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A case of thanatophore dwarfism

Karima M, Rania B, Syrine M, Ghofrane F, Chiraz E
Mahmoud El Matri, Ariana, Tunisia

Objective

We report a case of Thanatophore dwarfism (TD) collected in our department to share our experience and to highlight the value of regular prenatal follow-up and diagnosis.

Methods

A case report.

Results

This was a 34-year-old woman (Gravida 3, para 3, with two healthy living children) who came to our department for an obstetrical ultrasound at 26 weeks' gestation. It was a poorly monitored pregnancy. She had no family history of known congenital malformations. The obstetrical ultrasound obstetrical ultrasound revealed a female fetus (BIP = 66 mm, 27 SA). 66 mm, as 27 SA puts the percentiles, is better; a thorax that is little developed, an abdomen of normal volume (CA = 229 mm, 25 SA), puts the percentiles better, and especially a shortening of the bones of the four limbs with femurs, which are curved (LF = 19 mm, 16 SA), puts the percentiles better, and with a ratio LF/BIP = 29,75. There were no other associated morphological abnormalities. The diagnosis of type I TD was made based on these findings, and the couple was informed in the presence of a psychologist of the fatal outcome. We continued ultrasound monitoring of the pregnancy until 38 weeks' gestation, with the appearance of an excess of amniotic fluid. A caesarean section was scheduled at 38 weeks' gestation for a bi-scarred uterus. She gave birth to She gave birth to a dwarf female infant, 3000 g in weight and 36 cm long, who was transferred to the neonatal intensive care unit. She died in her second hour of life.

Conclusion

In view of the fatal outcome, efforts should be made to diagnose DT. The medical termination of the pregnancy is the decision of the couple after a fair and informed discussion.