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A case of acondrodysplasia punctata

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Objective

Prenatal diagnosis following a case report.

Methods

Review of scpecific literature on prenatal diagnosis in the case of acondrodysplasia punctata ans subsequent description of the clincal case.

Results

Chondrodysplasia punctata is a dysplastic syndrome which involves different groups of entities. All of them have in common the presence of multiple punctate calcifications in the joints. There are three different forms of chondrodysplasia. On the one hand, the autosomal recessive form, called rhizomelic, which is caused by mutations in the PEX7 gene (receptor for several peroxmal enzymes). On the other hand there are two other X-linked forms: The Conradi- Hünermann type chondrodysplasia (X-linked Autosomal dominant) and the autosomal recessive form caused by pathogenic variants in the arysulphatase gene (ARSE) or deletions or rearrangements of the short arm of the X chromosome. Rhizomelic form (AR): We can found the following physical features: severe short stature, distinctive craniofacial features with low hypoplasia of the nasal bridge and midface, rhizomelic (proximal) shortening of the limbs, wide joints, multiple joint contractures and abnormal spine with coronal clefts. Dotted calcifications can also be found in the spine and in the growth plates of some joints. Other features include congenital cataracts, intelectual disability and microcephaly. Conradi-Hünermann (X-linked AD). It occurs almost exclusively in females. They are characterised by the following physical features: The limbs are short and often asymmetrical, in this case the punctate calcifications are extraepiphyseal and variable joint contractures are also found. In this variant, patients have a low nasal bridge and midface hypoplasia. Other anomalies include severe cutaneous erythroderma and ichthyosis which may follow Blaschko lines. Scoliosis is common and often associated with multiple calcifications. X-linked recessive form: Occurs in males and is characterised by the following: punctate epiphyses (as seen in the other forms of chondrodysplasia punctata), shortening of the distal phalanges (brachycephalangia), and nasal and midface hypoplasia. Infants born to mothers with systemic lupus erythematosus or to women who were exposed to warfarine therapy during gestation may present with clinical features similar to the X-linked recessive form of chondrodysplasia punctata. Clinical case. A 40-year-old woman, expecting her third child, with no previous history of interest, who began prenatal care in the Canary Islands with the following results: - 12 weeks ultrasound: Calcifications in proximal femoral epiphysis and severe nasomaxillary hypoplasia (binder phenotype). - Screening for aneuploidy risk and normal QF-PCR XY genetic study. Normal arrays and exome without finding (ARSE without mutation). Subsequent controls were carried out in HUSE, and at 36 weeks we observed the following: - Low percentile growth with maxillofacial hypoplasia. Euthocic birth at 40 weeks, newborn was born with the following characteristics: birth weight 2740gr (p5), length 48.5cm (p15), head circumference 34cm (p45). In the postnatal study the following is observed: Fascies impression normoconfigured, typical racial features, minimal sensation of nasal and maxillary hypoplasia with inversion of upper lip. The nasal bones are palpable. Nasal tip not projected. Nasofrontal angle not flat, short columella, normal nasolabial angle. Palate intact. Different postnatal complementary tests were carried out: - X-ray of bone series with the following result: Hypoplastic distal phalanges of hands and feet. Punctate epiphyseal calcification in both calcaneus and some toes. Punctate epiphyseal calcification in the right hip. - Bone series X-ray. Hypoplastic distal phalanges of hands and feet. Punctate epiphyseal calcification in both calcaneus and some toes. Punctate epiphyseal calcification in the right hip. Management and treatment Genetic counselling and generally supportive treatment. Forecast Good, except if we found cervical canal stenosis with cervical cord compression which can lead to severe morbidity and early mortality.

Conclusion

- Prenatal ultrasound diagnosis: Hypoplasia of the nasal bridge and abnormal epiphyseal calcifications. Dotted calcifications are usually seen in the epiphyses of the tarsus, knee and distal phalanges, but may be even more generalised. - Postnatal diagnosis: Facial dysmorphia, quite similar to Binder maxillonasal dysplasia, reduced nasal tip protrusion but with normal nasal wings, and short columella. In this case, as a differential diagnosis we considered the following pathologies: the different types of chondrodysplasia punctata, embryopathies due to warfarin, embryopathies due to preudowarfarin, facial dysmorphic embryopathy, quite similar to that of Binder's maxillonasal dysplasia, reduction of the nasal tip protrusion but with normal nasal wings, and short columella.