

Wolf-Hirschhorn syndrome



What is Wolf-Hirschhorn syndrome?

Wolf-Hirschhorn Syndrome (WHS) is a rare genetic condition that occurs when a small part of chromosome 4 is missing (deleted). It is thought to affect 1 in 50,000 births with girls twice as likely to be affected as boys. It is likely that some individuals with the condition have never been diagnosed so this figure may be an underestimate [Battaglia GeneReviews® 2002/2015].

What are genes and chromosomes?

Our bodies are made up of trillions of cells. Most of these cells contain a set of around 20,000 different genes that carry the instructions that tell the body how to develop, grow and function.

Genes are made of a complex chemical called DNA and are carried on structures called chromosomes. Chromosomes (and hence genes) usually come in pairs with one member of each chromosome pair inherited

from each parent. A normal cell in the body has 46 chromosomes. 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.

In individuals with WHS part of the short (p) arm (from petit, the French for small) of chromosome 4 is deleted. The size of the deletion can vary but having a missing piece of chromosome 4 means that a number of important genes will be missing, and this can affect the development and intellectual abilities of a child, although there is considerable variability in the range and severity of features that are observed. The reasons for this are complicated and not yet fully understood, but include: the size of the deletion, which will affect the number of genes that are missing; the cumulative effects of the variation in the DNA sequence across a person's whole genome; the outcome of interactions between genetic variants across the genome; and the influence of environmental factors (both internal and external) on the individual's genome [Firth 2018].

When a particular set of developmental features occurs in a recognisable and consistent pattern as a result of a single cause, the condition is called a syndrome, and this is the case for WHS.



Chromosome pairs 1-22

X and Y (male)

chromosome 4 pair circled in red

Most common features of WHS

The most common features associated with WHS are:

- A characteristic facial appearance
- Learning difficulties
- Seizures
- Delayed growth
- Developmental delay

Other features

These features are also found in some people with WHS:

- Spinal curvature/skeletal abnormalities
- A cleft lip or palate
- Dental problems
- A heart condition
- Anomalies of the brain
- Eye conditions, including a squint (strabismus)
- Sleep disorders
- Hearing problems
- Frequent respiratory and ear infections
- Anomalies of the urinary tract
- Skin changes
- Minor anomalies of the genitals in boys

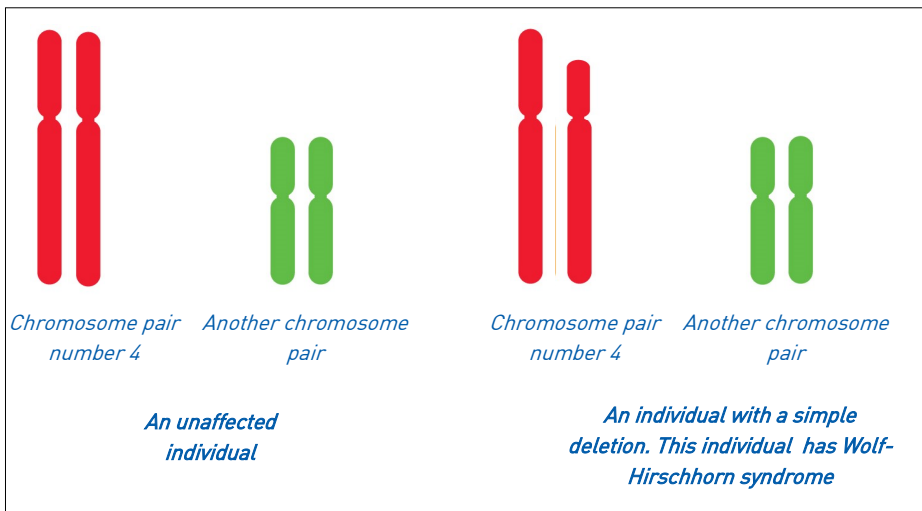
(Battaglia; Unique)

How did it happen?

Chromosome disorders can occur either as a result of rearrangements in one parent's own chromosomes that are passed on to the child or out of the blue (*de novo*), so the child with the chromosome disorder is the first person in the family to be affected.

Approximately 55% of individuals with WHS have a *de novo* (dn) deletion, which arises when during the formation of the egg or sperm from which the child was conceived, part of chromosome 4 is deleted. This is called a

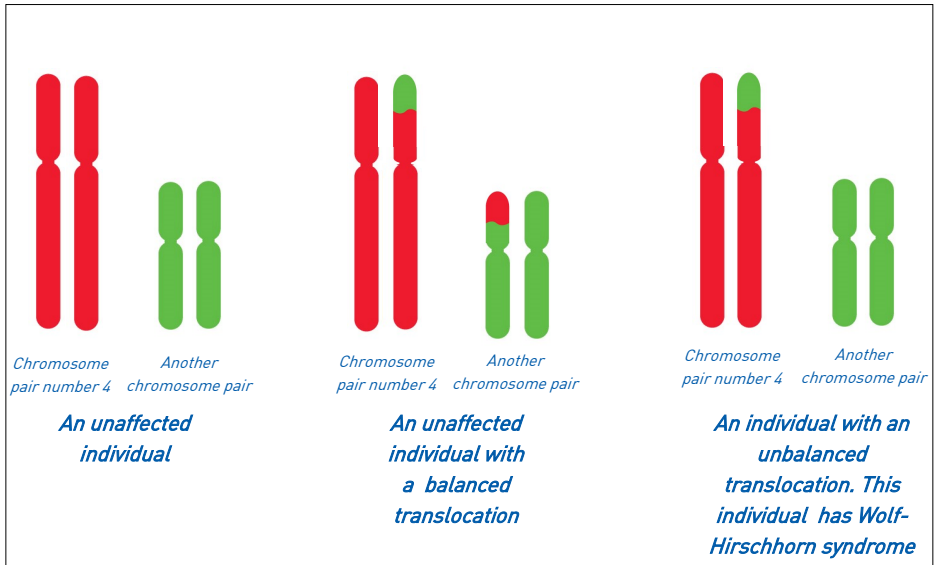
‘simple deletion’ as no other chromosomes are affected. The parents of these children do not have the deletion so the risk of having another child with the condition is very low.



Sometimes chromosomes break and sections of DNA swap places. This means that a section of chromosome 4 might move onto another chromosome. This is called a ‘translocation’. The individual will probably be healthy; they still have all of their genes, so they can make all of the proteins that they need. The only difference is that a group of genes that used to be on chromosome 4 are now on another chromosome. This is called a ‘balanced translocation’ (see *Unique’s* guide to [Balanced Translocations](#)).

However, when we make sperm and egg cells, we only put one copy of each of our chromosome pairs into the sperm or egg cell. This means that the individual with the balanced translocation will probably make some sperm or egg cells that contain the chromosome 4 with the deletion but not the other chromosome involved in the swap that the deleted section moved on to. As a result, a child could inherit a chromosome 4 that has a section of chromosome 4 deleted but too much (a duplication) of part of another chromosome. This is called an ‘unbalanced translocation’. If the “critical region” for WHS is deleted, the child will develop WHS. (They may also have additional features due to the presence of the extra duplicated material from another chromosome - often chromosome 8p (see *Unique’s* guide to [4p 8p translocation](#))). About 45% of patients with WHS have an unbalanced translocation. In some cases, the child inherits an unbalanced translocation

because one of their parents has a balanced translocation. However, in other cases the translocation is *de novo*: it happened due to a mistake during the formation of the sperm or egg cell.



Whatever the reason for a child inheriting WHS, it is important to understand that it was nobody's fault that this happened; nothing either parent did before, during or after pregnancy caused the deletion to happen.

Can it happen again?

To answer this question, it is necessary to examine the parents' chromosomes in order to predict the chances of them having another child with the condition. It is recommended that parents of children with WHS have genetic counselling to discuss the risks to their other children and any future children that they may have.

If the patient has a *de novo* deletion, the change was not inherited from either parent so the chances of having another child with WHS are very low; however, we are not aware of any cases in which recurrence has happened for *de novo* deletions and none have been described in the scientific literature.

If one of the parents has a balanced translocation involving chromosome 4, there is a greater chance that they could have another child with WHS; any future child could inherit an unbalanced translocation. There is also a risk that the parent could pass on a balanced translocation to any other children

that they have. These children will be healthy but are at risk of having a child themselves with WHS. This means that brothers and sisters of patients with WHS may be at risk of having children with WHS if they themselves carry a balanced translocation. Relatives of individuals with WHS can be offered a genetic test to determine if they have a balanced translocation and are therefore at risk of having children with the condition. This test involves giving a small blood sample.

If someone is found to have a balanced translocation involving chromosome 4 and is therefore at risk of having a child with WHS, they may wish to consider prenatal testing or preimplantation genetic diagnosis (PGD). In prenatal testing, a sample of the unborn child's cells can be tested to determine if they have the unbalanced translocation and are likely to develop WHS. PGD uses *in vitro* fertilisation (IVF) techniques: egg cells are extracted from the mother and fertilised in the laboratory using sperm from the father. The resulting embryos can then be genetically tested and only healthy embryos implanted into the mother's womb.

How is Wolf-Hirschhorn syndrome diagnosed?

If a doctor suspects that an individual may have WHS, a genetic test is used to confirm that the patient has the chromosome 4 deletion. Three types of genetic test may be used: cytogenetic analysis, fluorescence *in situ* hybridization (FISH), and genome-wide chromosomal microarray analysis (CMA or arr). In order for the tests to be performed, a sample of blood is taken from which the genetic material can be extracted for testing. These tests may take several weeks. The majority of *Unique* babies were diagnosed at birth or within the first few months (Unique).

Chromosome test results

Depending on the test that was carried out, someone with WHS might have a karyotype that looks like one of these examples:

46,XY,del(4)(p16.3)dn This result shows that the expected number of chromosomes (46) were observed. It also shows that an X and a Y chromosome were found, so this is a boy or a man. **del (4)** means there is a deletion of chromosome 4. **(p16.3)** shows the band in the chromosome that is deleted. The duplication occurred **dn** or *de novo* (as a 'new event'); the parents' chromosomes have been checked and no duplication or other chromosome change has been found on 4p.

46,XX,der(4)t(4;8)(p15.3;p22)mat This shows that there are 46 chromosomes and it's a girl or woman (XX). One chromosome 4 is a 'derivative' chromosome (**der(4)**). That means it has a change in its structure, in this case a piece of chromosome 4 missing, replaced by a piece of chromosome 8. There has been an unbalanced translocation (**t**) involving chromosomes 4 and 8 (**(4;8)**). An extra copy of the short arm of chromosome distal to p22 (e.g. from band 8p22 to the end of the short arm of 8p) is attached to the short arm of chromosome 4 at band p15.3 (**(p15.3;p22)**). The bands from 4p15.3 to the end of chromosome 4p are missing. This means that the girl has a duplication of part of chromosome 8p with a deletion of part of chromosome 4p. The translocation has arisen as a result of a balanced translocation in the mother (**maternal**).

arr 4p16.3(1665362_3135150)x1 This result shows that the analysis used microarray technology (**arr**). The analysis revealed a DNA anomaly involving band **4p16.3**. The DNA anomaly is identified by its base pair numbers (the points where the chromosomal change has occurred). In this example, the DNA anomaly lies between base pairs **1665362** and **3135150** (by taking the first number from the second, you can work out that this is 1,469,788 base pairs, or 1.47 Mb). There is one copy missing of this 1.47Mb section (**x1**; the normal copy number is two) so it is a deletion.

Pregnancy & Birth

Children with WHS grow more slowly than other children. This begins when they are still in their mother's womb; the baby will be much smaller than expected on ultrasound scans. Doctors call this 'intrauterine growth retardation' (IUGR) (Battaglia).

Birth weights were usually low (less than 2.5kg or 5lb 8oz), and a small number of babies are tiny at birth. Two *Unique* babies were delivered by caesarean section, one following a 32-week pregnancy scan that revealed reduced foetal growth (Unique).

Average birth weight (15 babies): 4lb 11oz (2.13kg)

Range: 2lb 1oz (0.94kg) – 8lb 1 oz (3.66kg) (Unique)

Feeding

Babies and children with WHS often have problems with feeding. They may have difficulties sucking or swallowing and food may often 'go down the wrong way'. This can cause chest infections. Children may also suffer from

reflux/vomiting.

Feeding problems might be due to a baby or child having under-developed muscles and, in some, the top lip and/or the roof of the mouth does not develop correctly resulting in a cleft lip and/or palate. Clefting can be corrected by surgery and special feeding devices or techniques, such as sitting babies up after feeds, can be used to help feed children who are struggling.

Babies who are struggling to swallow may be fed for a period of time using a tube that passes through the nose (a nasogastric (NG) tube) or mouth (an orogastric (OG) tube) into the stomach. In some cases, a feeding tube may be inserted through the baby's skin and into their stomach (a gastrostomy). Speech and language therapists can sometimes provide swallowing therapy (Unique).

“ She used to have severe gastro oesophageal reflux disease (GERD) but this has improved greatly at 15 years. A gastrostomy tube was placed at 19 months and removed at 12 years 8 months. She used to have behavioural feeding issues. At 15 years she has failure to thrive and is hypermetabolic. ”

“ He struggled to breastfeed and lost weight. He then had recurrent upper respiratory infections and bad reflux starting at seven weeks when he went on to formula milk. He remains on a high-calorie, mashed diet but is fed orally. ”

“ She has some feeding difficulties at 27 years but has no tube and feeds herself, but needs soft food. She has just started taking iron and vitamin supplements. My advice would be to show support and patience with food; be creative and only give small portions. ”

“ Cleft palate - surgically repaired at five years. ”

Facial appearance

Certain facial features are found more often in children with WHS than “typical” children, including: eyes that are wide apart (hypertelorism), a long forehead; highly-arched eyebrows; a mouth with down-turned corners; a small jaw; and ears that are low-set, under-developed and contain pits or tags. These features do not affect your child's health, but they may mean that you notice similarities between your child and others with WHS. They can also help doctors to identify individuals with WHS. If your child has these characteristic facial features, some may become less noticeable when they

reach puberty.

Delayed growth

Children with WHS grow more slowly than their peers and are usually shorter than other children of the same age. They typically struggle to gain weight even when they are given a balanced diet (Battaglia; Unique).

“ She has always been very small. At six years she weighed ~1st 10lb (10.9kg) and ate purees, although she could tolerate mash and soft sponge with custard and could eat a lot more and faster than when she was younger. At 8 years her growth is still very delayed – she weighs ~1st 13lb. ”

“ Very slow to put on weight. ”

Development (gross & fine motor skills)

Babies and children with WHS are typically delayed in reaching their developmental “milestones”, including rolling, sitting, moving and walking, and benefit from early intervention with occupational therapy and physiotherapy. Most children show slow improvements over time. Almost half of children learn to walk by 12 years, although this will not be possible for all and many will still require support.

Development of hand use and hand-eye coordination (fine motor skills), which are essential for tasks such as holding a bottle, using cutlery, playing with toys and fastening clothes, are also usually delayed, but some children develop the ability to feed themselves, dress, undress and perform simple tasks. A small number learn to control their toilet needs, usually between the ages of 8 and 14 years (Battaglia; Unique).

“ My son has a severe developmental delay. He rolled at one year and crawled at 18 months. As an adult he has trouble walking on uneven ground and has very little road sense. ” – 19 years

“ She has small hands which are curved thus making it difficult to grasp things. Her fine motor skills are not very strong, which makes daily living tasks hard for her. ” – 34 years

“ He has a mild to moderate developmental delay: he is one month behind at just over two years of age. ”

“ He is generally very healthy at 11 months but has a developmental delay. He is not yet holding toys, although he found his hands at ~9 months and will

now hold them together. He rolled from his stomach onto his back at 11 months and is starting to roll from back to stomach at 14 months. At 12 months he started to put his left thumb in his mouth but does not suck it, rather gently bites it. He has physiotherapy and occupational therapy and at 14 months is very playful; kicks legs a lot and swings his arms.”

“ My daughter smiled at two months, rolled at 21 months, sat at two years 6 months, crawled at five years 8 months, and walked at five years 10 months. She pulls herself up to stand from the floor using the sofa, commando crawls and gets everywhere. By 8 years she has started walking using her walker or holding hands and is becoming more mobile. She is lively, alert and very aware of her surroundings. She went to Florida for dolphin therapy at five years and loves hydro-, spa- and sensory-sessions at school. ”

Ability to Learn

Some degree of learning disability is to be expected, usually ranging from moderate to severe (Battaglia; Unique). Children benefit from additional support with their learning, in order to access the curriculum and develop to their full potential. Early intervention can prove beneficial and formal testing to assess specific, individual needs is recommended. In the UK, a tailored education, health and care (EHC) plan can be issued after a child has undergone an EHC needs assessment. This legally-binding document ensures that the educational, health and social provisions deemed necessary to support the child’s needs are delivered to the child (previously, a statement of special educational needs was issued to children with learning difficulties).

“ My son has a severe learning disability. He attends a special school, which is very sensory-based, with a maximum of six in the class with a teacher and two assistants. There are also a nursing team and therapists on-site. He will soon have a home teacher who will prepare him for special pre-school. The Jack and Jill Foundation were a great support providing advice and nursing help (www.jackandjill.ie). ”

“ He attended a special educational needs (SEN) nursery from the age of one and a SEN school from the age of three. He loves school. It is a very important part of his life and has no doubt aided his development. ”

“ My daughter has severe learning difficulties. At 8 years, she is doing really well at school full-time, and has a statement. ”

“ He has severe learning disabilities and does not read or write at 16 years. He is very happy but does not really enjoy academic work. He is quite active and sociable and is functioning at about five to six years of age. He can communicate verbally but will not talk about emotions. He has always attended a special school/college and needs to be supervised continuously as he is not aware of dangers. ”

Speech & Communication

Children with WHS often experience difficulties with speech and communication. Whilst many do not learn any spoken language, they may learn to express themselves through expressions and sounds. These skills often improve with time and children benefit from speech and language therapy to help them to communicate more effectively. Where individuals have no speech or very few words, communication can still be successful through augmentative/alternative communication (AAC) e.g. Makaton, signing, gesture, facial expression, Picture Exchange Communication System (PECS) and iPad communication (Battaglia; Unique).

Medical concerns

■ Seizures

Almost all children with WHS, both in the medical literature and the *Unique* series, experience seizures, with on-set usually between three- and 23-months. Seizures are caused by a change in electrical activity in the brain. Depending on the part(s) of the brain affected symptoms vary, but include temporary confusion, uncontrollable jerking movements, and loss of consciousness or awareness.

Seizures are often triggered by fever and can last over 15 minutes. Some children experience ‘seizure clusters’; they have several seizures within a two-hour period. Some children also have absence seizures, during which the child becomes unresponsive and vacant. Seizures can often be controlled using drugs, as was the case for most children in the *Unique* series, although it is sometimes difficult for doctors to work out the best treatment. If the seizures are treated properly, they often stop as the child gets older; over half of children with WHS stop having seizures at some point between the age of two and 13 years.

Seizures can be distressing to watch. If your child suffers a seizure for the first time, make sure that you remove any dangers in the surrounding area

and call an ambulance. Doctors may want to investigate the seizures further and they may perform an EEG (electroencephalogram) in order to look at electrical activity in the brain; however, most children with WHS show some abnormalities on an EEG (Battaglia; Unique).

“ He had his first seizure at 14 months and by 18 months he had had three prolonged seizures (up to 90 minutes), requiring hospitalisation for the first two. He coped better with his third seizure at 18 months. At seven years he has been seizure-free for 14 months, since his medications were optimised. At seven years he has had his best year so far health-wise. ”

“ He had seizures but only for the first three years. He had recurrent upper respiratory infections (which led to seizures) until he was three; since then he has not had any hospital admissions. ”

“ He was fitting but none since 18 months [at two years]. He had six febrile convulsions in four months prior to three years, up to having his tonsils out. His last febrile convulsion was at six years. ”

■ Low muscle tone (hypotonia)

Many babies and children with WHS appear to be more floppy than others of the same age. This is called hypotonia and can contribute to difficulties with carrying out gross motor skills. Many have underdeveloped muscles, particularly in their legs (Battaglia; Unique).

“ He has hypotonia and at 11 months still does not hold his head up completely on his own. ”

■ Head size & brain

Babies and children with WHS often have a smaller head (microcephaly) than typically-developing children. They can also have unusual findings on brain scans, including four cases among *Unique* members. Some brain regions may appear enlarged whilst others are smaller than expected or have not developed properly. A common finding is a thinning of the ‘corpus callosum’, a part of the brain that connects the left and right sides of the brain, allowing them to communicate. Different specific areas of the brain may be affected in different children. These areas include the cerebral cortex (the brain area responsible for higher thought processes including speech), the cerebellum (the brain area that coordinates movement) and the hippocampus (an area of the brain that is important for memory) (Battaglia; Unique).

■ Heart problems

Approximately half of children with WHS are born with heart problems. The most common problems are septal defects (holes between the left and right sides of the heart) (four cases in the *Unique* series); pulmonary stenosis (a narrowing of the tube that carries blood from the heart to the lungs); patent ductus arteriosus (PDA) (when a tube connecting two blood vessels of the heart fails to close properly at birth); aortic insufficiency (a problem with a heart valve) (one case in the *Unique* series); and a heart murmur (two cases in the *Unique* series). In *Unique's* experience, most of these conditions did not require treatment or resolved naturally over time, but some may require surgery.

If your child has not had their heart checked, you may want to discuss this with your doctor as they may wish to organize further investigations, including an ECHO (an ultrasound scan of the heart) (Battaglia; Unique).

“ Used to have a heart murmur but this has disappeared by 15 years. ”

“ He has a large atrial septal defect (ASD). His heart is being monitored and the hole will be closed when it becomes necessary - so far it is not really affecting him. When a heart murmur was heard at 8 weeks, blood was taken to do genetic testing - he also had bi-lateral talipes and a hypospadias there was concern that there was something that was linking his symptoms. ”

“ Mild heart defect: bicuspid valve without "gradient" and moderate dilatation of ascending aorta. He is to see a cardiac specialist at four years. ”

“ Has an ASD, but it is getting smaller and no surgery is required. ”

■ Hearing

Some babies and children are particularly prone to ear infections and glue ear (Unique) and over 40% of children with WHS have hearing problems (Battaglia). This is often because the inner parts of the ear haven't developed properly. Your doctor may decide to refer your child to a specialist for further investigations and hearing aids may be helpful (Battaglia; Unique).

“ Hearing was OK after grommets [aeration tubes] were fitted and her hearing was normal at five years. Grommets had to be removed due to on-going ear infections, leading to the glue ear coming back and hearing levels dropping so we tried hearing aids again. Left side grommet fitted at seven years but still having ear infections in right ear. Treated labyrinthitis [an inner ear infection] of the right ear under anaesthetic. ”

“ Has a moderate hearing impairment and ear infections, which are treated with a hearing aid and antibiotics. ”

■ Skeletal (bone) anomalies

Two-thirds (60-70%) of children with WHS have skeletal-related problems. These include conditions that affect the spine, such as scoliosis (a sideways curve of the spine) and kyphosis (an outward curve resulting in a hump), and conditions where the hands and feet don't develop correctly, including club foot (the feet turn inwards, with the soles pointing towards each other) and split hand (associated with muscle weakness in the hands). Some children have changes to their ribs; they may have an extra rib or two of their ribs may be joined together. Treatment options include physiotherapy, splinting, plaster casting and surgery in more severe cases (Battaglia; Unique).

“ Her bones are weak, and she has poor muscle tone, which makes activities harder for her. She cannot walk long distances; after about 400-500m her muscles start to hurt. ”

■ Eyes & vision

Up to half of children with WHS have problems with their eyes. These include exotropia, a form of strabismus (squint) where one or both eyes turning outward; excessive tears; drooping eyelids; coloboma, a condition where the eye(s) does not develop normally and there is a gap in part of the structures of the eye; and sight problems. Your child may be referred to an ophthalmologist in order to explore possible solutions.

■ Teeth

Dental problems are common among children with chromosome disorders, including WHS. Teeth often emerge later than would be expected and it is common for baby (milk) teeth to be late to fall out. It is also common for children to have abnormally-shaped teeth or missing teeth (Battaglia; Unique).

“ Eight missing adult (permanent) teeth at 15 years. ”

■ Infections

Children with WHS often experience more infections than other children their age, particularly chest and ear infections. Chest infections may occur if the child vomits and material accidentally enters the lungs. This is called 'aspiration'. Ear infections can be common if the child's ears have not

developed properly. Children may also suffer from a greater number of infections if they have problems with their immune system. If your child is experiencing more infections than you would expect for a child their age, or if the infections are often severe, you may wish to discuss this with your doctor and they may decide to carry out some further investigations; however, it is important to remember that colds and ear infections are common among young children and infections may become less frequent with age and maturity (Battaglia; Unique).

“ He had respiratory problems for the first three years, but they are not much of an issue now. ”

“ She was very susceptible to infections e.g. chicken pox, tonsillitis, chest infections. At six years she has lots of respiratory infections which aggravate her reflux and make her vomit. At 8 years, apart from ear infections, physically she is doing really well, although she still has some upper respiratory problems. ”

■ Urinary tract problems

Some children with WHS have problems with their urinary tract (the kidneys, bladder and tubes connecting them). They may be missing a kidney (renal agenesis) or their kidneys may be in an unusual position or poorly developed. There are also isolated cases of horseshoe kidneys and cysts in one kidney (Battaglia; Unique). Other conditions can include problems with the development of the bladder or abnormalities that result in urine flowing back into the kidneys and children may be susceptible to urinary tract infections (UTIs). Your child may be given tests in order to check their urinary tract.

“ She had small kidneys as a child which caused her to have 18 UTIs. Her last sonogram showed her kidneys to now be the right size for her frame at 15 years. ”

■ Minor genital anomalies in boys

A few boys in the *Unique* series had hypospadias, where the hole normally at the end of the penis is sited on the underside instead. In several cases this was corrected surgically (Unique).

■ Liver

Some children with WHS suffer from hepatic adenomas. This means that they develop benign (non-cancerous) tumours in their liver. These can be

detected by ultrasound and if found may be treated with surgery or chemotherapy. The treatment used is likely to depend on the number and size of the tumours (Battaglia).

■ Skin changes

It is common for children with WHS to have problems with their skin. Some children experience haemangiomas (raised marks on the skin that are usually red). Haemangiomas often increase in size initially but then shrink and may eventually disappear (Battaglia).

Sleep

Many children with WHS experience sleep problems in the early years. In many cases this improves as the child ages. In some children the lack of sleep is due to a medical problem such as ear inflammation, reflux, eczema or sleep apnoea (a condition in which the child briefly stops breathing whilst asleep).

If you feel that a medical problem is stopping your child from sleeping, you may wish to speak to a doctor about possible treatments. It can be extremely challenging for the whole family when a child does not sleep well. *Unique's* "Sleep problems in children with chromosome disorders" guide, in the practical guides for families section of our website, also has further information.

Personality & behavioural concerns

Parents describe their children as happy, sociable and contented. Some children with WHS demonstrate repetitive behaviours, including repeated hand-washing, hand-flapping and rocking or autistic traits. Any concerns should be discussed with a medical professional.

“ A very happy and contented child at 18 months with lots of smiles and laughs. At seven years he is generally a happy boy unless he is sick. ”

“ She gets on well with her younger sister and has learned a lot from her. Very independent at 15 years. She has a great personality, is very sociable and loves music. ”

“ He is very musical and can perfectly tap out a beat. Happiest when watching live music and loves to watch people play instruments and to dance; for that reason he loves all kinds of folk music and dancing. ”

“ He is a contented little boy. He is aware of his surroundings and smiles and laughs. He has intelligent eyes. ”

“ At 19 years, he enjoys the company of adults; fairs; crazy golf; bowling, etc. He is very happy but does not really enjoy academic work. He is quite active and sociable and can communicate verbally but will not talk about emotions. He enjoys watching YouTube and using his tablet to take pictures. ”

“ She is a very happy, contented little girl at four years, and awareness has improved even more since being in school. She likes to sit next to the washing machine and tumble drier to feel the vibrations and hear the noise. Likes to put her arms up to get a cuddle. At 8 years she is still a very happy and affectionate little girl. ”

“ Loves loud music and noises. A very happy child, who smiles and giggles much of the time, roars to get attention and throws herself backwards when annoyed. She hates showers but loves baths. ”

Puberty & Adulthood

The information we have relating to puberty and adulthood is limited. Two *Unique* families shared their experiences:

“ At 34 years, she still needs a lot of input and instant care in her life as she can be very vulnerable; she needs constant supervision in all aspects of her life. Now that she is an adult, she is a bit more confident but can be nervous of new situations and meeting new people. She has learnt to travel independently, can follow simple instructions and has shown good road awareness when out. She is frightened of dogs and will change direction upon meeting one. She loves to learn new things although it takes a long time for her to understand them. She has support during the week to help her gain more confidence in herself while out. She has no knowledge or appreciation of the value of money and therefore has to be supervised. ”

“ He has trouble carrying out personal care and cannot cook or prepare food. He is on high rate PIP for both components and is entitled to a Blue Badge. ”

Outlook

The most significant medical concerns that can affect lifespan are heart anomalies, uncontrollable seizures and repeated chest infections. In the majority of children these conditions can be controlled. Sadly, however, there

are some children who do not reach adulthood due to complications with these medical conditions.

“ The advice I would give to families is that you have to demonstrate great devotion in caring for your child. Care, love and tenderness will ease the way forward. For many years I have devoted my life to helping and supporting my daughter in all aspects of her life. I have sought professional health from people who have an understanding of coping with special needs children and adults. Treat them with love and respect and help them in finding their voice. Give them your time and special tools that will enhance learning. Praise them even when they achieve something small. Have compassion and love when other people stare. Build their confidence with nice clothes and things they like. Support them in their hygiene e.g. washing, dental care, brushing teeth, regular dental check-ups. Take them on outings to places of interest; provide outside help and support if possible. The list goes on but with time everything comes together. Most of all be happy and positive; that will help you and your child to grow and bond, no matter how old they are. ”

Management recommendations

Children with WHS should be closely monitored by a doctor. It may be necessary to measure their growth; blood count and kidney and liver function every year.

- Long-term follow up by a paediatrician
- Monitoring growth (specific growth charts have been developed for 0 to 4 years). Feeding difficulties and reflux are common and may need medical intervention
- An assessment should be made to check for a cleft palate
- An assessment should be made to check for heart conditions
- Vision and hearing should be assessed
- Kidney and liver function should be assessed
- Repeated infections are common and may require further investigation. Urinary tract infections and inner-ear abnormalities should be checked for by a medical professional
- Early input from a speech and language therapist
- Early input from a physiotherapist and/or occupational therapist
- An assessment of special education needs should be made allowing

appropriate provision of schooling

- Referral to clinical genetics for advice about inheritance and future pregnancies

Inform Network Support



Understanding Chromosome & Gene Disorders

Rare Chromosome Disorder Support Group

The Stables, Station Road West, Oxted, Surrey RH8 9EE, United Kingdom

Tel: +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

<http://whs4pminus.co.uk/> - Wolf
Hirschhorn Syndrome Trust (WHST)



This leaflet was made possible by a contribution
from the Sir Halley Stewart Trust



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.

This guide was written by Joanne Mumford, University of Sheffield, UK and Unique (CA) and reviewed by Dr. Alisdair McNeill (Honorary Consultant Clinical Geneticist), University of Sheffield, UK.

Copyright © Unique 2019

Rare Chromosome Disorder Support Group Charity Number 1110661

Registered in England and Wales Company Number 5460413