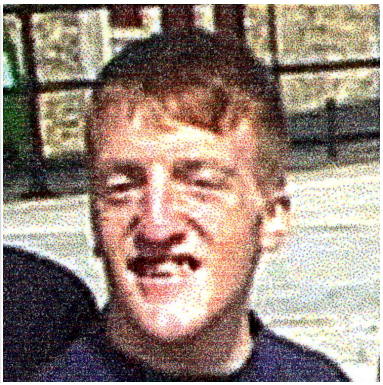
Understanding chromosome disorders

Unique

13q distal interstitial deletions



rarechromo.org

Sources & References

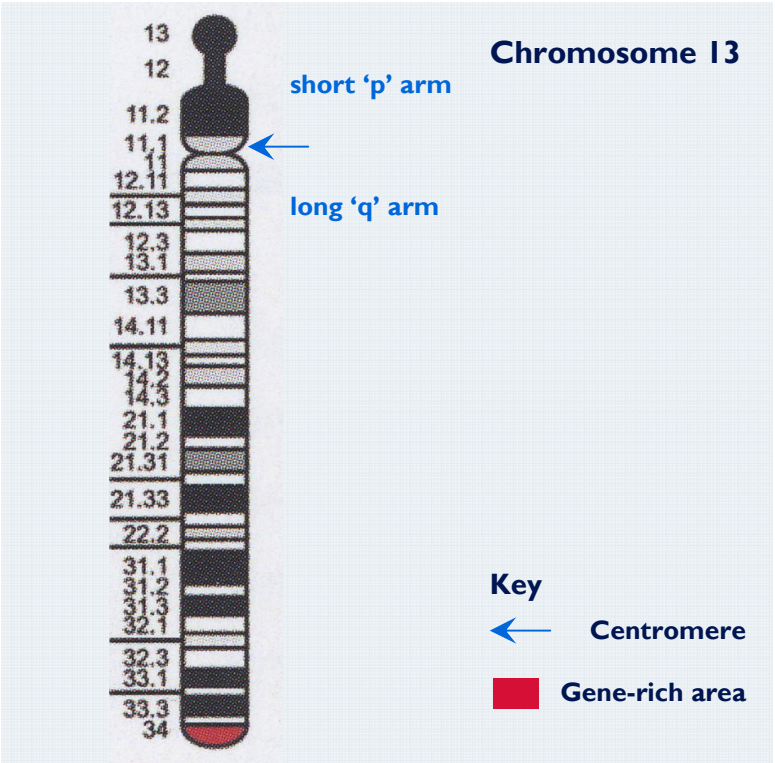
The text contains references to articles in the medical literature. The first-named author and publication date are given to allow you to look for the abstracts or original articles on the internet in PubMed. If you wish, you can obtain abstracts and articles from *Unique*.

References to information from the Unique database and a questionnaire completed by *Unique* families are marked U.

13q distal interstitial deletions: deletions within 13q21-q34

People with this rare chromosome disorder have part of the bottom (distal) end of the long arm of one of their chromosome 13s missing from the cells in their body. People with a single break have lost the end of the chromosome (see *Unique*'s leaflet **Deletions including the end of 13q**). People in whom the chromosome has more than one break, but the sticky broken ends of the chromosome have re-joined, leaving out a segment, have what is called an interstitial deletion.

This information sheet describes what we know about people who have lost chromosome material between bands 13q21 or 13q22 and bands 13q32 or 13q34. In the medical literature, people with deletions between 13q22 and 13q32 are generally considered to be mildly affected. Their level of learning disability is generally not severe, they often have no major organ defects and may only have minor oddities, such as low set ears, a squint (strabismus) or thumbs set oddly on the hand. However, the picture may change when more material is missing from near the end of the chromosome (Brown, 1993; Kotzot 1991; Onufer 1987; Peet 1987).



While the medical literature is helpful in identifying the distinctive features of people with 13q deletions, lifelong data is more helpful in describing development and in sketching possible outcomes. In addition to the published medical literature, we have therefore drawn on information, updated annually, about 17 members of *Unique.* Thirteen *Unique* members have lost a large segment of the chromosome from band 13q22 to band 13q32. Four *Unique* members have a smaller deletion within or near band 13q22 but their features are generally similar to the rest of the group.
The *Unique* sample includes 10 girls and seven boys. Seven families completed a detailed questionnaire in 2004.

Main effects

Effects are variable. People will not have all of these features. Some children will have a few and a few children will have many of them.

- Slow growth *in utero* and small for dates at birth
- Small head
- Developmental delay
- Learning disability (variable)
- Hypotonia – a floppiness of the muscles
- Finger and toe defects: absent or short thumbs, fused toes; short, incurving fifth finger

Other features

When your child is examined, the clinicians will be particularly alert to problems in these areas because they are typical of this chromosome disorder.

- Hearing
- Stability of the hip joints
- Anomalies in the formation of the brain
- The perineal area. The hole for the anus may be placed close to the genitalia or may not be visible. In boys, the testes may not be descended, the genitalia may not be fully developed or may be ambiguous. Occasionally, the penis and scrotum may be transposed so the scrotum appears above the penis
- The anatomy and function of the intestines. Hirschsprung's disease (a condition that causes acute constipation) may occur
- Heart – both simple and complex conditions may occur
- Eyes - the optic nerve may be underdeveloped. Coloboma of the iris (keyhole shaped defect) may also be seen
- Mouth, in particular a high palate
- Skeleton, in particular the hands. In addition to missing thumbs, a typical feature is fusion of the fourth and fifth metacarpal bones in the hand.

(Shanske 2001; Brown, 1993; Lamont 1989; Tranebjaerg 1988; Carnevale 1984; Brondum Nielsen 1981; Carukashansky 1979).

Appearance

There are facial similarities between people with 13q deletions. However, these are often quite subtle and may be more obvious to doctors than to parents. The similarities may be more obvious if you see photographs of others with a similar chromosome disorder. Characteristically, eyes slope downwards and are sometimes set unusually far apart. Ears may be set low on the head and appear large or to be tilted backwards. The bridge of the nose can be quite strong and once your child's upper teeth grow, they may be prominent.

Small head

Typically, children have a small head but it is usually in proportion to body size (Carnevale 1984; Brown 1993). In *Unique*'s experience this is an inconsistent feature and size does not relate to later function. Out of seven families, three reported a small head and four families said their child's head was not small. Most families in both groups had a q22q32 deletion, but one child with a small head had a deletion within band q22, while one member without this feature had a q22q34 deletion. One family (q22q32 deletion) has described a rapid drop in head circumference in the first six years: at birth their baby's head was at the 95th centile – near the upper line on a growth chart; by the age of one year it had dropped to below the fifth centile and by the age of six it was below the second centile – the lowest curve on the growth chart. *Unique*'s two highest-functioning adults with this deletion both have small heads, while one adult with a proportionate head size has a profound learning disability (U).

Slow growth in the womb and small for dates at birth

Research reports agree with the *Unique* experience that babies typically grow slowly *in utero* and are small for dates at birth (Carnevale 1984). Birth weights among *Unique* members generally range between around 2000 grams (4lb 6oz) and 3000 grams (6lb 10oz). But one baby – with a 13q22q32 deletion - was born weighing 3880 grams (8lb 9oz) (U).

Slow growth and short stature

The rate of growth after birth for most children is slow, so although people with 13q deletions may grow steadily, they tend to be short for their age and predicted final adult height may be below five foot (1m52). One *Unique* adult is 4'6" (1m37) tall. However, in *Unique*'s experience some children do reach average adult height. Children within *Unique* who have had their growth hormone levels checked by an endocrinologist have not been found to be deficient (U).

A childhood growth trajectory for one girl (q22q32 deletion) showed a steady difference of six inches between her height and an average height for a girl of her age: 2'1" (64 cm) at 12 months … 2' 10" (86 cm) at 4 years … 3' 3" (1m) at 6 years … 3' 9" (1.2m) at 10 years … 4' 9" (1.45m) - final adult height - at 14 years.
This compares with an average height on a UK standard child growth chart of 3' 4" (101cm) at 4 years … 3' 9" (1.15m) at 6 years … 4' 6" (1.38m) at 10 years …. and 5' 3" (1.59 m) at 14 years.

This pattern of slow but steady growth is, however, not uniform among *Unique* members. One baby was in the top five per cent of babies for length at birth but had

dropped by her first birthday to the lowest ten per cent. She then remained small throughout childhood until her growth spurt at puberty took her beyond the doctors' predictions to a final adult height of 5'6" (1.68m). Another child was of average height until her height increased rapidly when she started premature puberty around the age of seven. Another child grew very slowly in her first two years, then faster but still at a below average rate, while from the age of four she has grown at an average rate. At almost seven years old, she is 3' 7" (1.09m) tall.

Feeding difficulties and failure to thrive

Many babies with chromosome disorders find feeding difficult at first and struggle to put on weight. Initially their sucking reflex is weak and they cannot co-ordinate sucking with swallowing. From a practical point of view, this means that feeds by breast or bottle can take as long as two hours and babies tend to fall asleep in mid-feed. The *Unique* experience shows that while feeding problems are similar in type, they vary in severity. The most mildly affected babies may even breastfeed successfully, albeit slowly. They commonly still have a problem gaining weight but may be prescribed high-energy, vitamin-enriched supplements or milks. They may then be able to move on to solids with only minor difficulties coping with new textures.

"Mie was always a very small eater – she would taste anything but not eat a lot of it.

At the age of 26 Mie was 4' 9" (1.45m) tall and weighed 8 stone 3lb (52 kilos).

More severely affected babies need nutritional help, either being fed by nasogastric tube or direct into the stomach via a gastrostomy tube. This needs only to be a temporary step until the baby gains the strength and co-ordination to cope with his own feeds. Five babies in seventeen in the *Unique* series have needed a gastrostomy and one needed a central line for total parenteral nutrition.

Gastro oesophageal reflux, in which milk and stomach secretions flush up the food pipe from the stomach, and vomiting are common, with the risk of aspiration pneumonia. Careful positioning during and after feeds and feed thickeners may be enough to control reflux and medication may inhibit gastric acid that can otherwise cause pain in the foodpipe. But three in 17 *Unique* babies needed a fundoplication, an operation in which the top of the stomach is wrapped around the bottom of the food pipe and stitched in place. At the same time the hole in the diaphragm through which the food pipe passes is tightened.

Some babies have a medical cause for their slow weight gain. As examples from the *Unique* series, one baby's weight gain improved markedly after surgery for a heart condition, another when her tonsils were removed at the age of 14, and another after surgery for a chest malformation.

"Carrie continued to be short for her age and very thin-looking and mal-nourished. After pectus repair at the age of 10 she started gaining weight and being healthier.

Difficulties with weight gain do not appear to relate to the amount of chromosome that is missing. In *Unique*'s experience babies with small deletions within band 13q22 are as much affected as babies with much larger losses stretching from 13q22 to beyond 13q32 (U).

Constipation

Research has shown that some children who have lost all or part of band 13q22 will have Hirschsprung's disease, a condition in which the nerve cells that normally control the rhythmic contractions that lead to matter being pushed through the intestines are missing (Shanske 2001; Bottani 1991). Hirschsprung's disease results in severe constipation which is treated with bowel washouts or your child will have a temporary stoma, an operation to create an outlet from the bowel that bypasses the affected section of the gut. In time your child will have a further operation to pull through the non-functioning section of bowel, connecting the normal bowel to the anus.
Four out of 17 *Unique* families have noted severe or chronic constipation but none has been diagnosed with Hirschsprung's disease or required pull through surgery (U).

Sitting, walking, moving on …

"At 6, Hayley trampolined, ran, swam (very well), climbed and rode a horse, but she is not very co-ordinated.

"Carrie learned to ride a tricycle with foot straps at the age of 6 but could never ride a bike because of poor balance.

"Cycling took for ever to learn because of Mie's difficulties with balance; she loved horse riding and it helped her balance.

Babies are typically late to reach their developmental 'milestones' but they get there in the end. There is a broad range among *Unique* members, with the earliest sitting at six months and the latest at 24 months. Crawling started on average between eight months and three years and children started walking between 15 months and three and a half years. Once on their feet, children tend to be unco-ordinated and to have specific problems with balance but they make gradual, steady progress. Balance problems impact on the ability to cycle and ride a horse.

From the limited information available from *Unique*'s records there is a suggestion that children who have lost a small amount of chromosome material from around band 13q22 rather than a larger segment may have a distinctive pattern of delay. Their gross motor skills – sitting, standing, walking – may be as delayed as in children who have lost the bit from around band 13q22 to around band 13q32, but their fine motor skills (dexterity) may develop faster.

Hypotonia - floppy muscles with low tone – is very common but not universal in babies and young children. One family specifically said that their baby was never hypotonic and four have not recorded this as a special problem. It certainly affects some children very much more than others. In a newborn baby it may be so marked that it is the first sign that anything serious is amiss but two families said that it was only a problem of babyhood. One *Unique* member needed pillows to support her sitting into her third year and two children had ankle supports to enable them to stand.

Learning

The medical literature and *Unique*'s experience agree that families can usually expect their child with a 13q21-q34 deletion to have some degree of learning difficulty. But the extent of the difficulty is extremely variable. In the *Unique* series, the range is from a moderate learning difficulty to a severe one. One child with a q22q32 deletion has a very mild learning disability. One member with a q22q34 deletion has a profound learning disability. The snapshot views on page 7 give some idea of what individuals may achieve.

- Hayley attends a mainstream school with help. She started drawing at 3 and now likes to draw people, trains and her imaginary friends. She was also reading by the age of three and now reads comics and newspapers, generally preferring fact to fiction. Her writing is large and untidy and she is on level 1 of the UK mathematics curriculum (age appropriate). According to parental report, her memory is frighteningly good and her determination and curiosity help her achieve to her maximum ability. Her level of difficulty is described as very mild – age 6.
- Joe reads road signs and common notices and can write his name with a lot of help. He enjoys reading comics and has good mouse skills, and likes talking books on his computer. He is especially good at quickly mastering the equipment he needs for entertainment. He is good at remembering his home and familiar places. Joe's level of learning disability is described as profound – age 22.
- Carrie started to read when she was 9 but had specific difficulties with visual perception. She always found writing hard and today takes a long time to write. But she has used a keyboard since the age of 10 and is computer literate. She has an excellent memory for subjects that interest her – chiefly films (movies). At school her English was five grades ahead of her mathematics and her speech was – and still is – better than her writing. She attended a special class in a mainstream school and joined her peers for library, art and sometimes for physical education. Her IQ is assessed at 70 and her level of learning disability is described as moderate – age 23.
- Mie started to read when she was 7 and now, as an adult, she reads whatever she needs to. At the Rudolf Steiner School she attended for five years, she wrote before she could read and today although her spelling is not perfect, she sends text messages without difficulty. Her long term memory is good and she uses an appointments diary to compensate for gaps in short term memory. Mathematics and finance are areas of weakness but Mie speaks and understands English as well as her native Danish. She moved from the Rudolf Steiner School to a local mainstream school for two years, then into special education for two years before moving on for eighteen months to a 'finishing school' where she learned cooking, cleaning, washing and other household skills. Mie's level of learning difficulty is described as moderate – age 26.

To read about Mie's life, turn to pages 13-15.

Speech and language

Almost all children known to *Unique* have experienced delay in learning to talk but as adults most have developed speech and some are able to hold a conversation. Some children have a hearing impairment and many start to communicate using sign language before developing speech. First words typically emerged around the age of four or five but some children were eight years old before they started to speak. Two children (out of 15) were not speaking at nine years old and one was talking but unclearly. One child with a small deletion within band 13q22 was understanding at an age appropriate level at age 5 but still using single words to express himself.
The snapshots on the right hand page illustrate some characteristic features.

- Speech developed slowly and signing was needed to speed up communication. Getting eye contact took time.
- Speech was unclear and some sounds were incorrect. This child has a hearing impairment.
- Speaking made her breathless when she was young, so she used gestures when tired.
- Although she used long and complex sentences and spoke clearly, she did not understand jokes or anything that was not 'black and white'.

Medical concerns

Medical research suggests that most people with interstitial 13q deletions involving the segment of the chromosome between bands q22 and q32 do not have major health problems and the *Unique* experience supports this (Brown 1993; U). The most common problems experienced in *Unique* members were breathing disorders (5/17); kidney and urinary problems (4/17); dislocated or easily dislocatable hips and other loose or hypermobile joints (3/17); unusually angled or club feet (3/17); scoliosis (3/17); premature puberty (3/17).

Breathing disorders

Babies had a variety of respiratory problems including frequent infections, caused or made worse in many by inhaling milk. Two babies have needed a tracheostomy, the insertion of a tube into the windpipe to allow free flow of air and oxygen. One child was born with pectus excavatum, a severe chest deformity that does not allow the lungs to grow or expand properly. She suffered repeated respiratory infections until she had her chest rebuilt at the age of 10 and still has small lungs and gets quickly breathless when she walks. As babies' swallowing co-ordination improves, the risk of inhaling milk diminishes and aspiration pneumonia is less of a concern. By the teen years, the only *Unique* member left with a chronic breathing difficulty was the girl born with pectus excavatum.

Kidney and urinary problems

Four babies were born with a kidney abnormality or developed frequent urinary tract infections. In one baby the tube that takes urine from the kidneys to the bladder was incorrectly positioned and in another it was blocked, while another child was born with a single kidney and the fourth child had frequent urinary infections from the age of three, but these did no lasting damage.

Dislocated hips and other hypermobile joints

Three *Unique* members, all girls, were born with easily dislocated hip joints. Two needed surgery but both now have normal joint movement. The girl who did not require surgery as a baby is now an adult and has considerable pain from her hypermobile joints.

Talipes and other unusually angled feet

Three babies were born with unusually angled feet, but none needed surgery.

Scoliosis

Three girls have developed a spinal curvature but in two this is mild and is treated with physiotherapy and exercises. The scoliosis in the third girl is caused by two abnormal vertebrae and she wears a body brace to limit the curving and may need surgery.

Premature puberty

While puberty developed in the normal way in the three adults, three younger girls had signs of precocious puberty. One developed body hair at seven months and another started full puberty at the age of seven and is being treated with drugs to stop the ovaries from producing oestrogen.

Ano-genital anomalies

Anal displacement, bringing the anus close to the genitalia, was reported by two families out of seven and is a recognised feature of this chromosome deletion (Brown 1993). Genital anomalies, a recognised feature of this deletion, have been seen in only one *Unique* child who has a double vagina (Gershoni-Baruch 1996).

Heart conditions

A heart condition has been noted in only one *Unique* member. In the medical literature, heart conditions have affected a minority of children. Types of condition include Fallot's tetralogy with a 13q21.2q32 deletion and PDA and ASD with a 13q22q34 deletion. Fallot's tetralogy is a complex heart condition in which circulation to the lungs is reduced and de-oxygenated blood passes to the aorta, so the child appears blue. PDA is persistent ductus arteriosus, a persisting fetal structure which either resolves naturally in time or is corrected surgically, while ASD, atrial septal defect, is a hole between the upper chambers of the heart (Brown 1993).

Medical concerns described in the medical literature but not among *Unique* members include:
Intestinal anomalies These may include narrowing of the intestines and malrotation of the gut (Khong, 1994; Brown 1993)
Developmental brain defects (Towfighi 1987)
Defects in segmentation of the lung (Brown 1993)
Neural tube defects (Luo 2000)
Multiple spleens (Khong 1994)
Vertebral fusion, inguinal hernia, seizures (Lamont 1989)

Hearing

Hearing can be a major concern and a cause of speech delay. Seven out of 15 children in the *Unique* series have a permanent hearing impairment in one ear or both and three others have a fluctuating hearing loss caused by glue ear and repeated ear infections. One parent noted that as her daughter is very easy-going, she didn't cry with ear infections. She has required three surgical interventions to replace her eardrum. One child was educated at a school for the deaf (U). Hearing impairment is recognised in the medical literature and may be related to the absence of genes required for the normal development of the inner ear (Shanske 2001; Van Camp 1995; Roland 1989).

Vision

A high proportion of children in the *Unique* series have an important visual defect. Three have a coloboma, a gap in part of the structures of the eye that affects vision in two of them. A fourth child has a defect affecting the pupils that has required surgery to preserve vision. Three more children have strabismus, a squint, severe enough to need surgery. Iris coloboma is also recorded in the medical literature in association with a 13q22q32 deletion, as is underdevelopment of the optic nerve. It has been

suggested that a gene or genes responsible for normal eye development may be deleted (Shanske 2001; Khong 1994; Nichols 1979).

Behaviour

> "She would react strongly to sudden, unexplained changes: when she was 7 or 8, we once moved the furniture and took the pictures down from the walls to prepare for a party and she was very upset because she thought the walls would collapse. But we could always calm her by explaining things to her.

The *Unique* experience suggests that babies are typically happy and good-natured and this easy-going quality stays with some children into adulthood. However, a significant number of children develop behaviour that parents will find challenging and for which they will need expert guidance. Children may bite themselves or use aggression – biting, slapping, hair-pulling – as a means of defence or when thwarted. Two children in the *Unique* series are hyperactive and one adult has trialled treatment (methylphenidate – Ritalin) with mixed success. Four members are considered to be on the autistic spectrum although only one, an adult, has a formal diagnosis of an autistic spectrum disorder. His mother has wise advice for other parents: The features of autism diminish with appropriate management which we worked out for ourselves – a clear, consistent, no-nonsense approach, using praise to remind him of correct behaviours. One adult has in addition a diagnosis of obsessive compulsive disorder and has marked mood
swings (U).

There is very little information in the medical literature on behaviour. However, a boy with a small deletion between 13q32 and 13q33 is reported to have autism and aggression both towards himself and others (Bottani 1991).

Sleep

Most parents report that their children sleep well, but there are exceptions. Early waking and night waking may only be controlled with medicines and two families report what appears to them to be a disturbance in their child's body clock. The mother of one of the adults reports that she has an extended (36-hour) circadian rhythm and this has been confirmed by a sleep study (U).

Independence?

Of the three adults within *Unique* with this chromosome disorder aged between 22 and 25, one was living at home with her mother, one lived independently in her own flat and one lived in a group home with day and night carer support. One attended training workshops in computer skills and art and one was in full-time work, training to be a chef's assistant. None of the three drives a car, but two used public transport confidently alone. All had help with their finances.

The adult living at home spent her time eating, sleeping, watching television and being with her family. The adult in a group home spent his spare time on multi-media activities and his bicycle. He liked to please his carers and to know that his housemates were all right. The adult living in her own flat had a social life like anyone else – she went twice a week to a fitness club and enjoyed cross stitch and television. She has had

a boy friend in the past, would like to get married but is not yet sure if she wants to have children of her own.

What is special about children with this chromosome disorder?

This is what parents told us:

- “Hayley has an incredible memory and ability to read. Her school are constantly amazed. She reads way above her age - Hayley, age 6
- “Joe has helped us to appreciate ‘special’ behaviours. His unjudgmental involvement with us has been special - Joe, age 22
- “Carrie can be very loving and artistic - Carrie, age 23
- “Mie has given us so many happy moments every time she managed to do something new and we have always tried to live out lives as a ‘normal’ family. Her sweet personality makes it easy for people to love her and she knows exactly what to say and to do to wrap them around her little finger. She knows she is different but stands up for herself and has a good life now - Mie, age 26

Does it help to know the exact breakpoints in the chromosome?

Knowing more precisely the breakpoints in the chromosome allows determination of the presence or absence of key genes. However, at present, the value of knowing this is uncertain. Nonetheless, some bands of 13q are known to contain genes for particular conditions.

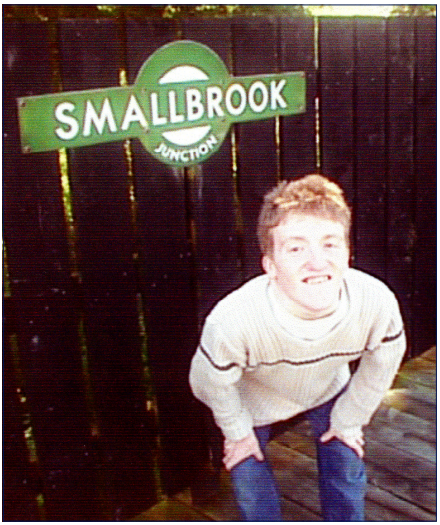
13q22.1q22.3 A gene (endothelin-B receptor gene – *EDNRB*) associated with Hirschsprung’s disease (see page) occurs in 13q22 (Shanske 2001; Sparkes 1984; Lamont 1989; Kiss & Osztovics 1989). Hirschsprung’s has also been associated with a 13q33 deletion (Bottani 1991).

Neighbouring genes may occur in 13q22 that are required for the normal development of the inner ear and the eye (Shanske 2001).

13q32 A small critical region of around 1Mb between D13S136 and D13S147 has been identified (Brown 1995, 1998). Anomalies include: thumb and/or big toe defects and various anogenital malformations.

Deletion of one copy of the *ZIC2* gene within the critical region appears to cause the holoprosencephaly sequence, resulting in incomplete development of the brain into two cerebral hemispheres. The eyes, nose and upper lip are usually also severely affected (Carukashansky 1979; Juberg 1984; Brown, 1993, 1995, 1998; Kuhnle 2000).

13q33.2 and 13q34 Evidence has been put forward for a separate critical region for neural tube defects (Luo 2000).



“He loves any kind of transport!
- 13q22q34 deletion

Mie has a deletion between 13q22 and 13q32. In 2004, when she was 26 years old, she wrote an article for the *Unique* newsletter about her life. Here it is.

A day in my life

by Mie Johansen, Denmark

My name is Mie, I am 26 years old and in February, 2004 I celebrated my first year in my very own apartment. I live in the same town I was born in and only a 30 minutes walk from my parents' house. But I live downtown where all the shops are and right next to the train station. This is very handy especially in the morning, as I have no problem catching my train to go to work. I have a nice kitchen, bathroom, bedroom and a combined dining room and sitting room plus a balcony – all is brand new and was ready to move into last year. I loved it from the first day and my parents sometimes complain that I do not come to visit them often enough.

I have decided to tell you about a Monday – just to start somewhere. My alarm clock goes off at 6am and I head directly for the kitchen to put the kettle on. Then I go to the bathroom for a quick shower. After the shower I set the table for breakfast and make toast or have cereals with milk. I listen to the radio in the morning as they keep me up to date with any delays or problems with the trains. IF the train is not running I can go to work by bus. It takes me an hour to get to work. I brush my teeth after breakfast, as I hate the taste of toothpaste and tea together.

I work as a cook's assistant at a café in the middle of a forest – it is such a cosy, old building with a thatched roof. It used to be an old watermill. We use ecological raw materials and everything we sell has been made from scratch – no fast food here! I have been working here for two years now and have been assigned to different jobs. I have been through baking, vegetables, meat, fish and cleaning. Right now I am the person dealing with customers – I like doing this as long as there is someone there to help me with the bill and giving back the correct amount. I expect to be ready to find a 'real' job later this year and I hope to find something closer to home as I am tired when I come home in the late afternoon.

Monday is also the day I go to the cashpoint to take out an amount of money to do my grocery shopping. My parents have helped me making an arrangement with the bank and I have a fixed amount every week on my card to help me not to overspend. If I need anything more expensive or if I have a dentist or hairdresser's appointment, I tell my parents and they ask the bank to transfer extra money that week. All this is to help me not to spend money on too many 'not necessary things' and it works fine – this way I have almost saved enough money to buy a computer some time this year. All my bills (rent, electricity, cable TV, telephone) are paid directly from another account I have with the bank.

Every second Monday I go to the physiotherapist because I have problems with my back. We discovered my back was not straight when my mother made a dress for me for my confirmation. I have a scoliosis problem and if I get these treatments an operation should not be necessary. Thank God for that – I have had enough operations due to my ear problems. Quite a few actually!!

Around 17:30 in the afternoon, I am back in my apartment ready to prepare my dinner. If I have had a cooked lunch I sometimes just have a couple of sandwiches for dinner – but I mostly cook something nice for myself. Sometimes I need to start my washing machine before I am ready to watch television. Tuesday is the day my social worker comes to visit me and help me with my letters, my cleaning or whatever I decide we should do and sometimes I need to spend an hour making my home look OK. Not that I am a messy person – BUT cleaning is not at the top of my 'to do' list!

This Monday I was alone, but sometimes I invite one of my colleagues home and then we cook together and hang out. Sometimes we rent a video, play a game or just chat.
I try to go to bed around 22:30 to be ready for work the day after.

It's a busy life – but it is **MINE** – and I love it!

Best wishes,
Mie

Two years later, when Mie was 28 years old, her mother write an update for this leaflet.

Mie has now qualified and received her 'green card' showing her ability to work with food and her understanding of the rules of good kitchen hygiene. She now works at her local school, preparing meals for the students. She works part-time, to allow time to have a social life and take care of things around her house.

This summer she will go to a Music festival and work there for a week, cooking and cleaning. This will be the fourth time she has done this with a couple of friends from her old school. They do this as a voluntary contribution. This is half of her summer holiday; the other half she will go with her family and friends to a house in France for two weeks.

Mie is still coping fine with her independent life.

Support and Information

Rare Chromosome Disorder Support Group,
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Tel/Fax: +44(0)1883 723356
info@rarechromo.org | www.rarechromo.org

Join Unique for family links, information and support.

Unique is a charity without government funding, existing entirely on donations and grants. If you can, please make a donation via our website at www.rarechromo.org Please help us to help you!

Chromosome 13 Facebook groups
www.facebook.com/chromosome13
www.facebook.com/groups/chromo13family
also
www.chromosome13deletion.com

Unique mentions other organisations' message boards and websites to help families looking for information. 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. It was compiled by Unique and reviewed by Professor Dorothy Warburton, Professor of Clinical Genetics and Development, Columbia University, New York, US and by Unique's chief medical advisor, Professor Maj Hultén, Professor of Reproductive Genetics, University of Warwick, UK. 2006 (PM)

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