Pediatrics & Therapeutics
ISSN: 2161-0665

Zahouani et al., Pediatr Ther 2016, 6:4
DOI: 10.4172/2161-0665.1000i120

Clinical Image Open Access

Type II Thanatophoric Dysplasia

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Received date: Sep 21, 2016; **Accepted date:** Sep 29, 2016; **Published date:** Oct 03, 2016

Clinical Image

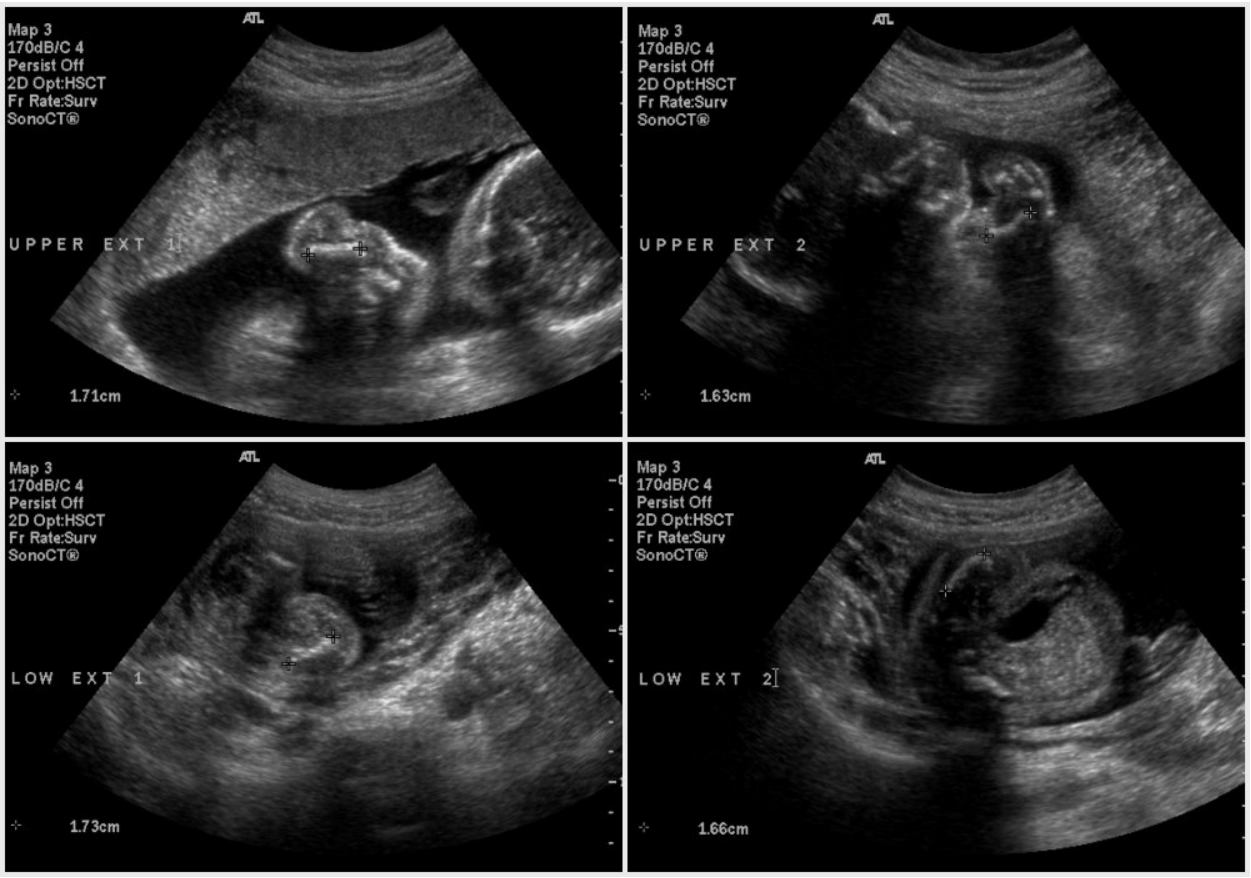

Figure 1: Ultrasound at 23 weeks showing Polyhydramnios and fetal morphology.

A baby girl was born at 32 weeks of gestation via vaginal delivery to a 28 y/o mother whose prenatal course was remarkable for ultrasound findings at 23 weeks suggestive of severe polyhydramnios and type II Thanatophoric Dysplasia and small compressed chest with severe pulmonary hypoplasia (Figure 1). MFM and Neonatologist counseled the mother about poor fetal neonatal outcome. The baby was born in footling breech presentation, was cyanotic with no respiratory effort, tone or reflexes but with few heart beats. The Apgar score was 1 at 1, 5 and 10 minutes. No resuscitation done, only comfort care provided and the infant died in mother's arm. In the delivery room, the support was provided and the mother refused autopsy. The infant weight was 1735 g, length was 34 cm, and head circumference was 31.5 cm. Physical examination revealed a large head with a prominent forehead in a cloverleaf shape, a dysmorphic face with a flat nose and very small extremities (Figure 2).

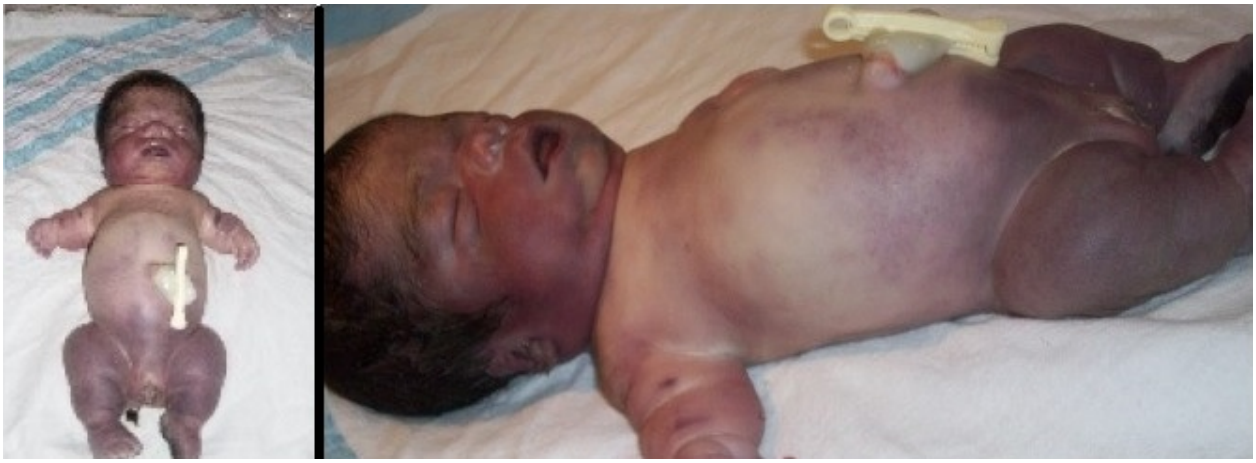

Figure 2: Infant born with typical features.

Thanatophoric Dysplasia (TD) is a severe congenital and lethal skeletal disorder with two different subtypes type I and type II. Both are lethal and are similar in clinical manifestations with minor variation particularly Type I with short and bowed long bones whereas in type II the head has a typical cloverleaf shape and the femur is short and straight [1]. In our case it was a type II and the cause of death was severe pulmonary hypoplasia. The incidence ranges between 1/20000 to 1/50000 births [2]. It is caused by de novo autosomal dominant mutation in fibroblast growth factor receptor 3 gene (FGFR3) located on chromosome 4, resulting in an activation of FGFR3 tyrosine kinase independently of ligands, leading to decreased apoptosis and increased proliferation [3]. Recurrence risk is not increased compared to the general population [1]. Nuchal brightness and limb shortness are ultrasonographic signs indicative of this condition during the first trimester [2]. Affected neonates are usually stillborn or die within the first few hours or days of life from respiratory failure with some rare cases of long term survival with respiratory support [3].

References

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