CONDITION

WHS

WHS is a rare genetic condition caused by missing material from chromosome 4.

WHAT IT IS
Rare chromosome condition affecting growth and development

BODY SYSTEM
Brain, growth, muscles, and several organs

MAIN SYMPTOMS
Developmental delay, slow growth, low muscle tone, seizures

WHAT CAUSES IT
Missing genetic material from chromosome 4

HOW IT IS DIAGNOSED
Chromosomal microarray and other chromosome tests

MAIN TREATMENT
Supportive care, seizure control, nutrition, and developmental therapies

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

IS IT AN EMERGENCY?
Sometimes - prolonged seizures or breathing problems

LONG-TERM OUTLOOK
Lifelong disability; health and abilities differ widely

Overview

WHS stands for Wolf-Hirschhorn syndrome. It is a rare genetic condition caused by a deletion, or missing piece, from the short arm of chromosome 4. Chromosomes are packages of DNA that carry the instructions used for growth and development.

The missing section can include several genes. This affects how the brain and body develop before and after birth. The size and location of the deletion help shape the condition, but they do not predict every feature or ability.

WHS commonly affects growth, muscle strength, learning, movement, and communication. Seizures are also common. Some people have differences involving the heart, kidneys, bones, eyes, hearing, immune system, or digestive system.

The condition is present from conception and lasts throughout life. It is not contagious, and it is

not caused by anything a parent did during pregnancy. Early medical care and developmental support can help manage symptoms, build skills, and improve comfort and participation in daily life.

Symptoms

WHS affects each person differently. Signs may be noticed during pregnancy, around birth, or as development is monitored during infancy.

Common features include:

Facial features may include widely spaced eyes, a broad or prominent area between the eyebrows, a short groove between the nose and upper lip, and a small lower jaw. These features can be subtle and may change with age. Facial appearance alone cannot confirm WHS.

Seizures often begin in infancy or early childhood. They may be triggered by fever in some children. Seizure type and frequency differ, and some people have more than one type.

Feeding problems may result from weak sucking, poor coordination of swallowing, reflux, or low muscle tone. Some children need extra nutritional support. A feeding tube may be recommended when eating by mouth does not provide enough nutrition or is unsafe.

Other possible features include heart or kidney differences, cleft lip or palate, dental problems, curvature of the spine, hearing loss, eye problems, undescended testicles, and repeated infections. Not everyone develops these problems.

- Slow growth before and after birth
- A small head size, called microcephaly
- Low muscle tone, meaning the muscles feel less firm
- Delayed movement, speech, learning, and self-care skills
- Intellectual disability
- Seizures
- Feeding difficulties or poor weight gain
- Distinctive facial features

Causes and risk factors

WHS results from missing genetic material near the end of chromosome 4, in an area called 4p. The condition is sometimes described as a 4p deletion syndrome.

Most cases occur because of a new chromosome change in the egg, sperm, or early embryo. This is called a de novo change. It usually happens by chance and is not related to a parent's health, age, behaviour, or pregnancy choices.

In some families, the deletion is connected to a balanced chromosome rearrangement in a parent. A balanced rearrangement means chromosome material has changed position but none is missing or extra in that parent. The parent is usually healthy, but a pregnancy may inherit an unbalanced arrangement with missing or extra genetic material.

The chance of WHS happening again is usually low when the deletion is new. It can be higher if a parent has a balanced rearrangement or certain other chromosome changes. Genetic testing of both biological parents can clarify the family pattern. A genetics professional can explain the results and what they may mean for future pregnancies.

Diagnosis

WHS is confirmed with genetic testing. A chromosomal microarray is commonly used because it can find missing or extra pieces of DNA and show approximately how large the deletion is.

Other chromosome tests may also be needed. A karyotype provides a broad picture of the chromosomes and may identify a translocation, which is a rearrangement between chromosomes. Fluorescence in situ hybridisation, or FISH, uses a targeted probe to look for a particular chromosome region. The choice of test depends on what has already been found and whether a rearrangement is suspected.

Testing the biological parents can show whether the chromosome change was inherited or occurred for the first time in the child.

During pregnancy, ultrasound may show slow growth or structural differences, but these findings are not specific to WHS. Screening tests may suggest a chromosome concern, but screening does not confirm the diagnosis. Diagnostic prenatal testing uses cells collected through chorionic villus sampling or amniocentesis for chromosome analysis.

After diagnosis, health assessments are used to identify features that need treatment. These may include an electroencephalogram, or EEG, to measure the brain's electrical activity; heart and kidney imaging; hearing and vision tests; feeding assessment; and evaluation of growth and development.

Treatment and management

There is no treatment that can replace the missing chromosome material. Care focuses on managing symptoms, preventing avoidable complications, and supporting development and quality of life.

Seizures are treated with antiseizure medicines chosen according to seizure type and the person's health. Some people need more than one medicine. Families may also receive a written seizure plan explaining what to do during a seizure and when rescue medicine or emergency care is needed.

Feeding support may include help with positioning, food texture, swallowing, reflux, and calorie intake. A dietitian and feeding or speech therapist may be involved. Tube feeding can support nutrition when swallowing is unsafe or growth remains poor.

Physical therapy can support posture, strength, sitting, standing, and mobility. Occupational therapy can help with hand use, daily activities, equipment, and sensory needs. Speech and language therapy may support speech, understanding, gestures, communication boards, or electronic communication devices.

Treatment may also address:

Follow-up is usually shared among primary care, genetics, neurology, developmental services, and other specialists based on the person's needs. Care plans should consider comfort, communication, family priorities, education, and participation in community life.

- Heart, kidney, eye, hearing, dental, or bone problems
- Repeated or unusually severe infections
- Sleep difficulties
- Constipation or reflux
- Cleft lip or palate
- Mobility, seating, and home accessibility needs

Complications

Seizures can sometimes last a long time or occur repeatedly without full recovery. This is called status epilepticus and requires urgent treatment. Poorly controlled seizures may also affect safety, sleep, and learning.

Low muscle tone and swallowing difficulties can allow food or liquid to enter the airway. This is called aspiration. It can contribute to coughing, choking, or chest infections. Feeding

difficulties can also lead to dehydration or inadequate nutrition.

Some people have congenital heart defects, meaning heart differences present from birth. Kidney differences may affect kidney function or increase the chance of urinary problems. Curvature of the spine and other bone or joint problems can affect posture and mobility.

Repeated infections may occur, particularly in younger children. An immune system assessment may be considered when infections are frequent, severe, or unusual.

WHS can be associated with serious health problems, especially in infancy and early childhood. Seizures, infections, aspiration, and major organ differences can affect survival. However, many people live into adolescence and adulthood. An individual outlook cannot be predicted from the diagnosis alone.

Prevention

WHS usually cannot be prevented because most chromosome deletions happen by chance at conception.

When a family has already had a pregnancy or child with WHS, chromosome testing and genetic counselling can help explain the chance of recurrence. If a parent carries a balanced chromosome rearrangement, options may include diagnostic testing during pregnancy or embryo testing with in vitro fertilisation. These are personal reproductive choices, not guarantees that a chromosome condition can always be avoided.

Nothing done during pregnancy causes the chromosome deletion.

Living with it and what to expect

WHS is lifelong. Development usually continues, but at a slower pace than expected for age. Skills may emerge over years rather than months. Some people learn to sit, walk, use words, or communicate in other ways. Others need extensive help with movement, communication, eating, and personal care.

Growth often remains below standard growth-chart ranges. Being small does not by itself show that treatment is failing. Clinicians look at the person's growth pattern, nutrition, swallowing safety, and general health rather than one measurement.

Seizures may change with age. They may become easier to control for some people, while others continue to need long-term treatment. Medicines should not be stopped suddenly unless directed by the prescribing clinician.

Most people need ongoing developmental, educational, and medical support. School plans may include adapted learning, communication tools, mobility equipment, nursing support, or help with feeding. During adolescence, planning gradually shifts toward adult medical care, decision-making support, daily living arrangements, and community services.

Regular follow-up is used to review seizures, nutrition, growth, hearing, vision, movement, bones, dental health, and any known heart or kidney concerns. New symptoms should be assessed rather than assumed to be part of WHS.

Families may find the level of care demanding, particularly during periods of frequent seizures, illness, or feeding difficulty. Respite care, social services, disability supports, and rare-condition organisations can help with practical and emotional needs.

When to get medical help

Arrange routine medical follow-up if there are changes in seizure pattern, feeding, growth, sleep, movement, hearing, vision, or behaviour. Increasing coughing during meals, repeated chest infections, constipation, pain, or loss of previously used skills also deserves assessment.

Contact the usual healthcare team promptly if seizures become more frequent, medicines are difficult to give, feeding takes much longer than usual, or there are signs of dehydration such as much less urine, a dry mouth, or unusual sleepiness.

Seek emergency help for:

Follow the person's seizure emergency plan when one is available. Do not place anything in the mouth during a seizure. Move nearby hazards away, protect the head, and turn the person onto their side when possible.

- A seizure lasting five minutes or longer, unless the seizure plan says otherwise
- Repeated seizures without returning to the usual level of awareness
- A first known seizure
- Trouble breathing, blue or grey lips, or severe choking
- Sudden unresponsiveness or difficulty waking
- A serious injury during a seizure

Common questions

What does WHS stand for?

WHS stands for Wolf-Hirschhorn syndrome. The name refers to the clinicians who first described the pattern of chromosome changes and physical features.

Is WHS inherited?

Usually not. Most cases result from a new chromosome deletion. A smaller number are related to a balanced chromosome rearrangement in a parent. Parental chromosome testing is needed to tell the difference.

Can a child with WHS learn to walk or talk?

Some children learn to walk and use spoken words, often later than other children. Others use wheelchairs, gestures, signs, picture boards, or electronic devices. The chromosome result cannot precisely predict an individual's future abilities.

How long can someone with WHS live?

Some children have life-threatening complications, particularly early in life. Many people survive into adolescence and adulthood. Outlook is influenced by seizure control, infections, swallowing safety, and heart or other organ differences.

Can WHS be detected before birth?

Yes. Diagnostic testing of placental or amniotic-fluid cells can detect the chromosome deletion. Ultrasound and prenatal screening may raise concern, but they do not confirm WHS on their own.

Does the size of the deletion predict how severe WHS will be?

Larger deletions often involve more genes and may be associated with more complex needs. However, people with similar deletions can have different symptoms and abilities. Medical assessments and development over time provide more useful individual information.

Sources

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