CONDITION

Hypercalcemia-supravalvar aortic stenosis

A genetic condition that can cause high blood calcium and narrowing above the heart's aortic valve.

WHAT IT IS
Genetic condition combining high calcium and aortic narrowing

BODY SYSTEM
Heart and circulation, kidneys, growth, and nervous system

MAIN SYMPTOMS
Feeding problems, poor growth, constipation, heart murmur

WHAT CAUSES IT
Usually a new deletion on chromosome 7

HOW IT IS DIAGNOSED
Blood calcium, echocardiogram, and chromosome microarray

MAIN TREATMENT
Calcium control, heart monitoring, and surgery when needed

CAN IT BE CURED?
No - the genetic change remains lifelong

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

LONG-TERM OUTLOOK
Many reach adulthood with lifelong multidisciplinary follow-up

Overview

Hypercalcemia-supravalvar aortic stenosis is a descriptive name for a rare genetic condition involving high blood calcium and narrowing of the aorta. The aorta is the main artery carrying blood from the heart to the body. "Supravalvar" means the narrowing is just above the aortic valve.

This combination is usually part of Williams syndrome , caused by a small missing section of chromosome 7. Not everyone with Williams syndrome develops high calcium or supravalvar aortic stenosis. The features and their severity can differ greatly from one person to another.

High calcium, called hypercalcemia , is most common during infancy or early childhood. It may affect feeding, growth, digestion, hydration, and kidney function. In some children it causes no clear symptoms and is found through a blood test.

Supravalvar aortic stenosis makes it harder for the heart to pump blood through the aorta. Mild narrowing may only need monitoring. More serious narrowing can strain the heart or reduce blood flow and may require surgery.

The chromosome change can also affect development, learning, growth, blood pressure, kidney health, hearing, and emotional well-being. Care therefore usually involves more than heart and calcium checks alone.

Symptoms

Symptoms depend on which features are present and how severe they are. A baby with high calcium may have:

These symptoms are not specific to hypercalcemia. Blood testing is needed to find out whether calcium is high.

Supravalvar aortic stenosis often causes a heart murmur, which is an extra sound heard through a stethoscope. Mild narrowing may cause no outward symptoms. More significant narrowing can cause shortness of breath, tiredness during activity, chest discomfort, dizziness, or fainting.

Some people also have narrowing in other arteries, including the lung arteries, kidney arteries, or coronary arteries that supply the heart muscle. High blood pressure may develop even when someone feels well.

Other possible features include short stature, sensitive hearing, dental differences, joint stiffness, kidney or bladder problems, and delays in speech or movement. The typical learning pattern may include strong social language alongside difficulty with visual-spatial tasks, such as judging distance or assembling shapes. Anxiety and attention problems can also occur.

No single symptom confirms this condition.

- Poor feeding or difficulty gaining weight
- Vomiting, constipation, or unusual irritability
- Excessive thirst or frequent urination
- Sleepiness, weakness, or reduced activity

Causes and risk factors

The usual cause is a deletion at 7q11.23 , meaning a small piece of chromosome 7 is missing.

This region contains several genes. Loss of one called ELN , which provides instructions for making the elastic protein elastin, is strongly linked to narrowing of the aorta and other arteries.

The reason high calcium develops is less clear. The body may absorb or process calcium and vitamin D differently, particularly during infancy. High calcium may be temporary, but calcium in the urine can remain high or return later.

Most chromosome deletions happen as a new genetic event in the egg or sperm. Nothing a parent did during pregnancy caused the deletion.

The condition can be passed on. A person with the chromosome deletion has a 1 in 2 chance of passing it to each child. This probability is the same in every pregnancy. When neither parent has the deletion, the chance of it happening again in another pregnancy is usually low, though not zero. A genetics professional can explain the risk for a particular family.

Eating calcium does not cause the chromosome change. However, excessive calcium or vitamin D supplements may worsen hypercalcemia in someone who is susceptible.

Diagnosis

Diagnosis considers the calcium problem, the cardiovascular findings, and genetic testing.

Blood tests measure total calcium or ionized calcium, the form that is active in the body. Results may be interpreted alongside albumin, a blood protein that affects total calcium measurements. Other tests may check kidney function, phosphorus, parathyroid hormone, and vitamin D.

A urine test can show whether the kidneys are releasing too much calcium. Kidney ultrasound may be used to look for stones or nephrocalcinosis , which means calcium deposits within the kidneys.

An echocardiogram , or heart ultrasound, shows the area above the aortic valve and measures how blood moves through it. An electrocardiogram records the heart's electrical activity. Blood pressure is often checked in both arms and sometimes a leg because artery narrowing can affect the readings. CT or MRI scans may be needed for a more detailed view of the aorta, coronary arteries, or other blood vessels.

A chromosome microarray can confirm the typical 7q11.23 deletion. Other targeted genetic tests may also detect it. If this deletion is not found, the medical team may investigate separate causes of high calcium and aortic narrowing.

Developmental, hearing, vision, kidney, thyroid, and dental assessments help identify other

support needs. Not every test is required at every visit or for every person.

Treatment and management

Treatment is matched to the calcium level, symptoms, heart findings, age, and kidney health.

Mild hypercalcemia may only require repeat blood and urine tests. Adequate fluid intake is important because dehydration can raise calcium further. A clinician or dietitian may recommend temporarily adjusting calcium or vitamin D intake, but long-term restriction should not be started without guidance. Children still need enough nutrients for bone development and growth.

More serious hypercalcemia may require treatment in hospital. This can include fluids given into a vein and medicines that lower calcium. The choice depends on the cause and severity rather than on the condition's name alone.

Supravalvar aortic stenosis is monitored by a cardiologist. Follow-up may include examinations, blood pressure measurements, echocardiograms, and other heart imaging. If narrowing is severe, is getting worse, causes symptoms, or places significant strain on the heart, surgery may be recommended to widen the narrowed area. Surgery can repair the narrowing, but it does not remove the underlying chromosome change.

Any procedure requiring sedation or a general anaesthetic needs careful planning. Some people with Williams syndrome have coronary artery narrowing or other cardiovascular features that increase anaesthetic risk. The anaesthesia team should know about the diagnosis and have access to an up-to-date heart assessment.

Management may also involve:

- Support for feeding, speech, movement, learning, or anxiety
- Monitoring of growth, thyroid function, hearing, and vision
- Kidney checks when calcium remains high in blood or urine
- Treatment of high blood pressure
- Dental and nutritional care suited to the person's needs

Complications

Persistent high calcium can cause dehydration, kidney stones, or calcium deposits in the kidneys. Severe hypercalcemia can affect alertness, heart rhythm, and kidney function,

although this is less common.

Ongoing aortic narrowing can make the heart's main pumping chamber work harder. Over time, its muscular wall may thicken. Some people also develop narrowing of the coronary, kidney, or lung arteries. High blood pressure can occur and may need lifelong treatment.

Cardiovascular problems can sometimes be serious even without obvious symptoms. This is why planned follow-up remains important after childhood and after any heart surgery.

Developmental or learning differences may affect school, work, and independent living. Anxiety, sensitivity to sound, and attention difficulties can also affect daily life. Appropriate educational and psychological support can reduce their impact.

Prevention

The chromosome deletion that usually causes this condition cannot currently be prevented through diet, exercise, or pregnancy choices.

Complications can sometimes be prevented or identified early through regular calcium, kidney, blood pressure, and heart monitoring. Avoiding unneeded high-dose calcium or vitamin D supplements may reduce the chance of worsening hypercalcemia. Prescribed supplements should not be stopped without medical advice.

Genetic counselling can explain inheritance, testing options, and what a result may mean for relatives or future pregnancies. It does not pressure families toward any particular decision.

Living with it and what to expect

High calcium often becomes less of a problem as a child grows, but this is not guaranteed. Calcium in the blood or urine may remain abnormal or rise again during illness, dehydration, or changes in supplement use. Periodic monitoring may continue even after earlier results return to normal.

Aortic and other artery narrowing generally does not disappear on its own. Mild narrowing may remain stable for years. Other narrowing progresses and eventually needs repair. Even after successful surgery, cardiology follow-up continues because narrowing, high blood pressure, or other blood vessel problems can develop elsewhere.

Development tends to move forward, though skills may develop unevenly. Early support can help with speech, movement, learning, and everyday tasks. Needs often change during school years, adolescence, and the move into adult services.

Many people take part in education, relationships, work, and community life. The level of practical support needed in adulthood differs. Some live with considerable independence, while others need regular help with health care, finances, transport, or daily routines.

A written care summary can be useful. It may include the genetic diagnosis, heart anatomy, operations, recent calcium results, medicines, and anaesthesia considerations.

When to get medical help

Arrange routine follow-up as advised for heart checks, blood pressure, calcium, kidney health, growth, and development. Contact the usual care team sooner if exercise tolerance declines, constipation persists, thirst or urination increases, or feeding and growth become more difficult.

Seek same-day medical advice for repeated vomiting, signs of dehydration, unusual sleepiness, marked weakness, confusion, or a clear reduction in urination. In a baby, poor feeding with fewer wet nappies also needs prompt assessment.

Seek emergency care for:

These symptoms do not always mean a severe complication is present, but they need urgent assessment.

- Fainting, especially during physical activity
- New or severe chest pain
- Serious trouble breathing
- Blue or grey lips or skin
- Collapse, a seizure, or difficulty waking
- A very fast, very slow, or irregular heartbeat with weakness or dizziness

Common questions

Is this the same as Williams syndrome?

The combination is most often associated with Williams syndrome, caused by a 7q11.23 chromosome deletion. The descriptive name focuses on high calcium and aortic narrowing, while Williams syndrome includes a wider pattern of cardiovascular, developmental, and physical features. Genetic testing can clarify the diagnosis.

Will the calcium level stay high forever?

Not necessarily. Hypercalcemia is most common in infancy and often improves with age. High urine calcium may last longer, and blood calcium can occasionally rise again. Follow-up is based on previous results, symptoms, kidney findings, and age.

Does supravalvar aortic stenosis always need surgery?

No. Mild narrowing may only require regular cardiology monitoring. Surgery is considered when narrowing is severe, progressing, causing symptoms, or placing too much strain on the heart. The decision also considers the coronary arteries and other blood vessels.

Should calcium and vitamin D be avoided?

Do not remove them from the diet without professional guidance. They are important for bones, teeth, and growth. Unnecessary high-dose supplements may be harmful when calcium is already elevated, but excessive restriction can also cause problems. A clinician or dietitian can advise based on test results.

Can it be inherited?

Yes. Most cases begin as a new chromosome deletion, so the parents do not have the condition. Once someone has the deletion, there is a 1 in 2 chance of passing it on in each pregnancy.

Can someone with this condition exercise?

Many people can be active, but safe activity depends on the severity and location of artery narrowing, blood pressure, symptoms, and any previous surgery. A cardiologist can advise whether particular competitive, high-intensity, or heavy-resistance activities should be limited.

Sources

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