A case of trisomy 13
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Objective
Trisomy 13 (T13), or Patau syndrome, has prevalence of 1 per 6,500 births. Most fetuses with trisomy 13 die in utero or are stillborn. Of those born alive, 20% will survive the first month of life and only 5% will survive the first six months. T13 may show various brain anomalies including holoprosencephaly (HPE), midfacial hypoplasia (cyclopia and proboscis), ventriculomegaly, enlarged cisterna magna, microcephaly and agenesis of corpus callosum.

Methods
The following describes a case report of a 30-year-old parous woman who underwent prenatal consultation at 11-14 weeks.

Results
The patient underwent first trimester screening according to the Fetal Medicine Foundation protocol. The results were as follows: CRL-59 mm, NT-3 mm, NB/-, TR/-, DV PI/0. 9/a-wave positive. Aneuploidy risk assessment gave risk T21 >1:4; T18 1:8 and T13 >1:4. Structural anomalies included: polydactyly, no eyes, proboscis, cleft palate and umbilical cyst. Patient underwent diagnostic amniocentesis. Karyotype was 46, XX, +13, der(13;14)(q10;q10). After genetic counselling, the patient decided to terminate the pregnancy.

Conclusion
Typical ultrasonographical findings in T13 include hygroma colli, craniofacial defects, cerebral malformations and problems with genitourinary tract. Cases of HPE, when maternal diabetes or fetal alcohol exposure are excluded; a genetic cause is most common. These include abnormalities in chromosomes 10, 13, 15, 18 and Klinefelter syndrome. For Patau syndrome, 75% cases suffer from HPE.