Unique

Understanding Chromosome & Gene Disorders

Wiedemann-Steiner Syndrome (WSS)

rarechromo.org

What is Wiedemann Steiner syndrome (WSS)?

Wiedemann-Steiner syndrome (WSS) is a rare genetic condition. WSS is named after the two doctors who first described children with features of this syndrome, firstly Dr Hans-Rudolf Wiedemann in Germany in 1989 and then subsequently Dr Carlos Steiner in Brazil in 2000 (Wiedemann 1989, Steiner 2000). The word ‘syndrome’ comes from a Greek word meaning ‘to run together’ and is simply used to describe the combination of features which can be seen together in people with a particular condition.

The main features of WSS are:

- developmental delay
- intellectual disability
- short stature
- behavioural difficulties
- increased body hair (hypertrichosis)

However, WSS is a variable condition and not everyone with WSS will have all of these features. Also, each person with a certain feature won’t be necessarily affected by it to the same level as other people with that feature.

What is the cause of WSS?

In 2012 a group of doctors and scientists carried out some research into the cause of WSS and showed that alterations (called ‘variants’ or ‘mutations’) in a gene called *KMT2A* cause Wiedemann-Steiner syndrome (Jones 2012). *KMT2A* was previously called *MLL.*

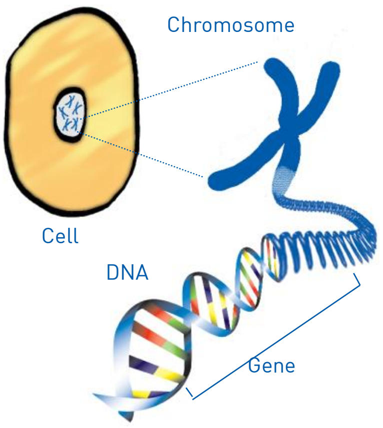
What are genes?

Our bodies are made of building blocks called cells. In most of these cells we store copies of our genes. Genes give our bodies instructions how to grow, develop and function.

Genes are made from a sequence of letters (also known as a ‘code’) called DNA. The DNA code uses the letters G, A, T and C.

DNA sequences are incredibly long, however, they need to fit inside the tiny cells of our bodies. DNA is therefore bundled into packages called chromosomes. We each have 23 pairs of chromosomes and we inherit one copy of each pair from our mother and the other copy from our father. The first 22 pairs of chromosomes are numbered 1 to 22 and are the same in men and women; they are known as autosomes. In addition, we have a pair of sex chromosomes; women have two X chromosomes and men have an X and a Y chromosome.

The *KMT2A* gene is located on chromosome 11, since we each have two copies of chromosome 11 we also usually have two copies of the *KMT2A* gene. Because men and women both have two copies of the *KMT2A* gene, both males and females can be affected by Wiedemann-Steiner syndrome.



Chromosome pairs 1-22, X and Y (male). Chromosome pair 11 is circled in red.

What does the KMT2A protein do?

Each of our genes usually gives our body instructions to make a specific protein. Proteins are large molecules that play important roles in the body. The *KMT2A* gene gives the body instructions to make a protein called KMT2A. The KMT2A protein is an enzyme. Enzymes are molecules that significantly speed up the rate of virtually all the chemical reactions that take place within our cells. They are vital for life and serve a wide range of important functions in the body, such as aiding in digestion and metabolism.

Some enzymes help break large molecules into smaller pieces, other enzymes help bind two molecules together. Enzymes usually work to speed up just one reaction. The KMT2A protein speeds up a reaction that involves the DNA itself. The KMT2A enzyme tells DNA to be ‘open’ (be less tightly wound up) or ‘closed’ (be more tightly wound up) by speeding up (catalysing) a reaction on the proteins that DNA wraps around. This affects the production of other proteins in the body. It is the effect on the other proteins in the body that is thought to cause the features of WSS.

How many people have WSS?

1 in 25,000 to 1 in 40,000 individuals are thought to have Wiedemann-Steiner syndrome across the world. These figures have come from calculations looking at how often we would expect to find alterations in the *KMT2A* gene and not by counting the number of people with the syndrome. However, the number of individuals who have been found to have WSS is much lower than this. This is because not all individuals with WSS will have had genetic testing because it might not be available to them where they live. Also, not all doctors will recognise WSS. However, with the increasing use of broad genetic testing, which looks across all of our genes, it is expected that many more people will be diagnosed with WSS in the future.

Why did this happen?

In most families there is only one affected child and the alteration in *KMT2A* most likely occurred out of the blue (*de novo*), this is no one's fault and there is nothing that happened during pregnancy or early life that caused this. We all have new alterations in our DNA which our parents don’t carry. These alterations happen by chance during the copying process that makes our eggs or sperm or very early on in development. Most people don’t know they have genetic changes since the alterations don’t affect important genes or important pieces of DNA in between our genes. However, if an individual has an alteration in the *KMT2A* gene, they have WSS.

Can it happen again?

Wiedemann-Steiner syndrome most commonly starts for the first time in the affected child, which means that if the parents are not found to carry the *KMT2A* gene alteration, the chance of them having another child with WSS is very low. The reason there is still a small chance is due to something called germline mosaicism, which is where the gene variant can be found in a few eggs or sperm, but is not found in the rest of the body’s cells (and so is not detected during a DNA test of a parent’s blood sample). There are families who have had two children with WSS.

Unique publishes a short general guide to mosaicism that covers this phenomenon.

If a person with WSS and a *KMT2A* alteration has children, the chance of them passing on the condition to each of their children is 1 in 2 (50%). This is known as ‘autosomal dominant’ inheritance (‘autosomal’ because the genetic change is on one of the numbered chromosomes (autosomes) and dominant since WSS occurs when only one gene copy (out of the two copies) is altered).

Each family situation is different and a clinical geneticist or genetic counsellor can give each family specific advice and talk through possible options for future pregnancies. If people with WSS or their parents are planning a pregnancy and would like to discuss their options they should contact their local genetics centre.

Unique publishes separate guides to Planning your next child, Prenatal genetic testing and diagnosis, A clinical genetics appointment and Supporting siblings of children with a rare genetic condition.

What are the features of WSS?

Below we will go through the variable health and behavioural difficulties which have been reported in individuals with WSS. WSS causes a range of features. However, it is important to know that not everyone with WSS will have all of the features; also each person with a certain feature won't necessarily be affected by it to the same level as other people with that feature. We will highlight those features which need special attention and family awareness.

"As Wiedemann-Steiner is a spectrum syndrome, meaning children can be affected mildly, moderately, or severely, it is not easy to diagnose. In addition, even top geneticists who have been practicing in the field for decades may know nothing, or very little about it. As a parent of two non-WSS children and one WSS warrior, I would say trust your instincts when they are very small. Some widely acknowledged features would be a lot of hair (especially on their elbows and backs), a deep sacral dimple, and feeding difficulties from very early on. It is also worth looking for some typical cranio-facial features such as wide flat nasal bridge, slightly pronounced forehead and wide set eyes. In our daughter, the first sign that something was wrong was that she lost weight after birth and vomited multiple times a day. It took a year for me to convince the (very well-meaning and kind) doctors to run genetic testing on her, I knew in my gut that something was wrong and if you have that feeling, keep pushing."

Pregnancy and Birth

The majority of babies with WSS have a normal birth weight and length, however most babies have birth weights below the 25th centile with some having a low birth weight (Baer 2018, Sheppard 2021). Some babies are noted to be floppier at birth than would usually be expected, this is due to low muscle tone (referred to as hypotonia).

"My pregnancy was mostly normal with no anomalies detected in the early scans, however at 38 weeks the sonographer notices that her ureter's were smaller than they should be. This, in itself, is relatively common and whilst specialists were brought in to observe, I was assured that this problem would very likely clear up on it's own (it did not). The birth was fine and although our daughter with Wiedemann-Steiner was the smallest of our three children by some way, she still measured in the normal range at birth (6lb 6oz). I would say it is in the first few months that we noticed things going wrong and more so than just colic, this was a very obvious decline. I would say to parents that in the early days it is really crucial to note everything down, too often medical professionals will look at the parents as failing in some way, or worse, suggest maltreatment of the child. We kept a meticulous log of all feeds, all wet and dirty nappies, all vomits etc. to clearly show the doctors what we were feeding her and what she could manage before inevitably vomiting. If you suspect anything is not right, keep a record."

Feeding

Failure to thrive and feeding difficulties are common in early life in individuals with WSS. Some babies require feeding through a tube that passes through the nose into the stomach (nasogastric tube / NG tube). Some children go on to need feeding by a PEG tube (percutaneous endoscopic gastrostomy tube). Most children with WSS grow out of their feeding difficulties over time.

Unique publishes a separate guide to Feeding.

"Feeding was by far our daughter's biggest hurdle in the first couple of years of life. She started to gain weight successfully after she had her tonsils and adenoids removed. Do push as much as you can, parents, for anything that might make it easier for your little one to gain calories without over-eating to the point of vomiting (not an easy task by any stretch). For us, that was infatrini formula milk prescribed by the NHS and, for a while, thickened fluids. Equally, I think there can be so much pressure on parents to try and do things a certain way, and if that is not working for you or your child, then do whatever does work, we are all just doing the best we can with the situation we find ourselves in and I think any form of feeding is wonderful."

Constipation

Constipation is also commonly seen in children with Wiedemann-Steiner syndrome and many take medication to help with constipation.

Growth

Many individuals with WSS have height and weight in the low normal range or just below the normal range. A small proportion of children with WSS have growth hormone deficiency and, for these children, growth hormone treatment is required.

Development and learning

All people with WSS generally acquire new skills at their own rate over time. Most people with WSS experience speech and motor delay with mild-to-moderate developmental delay and learning disability.

Most children require extra help at school or special schooling by the time they are 12 years old to help them fulfil their potential. The degree of learning disability varies considerably. A small number of individuals with WSS have severe or profound learning disabilities and a few individuals have only mild intellectual disabilities. There are a small number of individuals with WSS who have attended University / College.

Reading skills are highlighted as a strength, and a number of individuals with WSS may have a reading age above their chronological age. However, they may not understand all the words they are reading. Many individuals with WSS have difficulties with mathematics.

The level of abilities of adults with WSS is variable. There are adults with WSS who live and work independently and some of whom have their own children. Other adults with WSS have supported

work placements and live in supported housing. A small number of adults with WSS require full care.

Unique publishes separate guides to Education and Further education, training and work.

"Our daughter is still very young and so I am reticent to comment too much on her development and cannot speak of her learning, as she has not yet started school, however I would say that she has managed to come a really long way. Whilst still significantly delayed, she was able to walk with assistance from the age of 2.5 and now, at age 3, is starting to put words together to form short sentences. We really focused on trying to teach her the key words she would need to articulate desires and dislikes, so that she had more autonomy around her experience of life. We spent time learning yes and no, and used a mix of Makaton and spoken language to help her articulate what she did and did not want. One thing I observed is that, having older siblings around her who treated her "normally" seemed to help a huge amount. In a way, I am glad we did not get her diagnosis immediately as it meant we treated our daughter in the same way we treated our neuro-typical children when it came to things like sleep training and clearly showing her what behaviors were ok and what were not. She definitely learns in a different way to her siblings and some concepts most children find easy, like time (earlier / now / later) still elude her, but she is able to articulate things much more clearly now, so I guess my advice is to persevere where you can but don't rush your child, they will learn what they are capable of and will need patience to live in a world which is fundamentally not designed for them."

Behaviour: strengths and challenges

A study of behaviour in WSS by a team of psychologists showed that being confident in situations with other people (social confidence) was a strength in the majority of individuals with WSS. They also found that the majority of individuals with WSS are affectionate and happy (Yuill unpublished).

However, behavioural difficulties are common and can be severe in WSS and cause significant distress to parents. Behaviour can often be very good at school but very challenging for parents in the evenings. Difficulties with behaviour are often around having difficulties with changing from one task or situation to something else and having a low frustration tolerance, as well as hyperactivity. Some individuals with WSS receive a diagnosis of autism or ADHD. Some individuals have some of the features of autism, but not enough of them for a doctor to make a diagnosis of autism. From our experience, high levels of anxiety are often seen in individuals with WSS and this is not always recognised unless screened for.

Unique publishes separate guides to Challenging Behaviour.

"When fighting for an EHCP for our child, I made an appointment with a neuroscientist at a top children's hospital, to try and see if our daughter "fitted" into the autism spectrum disorder category. My reasoning was that it might be easier to explain her diagnosis as Autistic at school (a well-known disorder) rather than Wiedemann-Steiner (incredibly rare and not at all well-known). As it happens, in our case, she did not fit that description, but more importantly the very kind doctor gave me the advice to try my best not to label my child. This is all very well for some people to say, but as parents of SEN children, we know that the world works with labels, and often in order to receive any real support, a label can be crucial. Over time, however, I came to see more what she meant – our daughter has significant challenges in many areas of her life (terrible attention span, chronic meltdowns, sensory overloads, frustration and sometimes lashing out), and we try our best to rise to meet those needs. In other aspects, she thrives – she is a happy, sociable, loyal and loving child. Each child with WSS is different to the next, as each neuro-typical child is different to the next. Focus only on your child and don't worry too much about the labels."

Sleep disturbance

Sleep disturbance is very common in WSS and is possibly due to a disruption to the circadian rhythm, which is an internal process that normally tells us when to be awake and when to be asleep. Other features of this condition may also impact on sleep, including gastro-oesophageal reflux and behavioural difficulties.

Unique publishes separate guides to Sleep.

Seizures and the brain

Around 20% of individuals with WSS have seizures at some stage of their lives. From our experience, some individuals with seizures may have more significant developmental delay / intellectual disability.

The most common finding on brain imaging, if this is carried out, is an anomaly of the bundle of nerve fibres that connects the two halves of the brain (called the corpus callosum) or the protective layer around nerves (myelin sheath). However, a number of different findings have been reported, including pituitary anomalies (one of the hormone secreting glands in the brain), altered formation of outer layer of part of the brain (cerebral cortex) and Chiari type 1 spectrum malformations, which is where the lower part of the brain pushes down into the spinal canal (Sheppard 2022).

It is important to know that not everyone with WSS will have a Chiari malformation, and even if a person has a Chiari malformation, they won't necessarily have any symptoms as a result of this and if they don't have symptoms they might not need any treatment. Symptoms to look out for and to report to a doctor include: headaches, feeling and being sick, numbness / tingling in arms or legs, muscle weakness and visual disturbance.

Muscles and bones

Some, but not all individuals with WSS can have fusion of the spinal bones in the neck (cervical bone fusion, also known as cervical block vertebrae). This might mean that their neck is slightly shorter than it would be expected to be. This usually doesn't cause any problems, however this is something to look into further if a person has any symptoms in their arms and legs such as tingling or loss of sensation, weakness or if muscles looking wasted / smaller than usual.

Some children and adults with WSS have scoliosis (curvature of the spine) and some have had hip dysplasia, which is sometimes diagnosed later in life. It is common to have a sacral dimple (small pouch at the bottom of the spine) with this syndrome. A small number of individuals have been found to have 'hidden' spina bifida (spina bifida occulta), which is when the spine does not form as expected and a gap forms between vertebrae. Tethered cord, when the spinal cord attaches to tissue around the spine, has also been reported.

In adulthood, some people with WSS have problems with their kneecap bone (patellae) dislocating, requiring surgery.

Immunity and infections

Although a relatively large number of individuals with WSS have frequent infections in childhood, only a small number have a serious underlying problem with their immune system that might need treatment (immune deficiency). The types of immune deficiency reported are a low antibody count (hypogaammaglobulinaemia) and poor global response to the PPSV (pneumococcal polysaccharide) vaccine (Stellaci 2016, Bogaert 2017).

"Our child has had several bouts of pneumonia, one in particular was very severe and she ended up in ICU in a medically induced coma for 2 weeks during which time she had a cardiac arrest. If your child gets recurrent chest infections, I would advise that you tell the doctors as much as possible about Wiedemann-Steiner syndrome including hypotonia (low-muscle development) as this affects the child's ability to cough and clear mucus, especially as an infant. Never ever be afraid to ask for or insist on a second opinion, the worst that could happen is that a doctor's ego is bruised."

Eyes

Children and adults with WSS have a range of eye features; the most common is a squint (strabismus). Some individuals have drooping of the eyelids (ptosis) and some are short or long sighted.

ENT (ear, nose, throat)

Obstructive sleep apnoea, when breathing momentarily stops during sleep, is the most common ENT condition found in people with WSS. Some children require their tonsils and/or adenoids to be removed.

Teeth

Many children have premature eruption of their teeth or premature loss of their primary teeth.

Unique publishes separate guides to Looking after your child's teeth and Teeth: common concerns.

Heart

Around two thirds of babies reported with WSS were born with a healthy heart; around one third were born with a heart problem (congenital heart problem). Some of these heart anomalies may need monitoring but no treatment, some may require an operation. The commonest heart problems seen are patent ductus arteriosus and ventricular septal defect. Heart rhythm abnormalities have also been reported (Sheppard 2022).

Kidneys, genitals and urinary system

Children with WSS usually have normal kidney function but enlarged kidneys (hydronephrosis) and other structural anomalies of the kidneys have been seen as have anomalies of the uterus in females.

Facial features and body hair

Although children and adults with WSS look like their family members, they have some subtle facial features which are similar to other individuals with this genetic condition.

Individuals with WSS also often have increased body hair (hypertrichosis) compared to their family members.

Endocrine / hormonal anomalies

Short stature, or height at the bottom or below the normal range, is common in individuals with WSS. Some children have growth hormone deficiency and treatment with growth hormone is required.

In a small number of individuals, one of the hormone secreting glands in the brain (pituitary) is affected which means their hormone levels need monitoring and hormone replacement might be recommended / beneficial.

Some children with WSS have signs of puberty developing too early, such as early development of pubic and underarm hair (premature adrenarche).

Polycystic ovarian syndrome (PCOS), when cysts develop on ovaries, has been found in women with WSS.

Unique publishes a separate guide to Puberty.

There is NO known link between WSS and increased chance of cancer

Sometimes families worry, when they search for *KMT2A* on the internet, that there might be a link with WSS and an increased chance of developing cancer. To our current knowledge (2024) there is no known significantly increased chance of developing cancer for individuals with WSS, caused by an alteration within the *KMT2A* gene. In a group of 104 individuals, with the oldest in their 40s, none had developed cancer (Sheppard 2022). However, further studies are needed to look into this in more detail in the future.

Is there any treatment for WSS?

There is currently no cure for WSS since the effects of the genetic change take place during each baby's formation and development. However, knowing this diagnosis means that appropriate monitoring and treatment can be put in place as well as educational support to enable each child with WSS to fulfil their potential. Research is ongoing and in the future, there may be specific genetic treatments for some of the features of WSS.

“Our daughter is the happiest person I know, she finds joy in all the little things. Her favourite things in the whole world are having a shower and going in a lift. I think we could all learn a lot from her! She has smiles for everyone and loves with her whole heart. Life is complicated and her many appointments can be challenging but we wouldn't change her for the world. She brightens everyone's day whenever she is around and has made our lives infinitely better. Advocating for her needs has been one of the hardest things I've ever had to do, and I'm forever learning but it has made me into a much stronger and more confident person. Remember that, when speaking to professionals, you are the expert on your child (and probably on their condition as most haven't even heard of it!). Be confident and assertive and keep fighting for what your child needs.'

Management recommendations

Suggested clinical practice guidelines for WSS have been published (Baer and Sheppard 2021, Sheppard 2022).

Co-ordination of care

Children should be under the continued care of a general (hospital) or community paediatrician to assess and monitor their health, growth, development and behaviour. Input from other specialist teams may also be required.

Adults with Wiedemann-Steiner syndrome should undergo an annual check-up with their GP and may be under other specialists too, such as the adult learning disabilities team.

"My biggest tip for parents of children with WSS is, once you have a confirmed diagnosis, try to make sure you always take your child to the same hospital wherever possible and try to make sure that hospital is a big one with a really strong paediatric team and plenty of specialists. I know that may be difficult, and certainly I can attest to the fact that long stints in large NHS hospitals are 0% fun, but over time you may well find that your child has many health complications you were not aware of. As WSS is so unknown, you will have to educate medical professionals and you will have to do it often, sometimes in highly intense situations. It is a huge help to have all prior health data of your child to hand. In an ideal world, we would find paediatricians who can liaise with all the specialists your child will need, but in reality – that person is you. Keeping as much of their medical data as centralized as possible is really helpful."

Feeding and gastrointestinal

Monitoring weight gain in babies is important. A feeding assessment should be carried out and support offered. Some individuals may need to be under the care of a specialist feeding team.

Individuals should be evaluated for constipation.

Growth

Children should have their growth and growth velocity (increase in height over time) monitored during childhood.

If an individual's growth tails off, or if baseline growth hormone testing is abnormal, then referral to an endocrinologist for consideration of growth hormone dynamic stimulation testing should be considered, to look for growth hormone deficiency.

Heart

All individuals should have a heart scan (echocardiogram) to check for structural problems and an ECG (electrocardiogram) to look for heart rhythm disturbance.

Kidneys

All individuals should have a scan of their abdomen (an abdominal ultrasound) to check for structural alterations of the kidneys and bladder. An external pelvic ultrasound could be considered, if appropriate, and at an appropriate time, for example post puberty, to look for anomalies of the uterus.

Sacral dimple

A sacral dimple is a small indentation on the lower back near the crease of the buttocks. Most of these don't need any intervention but they should be looked for and be examined by a doctor. An investigation might be needed to look at the dimple in more detail in some individuals (e.g. by ultrasound).

ENT and sleep apnoea

Enquiry should be made about sleep disturbance and features of sleep apnoea. A sleep study and referral to ENT should be considered if there are concerns. Individuals should be reviewed by a dentist to evaluate for early loss of baby teeth (primary dentition) and early eruption of adult teeth (secondary teeth), as well as other tooth anomalies.

Immunology

All individuals should undergo an immunology assessment at diagnosis. This should include their immune system being assessed as well as their body's response to vaccinations. (This may include measuring antibodies (immunoglobulins), vaccine titres and lymphocyte profile). Individuals should be monitored for signs of frequent infection.

Endocrinology

An endocrinology assessment could be considered, for example, with regards to short stature and growth hormone deficiency.

Neurology

If there are concerns about seizures, referral to a neurologist and an EEG (electroencephalogram) and MRI (magnetic resonance imaging) could be considered.

An MRI brain scan could also be considered if doctors conclude this would be appropriate based on medical history.

Some individuals (but not all) with WSS have been found to have Chiari type 1 malformations. If there are symptoms suggested of a Chiari type 1 malformation causing increased pressure on the brain (this is rare) an MRI brain and spine scan should be considered.

Musculoskeletal

Individuals may benefit from physiotherapy to improve gross motor skills and or occupational therapy to improve fine motor skills.

Individuals should be assessed for hip dysplasia. Not all babies with WSS are born with hip dislocation (congenital dislocation of the hip(s)) but this should be excluded as it is seen in some. It is important to note that hip problems may occasionally be picked up in later childhood or early adulthood. Hip pain should be enquired about and how an individual walks (gait) observed.

Individuals should also be assessed for complications of having cervical vertebral fusion (fused neck bones). This rarely causes any problems at all but if there are any features suggesting nerve compression (for example: tingling or loss of sensation in arms or legs, weakness or muscles looking wasted / smaller than usual), spine imaging (scans) should be considered.

Vision and hearing

Children should have a formal examination by an ophthalmologist and a hearing test.

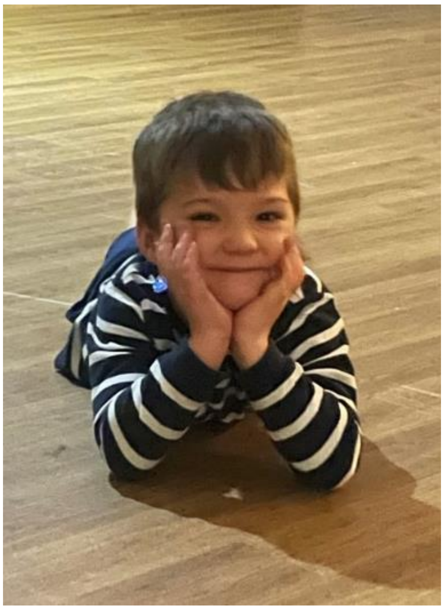
School and learning

Before starting school an assessment of special educational needs (SEN) should be carried out so that extra help can be put into place. Children with WSS may particularly need help with mathematics.

Behaviour and psychiatry

Individuals should be screened for concerns about behaviour, including sleep disturbance, anxiety and features of ADHD or autism. Parents or care givers may also benefit from psychological support in terms of adjusting to a WSS diagnosis and managing challenging behaviour / maintaining their own mental wellbeing when behaviour is challenging.

A psychiatry referral could be considered for adults with WSS.



Menstrual disturbance

Women with WSS and any of the following features should be reviewed by their general practitioner / family doctor with regards to whether they may have polyscytic ovarian syndrome: irregular periods or no periods at all, difficulties conceiving, excessive hair, e.g. on the face, chest or buttocks following puberty, thinning and hair loss from the head and acne.

Dermatology

In terms of increased body hair (hypertrichosis) for many individuals this does not cause them any concern. Some individuals choose to remove some of the additional body hair using conventional hair removal methods.

Genetic Counselling

Genetic counselling should be offered to individuals and their family members to discuss WSS, its inheritance and options for pregnancy.

Sources

The information in this booklet is drawn from the published medical literature and information from Unique members. In 2024, Unique had 37 members with Wiedemann-Steiner syndrome.

Families say …

“Having a child with WSS is very hard at times but also very rewarding. My boy can be so lovely, lots of hugs and funny, and clever at certain moments. The downside is the behaviour challenges which come nearly every evening, the medical problems and so many hospital appointments. The battle to get the right help and support at the right time is the hardest, as WSS is not known by many, so you have to educate medical staff, education staff and local councils. We go through so many emotions as a family but we always get through it and get ready for the next barrier.”

“If you ever doubted your ability as a parent, you will not after you raise a child with WSS, my child has given me the immense gift of knowing 100% that I am a good parent, and you will be too. Having a child with WSS is a huge shock, and it puts a great amount of pressure on you as parents and partners as well as on siblings and other caregivers. Whilst it is helpful to look for general issues you may face, I strongly advise to take it step by step; you can try to plan as much as possible but after a while that becomes exhausting and, as your child grows, so their needs and strengths change and evolve. Deal as best you can with the here and now, and know that you are not alone.”

References

Sheppard SE, Quintero-Rivera F. Wiedemann-Steiner Syndrome. 2022 May 26. In: Adam MP, Feldman J, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024.

Sheppard SE, Campbell IM, Harr MH, et al. Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. Am J Med Genet A. 2021;185(6):1649-1665. doi:10.1002/ajmg.a.62124.

Jones WD, Dafou D, McEntagart M, et al. De novo mutations in MLL cause Wiedemann-Steiner syndrome. Am J Hum Genet. 2012;91: 358-364.

Baer S, Afenjar A, Smol T, Piton A, Gérard B, Alembik Y, et al. WiedemannSteiner syndrome as a major cause of syndromic intellectual disability: a study of 33 French cases. Clin Genet. 2018;94(1):141–52.

Li N, Wang Y, Yang Y, Wang P, Huang H, Xiong S, Sun L, Cheng M, Song C, Cheng X, Ding Y, Chang G, Chen Y, Xu Y, Yu T, Yao RE, Shen Y, Wang X, Wang J. Description of the molecular and phenotypic spectrum of Wiedemann-Steiner syndrome in Chinese patients. Orphanet J Rare Dis. 2018 Oct 11;13(1):178.

Stellacci E, Onesimo R, Bruselles A, Pizzi S, Battaglia D, Leoni C, Zampino G, Tartaglia M. Congenital immunodeficiency in an individual with Wiedemann-Steiner syndrome due to a novel missense mutation in KMT2A. Am J Med Genet A. 2016 Sep;170(9):2389-93. doi: 10.1002/ajmg.a.37681. Epub 2016 Jun 20. PMID: 27320412.

Bogaert, D.J., Dullaers, M., Kuehn, H. et al. Early-onset primary antibody deficiency resembling common variable immunodeficiency challenges the diagnosis of Wiedeman-Steiner and Roifman syndromes. Sci Rep 7, 3702 (2017). https://doi.org/10.1038/s41598-017-02434-4

Stoyle G, Banka S, Langley C, Jones EA, Banerjee I. Growth hormone deficiency as a cause for short stature in Wiedemann-Steiner Syndrome. Endocrinol Diabetes Metab Case Rep. 2018 Aug 23;2018:18-0085. doi: 10.1530/EDM-18-0085. PMID: 30159147; PMCID: PMC6109209.

Yuill, N., Marshall, J, Viner, H. and Camilla Elphick. Wiedemann-Steiner Syndrome: An initial investigation of behaviour and family life.
Unpublished data.

Katada, S., Sassone-Corsi, P. The histone methyltransferase MLL1 permits the oscillation of circadian gene expression. Nat Struct Mol Biol 17, 1414–1421 (2010). https://doi.org/10.1038/nsmb.1961

Inform Network Support

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info@rarechromo.org | www.rarechromo.org

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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://www.rarechromo.org/donate Please help us to help you!

National Wiedermann-Steiner syndrome (UK)

Website: https://nwsswarriors.org.uk/
Facebook: https://www.facebook.com/NWSSWarriors

WSS Foundation (US)

Website: www.wssfoundation.org
Facebook: https://www.facebook.com/wssfoundation
Instagram: https://www.instagram.com/wssfoundation/
Twitter: https://x.com/wssfoundation

WSS Facebook Support group:

Parent Support Group: https://www.facebook.com/groups/667174239987278

Wiedemann -Steiner Syndrome: Info for Families, Friends, and Professionals
https://www.facebook.com/groups/339084866256825

Sleep advice:

https://www.cerebra.org.uk/help-and-information/sleep-service/
https://thesleepcharity.org.uk/
www.contact.org.uk

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.

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