CONDITION

Mccune-albright syndrome

McCune-Albright syndrome is a lifelong mosaic condition that can affect bones, skin, and hormone-producing organs.

WHAT IT IS
Mosaic condition causing abnormal bone and hormone activity

BODY SYSTEM
Bones, skin, and hormone-producing organs

MAIN SYMPTOMS
Bone pain or fractures, early puberty, café-au-lait patches

WHAT CAUSES IT
Random GNAS gene change after conception

HOW IT IS DIAGNOSED
Clinical features, hormone tests, bone imaging, sometimes genetic testing

MAIN TREATMENT
Hormone control, bone care, pain relief, and selective surgery

CAN IT BE CURED?
No - mosaic gene change remains lifelong

IS IT AN EMERGENCY?
Sometimes - sudden vision loss or suspected fracture

LONG-TERM OUTLOOK
Often manageable, though bone and hormone problems may continue

Overview

McCune-Albright syndrome is a lifelong condition that may affect the bones, skin, and hormone-producing organs. It is also called fibrous dysplasia-McCune-Albright syndrome, or FD/MAS. Morgagni-Turner-Albright syndrome is a less commonly used historical name.

The condition develops when a gene change occurs by chance shortly after conception. Only some cells carry the change, creating a pattern called mosaicism . The affected areas and symptoms can therefore differ greatly from one person to another.

The three best-known features are:

You do not need to have all three features. Some people have one affected bone and few other problems. Others have several affected bones and more than one hormone disorder.

McCune-Albright syndrome matters because fragile or misshapen bones can affect movement, while excess hormones can influence growth, puberty, metabolism, and heart health. Regular monitoring helps identify changes before they cause avoidable complications.

- Fibrous dysplasia, in which abnormal fibrous tissue replaces healthy bone
- Café-au-lait patches, which are flat areas of darker skin
- Early puberty caused by hormone production beginning too soon

Symptoms

Symptoms often begin during childhood, but mild cases may not be recognised until later. Which symptoms appear depends on where the affected cells are located.

Bone symptoms can include ongoing bone pain, repeated fractures, limping, or a difference in limb length. Affected bones may bend as a child grows. When the spine is involved, scoliosis-an abnormal sideways curve-may develop.

Fibrous dysplasia can affect one bone or many bones. The thigh bone, pelvis, ribs, skull, face, and upper arm are common locations. Skull or facial involvement may cause uneven facial shape, headaches, dental problems, hearing changes, or, less commonly, changes in vision.

Café-au-lait patches are usually present from birth or appear soon afterwards. They are light-to-dark brown and often have irregular borders. They do not usually cause discomfort or become cancerous.

Early puberty is particularly common in children with ovaries. Repeated ovarian cysts can release oestrogen, causing breast development or vaginal bleeding at an unusually young age. These episodes may come and go. Children with testes may develop testicular abnormalities, although obvious early puberty is less common.

Other hormone-related symptoms may include:

Rarely, babies develop excess cortisol, a condition called Cushing syndrome. This can cause poor growth, swelling, high blood pressure, or serious illness and requires specialist care.

- Rapid growth or unusually tall stature from excess growth hormone
- A fast heartbeat, sweating, tremor, or weight loss from an overactive thyroid
- Weak bones or muscle weakness from low blood phosphate
- Slowed growth after initially rapid puberty
- Enlarged hands, feet, or facial features when growth hormone remains high

Causes and risk factors

McCune-Albright syndrome is caused by a change in the GNAS gene. This gene helps control signals that tell cells when to produce hormones and how bone tissue should develop. The altered gene can leave these signals switched on.

The change happens randomly after an embryo has started developing. It is not caused by anything a parent did before or during pregnancy.

McCune-Albright syndrome is not usually inherited. A parent with the condition is not expected to pass it to a child because the gene change is typically absent from egg and sperm cells. There are no established lifestyle, environmental, or parental risk factors.

Because the change occurs after conception, some tissues carry it and others do not. An early change may affect more parts of the body, while a later change may involve only a small area. This helps explain why the condition has such a broad range of effects.

Diagnosis

Diagnosis is based on the pattern of bone, skin, and hormone findings. Having café-au-lait patches alone does not confirm the condition, and not everyone has the classic combination of symptoms.

X-rays can show the characteristic appearance of fibrous dysplasia. A bone scan or other whole-body imaging may be used to identify which bones are affected. CT or MRI scans can provide more detail when the skull, face, spine, or a complicated bone lesion is involved.

Blood and urine tests check hormone activity and bone health. Testing may include thyroid hormones, growth hormone markers, phosphate, vitamin D, and puberty-related hormones. Results are chosen according to your age and symptoms.

A bone biopsy, in which a small tissue sample is examined, is sometimes needed when imaging is not typical or another condition must be excluded. It is not always necessary.

Genetic testing can find a GNAS change and support the diagnosis. Testing affected tissue is more likely to detect it than a blood test. A negative blood or tissue result does not rule out McCune-Albright syndrome because the tested sample may not contain affected cells.

Diagnosis may involve an endocrinologist, who treats hormone conditions, and an orthopaedic or bone specialist. Dental, hearing, vision, and craniofacial assessments may be needed when the skull or face is affected.

Treatment and management

There is no treatment that removes the underlying gene change. Care focuses on controlling excess hormone activity, protecting bone function, relieving symptoms, and monitoring affected organs.

Hormone treatment is matched to the specific problem. Medicines may reduce the effects of early puberty, control an overactive thyroid, or lower excess growth hormone. Low phosphate may require specialist treatment to improve bone mineral strength. These treatments need regular blood tests because hormone activity can change over time.

Bone pain may be managed with suitable pain relief, physiotherapy, and activity adjustments. Some people with persistent moderate-to-severe bone pain are offered intravenous bisphosphonates, medicines that slow bone breakdown. They may reduce pain in selected people but do not cure fibrous dysplasia or reliably make lesions disappear.

Surgery may be considered to repair a fracture, stabilise a weak bone, correct a significant deformity, or relieve pressure on nearby structures. Operations on fibrous dysplasia can be complex because affected bone may regrow or continue to change.

Craniofacial fibrous dysplasia is often observed when it is stable and not affecting function. Surgery to open the optic canal-the bony passage around the vision nerve-is not routinely performed when vision is normal. New visual symptoms need prompt assessment.

Regular follow-up may include:

Café-au-lait patches do not require medical treatment. Cosmetic options can be discussed if their appearance is distressing.

- Growth, puberty, thyroid, and other hormone checks
- Blood tests for phosphate and bone health
- Review of new pain, fractures, posture, and mobility
- Hearing, vision, or dental checks when relevant
- Imaging based on known bone involvement and changing symptoms

Complications

Fibrous dysplasia can weaken bone, leading to fractures, progressive bending, arthritis near affected joints, or reduced mobility. Spinal involvement may cause worsening scoliosis. Severe deformity is not inevitable, but growing children need monitoring because bones can change as they lengthen.

Disease in the skull or face can affect tooth position, hearing, sinus drainage, or facial symmetry. Loss of vision is uncommon, but it is an important complication when bone or a cyst presses on the optic nerve.

Untreated hormone excess can create further problems. Early puberty may reduce adult height because bones mature too quickly. An overactive thyroid can affect heart rhythm and bone strength. Excess growth hormone can increase skull and facial bone growth. Low phosphate can cause bone pain, weakness, and poorly mineralised bone.

Ovarian cysts may cause pelvic pain or irregular bleeding. Testicular abnormalities are often painless but may require monitoring. Fertility is possible, although hormone problems can make menstrual cycles or conception less predictable for some people.

Cancer arising in fibrous dysplasia or another affected tissue is uncommon. Risk may be higher after radiation treatment, so radiotherapy is generally avoided in affected bone when another option is available. A rapidly enlarging area, new swelling, or a major change in pain needs medical review, but these symptoms do not automatically mean cancer.

Prevention

McCune-Albright syndrome cannot be prevented. The GNAS change occurs randomly after conception and is not linked to a known action, exposure, diet, or inherited family trait.

Complications can sometimes be reduced through regular hormone monitoring and timely treatment of weak or deformed bones. Physical activity is usually encouraged, but it may need adapting when a weight-bearing bone has a high fracture risk. A physiotherapist or bone specialist can help identify safer ways to remain active.

Vitamin D, calcium, or phosphate supplements should not be started specifically for this condition without advice based on blood tests. In particular, phosphate treatment requires specialist monitoring.

Living with it and what to expect

McCune-Albright syndrome follows a different course in each person because the gene change affects different tissues. New areas of fibrous dysplasia generally do not spread from one bone to another. However, existing lesions can enlarge during growth or become more active when certain hormone levels are high.

Bone lesions often become less active after skeletal maturity, but they do not disappear and may continue to cause pain, deformity, or fractures in adulthood. Hormone conditions can also

develop or change over time, so follow-up may continue even when you feel well.

Many people attend school, work, exercise, form relationships, and have children. Some need repeated operations, mobility support, hearing aids, dental care, or long-term hormone treatment. Severe widespread bone or hormone involvement can have a greater effect on daily life.

Pain and visible differences can affect sleep, mood, confidence, and social life. Rehabilitation, psychological support, school or workplace adjustments, and connection with a rare-condition support community may help.

Pregnancy is possible, but hormone and bone symptoms should be reviewed before and during pregnancy. Pregnancy does not affect everyone in the same way, so care is planned around the person's known bone lesions and hormone conditions.

When to get medical help

Arrange routine follow-up as recommended by your specialist team, even if symptoms are stable. Make a non-urgent appointment if you notice increasing bone pain, a new limp, gradual facial change, altered periods, signs of early puberty, or symptoms of thyroid or growth hormone excess.

Seek prompt medical assessment for:

Seek emergency care for sudden loss or major blurring of vision, a visibly deformed limb, or severe pain with an inability to stand or use the affected limb after an injury. These symptoms may indicate pressure on the optic nerve or a fracture.

If you have known adrenal insufficiency following treatment for cortisol excess, follow your emergency plan for vomiting, severe weakness, confusion, collapse, or inability to take prescribed steroid medicine.

- Sudden or rapidly worsening bone pain
- New swelling over an affected bone
- A new hearing change, severe headache, or facial numbness
- Persistent fast heartbeat, shaking, sweating, or unexplained weight loss
- New weakness or worsening difficulty walking
- Vaginal bleeding or breast development in a young child

Common questions

Is McCune-Albright syndrome inherited?

Usually not. The GNAS change happens randomly after conception and affects only some cells. It is generally not present throughout the body or passed from parent to child.

Can McCune-Albright syndrome be cured?

No current treatment removes the mosaic gene change. Individual hormone problems can often be controlled, and fractures or deformities can be treated. Long-term monitoring remains important.

Does everyone with McCune-Albright syndrome have early puberty?

No. Early puberty is common, especially in children with ovaries, but some people never develop it. Others mainly have fibrous dysplasia, skin patches, or a different hormone problem.

Does fibrous dysplasia keep spreading?

It does not spread between bones like an infection or cancer. The affected bones were determined during early development. Existing lesions can grow or change, particularly during childhood or with uncontrolled hormone excess.

Can I exercise with McCune-Albright syndrome?

Many people can and should remain active. The safest activities depend on which bones are affected, whether there has been a fracture, and how strong weight-bearing bones are. Low-impact exercise and tailored strengthening may be useful when fracture risk is higher.

What is the life expectancy with McCune-Albright syndrome?

Many people have a normal or near-normal lifespan, particularly when hormone problems and bone complications are monitored and treated. Rare severe complications involving widespread bone disease, major hormone excess, or infancy can affect the outlook.

Sources

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