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Prenatal diagnosis of malrotation of the knee joint leading to diagnosis of Nail-patella syndrome in the father and the fetus

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Objective

Nail-patella syndrome (NPS) is an autosomal dominant disease that is associated mainly with skeletal anomalies such as patellar hypoplasia/aplasia, elbow dysplasia and iliac horns, nail dysplasia, and less commonly ocular and renal involvement. Here we present a case with malrotation of the right knee joint leading to a diagnosis of Nail-patella syndrome in the fetus and the father.

Methods

This is a case report.

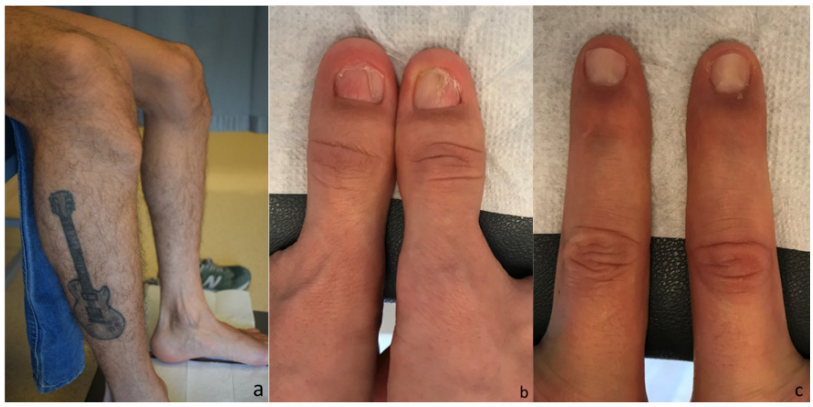
Results

A 26-year old woman in her first pregnancy underwent routine anomaly scan at 21 weeks of gestation which revealed an echogenic intracardiac focus and malrotation at the right knee joint with the dorsum of the foot facing the gluteal region. The fetal growth was consistent with dates and there were no other structural defects. First trimester scan was completely normal and the combined test revealed a risk 1/8300 for trisomy 21. Maternal medical history was unremarkable; however, when the parents were asked about possible joint abnormalities, the father mentioned that his patellar bones were small. Further genetic evaluation of the father demonstrated bilateral nail dysplasia in the thumbs, abnormal lunulae in the index fingers, normal toenails, normal elbows with no pterygium, no iliac horns, history of surgery for inguinal hernia and no signs of renal involvement, indicating NPS. Paternal genetic test results for LMX1B mutations are pending. After counseling, the parents declined invasive testing for further fetal assesment and confirmation of NPS.

Conclusion

Prenatal diagnosis of malrotation of the knee joint should raise the suspicion of abnormal patellae and prompt evaluation for possible additional findings of syndromes associated with absent/hypoplastic patellae in the fetus and family members.

Two dimensional image of the right leg with the dorsum of the foot facing the gluteal region at 21 weeks.



Photographs of the father demonstrating (a) hypoplastic and displaced patellae, (b) bilateral nail dysplasia of the thumbs, (c) abnormal lunulae of the index fingers.