A case of prenatal diagnosis of osteogenesis imperfecta type II

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Objective
Osteogenesis imperfecta (OI) is a heterogeneous group of diseases affecting type I collagen. It is characterized by bone fragility, multiple fractures, bowing and shortening of long bones. Both quantitative and qualitative abnormalities of collagen synthesis underlie the pathogenesis of osteogenesis imperfecta (OI). Typical features of the disease is micromelia associated with limb deformities. It is classified into types I, II, III, IV, V, and VI. Osteogenesis Imperfecta type 2 is the most serious form of the disease. Most of cases are due to the mutations in COL1A1 and COL1A2 genes, inherited in an autosomal dominant way.

Methods
We report a case of prenatally diagnosed lethal Osteogenesis Imperfecta type 2.

Results
A 28-year old primigravida was admitted to the outpatient clinic of Obstetrics and Gynecology on the 8th week of pregnancy. No significant previous history was documented, it was a spontaneous planned conception with a recommended preconception Folic acid administration. During the first trimester screening ultrasound, attention was paid to the short bones of the fetus. The examination demonstrated a bilaterally shortened femurs, but both the humerus bones were not visible. The fetal nuchal translucency thickness was 2.5 mm. First trimester serum markers were: PAPP-A: 0.269 MoM and free β-hCG: 1.605 MoM. The combined calculated risk was 1/25 [the sonographer was certified by The Fetal Medicine Foundation (FMF)].

The patient further underwent amniocentesis, which showed a diploid karyotype and no mutations that might suggest thanatophoric dysplasia. From the 16th week of pregnancy the upper and lower long bones in the limbs were difficult to visualize by ultrasound, however, both palms and feet were observed. The patient refused termination of pregnancy. The subsequent ultrasound scans showed both multiple, multi-level fractures of the fetal long bones, such as femurs or ribs, as well as also some extensive periosteal reactions. Other bone deformities were also observed such as foot distortion, arcuate bent limb bones with numerous thickening, indicating of bones scans. On the 34th week of pregnancy, due to premature rupture of membranes, the patient underwent a cesarean section and delivered a male neonate with body weight of 1200g. Fetus delivered in a bad condition, Apgar score was documented as 0/0 and unfortunately did not survive. A postmortem full body radiograph – “a babygram” was performed and confirmed the presence of distortion of the skeleton. Inhomogeneous spotted bones atrophy was also described.

Conclusion
Osteogenesis Imperfecta can be recognized prenatally. As intra uterine therapy of the disease is not plausible, obstetricians are forced to monitor the condition of the fetal skeleton or to offer termination of pregnancy. It is vital to properly recognize the type of skeletal dysplasia in order to prenatally outline the appropriate care. Type II OI is lethal, however, other variants of the disease are not and hence the importance of a careful multidisciplinary care to be outlined for these fetuses.