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Case Report

Short femur on antenatal ultrasound reveals chondrodysplasia in two twins: a case report

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ABSTRACT

Chondrodysplasia is a rare autosomal recessive disorder with diverse clinical manifestations, including rhizomelic short limbs, facial anomalies such as a broad nasal bridge, epicanthus, dysplastic ears, and micrognathia. While mental impairment is uncommon, spasticity and seizures may occur. The fetal prognosis is generally favorable in the absence of additional malformations. In cases with a family history of chondrodysplasia, early morphological assessment, especially of the limbs during the first trimester, is crucial for identifying suggestive ultrasound signs. We present a rare case of chondrodysplasia in a twin pregnancy in a multiparous woman. This case underscores the importance of antenatal diagnosis and comprehensive genetic counselling to reduce the risk of transmission to future generations.

Keywords: Chondrodysplasia, Ultrasound, Genetics, Femur

INTRODUCTION

Chondrodysplasia is a multi-systemic disease.[1] The disease has a low occurrence rate, making diagnosis very difficult, although a family history suggests autosomal genetic inheritance, which alters the function of cartilage and bone.[2] The pathophysiology of chondrodysplasia is poorly elucidated; it involves a disorder in the biogenesis of peroxisomes responsible for neutralising cellular peroxides.[3] The prognosis is generally unfavourable, associated with high morbidity and mortality.[4] Postnatal management of chondrodysplasia is essentially based on relieving symptoms and preventing complications.[1] In our case, this condition is described both antenatally and postnatally.

CASE REPORT

Mrs L.S, aged 38, G6 P5 A2, with a history of 3 uneventful vaginal deliveries, her husband suffering from chondrodysplasia, was admitted at a term of 35 days' gestation +6 days, to go into labour. This was a twin pregnancy with a single amniotic chorion. During antenatal follow-up, and at morphological ultrasound, her general practitioner noted disharmonious growth retardation with short femurs in both male foetuses (Figure 1). A geneticist's opinion was sought and the diagnosis of chondrodysplasia was suggested, with a 50% risk of transmission with each pregnancy. The parents were informed of the possibility of foetal damage and the likely complications.

Figure 1: Morphological ultrasound at 23 + 6 days, femur length of 18 days.

The patient underwent emergency caesarean section on D1 with transverse presentation. These were two male newborns. The birth weight for J1 was 2500 g, Apgar 7-8-9 and for J2, who was in breech presentation, the birth weight was 2400 g, Apgar 8-8-9. Initially, the twins presented with a bradycardia that rapidly recovered after stimulation at M2, with no immediate respiratory distress. Skull dysmorphia (Figure 2), facial dysmorphia with epicanthus, wide nasal bridge (Figure 3) and dysplastic ears; femurs and humeri that are short with reducible club feet (Figure 4).

Figure 2: Dysmorphic skull, dysplastic ears.

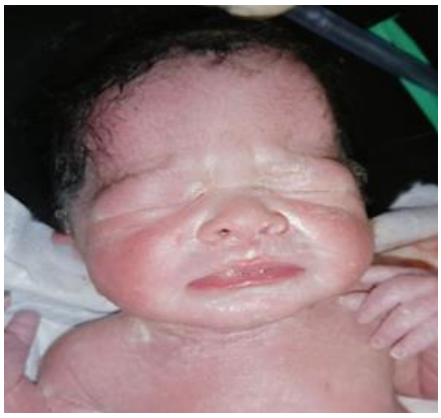

Figure 3: Prominent forehead, epicanthus, wide nasal bridge.

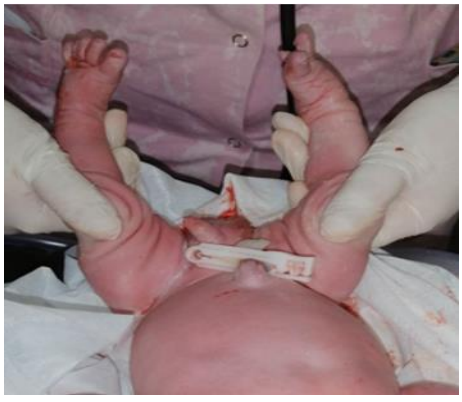

Figure 4: Reducible club feet.

DISCUSSION

Chondrodysplasia is a rare primary bone dysplasia characterised by rhizomelic shortening of the limbs, punctate calcifications in the cartilage with epiphyseal and metaphyseal abnormalities.[1] The prevalence of rhizomelic chondrodysplasia punctata (RCDP) is less than 1/100,000, with an incidence of around 0.7 and 0.5 cases per 100,000 births in Europe and the USA, respectively.[5]

Chondrodysplasia is caused by defects in the genes encoding the peroxisomal proteins required for plasmalogen (PL) biosynthesis, in particular the PEX7 and PEX5 receptors, or the GNPAT, AGPS and FAR1 enzymes. The severity of the disease is correlated with erythrocyte PL levels, which are almost undetectable in severe (classic) CDDR. In milder (non-classical) forms, residual PL levels are associated with improved growth and development.[6]

Clinically, most patients have congenital cataracts and skeletal dysplasia. In the classic form, there is profound growth restriction and psychomotor retardation, with most patients not progressing beyond the stages of infantile development, and occasionally other major symptoms such as learning disabilities, behavioural problems, convulsions and cardiac malformations have been observed. The genetic abnormalities responsible for chondrodysplasia are autosomal recessive linked to chromosomes 6, 1 and 12 but can also be gonosomal X-linked.[7]

Prenatal diagnosis may be suggested by nasomaxillary hypoplasia, known as the Binder phenotype, short limb length, epiphyseal stippling or prenatal irregularity of the spine.[8] Prenatal diagnosis of these findings is possible in high-risk pregnancies if the type of deficit has been previously identified in the family.

Isolated rhizomyelic chondrodysplasia is not an indication for medical termination of pregnancy except at the request of the family; the woman requires special follow-up and close monitoring in the face of foetal growth retardation.

The postnatal diagnosis is suspected on the basis of clinical signs: symmetrical proximal shortening of the extremities, facial dysmorphia, cataracts and severe growth retardation, and radiological signs such as punctate calcifications of the epiphyseal cartilages of the knees, hips, elbows and shoulders, which may also affect the hyoid bone, larynx, chondrocostal junction and vertebrae.[9]

In Ranza et al series, molecular studies of patients with chondrodysplasia enabled pathogenic variants to be identified in 60% of patients (18/30); when a clinical diagnosis was suspected, this was molecularly confirmed in 53% of cases. However, 40% of patients remain without a molecular aetiology.[10]

Management is symptomatic and individualised: orthopaedic management of leg length discrepancy; frequent assessment of kyphoscoliosis; management of respiratory impairment by a pulmonologist; dermatological management with emollients and keratolytics; cataract management and vision correction, with emphasis on family support.

The prognosis for chondrodysplasia is often unfavourable. In addition to osteoarticular development problems, these children present with psychomotor disorders, epileptic seizures of all types and a pyramidal syndrome. Most children also present with congenital bilateral cataracts, often requiring surgery at an average age of 5 months. congenital bilateral cataracts or early in childhood, often requiring surgery at an average age of 5 months. As a result of abnormal facial structures, one child in two suffers from recurrent otitis media.

In terms of the lungs, two-thirds of patients experience respiratory distress after birth, followed by recurrent respiratory infections.

Chondrodysplasia is generally fatal, mainly due to respiratory problems, in the first few years of life, with 50% survival at 6 years.[9]

CONCLUSION

Chondrodysplasia is a rare genetic disorder. Its pathophysiology is poorly understood and complex. The diagnosis is made antenatally on the basis of suggestive ultrasound findings and confirmed postnatally by molecular genetic studies. The prognosis is generally unfavourable and treatment is essentially symptomatic.

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