anomaly. This can be quite mild, like overlapping toes, or more serious, and need correction for walking to be possible, like a marked club foot.

Two babies did indeed have clubfoot, both of them with large duplications of 5p, and one with a large deletion as well. The type of clubfoot seen most commonly in 5p duplications is talipes equinovarus, where the foot points downwards and inwards. The foot is usually short and broad and the heel points downward while the front half of the foot turns inwards. Treatment is individually tailored and aims to straighten the foot so that it can grow and develop normally.

Another baby had curved soles known as rocker-bottom feet, and a 5-year-old child had hypermobile flat feet, needing support with specially-shaped insoles. Two children had unusual toes, one of them unusually broad. (Vetro 2008; Wang 2008; Mosca 2011; Izzo 2012; Unique)

## ■ Behaviour difficulties, including autism

We only have information on behaviour in 5 children, but of these at least 3 have a diagnosis of autism, so this appears to be common. Signs appear early, and one boy was diagnosed by the age of 4. Autism can exist alongside endearing character traits: one child diagnosed autistic is described by his parents as 'even-tempered, getting on well with other people, and very loving'. He responds well to Applied Behaviour Analysis (ABA therapy), a behaviour modification approach that has had great success with children with autistic features and classical autism.

Other character traits are described in one child only. They include aggression in a child of 2 years; and sensory dysfunction – problems in organising responses to information received by the senses. But one child, as you can see below, is much like any other 3-year-old. (Wang 2008; Unique)

"She is a happy, quiet and playful child who is really attached to her parents, but loves to go to the revalidation center. After initially being cautious towards other children, and preferring to play by herself, she gained their trust and now loves playing with other children. This has given her development a great boost. She also loves playing with her toys (dolls, little people, Lego, Dora) and reads books. She is now starting to draw more and more, and showing fantasy-play etc.

"On the problem side, she makes contact with adults easily and is so trusting that she could walk away with them. She also shows a lot of frustration when she doesn't understand things or questions and that can lead to anger." 3½ years

"The diagnosis of autism was very helpful." 12½ years

## What do we know about people with an inv dup del 5p?

Looking at the information we have on 7 babies and children with inv dup del 5p, with differing sizes of duplication and deletion, the most common features are:

- Developmental delay, including delay in moving, speech and learning
- Epilepsy
- Feeding difficulties
- Normal height and build
- Unusual feet
- Behaviour difficulties, including autism
- Brain and head
- Slightly unusual facial features

Other less common features include:

- Squint
- Kidneys
- Hands

- Developmental delay, including delay in moving, speech and learning

"We first noticed the delay in her motor as well as her language/speaking abilities. We roughly estimate it between 6-12 months of delay at 3½ years."

Some degree of developmental delay has been observed in all 7 babies and children with an inv dup del 5p. A 4-year-old girl with a deletion of most of the 5p15.3 band and a duplication between 5p14 and 5p15.3 showed only mild delay, sitting, crawling and walking on time, but then developing slightly more slowly; and starting to talk rather late and only stringing sentences together by the age of 3 (Sreekantaiah 1999). Unique children with deletions of different sizes within 5p15.3 and duplications reaching as far as 5p14 or 13 showed more obvious delay affecting every area of development. Babies were late to hold their head steady (5 months); to sit (8-11 months); to crawl or otherwise get mobile (around 14 months); and to walk. However, their mobility was only mildly delayed, and they were generally able to walk with support from 17-18 months, although unsupported walking did not develop until several months later.

In most children low muscle tone underlies the delay in sitting, walking and moving, and some children have unusually bendy, loose joints as well. Three out of four children that we know of have low muscle tone (hypotonia).

(Sreekantaiah 1999; Wang 2008; Unique)

"To get around, she walks, runs or uses her favourite 4-wheeled bike. Her favourite activities are walking, and going to the playground, especially the swing. Both her gross motor skills and her ability to speak appear to have been delayed due to the low muscle tone and hyperlax joints." 3½ years

"He has slightly low musculature, and is less strong than a normal child but is not floppy. He most enjoys walking and swimming." 12½ years

Starting to talk has been delayed in all the children we know about, but the amount of delay varies, with some children talking reasonably fluently while others only use one or two words, if any. Babies started to smile socially between the ages of one and 9 months, and the earliest age at which a child started to use words is 2 years. Children use many alternative ways of communicating, including crying, pushing, pulling, pointing, pictures and in particular signing, usually understanding many more signs than they can make themselves. While some children really try to communicate, there is evidence from Unique that others are quiet and somewhat unresponsive. (Sreekantaiah 1999; Wang 2008; Unique)

“She was a quiet, shy baby and didn’t babble a lot, but her speech is improving rapidly now and she also uses signing, pushing and pulling and pointing. She has speech therapy, but what boosted her speech was being part of a group of children of her own age, and now she uses 2-3 word phrases, and is starting to use verbs in a proper way, using past and present tenses. She does seem to be frustrated more often as she appears to understand very much of what we say and ask, but lacks the ability to respond verbally, and she has difficulty saying the sounds at the beginning of words, such as: *s, sch, k, g, v, f, z* and *l*.” 3½ years  
 “He uses one or two words.” 12½ years

#### ■ Seizures

Every child with an inv dup del 5p apart from a 4-year-old with a duplication between 5p14 and 5p15.3 has had seizures, but they have not all been diagnosed with epilepsy. The child who had no seizures also had normal brain scans and normal recordings of electrical activity in the brain (EEG) ([Sreekantaiah 1999]. Seizure types and age of onset varied in the other children. One baby had 2 seizures at 3 months of age, was immediately prescribed an anti-epileptic drug, and over 3 years later remains seizure-free, so it is now not known whether she would have been seizure-free without the medication. Another baby started to have myoclonic seizures [jerky muscle contractions usually in the arms and legs and lasting at most for a second] at 1 year of age; seizures increased at puberty but are well controlled with anti-epileptic medications. One child has had reflex anoxic seizures, which are not epilepsy, but a sudden response to fear or pain where the child cries, turns pale and may stiffen and then go limp, before recovering spontaneously after a few seconds or a minute. Another has tonic-clonic seizures, where the child suddenly stiffens and if on their feet may fall, but then their muscles contract repeatedly and rhythmically. This type of seizure used to be called grand mal. Seizures of this type usually last no longer than three minutes. (Wang 2008; Unique)

“He is very loving.” 12½ years

#### ■ Feeding difficulties

At least 5 babies had initial feeding problems, but in most cases we know about the difficulties were fairly mild, to do with breastfeeding, and by the toddler stage children were eating a normal diet in a more or less normal way. This is not possible for all, however; at least one child was not able to feed himself at the age of 4. Three Unique families report difficulty establishing breastfeeding: one baby fed at the breast, but less energetically and for shorter periods than his unaffected siblings; another simply refused to breastfeed; while another was fed for the first few days by syringe, and once breastfeeding got underway, was still given extra milk to ensure that she grew well.

One baby experienced a very sharp drop in weight after birth until feeding was properly established, and another failed to thrive, meaning that he was unable to drink enough to properly maintain his growth rate. Two babies had reflux, where feeds aren’t efficiently processed into the stomach but instead come back up the food pipe and can be inhaled (gastro oesophageal reflux/ GORD/ GERD). Managing mild reflux can involve sitting a baby semi-upright during and after feeds; raising the head end of the cot or bed; or, if it is more persistent, using prescribed anti-reflux milks or medications. (Wang 2008; Unique)

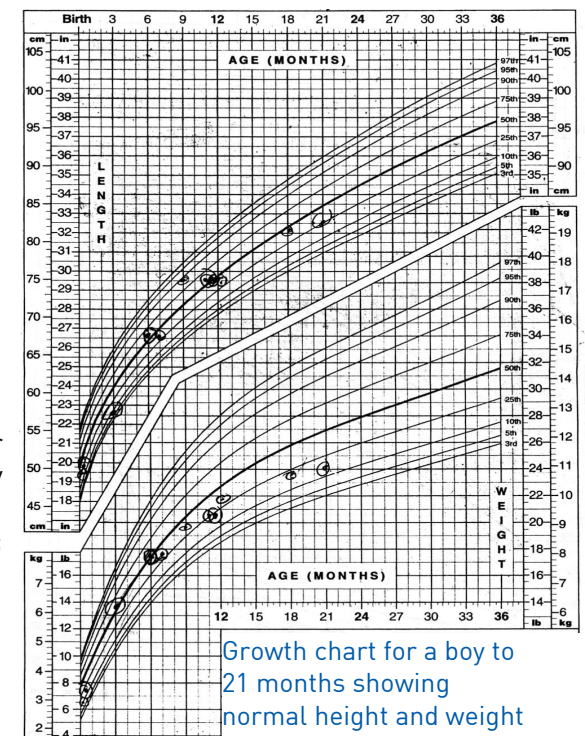
“She eats & drinks like a 3-year-old is supposed to, and basically eats everything that we eat. We do eat quite healthy food - low fat, fresh meat & vegetables, and fruit.” 3½ years

#### ■ Normal growth

Babies are on the small side at birth. Among 5 babies whose weight at birth is known, the average for babies born around term was 2.75kg (just over 6 pounds), which is small but within the normal range. Just one baby is known to have been small while growing in the womb.

From what we know about babies and young children, their height and weight seems to stay within the normal range. (Sreekantaiah 1999; Wang 2008; Izzo 2012; Unique)

“A bit small, but within the normal range. Based on her clothing size I would estimate her height at 1 metre (3’3”).” 3½ years



Growth chart for a boy to 21 months showing normal height and weight