CONDITION

Williams syndrome

Williams syndrome is a lifelong genetic condition affecting development, learning, the heart, and several other body systems.

WHAT IT IS
Lifelong genetic condition affecting development and several body systems

BODY SYSTEM
Heart, brain, kidneys, hormones, hearing, and growth

MAIN SYMPTOMS
Heart narrowing, developmental delay, learning differences, sound sensitivity

WHAT CAUSES IT
Usually a new chromosome 7 gene deletion

HOW IT IS DIAGNOSED
Chromosomal microarray or targeted deletion testing

MAIN TREATMENT
Coordinated medical, developmental, educational, and heart care

CAN IT BE CURED?
No - the chromosome deletion cannot be reversed

IS IT AN EMERGENCY?
Sometimes - fainting, chest pain, or severe breathing trouble

LONG-TERM OUTLOOK
Lifelong needs, often with active and fulfilling adult lives

Overview

Williams syndrome, also called Williams-Beuren syndrome or WMS, is a genetic condition present from birth. It is caused by a small missing section of chromosome 7. This section contains several genes, including one involved in making elastin, a protein that helps blood vessels stretch.

The condition can affect many parts of the body. Common areas include the heart and blood vessels, learning and development, growth, hearing, hormones, kidneys, joints, and teeth. The pattern differs from person to person. Some people need substantial daily support, while others become partly or largely independent as adults.

Williams syndrome is also linked with a distinctive learning and behavioural profile. A person may have strong spoken language or social interest alongside difficulty with numbers,

planning, attention, or understanding where objects are in space. Being outgoing is common, but it is not universal and should not be used to define someone's personality.

Early diagnosis matters because some associated problems, especially narrowing of blood vessels and high blood pressure, may not cause obvious symptoms at first. Regular care can identify these concerns and support development, education, and wellbeing.

Symptoms

Features may become noticeable in infancy or early childhood, although some are not recognised until later. No one has every possible feature.

Babies may have feeding difficulty, slow weight gain, reflux, constipation, low muscle tone, or delayed milestones such as sitting and walking. Some infants have high calcium levels in their blood. This can contribute to irritability, vomiting, constipation, poor feeding, or unusual sleepiness, but these symptoms have many other possible causes.

Heart and blood vessel findings can include:

Developmental delay and learning disability range from mild to more significant. Many people find visual-spatial tasks difficult. These include drawing, puzzles, judging distance, or navigating unfamiliar places. Attention-deficit/hyperactivity disorder, anxiety, specific fears, and sleep problems are also more common.

Sound sensitivity, known as hyperacusis, can make ordinary noises painful or upsetting. Hearing loss may develop as well. Vision concerns can include crossed eyes or difficulty focusing.

Other possible features include short stature, a distinctive facial appearance, dental differences, a hoarse voice, joint stiffness or loose joints, curvature of the spine, urinary problems, thyroid problems, and early puberty. Adults have an increased tendency to develop problems with blood sugar.

- Narrowing above the aortic valve, called supravalvular aortic stenosis
- Narrowing of arteries leading to the lungs
- Narrowing of other arteries, including those serving the kidneys
- High blood pressure

Causes and risk factors

Williams syndrome results from a deletion of genetic material at chromosome region 7q11.23. The deletion usually includes 25 to 27 genes. Loss of the ELN gene, which provides instructions for making elastin, contributes to narrowing and stiffness in some blood vessels.

Most cases are caused by a new genetic change in the egg or sperm, or very early after conception. This is called a de novo deletion. It is not caused by anything a parent did during pregnancy, and no lifestyle or environmental risk factor is known to cause it.

Less often, a parent with Williams syndrome passes the deletion to a child. The condition follows an autosomal dominant inheritance pattern. This means a person with the deletion has a 1 in 2 chance of passing it on in each pregnancy. The features in a child cannot be predicted from how mildly or strongly the parent is affected.

Parents without the deletion usually have a low chance of having another child with Williams syndrome, although the chance is not zero. A genetics professional can explain recurrence risk for a particular family.

Diagnosis

Williams syndrome is confirmed with a genetic blood test that shows the missing chromosome 7 region. Chromosomal microarray is commonly used because it can detect the deletion and show its approximate size. Targeted tests, such as fluorescence in situ hybridisation, or FISH, can also detect the typical deletion.

Testing may be considered when a child has characteristic heart findings, developmental differences, growth concerns, high calcium, or a combination of recognised features. Physical appearance or behaviour alone cannot confirm the condition.

After confirmation, an assessment usually checks which body systems are affected. This may include an examination by a heart specialist, an echocardiogram to create ultrasound pictures of the heart, blood pressure measurements in more than one limb, and blood and urine tests. Hearing, vision, thyroid function, growth, calcium levels, kidneys, feeding, and development may also be assessed.

Diagnostic testing during pregnancy can identify the deletion if chromosome tissue is tested. Screening tests do not confirm Williams syndrome. A genetics specialist can explain the meaning and limits of prenatal results.

Treatment and management

There is no treatment that replaces the missing genes. Care focuses on specific health

concerns, development, education, comfort, and independence.

A heart specialist monitors the heart, arteries, and blood pressure. Some narrowed blood vessels only need observation. More serious narrowing may require surgery or another procedure. High blood pressure may be treated after its cause and the person's full cardiovascular anatomy have been assessed.

Developmental support may include physical therapy, occupational therapy, and speech and language therapy. Early support can help with movement, communication, feeding, self-care, and participation at home or school.

Educational plans often work best when they build on verbal and social strengths while directly supporting visual-spatial skills, mathematics, attention, and planning. Psychological or behavioural care can help with anxiety, fears, attention problems, sleep, or social boundaries.

Other management may include treatment for thyroid problems, hearing or vision care, dental treatment, constipation support, and care for joint, spine, kidney, or urinary concerns. Calcium and vitamin D supplements should not be started or stopped solely because of the diagnosis; decisions should be based on diet, blood tests, bone health, and professional advice.

Anaesthesia and sedation need advance planning. Williams syndrome can increase cardiovascular risk during these procedures, particularly when significant artery narrowing or heart problems are present. The anaesthesia team should know about the diagnosis and review current heart and blood pressure information before a planned procedure.

Complications

Cardiovascular complications are among the most important concerns. Narrow arteries can place extra strain on the heart or reduce blood flow. High blood pressure may be related to stiff arteries or narrowing of the kidney arteries. Rarely, narrowing affecting the coronary arteries, which supply the heart muscle, can contribute to serious events.

High blood calcium is most often recognised in infancy or early childhood. Persistent or marked elevation can affect hydration, the kidneys, feeding, and alertness. Kidney stones or calcium deposits in the kidneys can occur.

Repeated middle-ear problems, progressive hearing loss, dental abnormalities, and vision difficulties may affect communication and learning. Joint stiffness, altered walking, or spinal curvature can affect comfort and mobility over time.

Anxiety can become a major difficulty in adolescence or adulthood. Some people also need support with attention, daily living skills, personal safety, money, employment, or relationships.

These complications are possibilities rather than a prediction. Regular follow-up is intended to find treatable problems before they have a larger effect.

Prevention

Williams syndrome cannot currently be prevented. Most cases happen because of a new chromosome deletion, not because of behaviour, diet, medication use, or an avoidable exposure.

If you or your child has Williams syndrome, genetic counselling can explain inheritance and reproductive testing options. This does not prevent the condition, but it can help with informed family planning.

Medical complications may sometimes be reduced through planned monitoring. Blood pressure checks, cardiovascular care, hearing and vision assessments, and appropriate blood or urine tests help identify changes that may benefit from treatment.

Living with it and what to expect

Williams syndrome is lifelong, but needs change over time. During infancy and childhood, feeding, growth, heart care, movement, communication, and education are often the main priorities. Development usually continues, although milestones may be reached later than expected.

School-age children may become fluent speakers while still having difficulty with abstract language, numbers, writing, visual-spatial tasks, or independent organisation. A person's social confidence can sometimes hide how much support they need to understand complex information or judge risk.

Teenagers and adults may need ongoing help with anxiety, health care, travel, budgeting, employment, and relationships. Some live with family or in supported housing. Others achieve greater independence. Vocational training and practical teaching can support meaningful work and community involvement.

Heart and blood vessel monitoring remains important in adulthood, even if treatment was completed in childhood. Blood pressure, hearing, thyroid function, kidney health, blood sugar, joints, and emotional wellbeing may also need continuing review.

Most changes are managed through routine appointments rather than emergency care. A clear health summary can be useful when changing schools, moving to adult services, travelling, or meeting a new care team.

When to get medical help

Arrange a routine appointment if there are concerns about growth, feeding, constipation, sleep, hearing, vision, learning, anxiety, mobility, or changes in daily skills. New headaches, reduced exercise tolerance, palpitations, or consistently raised blood pressure also deserve medical review.

Seek prompt, same-day advice for repeated vomiting, inability to keep fluids down, much less urination, unusual drowsiness, or marked weakness. These symptoms can have several causes, including dehydration or high calcium.

Call emergency services for:

Before planned surgery, dental sedation, or an imaging test requiring sedation, tell the team about Williams syndrome well in advance. This allows time for cardiovascular and anaesthetic assessment.

- Fainting, especially during exercise
- Chest pain or severe breathing difficulty
- Blue or grey lips or skin
- Sudden weakness, loss of speech, or a seizure
- Collapse or failure to respond normally

Common questions

Is Williams syndrome inherited?

Usually not from a parent. Most people have a new chromosome deletion that occurred around conception. A person who has Williams syndrome can pass the deletion to a child, with a 1 in 2 chance in each pregnancy.

Can Williams syndrome be cured?

No. The missing chromosome material cannot currently be restored. Treatment can address many related health concerns and provide developmental, educational, and emotional support.

What is the life expectancy with Williams syndrome?

There is no single life expectancy that applies to everyone. The outlook is influenced mainly by the type and severity of heart and blood vessel problems and access to ongoing care. Many people live into adulthood, but lifelong cardiovascular monitoring remains important.

Do all people with Williams syndrome have a very friendly personality?

No. Strong social interest and an outgoing manner are common, but personalities differ. Some people are shy or cautious, and many experience anxiety. Apparent friendliness does not mean a person understands every social situation or is safe with strangers.

Can someone with Williams syndrome live independently?

Some adults live partly or largely independently, while others need continuing support. Independence is shaped by learning needs, health, anxiety, practical skills, personal safety, and available services. Skills can continue to develop in adulthood.

Why are ordinary sounds so uncomfortable?

Many people with Williams syndrome have hyperacusis, meaning the brain experiences certain sounds as unusually intense or distressing. Hearing assessment and gradual, individualised coping strategies may help. Constantly avoiding all sound can make daily participation harder.

Sources

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