

Metatropic Dysplasia

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Abstract

Metatropic Dysplasia is a severe spondyloepimetaphyseal dysplasia characterised in infancy by long trunk and short extremities. Due to progressive kyphoscoliosis there is reversal of proportions in childhood – shortening of trunk with relatively long extremities. The face in the newborn is slightly dysmorphic, grossly normal later in life. The diagnostic radiographic findings consist of marked platyspondyly, widened metaphyses and small epiphyses and a specific pelvic shape. Inheritance is recessive or dominant. This is a very rare disease. About 80 cases have been reported in the literature. Prevention is not known. Antenatal diagnosis is possible in a fetus with long trunk and platyspondyly, short extremities and widened metaphyses.

Keywords

Spondyloepimetaphyseal dysplasia – platyspondyly – kyphosis

Disease name

Metatropic dysplasia

Definition/Clinical description/Diagnostic criteria

Metatropic dysplasia (metatropic-changing) (hypertrophic achondroplasia) is a severe spondyloepimetaphyseal dysplasia with characteristic clinical and diagnostic radiographic findings. In the newborns and infants there is a long trunk with disproportionately short extremities. Length is usually normal. The chest is narrow and often there is a 1-3 cm long tail-like appendage at the level of coccyx. The joints are prominent with decreased mobility. The fingers

and toes are long. The face is uncharacteristic although a prominent forehead and flattened

nasal bridge may be present in infancy. The proportions change during childhood with relative shortening of the trunk due to progressive kyphosis and relatively long extremities. Chest deformity and flexion contractures of major joints are associated findings. Hyperextensibility of the fingers may be present. The adult height is up to 120 cm. Intellectual development is normal.

The diagnostic radiographic findings in newborns consists of marked platyspondyly (flattening of all the vertebral bodies), short tubular bones with broad concave metaphyses and small epiphyses (delayed ossification – retarded bone age). The

proximal end of the femur is club shaped and the lesser trochanter is prominent.

The shape of the iliac bone is unique – a notch between the shortened iliac body and prominent crescent shaped iliac wing resembles a halberd. Dens hypoplasia with C1/C2 instability is a feature in severe case. Narrowing of the cervical and thoracic spinal canal may be present. The posterior part of the vertebrae is well developed and the sacrum and coccyx are long.

With advancing age osteoarthritic changes are superimposed on regressive metaphyseal changes. Progressive kyphoscoliosis with decreasing flattening of the vertebral bodies makes the diagnosis more difficult.

There is a high mortality in the severe forms subsequent to chest and spinal deformities (respiratory infections).

Differential diagnosis

The differential diagnosis in infancy is with other platyspondylic spondyloepimetaphyseal dysplasias. None of them shows such marked disproportion between the long trunk and short extremities. The radiographic pattern of the spinal, metaphyseal and pelvic changes is unique. Later in childhood and adults the differential diagnosis is with platyspondylic bone dysplasias, especially SMD Kozlowski and Morquio disease (Mucopolysaccharidosis IV). The spinal changes may be indistinguishable from those of the common types of spondylometaphyseal dysplasia and similar to some spondyloepimetaphyseal dysplasias. The distinctive shape of metaphyses and pelvis are useful differential diagnostic signs. Dysostosis multiplex is a feature of Morquio disease. Patients without fully developed clinical and radiographic signs of metatropic dysplasia have been reported. Their relationship to the “standard forms” of metatropic dysplasia is uncertain.

Diagnostic methods

Clinical examination is suggestive. Skeletal survey is a diagnostic examination.

Epidemiology

No racial or regional preponderance. Mostly single cases in normal, healthy families. About 80 cases have been reported in the literature.

Genetic counselling

Autosomal dominant and autosomal recessive inheritance. Gene mapping and linkage unknown. Prevention not known.

Antenatal diagnosis

Possible in a fetus with long trunk and platyspondyly, short extremities and widened metaphyses.

Management including treatment

Kyphosis is usually rapidly progressive after the first 6 months of age. Current best practice is for patients to be cast under anaesthesia and braced in hyperextension in a clam shell polypropylene body orthosis. This will need to be replaced as the child grows. Bracing should be maintained until skeletal maturity or until spinal fusion is feasible. There is a very high rate of non-union when anterior and posterior spinal fusion is attempted in Metatropic dysplasia and surgery is best avoided. Complete rehabilitation including physiotherapy and occupational therapy is important as there is a risk of progressive knee and hand arthropathy.

Unresolved questions

The relationship between metatropic like dysplasias and the “standard cases” is uncertain.

References

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