Introduction
Beare-Stevenson cutis gyrata syndrome is rare genetic disorder characterized by skin abnormalities (cutis gyrata, which causes a furrowed and wrinkled appearance) and the premature fusion of certain bones of the skull. This early fusion prevents the skull from growing normally and affects the shape of the head and face. Beare-Stevenson cutis gyrata syndrome is inherited in an autosomal dominant pattern.

Case report
A 26 year old woman, gravida 1, para 0 was referred to our center due to suspected fetal brain anomaly at 27+6 weeks of gestation. The nuchal translucency was above the 95th percentile (3.4mm at 13+2 weeks) and the result of maternal serum screening for Down syndrome was positive (1:27). The result of non-invasive prenatal testing was negative for trisomy 13, 18 and 21 at 16 weeks of gestation.

Her medical history and family history were unremarkable. Fetal ultrasound showed a cloverleaf-shaped skull, exophthalmos, frontal bossing and depressed nasal bridge. Genetic amniocentesis identified activation pointing mutation in c.1124A>G (p.Y375C) of FGFR2. Therefore, Beare-Stevenson cutis gyrata syndrome was confirmed.

Conclusion
Beare-Stevenson cutis gyrata syndrome can be diagnosed by prenatal ultrasonography and genetic study.