

# Acrodysostosis

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## Keywords

Disease name / synonyms

Definition / Clinical description

Radiological features

Etiology

Prevalence

Genetic counseling

Antenatal diagnosis

Management including treatment

Unresolved questions

References

## Abstract

*Acrodysostosis is an extremely rare disorder characterized by abnormally short and malformed bones of the hands and feet (peripheral dysostosis), nasal hypoplasia and mental retardation. Other findings include progressive growth delays, short stature, unusual head and characteristic facies. Occasionally, abnormal interpedicular spinal spaces, increased mandibular angle and hearing loss have been observed. Widespread epiphyseal stippling and metacarpophalangeal pattern profile could be useful as diagnostic tools. The aetiology and the prevalence are unknown. Many reported cases are sporadic; some are familial and are transmitted in autosomal dominant manner. Prenatal diagnosis relies on high-resolution real-time ultrasonography. There is no treatment or specific preventive measures. Consultation with an orthopedist and podiatrist as well as learning support should be offered.*

## Keywords

Peripheral dysostosis, epiphyseal stippling, nasal hypoplasia, mental retardation

## Disease name / synonyms

Acrodysplasia;

Arkless-Graham syndrome;

Maroteaux-Malamut syndrome;

Peripheral dysostosis-nasal hypoplasia-mental retardation (PNM) syndrome.

## Definition / Clinical description

Acrodysostosis (OMIM 101800) is an extremely rare disorder characterized by abnormally short and malformed bones of the hands and feet (peripheral dysostosis), nasal hypoplasia and mental retardation. Other findings may include progressive growth delays, short stature, unusual head and characteristic facies. Occasionally abnormal interpedicular spinal

spaces, increased mandibular angle and hearing loss have been observed.

Affected infants exhibit premature maturation of bones of the hands and feet, malformation and shortening of the forearm bones (radius and ulna) near the wrist, abnormally short fingers and toes (brachydactyly).

Characteristic facial features may include a flattened, underdeveloped (hypoplastic) "pug" nose, an underdeveloped upper jaw bone (maxillary hypoplasia), mouth malocclusion (open mouth), widely spaced eyes (ocular hypertelorism), and/or an extra fold of skin on either side of the nose that may cover the eyes' inner corners (epicanthal folds). Abnormalities of the skin appendages (broad and short nails) can occur.

### Radiological features

The radiological features are brachycephaly, hypoplasia of the mid face, prognathism and delayed dentition. The hand and feet abnormalities are shortening of the metacarpals, metatarsals, phalanges and cone-shaped epiphyses. Widespread epiphyseal stippling as well as metacarpophalangeal pattern profile could be useful as diagnostic tools in patients suspected to have acrodysostosis (1, 2). Irregularity of the vertebral end plates may also be observed. Varying degrees of spinal stenosis are characteristic of acrodysostosis (3).

### Etiology

It remains unknown. It may be difficult to distinguish acrodysostosis from pseudohypoparathyroidism (Albright hereditary osteodystrophy; OMIM 103580). In contrast to pseudohypoparathyroidism, patients with acrodysostosis had normal bioactivity of the alpha subunit of the Gs protein, indicating that acrodysostosis has a different pathogenesis from pseudohypoparathyroidism (3, 4).

### Prevalence

Acrodysostosis is an extremely rare disorder. The exact prevalence is unknown. Overall incidence of skeletal dysplasias is approximately 1 case per 4000-5000 births.

### Genetic counseling

Many reported cases are sporadic but some are familial and are transmitted as an autosomal dominant trait, although no gene has yet been identified. The disorder tends to occur with older parental age (2, 5) and affects children of both sexes.

### Antenatal diagnosis

Prenatal diagnosis relies on high-resolution real-time ultrasonography of the fetus. Prenatal diagnosis may be made in babies whose mother has the condition, but routine screening is not done.

### Management including treatment

There is no treatment or cure for acrodysostosis, neither specific preventive measures. Consultation with an orthopedist and podiatrist may prevent progression of skeletal problems by appropriate braces, shoes and early surgery. Appropriate support, especially for learning disabilities, is important.

### Unresolved questions

The molecular basis of this syndrome remains to be determined.

### References

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