Shprintzen-Goldberg syndrome: a case report

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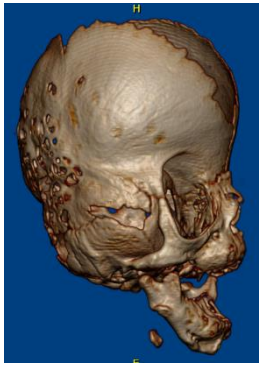
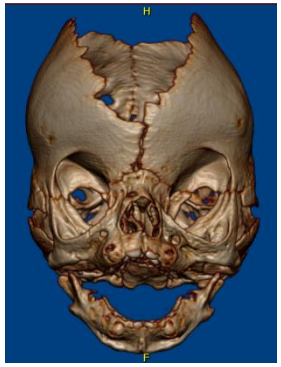
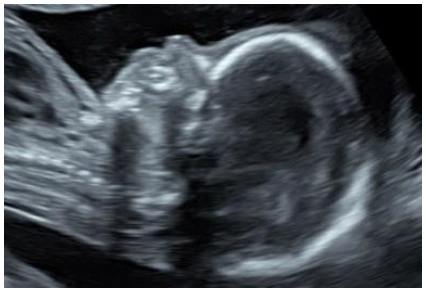
Objective: Shprintzen-Goldberg syndrome (SGS) is an extremely rare disease with the indidence of 1 in 1,000,000. SGS is autosomal dominant and was first described in 1979. A common features of SGS include premature fusion of skull bones, long and narrow face, hypertelorism, exophthalmos, broad nose bridge, micrognathia, marfanoid habitus, skeletal malformations, hypotonia, arachnodactyly, omphalocoele and cardiovascular abnormalities. It is caused by mutation in the SKI gene located on 1p36.33-p36.32 and FBN1 gene located on 15q21.1. Diagnosis is based on clinical findings and confirmed if mutation is found. The aim of the case report is to present this extremely rare condition.



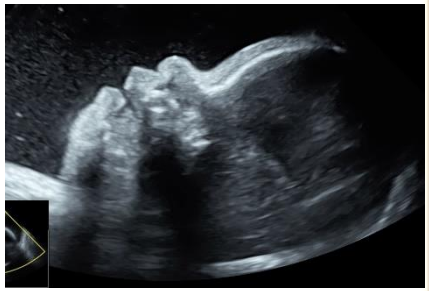


3D CT of head in the third month

Results: A boy with the weigth of 3360 g was born at 40 weeks. He presented with kraniosynostosis, turricephaly, microcephaly, skeletal anomalies, marfanoid habitus, hypotonia and kryptorchism. He underwent calvarial remodeling and shunting of the hydrocephalus at the age 4 and 5 months, respectively. Molecular genetic examination found a known pathogenic mutation c. 95T>C (p. Leu32Pro) in SKI gene. The boy died at the age of 10 month.



Second trimester ultrasound image (20 weeks)



Third trimester ultrasound image (30 weeks)

Methods: An 18-year-old primigravida underwent the first trimester combined test with the risk for T21 1/32, T18 1/257 and T13 1/159 (PAPPA-a 0.23 MoM, free β-hCG 0.81 MoM and NT 2 mm). No ultrasound anomalies were found. CVS was performed with normal array results. Subsequent ultrasound scan at 16 and 20 weeks revealed normal fetus with no organ anomalies. However, an ultrasound scan at 30 weeks found abnormal shape of fetal head (turricephaly), flat fetal profile, hypertelorism, a 9-mm dilatation of cavum septi pelucidi and asymmetric lateral ventricles. MRI confirmed the findings with the dominant turricephaly and hypertelorism. In addition to known anomalies MRI suspected a cleft palate.

At the age of five month