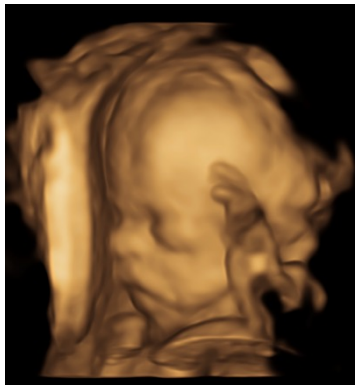
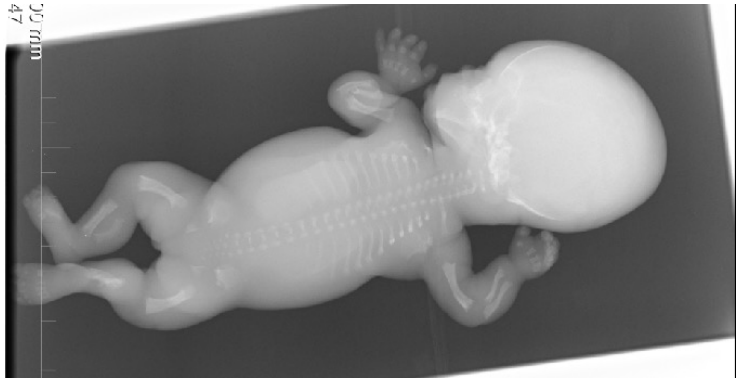
Thanatophoric dysplasia

Holub M.[1], Sekowská M.[1], Smetanová D.[1], Sobolová K.[1], Koudová M.[1]
1) Gennet, Center for Fetal Medicine and Reproductive Genetics, Prague, Czech Republic



Abstract
We present a case of thanatophoric dysplasia(TD). This case was diagnosed by ultrasonography and whole exom sequencing at our department.

Objective
Thanatophoric dysplasia is a rare skeletal dysplasia, but it is the most common lethal skeletal dysplasia. Skeletal dysplasias are a wide, heterogeneous group of diseases. The incidence of thanatophoric dysplasia is reported to be 1:20000 – 1:50000 at birth. TD is inherited in an autosomal dominant manner, it caused by mutation in FGFR3 gene on the short arm of chromosome 4. Typical ultrasound signs of TD are short limbs, narrow chest, short ribs, frotnal bossing, makrocephally, abnormalities of scul and spine. The major role in diagnostics have high-quality ultrasound examination in the 2nd trimester and modern genetic testing. Prognosis of TD is poor, most infants die after birth because of respiratory failure.



Results
We present a case of a 29 years old woman with a fetus in 20+6 weeks of gestation. She was gravida 1, para 0. The medical history of the woman was normal. She was not taking any medication. She underwent 1.st trimester screening with positive results, but NIPT was negative. Than, she continue for an ultrasound scan in 20+6 weeks of gestation, whitch showed short and abnormal limbs, narrow chest, atypical face with frontal bosing. We suspected a skeletal dysplasia. AMC was performed with normal array results, but we performed the exom. After week, findings of fetus did negative progresion. The couple decided to terminate the pregnancy. The results from exom confirm thanatoforic dysplasia. The autopsy and RTG of fetus agree with this diagnosis.

Conclusion
The major role in diagnostics of skeletal dysplasia has high-quality ultrasound examination in the 2nd trimester and modern genetic testing. The aim of the case report is showed that whole exom sequencing is method of choice, how to diagnose the type of skeletal dysplasia. Knowing the correct diagnosis is crucial for the future of the fetus and the couple.