A case of 6p 24 deletion syndrome (OMIM #612582)
Ergin RN, Yayla M, Cigerciogullari E, Alanay Y
Bahçeşehir University Faculty of Medicine, Istanbul, Turkey

Objective
OMIM #6125826, namely 6p 24 deletion syndrome, is a contiguous gene deletion syndrome characterized by a wide spectrum of clinical presentations. Forkhead box protein C1 gene (FOXC1/FKHL7), encoding a member of the winged helix/forkhead family of transcription factors resides in the 6p 24 region and plays major role in embryonic developmental processes including somatic, cardiovascular, renal, ocular, skull, and cerebellar development. In this case report we present an antenatal case of 6p 24 deletion syndrome variant involving FOXC1 gene.

Methods
Case report.

Results
This is a case report of a 34 year old primergravida with uneventful past history who presented at 13 weeks of gestation for first trimester screening test. The nuchal translucency was increased (NT 4.4 mm by CRL 69mm) and a VSD was suspected. A CVS was performed and the karyotype was normal. (46 XY). At 16 weeks gestation cleft lip and palate was detected. Fetal echocardiography confirmed the suspected VSD at 18 weeks. The anatomy scan at 20 weeks showed, additional to the cleft lip and palate and the already diagnosed subaortic VSD, low set ears and nuchal oedema of 7 mm. Based on these findings an amniocentesis was performed. Array-CGH revealed a 9, 6 Mb interstitial deletion in the 6p25. 2p24. 1 region containing the gene FOXC1 and 119 Kb interstitial deletion at 9q22. 31. The pregnancy was terminated thereafter. The postmortem examination showed were turricephaly, hypertelorism, depressed nasal bridge, broad nasal tip, left sided cleft lip, low-set small ears, micrognathia, short neck, increased nuchal fold, short broad distal phalanges, broad thumbs, broad halluces and broad toes.

Conclusion
An amniocentesis should be performed by structural abnormalities even by normal karyotype in CVS. This is the only diagnostic option of rare syndroms, for example as in our case due to deletions.