A case of Mucopolysaccharidosis type VII (Sly Syndrome)
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Objective
Mucopolysaccharidosis type VII (MPSV II), also known as Sly Syndrome, is a progressive metabolic deficiency which affects most tissues and organs. The exact incidence of MPS VII is unknown, although it is estimated to occur in 1: 250,000 newborns. It is one of the rarest types of mucopolysaccharidosis and is inherited in an autosomal recessive pattern. The objective of this presentation is to mention the importance of investigating metabolic syndromes in severe second trimester cases of hydrops fetalis.

Methods
This is a case report presentation.

Results
We present a case of antenatal diagnosis of MPS VII in a fetus with severe early onset hydrops fetalis. The mother was a 19 years old primiparous with unremarkable medical and family history. Of note the parents were second degree relatives. The first trimester combined screening test came back as low risk for all three main chromosomal abnormalities, with a NT measuring 2.1 mm. The pregnancy progressed uncomplicated till the anomaly scan at 20 weeks when we detected isolated severe hydrops fetalis with normal amniotic fluid. The initial investigation excluded the immune factor, as the maternal blood group was A Rhesus +, no atypical red cell antibodies were present and the MCA-PSV was normal. The TORCH investigation revealed a recent Toxoplasmosis maternal infection. An amniocentesis performed and amniotic fluid was sent for genetic analysis and culture. Fetal echocardiography did not show any major cardiac defect. The DNA microarray analysis showed that the fetus was homozygous for MPS VII, while the AF culture excluded the congenital Toxoplasmosis. A termination was offered but the parents were committed to the pregnancy. The non-immune hydrops fetalis progressed significantly during the next weeks of gestation, till 32 weeks when intrauterine demise occurred. The fetus was sent for post-mortem investigation which confirmed the antenatal diagnosis of MPS VII. Both parents are carriers of the mutation.

Conclusion
In cases of non-immune fetal hydrops, rare genetic metabolic syndromes should be part of our differential diagnosis, especially when consanguinity is present. MPS VII has an unfavourable course, even in live-born fetuses. Our counselling should be based on the so far knowledge on the consequences of the syndrome. Future pregnancies should be investigated accordingly.