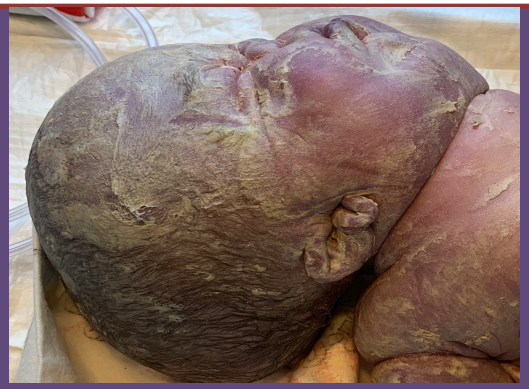
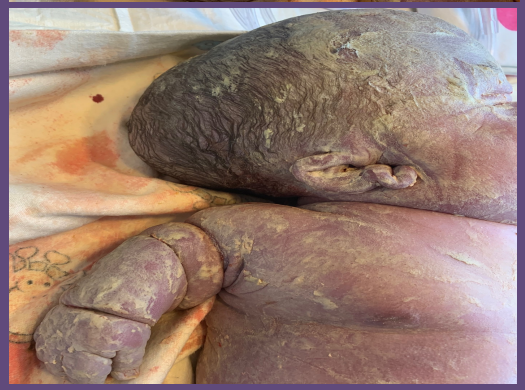
Thanatophoric dysplasia- case report ID 4243

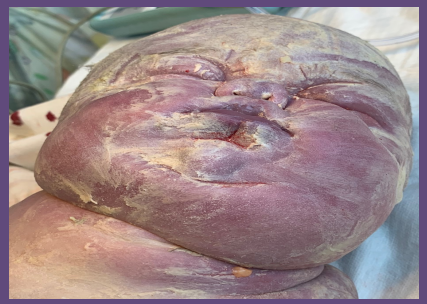
Migda Michał, Migda Marian
[1]Clinical Unit of Obstetrics, Women's Disease and Gynecological Oncology, United District Hospital, Collegium Medicum University of Nicolaus Copernicus in Toruń, Poland , [2]Civis Vita Medical Center Toruń, Poland,

Objective

Thanatophoric dysplasia is a severe skeletal disorder characterized by extremely short limbs and folds of redundant skin on lower and upper extremities. Other features are narrow chest, short ribs, underdeveloped lungs and enlarged head with large forehead and wide-spaced eyes. There are two major forms of this conditio. Type I – mostly distinguished by curved thigh bones and flattened bones of spine (platyspondyly). Type II is characterized by straight thigh bones and moderate to severe skull anomalies called cloversleaf skull. The frequency of this condition is 1:20000 to 50000 newborns. Type I is more common than type II. The cause of this dysplasia is mutation in FGR3 gene







Pict.1,2,3 Postpartum images

Case report

24-year old patient was admitted to our Unit in 39 weeks of gestation for delivery. Fetus was diagnosed with lethal thanatophoric dyplasia type I. Due to legal issues patient could not undergo termination of pregnancy and had to continue the pregnancy. At the admission ultrasound was performed. Fetus was in pelvic presentation. Unfortunately due to extreme head and abdomen size patient was consulted to delivery by cesarean section. We delivered a female fetus 4470g. Newborn died 1 hour after delivery

Conclusion

Thanatophoric dysplasia is a rare and severe condition and still most common life-threatening form skeletal dysplasia. Is and autosomal dominant condition but the majority of probands are de novo FGR3 mutations. The diagnosis is established with clinical and/or radiologic features. Most individuals die in the perinatal period because of the multisystem complications of the disorder. Newborns who survive require long-term respiratory support (typically with tracheostomy and ventilation). Treatment goals in perinatal period are to avoid pregnancy complications like prematurity, polyhydramnios, delivery complications from macrocephaly and rigid neck. A cesarean section may be considered to avoid maternal complications.