A case of Neu Laxova syndrome

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Objective

Neu-Laxova Syndrome (NLS) is a lethal, autosomal recessive multiple malformation syndrome characterized by ichthyosis, marked intrauterine growth restriction (IUGR), microcephaly, short neck, central nervous system (CNS) anomalies, limb deformities, hypoplastic lungs, edema, and abnormal facial features. We report a case of NLS diagnosed prenatally by ultrasonography examination.

Methods

Case presentation: A 31 years old gravida 6 para 4 woman was referred to our clinic at 34 weeks gestation because of severe IUGR. The parents were first cousins and their medical history was unremarkable. No history of any genetic or structural anomalies was noted in herself, her husband, and in the family. Ultrasound obtained at 34 weeks weeks revealed a live fetus with severe IUGR (fetal measurements were consistent with 24 weeks), subcutaneous edema, micrognathia, polyhydramnios, microcephaly, lissencephaly, cerebellar hypoplasia, limb deformities, face anomalies including flattened nose, severe proptosis with ectropion and bilateral cataract.

Results

The mother presented at 34 weeks of gestation in active labor. The fetal heart monitor revealed late decelerations, the decision was made to deliver the infant by cesarean section. She delivered a 1000 gram female fetus. The baby died 2 days later. Postmortem examination by genetic specialists confirmed the diagnosis of NLS.

Conclusion

Prenatal diagnosis of NLS is important because of extremely poor prognosis. Parents should be informed about inheritance therefore genetic counseling is mandatory.