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Hajdu-Cheney Syndrome: could it be detected on prenatal ultrasound?

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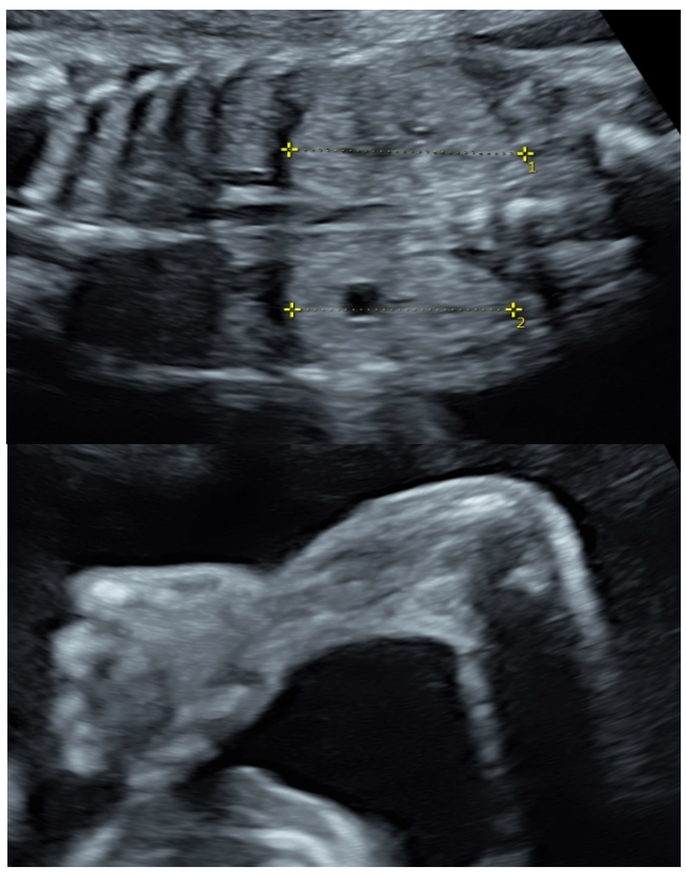
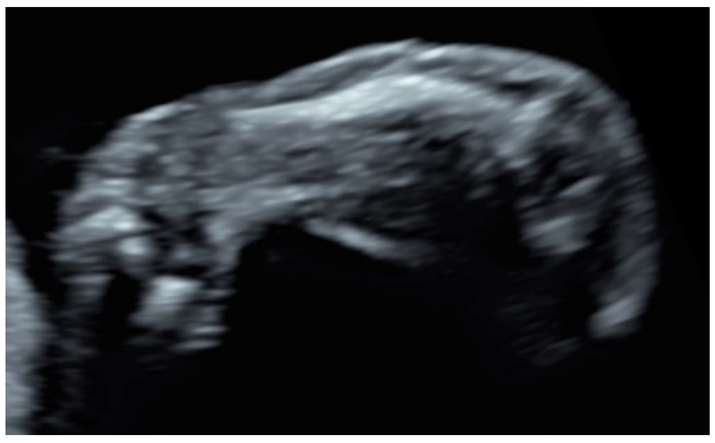
Introduction

Hajdu-Cheney Syndrome (HCS) is a rare genetic disease caused by an heterozygotic mutation of gene ***NOTCH2*** located on chromosome 1p13-p1 and it follows an autosomal-dominant inheritance pattern. In HCS there is a **bone density deficit** that leads to a general skeletal dysplasia. There is a subgroup of patients that present two distinctive signs: serpentine fibula and polycystic kidneys (SFPKS).

Case Report

A 36-year-old secundiparous (2 CST) with no other risk factors had an intermediate risk of Pataw syndrome and a high risk of NTDs on the 2nd trimester aneuploidy screening. It was found **bilateral choroid plexus cysts** and **mesomelia** of both superior limbs at 20w scan. Initial genetic study was ordered with a normal QF-PCR, array and negative study to hypochondroplasia, achondroplasia and osteogenesis imperfecta. Follow-up US were performed with findings consisting with severe mesomelia, **radius fracture** and bowed ulnae and fibula (**serpentine fibula**), signs of bone hypomineralization, enlarged **hyperchogenic kidneys with a 6 mm simple cyst**, as well as, bilateral cryptorchidism.





Exome sequencing was achieved and a *NOTCH2 de novo* mutation *(NOTCH 2 1:120458537 NM_024408.4c.6804_6807dupTGGT p. (Met2270Trpfs*8))* was found.

A caesarian section was scheduled at 38.5w. On admission, a loss of fetal well-being was observed, that's why an emergency CST was performed extracting a live male fetus weighing 2625 g with an Apgar test of 1/6/7. A cerebral palsy caused by an acute hypoxic-ischemic lesion was described. The newborn is currently being monitored for palliative care.

Conclusion

Exome sequencing allows us to obtain diagnoses that until now would have been unknown at the prenatal stage. Is important to describe accurately ultrasound findings to guide obstetricians, geneticist amb peditricians to do a precise diagnosis.