Introduction
Achondroplasia is the most common non-lethal skeletal dysplasia, with the prevalence ranging from 1:10,000 to 1:50,000 births. It is a genetic bone disorder and autosomal dominance is its pattern of inheritance. Individual with achondroplasia has mutation in fibroblast growth factor receptor-3 gene (FGFR-3), which is located on short hand of chromosome 4. Other skeletal dysplasias caused by same gene mutation are thanatophoric dysplasia and hypochondroplasia, therefore they share similar phenotypic abnormalities which can be used as differential diagnosis. Although other common skeletal dysplasias such as osteogenesis imperfecta and achondrogenesis can also be a differential diagnosis to achondroplasia. The gold standard method of diagnosis is DNA testing for mutations of FGFR-3 gene.

Case Presentation
A 27 years old G3P2 came to our clinic in her 38 weeks of gestation. Both of her previous pregnancies were unremarkable and also her past and family history. She came to our clinic with second opinion because her fetus has been diagnosed with shortened limbs, enlarged head and anomaly in fetal heart.

We did an ultrasound examination and showed a live male singleton pregnancy with breech presentation, shortened extremities and enlarged head. The transversebell diameter was equal to 38 weeks. The biparietal diameter was in accordance with 40±2 weeks and head circumference was 41±5 weeks, with enlarged lateral ventricle (15 mm). The distance of fetal orbital was in accordance with 36±5 weeks (biparietal diameter 59.3 mm and interorbital diameter 19.5 mm). Abdominal circumference was in accordance to 38±3 weeks. Femur length was shortened in accordance with 27±2 weeks. Tibial and fibular length were shortened, in accordance to 28±5 and 27±4 weeks. Radial and ulnar length were equal to 30±4 and 31±5 weeks respectively. Fetal heart was enlarged with cardiotrophic ratio of 0.64 without any significantly noted anomaly in its anatomy and heart rate was 136 beats per minute. Ratio of femoral length to abdominal circumference was 0.15 and ratio of thorax circumference to abdominal circumference were 0.59. Umbilical artery were in normal limit. The estimated fetal weight was 2,952 grams.

A term male neonate was subsequently delivered operatively at his 40 weeks with the presence of pediatrician, weighing 3350 grams and birth length of 46 cm with APGAR score of 9 at 1st minute and 10 at 5th minute. Neonate showed a prominent forehead with shortened limbs (Figure 1). The sign of demineralization is prominent falls in the fetal brain and the absence or decreased echogenicity of the fetal spine, which is usually found in hypophosphatasia and osteogenesis imperfecta.

Figure 1 - Term neonate delivered in operating theater (left) and in neonatal intensive care unit

Then the neonate was referred to neonatal intensive care unit for thorough examination.

Discussion
Achondroplasia is the most common non-lethal skeletal dysplasia with a prevalence of 1:10,000 to 1:350,000 births (1, 2). It is caused by mutation of fibroblast growth factor receptor-3 (FGFR3) gene that is located on the short arm of chromosome 4, which is the physiological regulator of linear bone growth. The receptor mutation affects bones derived from endochondral ossification. Its inheritance follows Mendelian autosomal dominant trait, although 80% of achondroplasia result from de novo mutation (2). Prenatal diagnosis of achondroplasia can be done by ultrasound usually third trimester when the length of the bones is below 5th percentile (1, 3). Prognostic finding of achondroplasia is late-onset rhizomelic shortening of the limbs and trident hand (4). Macrocephaly with frontal bossing can also be found in fetus with achondroplasia. Shortened femur can be used as initial step to distinguish between five most common skeletal dysplasia, such as thanatophoric dysplasia, osteogenesis imperfecta type II, achondrogenesis, achondroplasia, and hypochondroplasia (4, 5). After the shortened femur has been found in the initial fetal biometry examination, then complete fetal long bones measurement should be done to evaluate the type of short limb dysplasia (rhizomelia, mesomelia, acromelia, or micromelia).

Degree of bone mineralization should be assessed by examining the acoustic shadow below the bone as well as echogenicity of the bone.

Conclusion
Achondroplasia is the most common non-lethal skeletal dysplasia. It characterized by late-onset rhizomelic bone dysplasia and trident hand, also macrocephaly with frontal bossing, which can be detected by ultrasound. Fibroblast growth factor receptor-3 (FGFR3) which is located on short arm of chromosome 4, is the gene that is responsible for Achondroplasia. Its inheritance pattern follows Mendelian autosomal dominant, although 80% of the cases results from de novo mutation. The prediction of pulmonary hypoplasia by calculating the ratio of femur length and abdominal circumference that is formulized by Rahemtullah et al, is not reliable for this case.

Conflict of Interest
The authors declare no conflict of interest.

Reference