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Antenatal Diagnosis of a Case of Thanatophoric Dwarfism

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Abstract:

Maroteaux et al. described for the first time, in 1967, thanatophoric dwarfism (NT), an osteochondrodysplasia classified into two types: I and II. [1]. It is an extremely rare anomaly, the incidence of which is estimated at between one case in 20,000 and one case in 40,000 births. It is due to a mutation in the FGFR3 gene (fibroblast growth factor receptor 3) which is located on the short arm of the chromosome. Micromelic dwarfism, macrocrania and pulmonary hypoplasia due to thoracic constriction are clinically observed, as well as an appearance cranial latticeshaped.Training performed on data through October 2023. For type I, there is a reduction and curvature of the long bones, while they remain straight for type II. We also observe a sagging of the vertebral body accompanied by an expansion of the intervertebral spaces
Key words: micromelic dwarfism; psychological; ultrasound; pregnancy

Introduction:

Maroteaux et al. described for the first time, in 1967, thanatophoric dwarfism (NT), an osteochondrodysplasia classified into two types: I and II. [1]. It is an extremely rare anomaly, the incidence of which is estimated at between one case in 20,000 and one case in 40,000 births. It is due to a mutation in the FGFR3 gene (fibroblast growth factor receptor 3) which is located on the short arm of the chromosome. Micromelic dwarfism, macro crania and pulmonary hypoplasia due to thoracic constriction are clinically observed, as well as an appearance cranial lattice shaped. Training performed on data through October 2023. For type I, there is a reduction and curvature of the long bones, while they remain straight for type II. We also observe a sagging of the vertebral body accompanied by an expansion of the intervertebral spaces.4]. Ultrasound, often combined with x-rays of the uterine contents, facilitates early diagnosis. Its course always remains fatal and medical termination of the pregnancy should be considered in the event of diagnosis. Antenatal [5]. We then find ourselves faced with a double problem. This involves the differential diagnosis on medical imaging with other types of non-lethal fetal dysostoses, as well as psychological intervention with parents faced with situations where the desire is strongly pronounced. We report here a case of type I NT identified by ultrasound at the 26th week and followed until delivery in a 33-year-old woman, without children, who refused therapeutic interruption. pregnancy (ITG) Our objective is to recall the ultrasound and radiographic signs of this rare fetal malformation with a view to its early antenatal diagnosis.

Observation:

A 39-year-old woman consulted our structure for chondrodysplasia dwarfism in a pregnancy of 24 weeks + 5 days This patient had no medical or surgical history, it is a 2nd part having a notion of spontaneous miscarriage dating back 5 years and a child living vaginally aged 6 years with a birth weight of 2500 grams This pregnancy was not followed, unwanted, the date of the last period was July 28, 2022 the first ultrasound done on January 9, 2023 revealing an anomaly in the upper and lower limbs, funnel-shaped thorax with absence of visualization of the lungs Examination on admission found a conscious patient, normal-colored conjunctiva, no apparent physical abnormalities, height: 1.67 cm, weight 75 kg, Blood pressure: 12/07 normal tense, arterial pulse at 81 bpm, urine strips without abnormalities, without edema, afebrile Gynecological examination: no uterine contractions, firm cervix, intact membranes, pelvis and perineum without abnormalities An ultrasound was done finding: Biometry: bi-parietal diameter of 60mm, DAT: 56mm, abdominal circumference: 209 mm Anomaly in the upper and lower limbs, very narrow funnel-shaped thorax, absence of lung

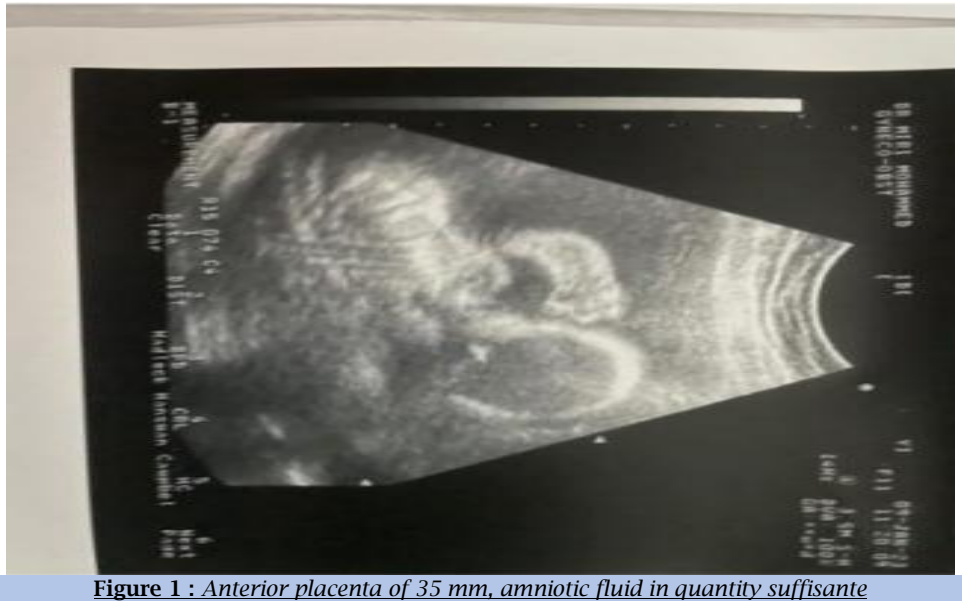
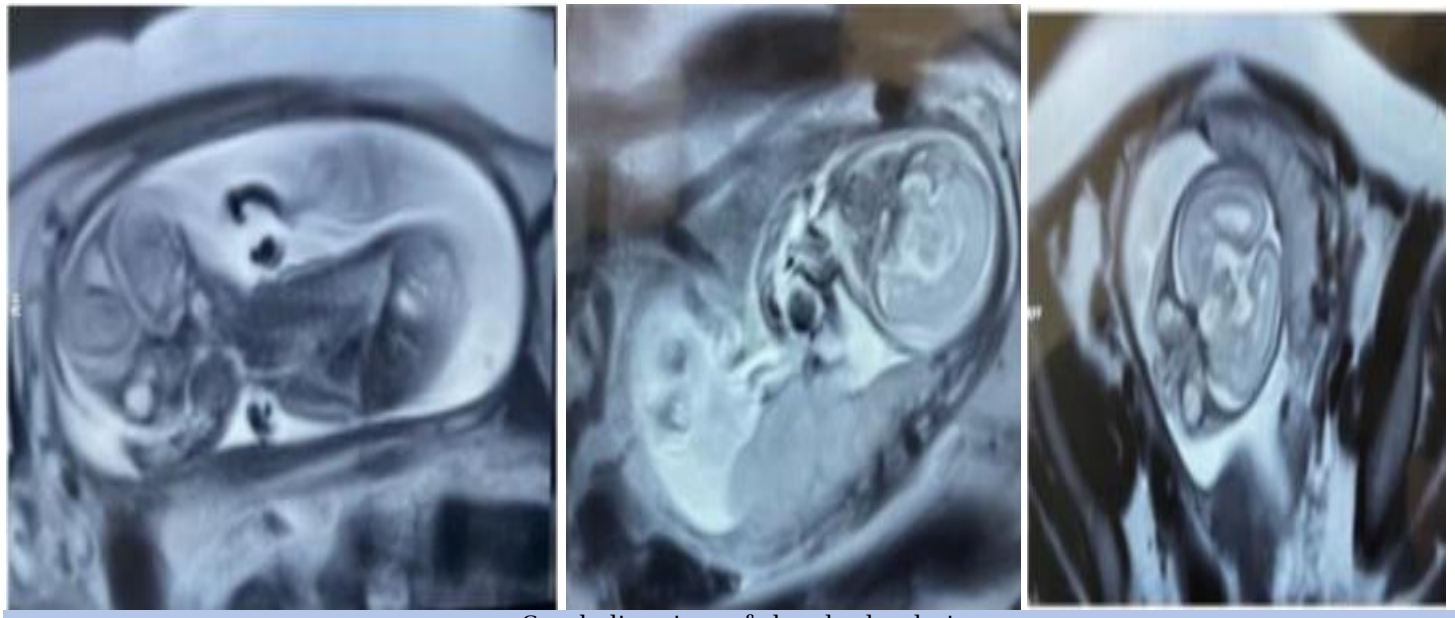


Figure 1 : *Anterior placenta of 35 mm, amniotic fluid in quantity suffisante*

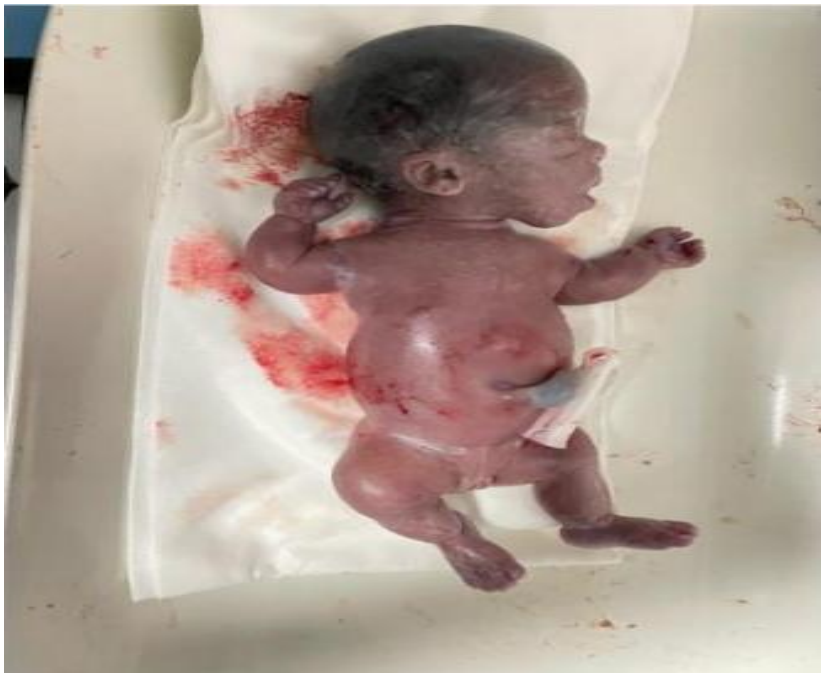
Additional fetal MRI:

objectifying a dysmorphic fetus by skeletal abnormality, namely a macro crania, a narrow thorax, rhizomelic type shortening of the upper and lower limbs Sufficient anterior placenta and amniotic fluid



Concluding signs of chondrodysplasia

After referring the parturient to the obstetric staff, a therapeutic termination of the pregnancy was decided.
The delivery was eutrophic without anomaly giving birth to a new female desex child weighing 952 g who died after thirty minutes



Discussion:

NT is a major and lethal fetal malformation. According to Machado et al. [4], the term thanatophore would be taken from the Greek mythology "thanatos" and would mean death personified. The life expectancy of newborns with NT is assessed at approximately one hour after birth by Noe et al. [3] who mentioned some rare cases of survival up to five and eight years. In our observation, the newborn with NT survived only 30 minutes. Severe respiratory distress linked to the narrowness of the rib cage with a lack of development of the lungs would be the cause according to Pietryga et al. [6]. The notion of consanguinity or related couple noted in the literature was not found in our case. In LahmarBoufaroua's study in Tunisia, consanguinity was found in 61% of cases.
Antenatal diagnosis of fetal malformations, whether lethal or not, should be the dread of every sonographer. For this, some authors recommend looking for early morphological abnormalities on first trimester ultrasound [7]. This ultrasound should emphasize nuchal translucency. According to Tonni et al. [8], nuchal hyperlucency is an early sign of TN. In our observation, the sonographer had only measured the thickness of the nuchal translucency without mentioning its echogenicity. We believe that the quantity of amniotic fluid must also be taken into account in the ultrasound exploration during the prenatal assessment. Indeed, polyhydramnios is a clinical and ultrasound sign constantly found in the literature as part of the signs associated with Nt The signs of NT as well as the classification into type I or II appear evident from the second trimester on two-dimensional ultrasound. This involves highlighting a macrocrania, a narrowness of the thorax, a prominent abdomen and extremely shortened limbs. In type I, the femur is curved while it is not in type II. Recent advances in technology with the advent of three-dimensional ultrasound have further facilitated the diagnosis of TN [9]. But the fear of making a mistake or confusing NT with achondroplasia, osteogenesis imperfecta, achondrogenesis or any other non-lethal micromelia would require coupling ultrasound with molecular biology which provides a definitive diagnosis [3,10].
In its absence, it would be desirable to associate an x-ray of the uterine contents with ultrasound [10]. Vertebral compressions and shortening bones of the limbs with their curvature in the shape of a "telephone handset" [11].

Conclusion:

NT is a major fetal morphological anomaly for which antenatal diagnosis is essential. In the absence of molecular biology, obstetric ultrasound sometimes coupled with radiography of the uterine contents makes it possible to make the diagnosis of NT and to eliminate other types of micromelic dwarfism. Certain accompanying signs such as polyhydramnios should attract the attention of both the clinician and the sonographer for a more in-depth and earlier search for NT. The therapeutic attitude

which consists of the medical termination of the pregnancy in front of a NT requires adequate psychological care.

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