

Case report of a live-born infant with prenatally diagnosed partial trisomy

17, structural abnormalities, and growth restriction.

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Introduction

We report a rare case of a live-born singleton with prenatally diagnosed partial trisomy 17.

Material and Methods

A 21-year-old woman (2G 1P) presented at 23+5 weeks of gestation for detailed diagnostic evaluation. A hypotrophic female fetus was observed with normal amniotic fluid volume and placenta. A Blake’s pouch cyst was detected; in addition, aortic isthmus stenosis was suspected. Maternal Doppler studies were unremarkable; fetal Doppler revealed increased umbilical resistance. Invasive diagnosis was recommended. Amniocentesis was performed at 29+0 weeks of gestation. Cytogenetic analysis revealed partial trisomy 17 (mos 47,XX+17[17]/46,XX[25]). The couple received genetic and neonatal counseling, and due to fetal growth restriction (FGR, 1910 g, 2nd percentile, increased umbilical resistance), elective prostaglandin induction was agreed upon for 37–38 weeks of gestation.

Results

The female newborn was delivered at 38+0 weeks of gestation via spontaneous delivery (APGAR 9/10/10, umbilical cord blood pH 7.31, BE 2.90 mmol/L, birthweight 2350 g). The prenatally diagnosed BPC was confirmed. In addition, a left ear tag and a muscular VSD as well as mild tricuspid insufficiency were observed. The child’s adaptation was delayed, but she was discharged home with follow-up care by a pediatrician.

Summary

Partial trisomy 17 is extremely rare, with a prevalence well below 1 in 1,000,000; the most commonly described forms involve segmental duplications of the short arm (trisomy 17p) and, less frequently, of the long arm (17q), as well as mosaic forms. Prenatal signs include, among others, increased nuchal translucency (NT), fetal growth restriction (FGR), central nervous system (CNS) abnormalities, heart defects, and urogenital malformations. The postnatal course ranges from mild presentations to severe intellectual and motor developmental delay.

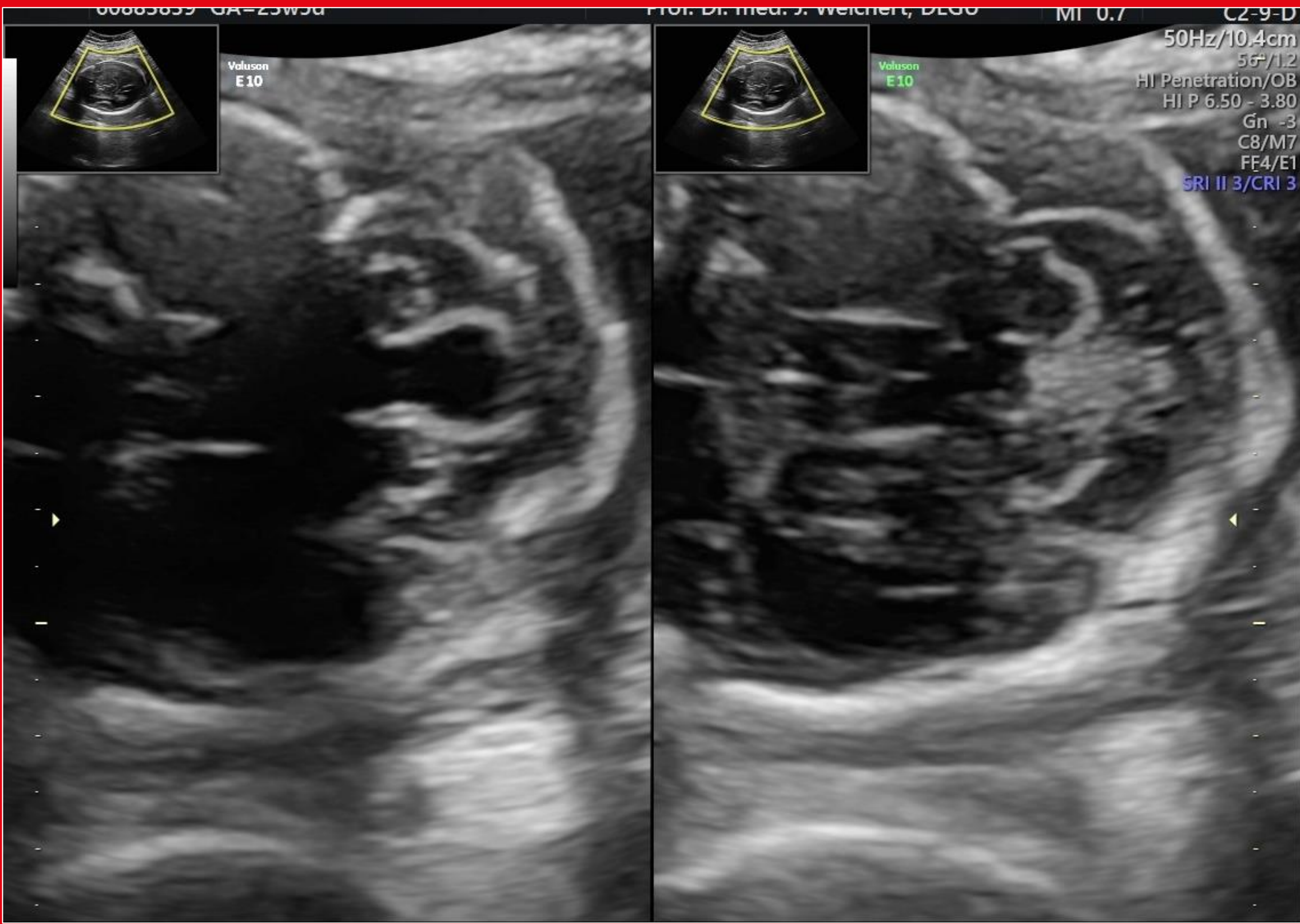


Figure 1. Blake’s pouch cyst in 23+5 weeks.



Figure 2. Aortic isthmus stenosis in 27+3 weeks.