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Galactosialidosis caused by CTSA gene mutation: a rare cause of late second trimester fetal hydrops

Salwan Al-Nasiry, Anne van der Velden, Femke Slaghekke, Mijntje Pieters.

Maastricht UMC. Maastricht. The Netherlands

Background

Fetal hydrops occurs in approximately 1 in 2000 pregnancies in the third trimester. Although most cases are non-immune in nature, only 1–2% are caused by inborn metabolic disorders with a poor prognosis of around 70% mortality.

Case report

A 27-year-old multiparous woman was referred to our center at 27+1 weeks with suspected fetal hydrops. NIPT, first and second trimester anomaly scans were normal. Ultrasound revealed severe ascites, significant skin edema, cardiomegaly and a hydrocele. Fetal movements and umbilical artery Doppler were normal. However, an elevated peak systolic velocity in the middle cerebral artery (MCA) was noted. Screening for TORCH infections as well as fetal cell count were negative. The patient was referred to a fetal therapy center and amniocentesis was performed, and parvovirus B19 and cytomegalovirus infection were excluded. Ultrasound 11 days later demonstrated worsening fetal ascites and a further increase in MCA peak systolic velocity.

A diagnostic cordocentesis from the intrahepatic umbilical vein showed no fetal anaemia. Creatinine levels in fetal ascites were similar to those in fetal blood. At 29+0 weeks the patient went into spontaneous labour and delivered a male infant who died shortly after birth. Whole exome sequencing revealed a homozygous mutation in the CTSA gene (CTSA;Chr20(GRCH37): g.44520359C>A: NM_000308,4:c,152C>Ap.(Ser51Tyr)), confirming the diagnosis of galactosialidosis, a lysosomal storage disorder. Both parents were identified as mutation carriers. The infantile form of galactosialidosis is consistent with fetal hydrops and fetal or neonatal demise. Post-mortem examination showed enlarged islets of Langerhans, retrognathia and absence of the right nipple.

Conclusion

We present a rare case of galactosialidosis presenting as fetal hydrops in the late second trimester, caused by a homozygous mutation in the CTSA gene and diagnosed antenatally using whole exome sequencing.



Figure 1. Abdominal ascites



Figure 2. Skin edema