Objective
The measurement of fetal NT at 10–14 weeks of gestation has been established as a sensitive, accurate and effective method of screening for chromosomal abnormalities. Additionally, increased NT has been reported to be associated with other structural defects, rare genetic syndromes and skeletal dysplasia. There is substantial evidence that in fetuses with increased NT and normal karyotype, the prevalence of skeletal dysplasia is greater. Achondrogenesis type II is characterized by severe micromelic dwarfism with small chest and prominent abdomen, incomplete ossification of the vertebral bodies, and disorganization of the costochondral junction. This form is an autosomal dominant trait occurring mostly as new mutations. However, somatic and germline mosaicism have been reported. We present a case of achondrogenesis with first trimester increased NT at our institute.

Methods
This is a case report.

Results
32-year-old, G3 P2 pregnant woman was referred to our clinic because of increased nuchal translucency. The sonography was performed at 11th gestational weeks. The fetal was CRL 44 mm compatible with 11 weeks 2 days and NT was measured as 3 mm. CVS (chorion villus sampling) was performed. The result of conventional karyotyping was normal. At 19 gestational weeks, mid-trimester fetal ultrasound scan was performed and micrognathia, micromelia, pyelectasia, low ear position were observed. All long bones measurements were compatible with 15 gestational weeks. At the request of the family, termination of pregnancy was performed at 20 gestational weeks.

Conclusion
Sonographers should be aware that an increased NT measurement at the first routine ultrasound examination should lead to a meticulous analysis of the fetal skeleton, after exclusion of chromosomal abnormalities.