A case of I Cell disease

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Objective

To report a case of a fetus affected with I Cell disease, with a known family history.

Methods

We reviewed the family history, the ultrasound scans and the outcome of this pregnancy and compared to the literature of I Cell disease.

Results

A 24 year old woman, with a previous history of a child diagnosed with I Cell disease, was referred to our unit at 30 weeks of gestation. Genetic counselling was offered to the couple before the second pregnancy and declined. The pregnancy was redated in the first trimester scan and invasive testing was again offered and declined. At the 30 weeks growth scan there was polyhydramnios, from the 3D evaluation of fetal face there were signs of coarse facial features , micrognathia and flat nasal bridge and the femur and the humerus were below the 5th centile and hypo echogenic. The woman had a spontaneous vaginal delivery of a female baby at 37 weeks. Metabolic tests were performed and showed elevation of Beta-glucuronidase and other metabolic products suggestive of I Cell disease. Histological examination of cultured fibroblasts with a phase contrast microscope demonstrated the presence of numerous granular inclusions. Lees et al. and Yuksel at al. showed that short long bones, with periosteal cloaking and polyhydramnios are ultrasound markers for I cell disease. In our case, we also noticed abnormal ossification of long bones.

Conclusion

I-cell disease is inherited as an autosomal recessive genetic trait and is caused by a mutation in the GNPTA gene that leads to a deficiency in the enzyme UDP-N-acetylglucoseamine-1-phosphotransferase and low levels of intracellular lysosomal enzymes. Prenatal diagnosis is possible using enzyme analysis of samples from chorionic villus or amniotic fluid. The life expectancy of children with I-cell disease is poor, as death occurs between the fifth and seventh year. Progressive mental retardation, coarse facial features and skeletal abnormalities are the main characteristics. Without family history there no specific ultrasound findings. However, in cases of family history, like ours, ultrasound findings, like short femur and humerus, polyhydramnios and coarse facial features become more significant.