CONDITION

WBS

WBS is a genetic condition caused by a small chromosome 7 deletion that affects development, the heart, and other body systems.

WHAT IT IS
Genetic condition affecting development and several body systems

BODY SYSTEM
Development, heart and circulation, kidneys, hearing, and hormones

MAIN SYMPTOMS
Developmental delay, sound sensitivity, distinctive facial features, artery narrowing

WHAT CAUSES IT
Missing genes on chromosome 7, usually a new change

HOW IT IS DIAGNOSED
Chromosomal microarray or targeted deletion testing

MAIN TREATMENT
Developmental support and treatment of specific health problems

CAN IT BE CURED?
No - the chromosome deletion is lifelong

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

LONG-TERM OUTLOOK
Many reach adulthood; lifelong health monitoring remains important

Overview

WBS stands for Williams-Beuren syndrome. It is a genetic condition caused by a small missing section of chromosome 7. Chromosomes are packages of DNA that contain the body's genes.

The missing section usually includes several genes. One is the ELN gene, which helps make elastin, a protein that allows blood vessels and other tissues to stretch. Losing this gene contributes to the blood vessel narrowing seen in many people with WBS.

WBS can affect development, learning, behaviour, the heart and blood vessels, growth, hearing, kidneys, and hormone balance. Its effects differ from person to person. Some people have major heart or feeding problems early in life, while others have milder physical concerns but need ongoing learning and emotional support.

WBS is present from birth and lasts throughout life. It is not contagious, and it is not caused by anything someone did during pregnancy.

Symptoms

The signs of WBS may appear at different ages. A baby may first come to medical attention because of a heart murmur, feeding difficulties, slow weight gain, or unusually high calcium levels. In other children, developmental differences lead to testing.

Possible features include:

A common heart problem is supravalvar aortic stenosis . This means the main artery leaving the heart is narrowed just above the aortic valve. Other arteries, including those leading to the lungs or kidneys, can also narrow.

Some babies develop hypercalcaemia , meaning too much calcium in the blood. This may cause poor feeding, vomiting, constipation, irritability, unusual sleepiness, or increased thirst and urination. Not every baby with WBS develops it.

No single behaviour, facial feature, or heart finding confirms WBS. The pattern can suggest the condition, but genetic testing is needed for confirmation.

- Delayed sitting, walking, speech, or other developmental skills
- Mild to moderate intellectual disability or learning difficulties
- Strong social interest and relatively fluent spoken language
- Difficulty with visual-spatial tasks, such as drawing or judging distances
- Anxiety, specific fears, attention problems, or sensitivity to change
- Strong sensitivity to certain sounds, called hyperacusis
- Feeding difficulties, reflux, constipation, or slow growth
- Characteristic facial features that may become more noticeable with age
- Heart murmurs, high blood pressure, or reduced exercise tolerance
- Joint stiffness, low muscle tone, or changes in posture
- Hearing, vision, dental, kidney, or urinary problems

Causes and risk factors

WBS results from a deletion at a location called 7q11.23 on chromosome 7. The deletion

removes one copy of several neighbouring genes.

In most people, the deletion is a new genetic change. It usually happens while an egg or sperm is forming, or very early after conception. The parents generally do not carry the deletion, and ordinary activities, medicines, foods, or events during pregnancy do not cause it.

Less often, a person with WBS passes the chromosome deletion to a child. This follows an autosomal dominant pattern, meaning each pregnancy has a chance of inheriting the altered chromosome. A genetics professional can explain this in relation to a particular family.

WBS affects people of any sex or ethnic background. There are no lifestyle-related risk factors.

Diagnosis

WBS is confirmed with a blood or saliva test that can detect the missing chromosome segment.

A chromosomal microarray looks across all the chromosomes for missing or extra DNA. It can confirm the typical WBS deletion and may show its approximate size. Targeted tests, such as fluorescence in situ hybridisation, may also detect the deletion when WBS is already strongly suspected.

A diagnosis is sometimes made before birth if prenatal chromosome testing identifies the deletion. Ultrasound findings alone cannot confirm WBS.

After confirmation, further assessments help identify which body systems are affected. These commonly include:

These checks do not mean that every listed problem is present. They establish a starting point and help plan follow-up.

- Examination by a heart specialist, with an echocardiogram and electrocardiogram
- Blood pressure measurements in more than one limb
- Blood and urine tests for calcium, kidney function, and thyroid function
- Hearing, vision, dental, growth, and developmental assessments
- Kidney or urinary tract imaging when appropriate

Treatment and management

There is no treatment that replaces the missing chromosome segment. Care focuses on

development, quality of life, and the health concerns found in each person.

Heart and blood vessel narrowing may only need monitoring, or it may require a procedure or surgery. High blood pressure also needs assessment because it may relate to narrowed arteries as well as other causes.

Developmental support can begin in infancy. Depending on the person's needs, this may involve physical therapy, occupational therapy, speech and language therapy, and support with feeding. At school, teaching may build on verbal and social strengths while providing extra help with numbers, handwriting, visual-spatial tasks, attention, and changes in routine.

Anxiety, fears, attention difficulties, and sleep problems can affect daily life. Psychological therapies, environmental adjustments, and sometimes medication may be considered after an individual assessment.

Hearing should be checked over time because sound sensitivity and hearing loss can both occur. Vision, dental health, thyroid function, blood calcium, kidney health, growth, and joint movement may also need ongoing review.

Calcium or vitamin D should not be restricted without medical advice. Unnecessary restriction can harm bones and nutrition. If blood calcium is high, management is based on the test result, symptoms, age, and underlying cause.

WBS can increase the risk associated with sedation and general anaesthesia, particularly when serious heart or artery narrowing is present. Before a planned procedure, make sure the anaesthesia team knows about the WBS diagnosis and has access to current heart information.

Complications

Potential complications are not the same for everyone. Regular follow-up is intended to find treatable problems before they cause lasting harm.

Heart and circulation complications can include worsening artery narrowing, high blood pressure, reduced blood flow to the heart, abnormal heart rhythms, or strain on the heart. Serious cardiac events can occur, which is why lifelong cardiovascular review is important even when someone feels well.

Other possible complications include:

Adults may need support with money, appointments, employment, housing, and personal safety. The level of support can range from occasional help to daily assistance.

- Ongoing feeding, reflux, constipation, or abdominal problems

- Kidney or urinary tract abnormalities
- Hearing loss or recurring middle-ear problems
- Thyroid underactivity
- High blood calcium, especially in infancy
- Early puberty or metabolic problems later in life
- Joint stiffness, spinal curvature, or reduced mobility
- Anxiety that limits education, work, travel, or social activities

Prevention

WBS usually cannot be prevented. The chromosome deletion most often happens as a new genetic change, and there is no known action during pregnancy that prevents it.

After a diagnosis, genetic counselling can clarify whether parental testing is useful and explain the chance of WBS in a future pregnancy. Adults with WBS who are considering pregnancy can also discuss inheritance, prenatal testing, and reproductive options.

Although the genetic condition cannot be prevented, some complications can be reduced or treated through regular heart, blood pressure, hearing, development, and general health checks.

Living with it and what to expect

WBS changes over time, but the chromosome deletion itself does not progress or disappear.

During infancy and early childhood, feeding, growth, calcium levels, heart problems, and developmental delay may receive the most attention. Children continue to learn new skills, although they may reach milestones later and have an uneven learning profile.

Many children are socially motivated and develop useful spoken language. They may still find non-verbal communication, personal boundaries, numbers, planning, or visual-spatial work difficult. Friendly behaviour does not mean a person understands every social situation or is protected from loneliness and exploitation.

Anxiety often becomes more noticeable in later childhood or adolescence. Predictable routines, preparation for changes, and suitable mental health support can help. Sound sensitivity may improve, stay similar, or remain troublesome in particular settings.

The transition to adult services requires planning. Heart and blood pressure monitoring must

continue throughout adulthood. Hearing, thyroid, kidney, dental, digestive, and mental health concerns may also continue or emerge later.

Many adults with WBS take part in education, supported or independent work, relationships, hobbies, and community life. Some live with family or in supported housing, while others manage more independently. Their support needs are shaped by learning ability, physical health, anxiety, and practical life skills.

Life expectancy can be affected by serious heart and blood vessel complications. An individual outlook cannot be predicted from the diagnosis alone, so ongoing cardiovascular care remains important even after a stable childhood.

When to get medical help

Arrange routine follow-up for heart and blood pressure checks, developmental support, hearing assessments, and other monitoring recommended for age and known health concerns.

Contact a healthcare professional promptly for new or worsening symptoms such as reduced exercise tolerance, persistent palpitations, repeated vomiting, unusual thirst or urination, severe constipation, unexplained weight loss, hearing changes, or anxiety that is disrupting daily life.

Seek urgent assessment for persistent vomiting with dehydration, marked sleepiness, or confusion, particularly if high calcium is a known concern.

Call emergency services for:

These symptoms do not always mean a WBS-related complication, but they need immediate assessment.

- Chest pain
- Fainting, collapse, or failure to recover normally after fainting
- Severe or sudden breathing difficulty
- Blue or grey lips or skin
- A sustained very fast or irregular heartbeat with weakness or dizziness

Common questions

Is WBS the same as Williams syndrome?

WBS is the shortened form of Williams-Beuren syndrome. "Williams syndrome" is commonly

used for the same genetic condition.

Can WBS be mild?

Yes. The effects range widely. Some people have less severe developmental or physical concerns, but even a person who appears mildly affected may need lifelong heart and blood pressure monitoring.

Will my child with WBS be able to talk and learn?

Most children develop spoken language and continue learning throughout childhood and adulthood. Development is usually delayed, and the pattern of strengths and difficulties is often uneven. Early therapy and suitable educational support can help a child use their abilities.

Is WBS inherited from a parent?

Usually not. Most cases result from a new chromosome deletion. A parent with WBS can pass the deletion to a child, so genetic counselling can provide family-specific information.

Can someone with WBS live independently?

Some adults live with limited support, while others need regular or daily help. Independence is influenced by learning skills, physical health, anxiety, money management, personal safety, and access to suitable services.

Can people with WBS have surgery or anaesthesia?

Yes, when needed, but planning is important because heart and artery problems can increase anaesthetic risk. The surgical and anaesthesia teams should know about WBS and review up-to-date cardiovascular information before the procedure.

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