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Tanatophoric Nanism: A Case Report and Review of The Literature

I.Alilou; K. Saoud; M. Mahmoud; N. Mamouni; S. Errarhay; C. Bouchikhi; A. Banani.

Corresponding author: Dr Alilou Ismail, dralilouismail@yahoo.com,
Service de gynécologie et obstétrique 1, CHU HASSAN II FES**,** MAROC.

Abstract*: Tanatophoric nanism is a very rare osteochandrodyasplasia, it affects one birth in 4000. It was first described by Marotaux et al in 1967[1]. Its evolution is towards a rapid death at birth. Its antenatal diagnosis is done by imaging which will allow a psychic preparation of the mother and propose an medical interruption of pregnancy. We report the case of a 29-year-old nulliparous woman who during the follow-up of her pregnancy, a routine ultrasound scan in the framework of the antenatal surveillance of her pregnancy carried out at 28 SA allowed to suspect the diagnosis of tanatophoric nanism, in front of very evocative images associating a macrocephaly, a shortened aspect of the limbs, a narrow thorax contrasting with a bloated abdomen. The course of the pregnancy was uneventful with delivery at 37 weeks of age of a male infant weighing 3100 grams who died after 2 days of life.*

Keywords: Tanatophoric nanism; micromyelia; antenatal diagnosis;

1. INTRODUCTION

Tanatophoric nanism is an osteochandrodyasplasia that is classified into 2 types I and II [1]. It is a rare malformation due to a genetic mutation that affects the FGFR3 gene located on the short arm of chromosome 4 [7]. Fibroblast growth factors (FGR) are proteins related to cell growth. Membrane receptors or FGFRs modulate their functions. FGFR3 is a bone growth inhibiting factor but stimulates bone differentiation.

They cause a very important shortening of the femurs and humeri, a very narrow thorax, a normal length of the trunk, a macrocephaly and a hydramnios.

The diagnosis is often made antenatally and is based mainly on ultrasound.

We report in our observation the case of a tanatophoric nanism of type II discovered in a nulliparous patient in a pregnancy of 28 SA during an obstetrical ultrasound.

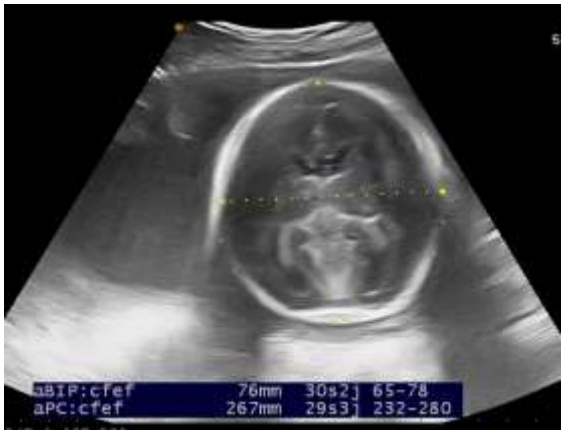
Our objective is to recall the modalities of antenatal diagnosis of tanatophoric nanism.

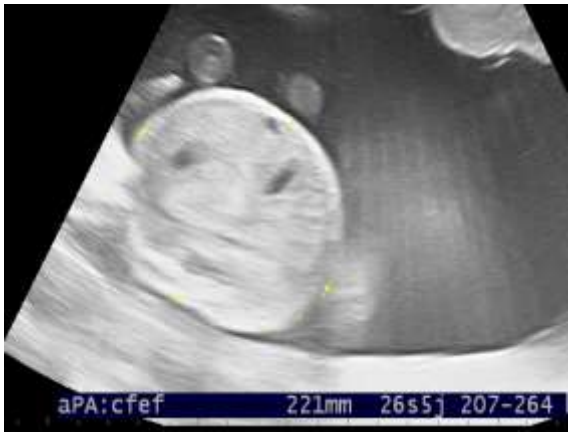
2. Medical observation

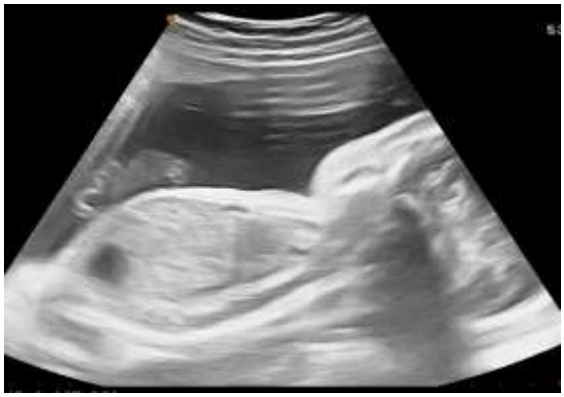
Mrs. I.L., 29 years old, G1P0, was followed up at the Gynecology and Obstetrics Department of the University Hospital of FES for her pregnancy. An ultrasound scan performed at 12 weeks' gestation was in favor of an intrauterine pregnancy with a CCL corresponding to the gestational age calculated from the date of the last period.

The nuchal translucency was of normal thickness. A second morphology ultrasound performed at 28 days of gestation showed a BIP at 76mm (figure 1a) with a PC at 267mm (figure 1b), an AC at 221mm (figure 1c), a shortened LF at 31mm, and an excess of amniotic fluid with a large cistern at 113mm. No other malformations were noted.

At 32 weeks of age, the hydramnios was still present on ultrasound, with an increased BIP in relation to the gestational age and shortened limbs. The RCF was oscillating and reactive. At 37 days of gestation, the patient delivered a dwarf male infant weighing 3100g, with a height of 35cm, and died at 2 days of age in a context of respiratory distress.







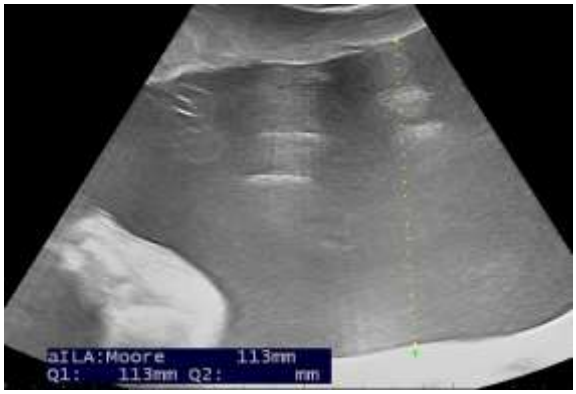


Figure 1 a, b, c, d : Patiente de 29 ans G1P0 à 28 SA selon la date des dernières règles. Échographie obstétricale bidimensionnelle réalisée dans le cadre d'un bilan morphologique classique. L'échographie objective un diamètre biparietal (a) mesurant 76 mm, soit 30 SA et un périmètre abdominal (b) 221mm soit à 26 SA, un hydramnios grande citerne(c) à 113mm, une coupe sagittale du fœtus(d) montrant le contraste de taille entre le tronc et l'extrémité céphalique.

3. Discussion

Tanathophoric nanism is a lethal fetal malformation, the time from birth to death of the affected neonate is about 1 hour [1], some rare cases of survival to 8 years have been reported [1], in our case the survival was 2 days. The explanation reported by Pietryga et al. was the poor thoracic ampliation necessary for the development of the respiratory system responsible for lethal respiratory failure [2].

Ante-natal research of fetal malformations is an essential element in the surveillance of any pregnancy, especially in the presence of a particular context. In our case, the notion of consanguinity was absent. According to the Tunisian study by Lahmar-Boufaroua, consanguinity was present in 61% of cases [3]. This search requires a meticulous ultrasound examination, the search for the thickness of the nuchal translucency and its echogenicity in the first trimester: hyperclarity is a sign in favor of NT [4]. The T1 ultrasound performed in our patient did not provide any information on its echogenicity. Subsequently, the signs in favour of a NT appeared obvious on the second and even more so on the third trimester ultrasound. We find a particularly severe micromelia, with a curved aspect in type 1, a trunk of normal length, a macrocephaly associated sometimes with a "cloverleaf" skull and an excess of amniotic fluid [5]. The diagnosis suspected on ultrasound must be confirmed by molecular biology [6]. In its absence, the risk is to take for NT another non-lethal micromelia. The realisation of an X-ray of the uterine contents is particularly interesting in these cases.

Once the diagnosis is made, an informed explanation to the mother of the prognosis and the risk of dystocia must be provided.

Figure 2 a, b: same patient. Delivery by vaginal route at 37th SA of a dwarf baby with the same characteristics as those described on the ultrasound.

4. CONCLUSION:

This foetal malformation whose diagnosis must be made in ante-natal is lethal and calls for ultrasound and molecular biology failing an X-ray of the uterine contents. In our context where the realisation of a medical interruption of pregnancy is difficult, the aim of this diagnosis is especially to prepare the mother psychologically and to ensure an adequate obstetrical and psychological follow-up.

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