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Prenatal diagnosis of Simpson-Golabi- Behmel syndrome type1: a case report

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Introduction

Simpson-Gobali-Behmel syndrome type 1 (SGBS1) is a rare X-linked genetic disorder characterized by pre-and postnatal overgrowth, distinctive craniofacial features, variable congenital malformations, organomegaly and increased tumor risk.

Casereport

We present a primigravida at 19 weeks GA with mild bilateral ventriculomegaly of 10,5 mm, macrocephaly and macrosomia (growth percentile 99) in a male fetus(fig1-2). Whole exome sequencing on amniotic fluid and concurrent testing of the couple revealed a pathogenic variant in the GPC3 gene (located on Xq26) witch encodes for Glypican-3, maternally inherited. After counselling, the couple decided to terminate the pregnancy.

SGBS1 can present with pre-and postnatal overgrowth, macrocephaly, macroglossia, organomegaly, genitourinary anomalies and facial dysmorphism (hypertelorism, frontal bossing, anteverted nares, high-arched or cleft palate) and variable degree of intellectual disability, increased predisposition for embryonal tumors such as Wilms tumor and hepatoblastoma. Most clinical diagnoses are made postnatal.Early onset macrosomia as reported is rare .

SGBS1 is inherited X-linked recessive. Males inheriting the pathogenic variant are affected. Females who inherit the pathogenic variant are carriers and may have milder manifestations. Type 1 differs from the rare lethal form, type 2 associated to a different region of chomosome X. (Xp22.2)

Conclusion:Prenatal early-onset macrosomia and visceromegaly should prompt genetic testing focusing on overgrowth disorders and include Simpson-Gobali-Behmel syndrome type 1 (SGBS1)

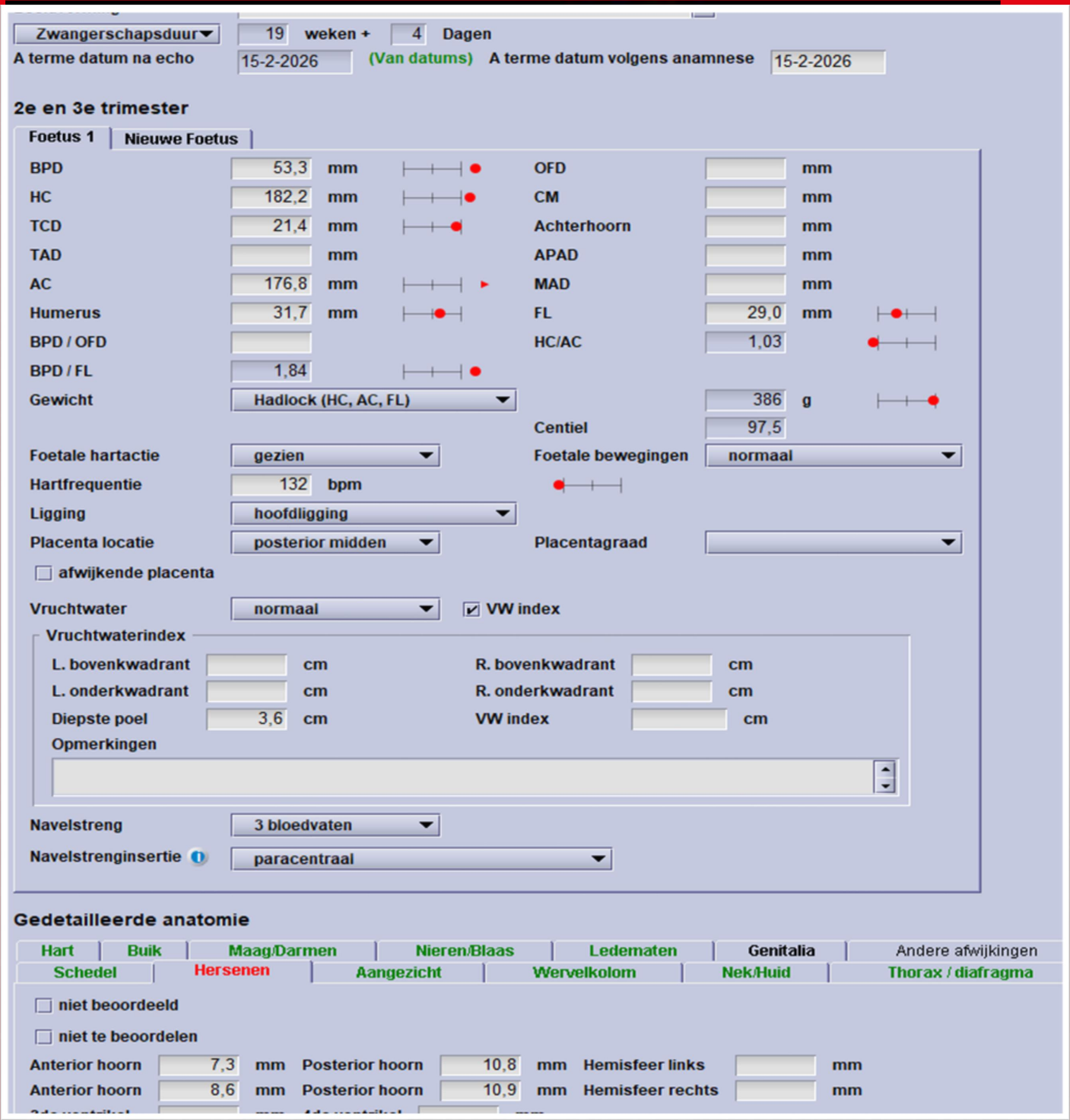


Figure 1. ultrasound 19+4 wks with early onset macrosomia and mild ventriculomegaly

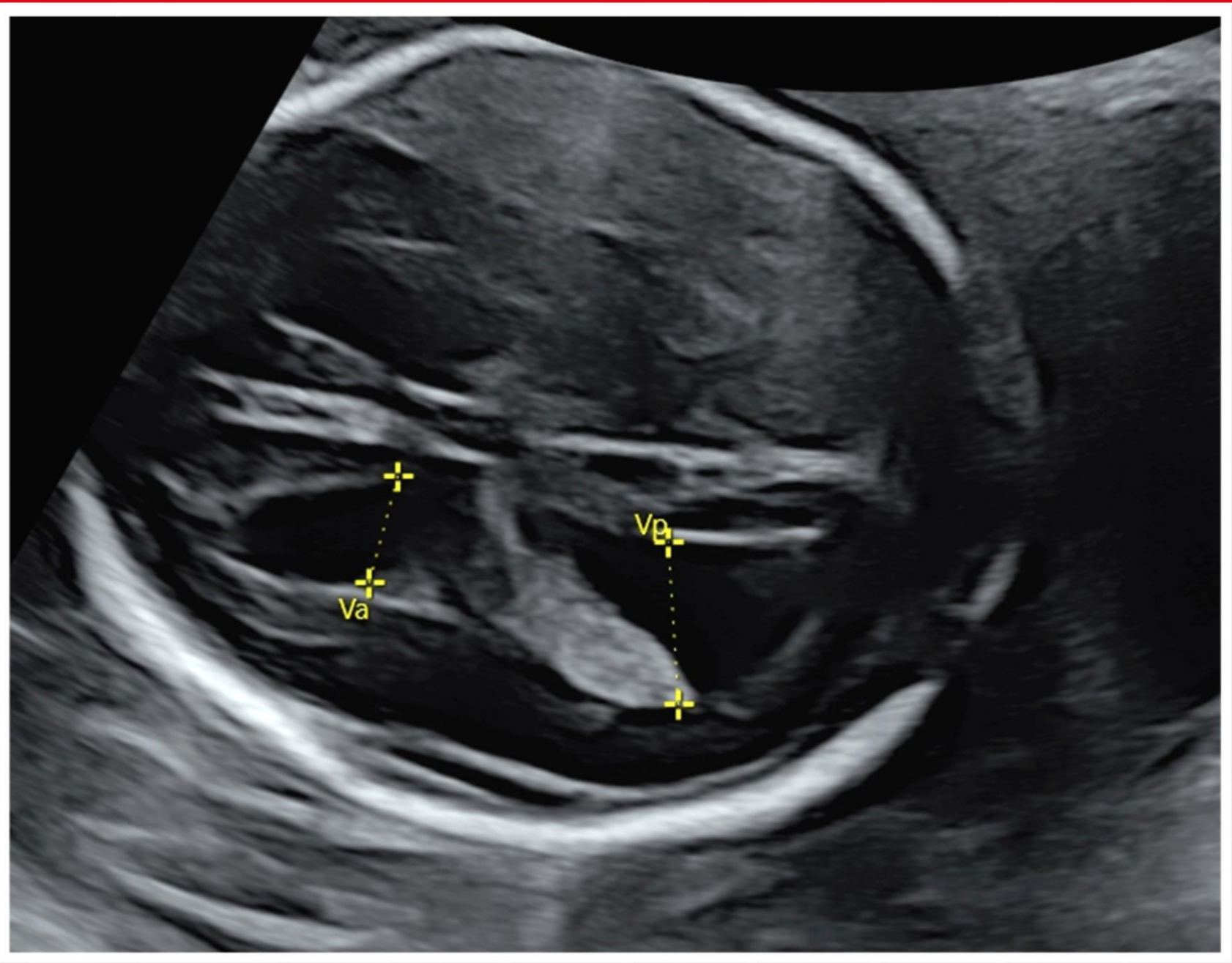


Figure 2. axial scan with mild ventriculomegaly