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Prenatal sonographic diagnosis of mucolipidosis type II in a family with a previous history of unexplained neonatal death

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Objective

Mucolipidosis type II (ML II) or Leroy I-cell disease is an autosomal recessive lysosomal storage disease that results in death within the first decade due to cardiorespiratory failure. Prenatal diagnosis of I-cell disease is possible using enzyme or DNA analysis of chorionic villi or amniotic fluid. Thus far, prenatal testing has been succesfully performed in those with a previous history of an affected child and our case is the first prenatally diagnosed one due to the associated sonographic findings.

Methods

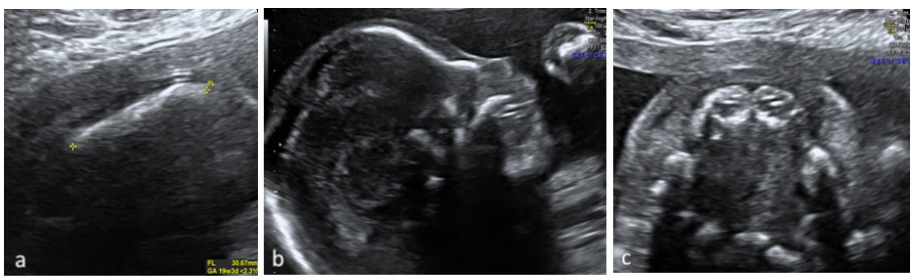
This is a case report.

Results

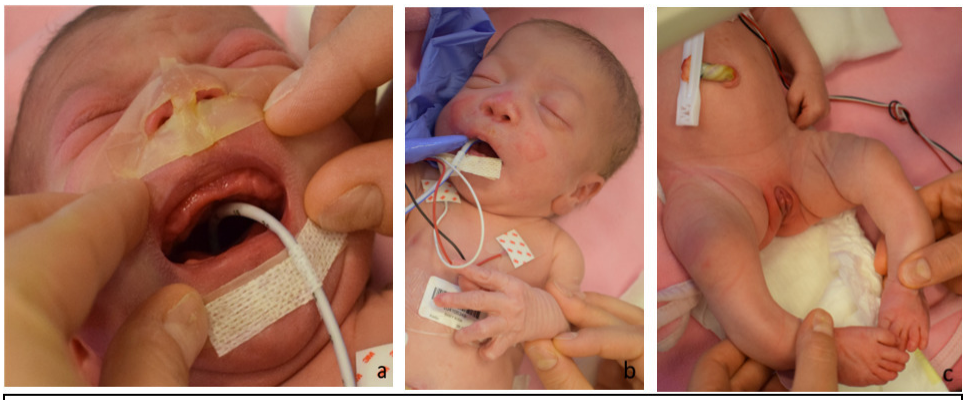
A 36-year old woman was referred at 23 weeks of gestation as the femur length was below the fifth centile. She had a history of one first trimester miscarriage and one caeserean section at term for a male baby who died at around 40 days of age. The cause of death was unknown and neither autopsy nor further diagnostic tests were performed. The couple were first degree cousins. At 23 weeks' gestation, the lengths of all tubular bones were below the first centile whereas head measurements, abdominal and thoracic circumferences were within the normal range. The diaphyses of both femora were broad and there was slight bowing. The facial profile was flat with a long philtrum. On axial scan, hypertelorism was noted and the maxilla appeared depressed in the middle part. There were no other structural abnormalities. As cordocentesis revealed normal karyotype, the parents were asked about the postnatal phenotypical traits of the first child. The history of probable gingival hypertrophy in the first child led to genetic testing for ML II which revealed homozygous c. 3503_3504delTC mutation in exome 19 of GNPTAB gene confirming the diagnosis. After counseling, the parents opted to continue with the pregnancy and a female baby with a birthweight of 2240 g was delivered by repeat caeserean section at 39 weeks. Coarse facial features were noted along with proptosis, long philtrum, gingival hypertrophy and retrognathia. Furthermore, postnatal ultrasound demonstrated the bowing, periosteal cloaking and dysplasia of the diaphyses of the long bones, that are characteristic of I-cell disease.

Conclusion

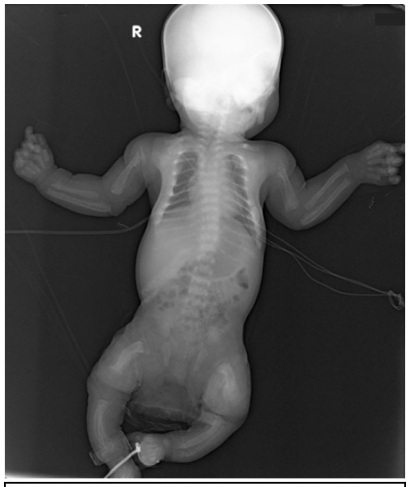
ML II is a rare lysosomal storage disease that may have perinatal manifestations such as non-immune hydrops fetalis, growth restriction, cardiomyopathy, short and broad femora, and dysmorphic facial features. In view of this case and a small number of other cases presenting with prenatal findings, ML II should be considered in the differential diagnosis of short and/or abnormal (broad and/or bowed) femora, particularly in the absence of characteristic findings associated with other skeletal dysplasias and chromosomal abnormalities.



Two dimensional images demonstrating (a) broad and short femur, (b) flat facial profile, and (c) axial scan of depressed maxilla of the fetus.



Images of the neonate; (a) gingival hyperplasia, (b) coarse facial features, (c) short and bowed bones.



Babygram demonstrating characteristic periosteal cloaking of the long bones.