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Lethal Thanatophoric Dwarfism: A Case Report

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Abstract

Thanatophoric dwarfism is a rare and lethal congenital bone dysplasia characterized by a narrow thorax, extreme limb shortening, and severe pulmonary hypoplasia. We report the case of a 41-year-old patient who gave birth to a newborn with these abnormalities.

Keywords: Thanatophoric dwarfism, Skeletal dysplasia, Lethal, Prenatal diagnosis, FGFR3

Introduction

Thanatophoric dwarfism (or thanatophoric dysplasia) is a lethal condition belonging to the group of skeletal dysplasias, linked to a mutation in the FGFR3 gene (Fibroblast Growth Factor Receptor 3). It manifests as a very narrow chest, severe shortening of the long bones, macrocephaly, and often spinal abnormalities. The prognosis is poor, with most newborns dying within the first few hours or days of life due to acute respiratory failure caused by pulmonary hypoplasia.

Observation

Ms. F.H., aged 41, IGIP, was admitted to the maternity ward of Abderrahim El Harouchi University Hospital for delivery of a malformed fetus. The morphological ultrasound performed during the third trimester revealed: ventriculomegaly measuring 10.33 mm, a short spine, a narrow thorax, hypoplastic lungs, and short limbs: short and curved femur, suggesting lethal thanatophoric dwarfism.

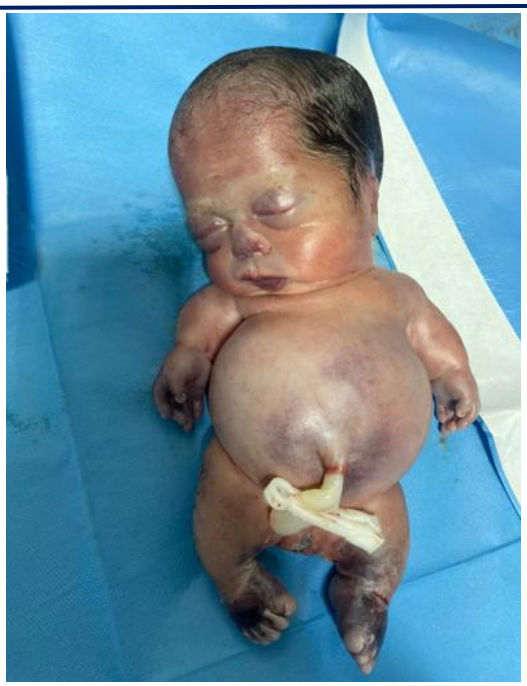
The patient gave birth vaginally to a premature female newborn weighing 1050 g at 34 weeks + 6 days of amenorrhea, with an Apgar score of 1/10 at birth. The newborn died at 5 hours of life, with severe respiratory distress.

The newborn's clinical appearance was characteristic of thanatophoric dwarfism: very short limbs, barrel chest, prominent abdomen, and macrocephaly. The diagnosis was confirmed clinically.

An autopsy was not performed at the request of the family.

Discussion

Thanatophoric dwarfism is one of the most severe skeletal dysplasias, with an estimated incidence of 1/20,000 to 1/50,000 live births. It is transmitted in an autosomal dominant pattern, but the majority of cases are due to de novo mutations **[1]**.



Two types are described: Type I: bucket-handle femurs, without cloverleaf skull; Type II: straight femurs, with characteristic cloverleaf skull **[2]**.

The diagnosis is usually possible from the second trimester onwards by morphological ultrasound, which reveals severe shortening of the long bones, a narrow thorax, and sometimes macrocephaly or ventriculomegaly.

In our case, the ultrasound abnormalities strongly suggested this condition. The association with a polymyomatous uterus may have complicated ultrasound monitoring, but there was no direct etiopathogenic link with the dysplasia **[3]**.

The prognosis is invariably fatal due to major pulmonary hypoplasia. No curative treatment exists to date. Management is essentially palliative and multidisciplinary, focusing on parental support and genetic counselling **[4]**.

Conclusion

Thanatophoric dwarfism remains a rare and devastating condition, with prenatal diagnosis based on morphological ultrasound and confirmed by molecular analysis of the FGFR3 gene. Appropriate psychological and genetic support is essential for affected couples, in order to anticipate the risk of recurrence and provide support during the period of perinatal bereavement.

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