CONDITION

Fahr disease

Fahr disease is a rare condition involving calcium deposits in brain areas that control movement, thinking, and behavior.

WHAT IT IS
Rare condition causing calcium deposits in deep brain areas

BODY SYSTEM
Nervous system

MAIN SYMPTOMS
Movement problems, balance trouble, thinking changes, mood changes

WHAT CAUSES IT
Usually inherited gene changes or an unknown cause

HOW IT IS DIAGNOSED
Brain CT, blood tests, sometimes genetic testing

MAIN TREATMENT
Symptom-based medicines, rehabilitation, and mental health support

CAN IT BE CURED?
No - existing brain deposits cannot currently be removed

IS IT AN EMERGENCY?
Sometimes - first seizure, stroke signs, or severe confusion

LONG-TERM OUTLOOK
Often slowly progressive, but severity is unpredictable

Overview

Fahr disease is a rare condition in which calcium and other minerals collect in certain parts of the brain. The deposits usually affect the basal ganglia, which are deep brain structures involved in movement. Other areas that control balance, thinking, mood, and behavior may also be affected.

The preferred medical name is primary familial brain calcification , or PFBC, when the condition is inherited or has no other identifiable cause. The historical name "Fahr disease" is still widely used.

Brain calcification does not affect everyone in the same way. Some people have no symptoms and discover it during a scan performed for another reason. Others develop movement problems, changes in thinking or mood, seizures, or difficulty with everyday tasks. The amount

of calcification on a scan does not reliably predict how severe symptoms will be.

It is important to distinguish primary brain calcification from deposits caused by another medical problem. Calcification related to a hormone disorder, infection, toxin, or another condition is sometimes called Fahr syndrome or secondary brain calcification. Finding and treating an underlying cause may influence care.

Symptoms

Symptoms can begin at almost any age, although they often appear in adulthood. Even members of the same family may develop different symptoms or remain unaffected for many years.

Movement symptoms may include:

Some people experience changes in memory, attention, planning, or judgment. These may be mild, or they may eventually interfere with work, finances, medication management, or other daily activities.

Mood and behavior can also change. Possible symptoms include depression, anxiety, irritability, loss of motivation, hallucinations, or other symptoms of psychosis. Psychosis means losing some contact with reality, such as hearing voices or developing strongly held beliefs that are not based in fact.

Other possible symptoms include headaches, seizures, urinary problems, sleep disturbance, and difficulty speaking or swallowing. None of these symptoms is unique to Fahr disease, and many have more common causes.

Some people with extensive calcification have few or no symptoms. Others have significant symptoms despite less striking changes on a scan.

- Slowness, stiffness, or shaking similar to Parkinson's symptoms
- Poor balance, unsteady walking, or frequent falls
- Muscle spasms or twisting movements, known as dystonia
- Jerky, dance-like movements, known as chorea
- Poor coordination, tremor, or unclear speech

Causes and risk factors

In primary familial brain calcification, the deposits are linked to changes in genes involved in

how brain cells and small blood vessels manage minerals and chemical signals. Several genes are known to cause the condition, but genetic testing does not find a cause in every affected person.

Some forms follow an autosomal dominant inheritance pattern. This means a person needs one altered copy of a gene to inherit the condition. Other, less common forms are autosomal recessive, meaning both copies of a gene are altered.

Inheriting a gene change does not predict exactly when symptoms will begin or how severe they will become. Some people with a disease-causing gene change never develop noticeable symptoms.

A family history of brain calcification, unexplained movement problems, or similar neurological symptoms increases the likelihood of an inherited form. However, the condition can be identified in someone without a known family history.

Secondary brain calcification can result from other medical problems. Important examples include disorders of the parathyroid glands, which control calcium and phosphate levels, as well as certain genetic, mitochondrial, infectious, or toxic conditions. These are not the same as primary familial brain calcification.

The deposits are not thought to result from eating calcium-rich foods or taking in a typical amount of dietary calcium.

Diagnosis

A non-contrast CT scan of the head is the main imaging test used to detect brain calcification. CT usually shows calcium more clearly than MRI. The location, pattern, and symmetry of the deposits help with interpretation.

Calcification alone does not confirm primary familial brain calcification. Small deposits can occur with aging, and several other conditions can produce a similar scan. Your medical history, symptoms, family history, examination, and blood test results all matter.

Blood tests commonly assess calcium, phosphate, magnesium, parathyroid hormone, vitamin D, and kidney function. Further testing may be guided by your age, medical history, scan findings, and possible exposure to infection or toxins.

Genetic testing may identify a disease-causing gene change. A positive result can support the diagnosis and clarify how the condition may run in a family. A negative result does not rule it out because not every genetic cause is known or detectable.

Genetic counseling can help you understand what a result means for you and your relatives.

Testing relatives is usually considered after the family's specific gene change has been identified, rather than using scans alone without discussion of the possible consequences.

Treatment and management

There is currently no treatment proven to remove the brain deposits or stop primary familial brain calcification from progressing. Care focuses on the symptoms that affect you and on maintaining safety and independence.

Treatment may include medicines for movement symptoms, seizures, depression, anxiety, psychosis, sleep problems, or pain. Responses differ, and some people may be sensitive to side effects. Regular medication review is useful, especially if balance, alertness, or swallowing changes.

Physical therapy can support strength, walking, balance, and fall prevention. Occupational therapy can suggest ways to make work, self-care, and home activities easier. Walking aids or home modifications may improve safety when needed.

Speech and language therapy can help with communication and swallowing. If swallowing becomes difficult, an assessment can identify safer food textures, eating techniques, or other forms of nutrition support.

Mental health care is an important part of treatment when mood, behavior, or perception changes. Support may include talking therapies, practical help, and carefully monitored medication.

If calcification is secondary to another condition, treatment targets that cause. For example, correcting a parathyroid or mineral disorder may prevent further complications. Existing deposits may remain even after the underlying problem is treated.

Complications

Possible complications depend on which symptoms develop. Movement and balance problems may lead to falls or injuries. Swallowing problems can cause weight loss, dehydration, choking, or aspiration, in which food or liquid enters the airway.

Cognitive or psychiatric symptoms may affect relationships, employment, financial decisions, and independent living. Seizures may cause injuries and can affect driving eligibility.

These complications are not inevitable. Early rehabilitation, treatment of specific symptoms, and practical support can reduce their impact.

Prevention

Inherited primary familial brain calcification cannot currently be prevented. Healthy eating, exercise, and avoiding tobacco support general health, but they have not been shown to stop these brain deposits.

Secondary calcification may sometimes be limited by identifying and treating the underlying medical condition. This is one reason blood tests are important when brain calcification is first found.

If a disease-causing gene change is present in your family, genetic counseling can explain inheritance and family-planning options. This does not prevent the condition in someone who already carries the gene change.

Living with it and what to expect

The long-term course is difficult to predict. Some people remain free of symptoms or have mild, stable problems for many years. Others gradually develop more difficulty with movement, thinking, communication, or daily activities. Symptoms do not always progress at the same rate.

Routine follow-up is usually guided by your symptoms. Appointments may review walking, falls, mood, memory, seizures, swallowing, daily function, and medication side effects. Repeated CT scans are not always helpful because changes on imaging may not match changes in symptoms.

You may need increasing support over time, but significant disability is not certain. Many people continue working, caring for family, exercising, and taking part in social activities, especially when symptoms are mild or well managed.

Planning ahead can help if symptoms begin to affect complex decisions. This may include discussing workplace adjustments, transport, financial arrangements, or future care preferences. Family and caregiver support may also need to change as symptoms change.

When to get medical help

Arrange a routine medical appointment if you develop gradual changes in balance, movement, memory, mood, speech, or coordination. You should also discuss an incidental finding of brain calcification, particularly if close relatives have similar scan findings or neurological symptoms.

Seek prompt medical advice for increasing falls, new hallucinations, worsening swallowing, unexplained weight loss, medication side effects, or a clear decline in daily function.

Call emergency services for:

These symptoms may have causes unrelated to Fahr disease, but they need urgent assessment.

- A first seizure, repeated seizures, or failure to recover normally afterward
- Sudden facial weakness, arm weakness, speech difficulty, or other stroke signs
- Sudden severe confusion, collapse, or loss of consciousness
- Choking with difficulty breathing
- Thoughts or actions suggesting an immediate risk of self-harm or harm to others

Common questions

Is Fahr disease the same as Fahr syndrome?

The terms have been used inconsistently. Fahr disease usually refers to primary familial brain calcification caused by an inherited gene change or without another identified cause. Fahr syndrome often refers to similar calcification caused by another medical condition. Some sources use the names interchangeably.

Does brain calcification mean I have too much calcium in my diet?

Usually not. Primary familial brain calcification involves changes in how minerals and signals are handled around brain cells and small blood vessels. It is not known to be caused by eating dairy products or other calcium-rich foods. Do not restrict calcium unless a healthcare professional has identified a separate reason.

Will Fahr disease cause dementia?

It can cause problems with memory, attention, planning, or judgment, but dementia is not inevitable. Some people have no cognitive symptoms, while others develop gradual impairment. A cognitive assessment can help identify the type and extent of any changes.

Can the calcium deposits be removed or reversed?

There is no established treatment that removes the deposits in primary familial brain calcification. When another medical condition is responsible, treating it may correct abnormal mineral levels and reduce further harm, although existing deposits may remain.

Should my family members have genetic testing?

Testing may be useful when a disease-causing gene change has already been identified in the

family. Because a result can affect health planning, insurance, and family decisions, genetic counseling is recommended before testing relatives who do not have symptoms.

Can I drive with Fahr disease?

A diagnosis alone does not always prevent driving. Seizures, slowed reactions, movement problems, impaired vision, or cognitive changes may affect safety and legal eligibility. Driving rules differ by location, and new seizures or significant neurological symptoms should be reported according to local requirements.

Sources

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