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Prenatal diagnosis of congenital clubfoot: role of genetic testing in isolated cases

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Objective

The aim of this retrospective study was to assess the accuracy of prenatal diagnosis of isolated congenital clubfoot and to determine the value of genetic testing in isolated clubfoot cases.

Methods

Retrospective analysis of all fetuses prenatally diagnosed with clubfoot (n=742) identified during the second trimester routine screening ultrasound examinations between 2007 and 2021 in the North-West region of the Netherlands. All parents were offered genetic testing. Sonographic characteristics, prenatal follow-up and postnatal outcomes at least until one year of age were evaluated.

Results

There were 742 cases of clubfoot of which 474 (64%) were classified as isolated after the first ultrasound examination. The prevalence of isolated clubfoot was 0.98 per 1,000 births. Invasive prenatal testing in 130/474 (27%) yielded 12 (9%) fetuses with genetic or chromosomal abnormalities. In total 36 (8%) cases the prenatal diagnosis of isolated clubfoot changed or was rejected during prenatal follow-up. Diagnosis was confirmed postpartum in 317/439 (72%) cases. In 53 (12%) children associated anomalies were found during postnatal follow-up and in 18/53 children (34%) a genetic cause for the congenital anomalies or neurodevelopmental delay was found. In 59 (13%) children there was no clubfoot present postpartum.

Conclusion

Genetic testing should be considered in isolated clubfoot cases to reduce the risk of misdiagnosis and to provide appropriate counseling and management. Prenatal diagnosis of complex clubfoot is crucial as these cases have a high risk of poor prognosis.