A case of chondroectodermal dysplasia and comparison between the results of prenatal ultrasound, fetal autopsy and body X-Ray

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Objective

To insist on the importance of knowing Ellis Van Creveld Syndrome for better genetic counseling.

Methods

A case report of a termination of pregnancy for Ellis Van Creveld Syndrome suspected during prenatal ultrasound confirmed during fetal autopsy and X-Ray of the entire body.

Results

We report a case of a woman of 38 years old, blood group B positive, first-degree consanguineous marriage, G7 P2 (fetal death in utero for malformative syndrome, 3 pregnancies arrested, 2 children in apparent good health) admitted to our service after the discovery of fetal malformations during morphological ultrasound. The ultrasound showed: agenesis of the vermis, ventriculomegaly, clubfoot, short tibia and fibula. Fetal MRI confirmed the lesions found in ultrasound. It also showed polydactyly of hands and feet and dwarfism rheumatica. Termination of the pregnancy was then decided by the patient. The fetal autopsy found a narrow thorax, polydactyly of hands and feet with syndactyly, gingival hypertrophy with irregular gums and presence of teeth. To the notion of consanguinity and the recurrence of this malformation, the diagnosis of Ellis Van Creveld syndrome was strongly suspected. Body X-Ray of the fetus confirmed the diagnosis of this chondroectodermic dysplasia.

Conclusion

Ellis Van Creveld syndrome is certainly very rare, but it is interesting to know, when there are similar cases in the family, for a better genetic counseling. Prenatal ultrasound is interesting in the inventory of abnormalities and indication of a termination of pregnancy. Definitive diagnosis is made on fetal autopsy.