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Prenatal diagnosis of Holt–Oram syndrome at 16 weeks in a pregnancy at risk: a case report

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Introduction

Abstract Body Holt–Oram syndrome is a rare autosomal dominant disorder caused by pathogenic variants in the TBX5 gene and characterized by congenital upper limb abnormalities associated with cardiac defects or conduction abnormalities. Prenatal diagnosis may be suspected in the presence of radial ray defects, particularly in pregnancies with a known familial history.

Case

We report a case diagnosed at 16 weeks of gestation in a fetus of a mother affected by Holt–Oram syndrome (figure 1). The ultrasound examination revealed severe upper limb abnormalities, including shortening of the forearms with radial ray defect and abnormal hand positioning associated to a ventricular septal defect. Given the maternal history of Holt–Oram syndrome and the characteristic limb findings,

the diagnosis was strongly suspected during the first-trimester scan. Genetic counseling was provided considering the 50% recurrence risk associated with autosomal dominant inheritance and the potential association with congenital cardiac defects. After multidisciplinary counseling regarding prognosis and possible outcomes, the patient opted for termination of pregnancy. Fetopathological examination confirmed the presence of severe radial ray and heart abnormalities supporting the prenatal diagnosis (figure 2).

Conclusion

This case highlights the value of careful first-trimester ultrasound evaluation of the fetal upper limbs, especially in pregnancies at risk for inherited syndromes. Early recognition of radial ray abnormalities can allow very early suspicion of Holt–Oram syndrome, enabling timely genetic counseling and informed parental decision-making.



Figure 1 : the mother's hands



Figure 2: fetopathological examination