

Desbuquois syndrome

Authors: Doctors Laurence Faivre¹ and Valérie Cormier-Daire

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¹Service de génétique, CHU Hôpital d'Enfants, 10 Boulevard Maréchal de Lattre de Tassigny BP 77908, 21079 Dijon Cedex, France. laurence.favre@chu-dijon.fr

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Abstract

Desbuquois syndrome is a rare autosomal recessive osteochondrodysplasia first described by Desbuquois in 1966. It is characterized 1) clinically, by short stature of prenatal onset with rhizomelic and mesomelic shortness, marked joint laxity, kyphoscoliosis and characteristic facial dysmorphism, and 2) radiologically, by Swedish key or monkey wrench appearance of the femoral heads and advanced carpal bone age. Radial deviation of the second finger with a supernumerary ossicle at the base of the second phalanx is a radiological hallmark of this condition but is not essential. The disease is a clinically heterogeneous with variable severity. A gene for the disease has been mapped to chromosome 17q25.3 in the subgroup of patients with typical hand abnormalities only. The main differential diagnosis is Larsen syndrome in the La Réunion Island form.

Keywords

Osteochondrodysplasia, micromelic dwarfism, advanced carpotarsal ossification, 17q25.3 locus, autosomal recessive inheritance

Disease name and synonyms

- Desbuquois syndrome
- Desbuquois dysplasia
- Micromelic dwarfism with vertebral and metaphyseal abnormalities and advanced carpotarsal ossification

Prevalence

The prevalence is unknown. More than 30 cases have been reported to date.

Diagnosis criteria / Definition

Association of:

- short stature of prenatal onset with rhizomelic and mesomelic shortness, marked joint laxity and characteristic facial dysmorphism at physical examination;
- Swedish key or monkey wrench appearance of the femoral heads and advanced carpal bone age at radiological examination. A supernumerary ossicle at the base of the second phalanx is a

radiological hallmark of this condition but is not essential.

- autosomal recessive inheritance.

Clinical description

Desbuquois syndrome is characterized by:

- intrauterine growth retardation with a mean birth length of 43 cm;
- severe dwarfism with rhizomelic and mesomelic shortness. Adult height often - 10 SD;
- generalized joint laxity with possibility of subluxations or luxations of large joints;
- radial deviation of fingers at the metacarpophalangeal and proximal interphalangeal joints. Thumbs are often broad, proximally placed in a hitchhiker position;
- narrow chest with frequent pectus carinatum deformity leading to a propensity for respiratory infections and early death, and kyphoscoliosis contributing to the short stature;
- facial dysmorphism includes round flat face, prominent eyes, micrognathia, saddle nose, long upper lip, flat philtrum, short neck;
- mental retardation of various degrees has been reported in several instances;
- inconsistent clinical features or complications include: obstructive sleep apnea, frequent respiratory distress, strabismus, cryptorchidism, cleft palate, clubfeet.
- It is worth noting that there is significant variability in the expression of the disease, and that some patients have been reported as being variants of Desbuquois syndrome.

Diagnostic methods

Diagnosis currently depends upon recognition of clinical and radiographic findings, as no specific biochemical abnormalities or gene have been identified.

X-ray features are characteristic, with distinctive findings in the hand.

1) Hands: the metacarpals are short, especially in the first and second digits. The first fingers show hypoplastic proximal and distal phalanges. Advanced carpal bone age is a constant feature. There are extraossification centres causing deviation of the fingers, which ultimately fused to the epiphyses. Deviation is generally located between the proximal phalanx of the index finger and the second metacarpal. The thumb can be either duplicated with a delta or broad phalange. Although these findings are very distinctive, they are not constant findings. Similar extra ossification centres can also be present in the

feet and tend to be symmetrical. In a recent review, Desbuquois dysplasia was divided into two subgroups, based on the presence or absence of the supernumerary ossicle at the second phalanx base: 19 patients had a supernumerary ossicle, whereas 14 did not.

2) Pelvis: the femoral neck shows enlargement of the lesser trochanter with metaphyseal beaking, producing a characteristic monkey wrench or Swedish key appearance. There is flared iliac and a flat acetabular roof are present.

3) Long bones are extremely shortened, as compared to the trunk. The distal femoras and tibias have metaphyseal flaring, the fibulas show proximal and distal overgrowth, and similar changes are noted in the respective long bones of the upper limbs. The epiphyses are normal but the diaphyses are extremely short. There is generalized osteoporosis.

4) Thoracic and lumbar spine: there are coronal and sagittal clefting of the vertebrae under 1 year of age. The cervical spine sometimes is excessively mobile on flexion/extension.

Differential diagnosis

[Larsen's syndrome](#) of Reunion Island: Short stature of prenatal onset with rhizomelic and mesomelic shortness are present in both entities, whereas joint laxity is more marked with multiple luxations, major orthopedic problems, and frequently club feet in Larsen's syndrome. Concerning radiological features, an extraossification centre is absent in patients with Larsen's syndrome of Reunion Island, Swedish key appearance of femoral head and advanced carpal bone age are not constant features, whereas radiocubital synostosis is frequent.

[Catel-Manzke syndrome](#): There are similar characteristic extra bones at the bases of some fingers in this condition but, in comparison to Desbuquois syndrome, patients with Catel-Manzke syndrome are of normal birth length, and are not as short thereafter, and do not present joint laxity or femoral neck changes. On the other hand, they frequently have a cleft palate and heart malformations.

[Diastrophic dysplasia](#) and pseudodiastrophic dysplasia can be misdiagnosed as Desbuquois syndrome.

Etiology

A gene for the disease has been mapped to chromosome 17q25.3 in the subgroup of patients with typical hand abnormalities only. It has been shown that this locus does not account for patients with Desbuquois dysplasia and "normal hands", suggesting genetic heterogeneity of the disease.

Genetic counseling

Autosomal recessive inheritance is suggested by the observation of multiple affected members of sibships and consanguinity of some parents. The risk of having another affected child is 25% for a couple who had a first child with Desbuquois syndrome. Genetic counseling for other members of the family can be reassuring in light of the low frequency of the disease, unless the couple is consanguineous. Similarly, an affected person does not run the risk of transmitting the disease to a child, unless his/her partner is a blood relative.

Antenatal diagnosis

Recurrence of the disease can only be suspected based on the ultrasonographic observation of intrauterine growth retardation and short limbs during the second trimester of pregnancy if a first child was born with Desbuquois syndrome. Linkage analyses are difficult even in patients with typical hand abnormalities, regarding genetic heterogeneity of the disease.

Management including treatment

Some of the patients present lethal respiratory distress at birth or later during infancy. Only symptomatic treatment is available for Desbuquois syndrome. Severe scoliosis is a frequent complication of the disease and physiotherapy should be proposed to control the progression of spine abnormalities. Arthrodesis can be proposed for dislocations. Surgery can be proposed for glaucoma. The degree of shortness is a major handicap, and patients sometimes had social problems for which psychological support should be proposed.

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