عنوان مقاله:

New Insights on Genetic Features of Neu-Laxova Syndrome

محل انتشار:

مجله علمی ناباروری ایران, دوره 8, شماره 1 (سال: 1396)

تعداد صفحات اصل مقاله: 3

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خلاصه مقاله:

ABSTRACTBackground: This study aimed to present a rare case of Neu-Laxova syndrome (NLS) and review the newly revealed genetic features of the disease in hopes to find a way for early interventions.Case report: Female newborn with NLS was born at 30 weeks of gestation to consanguineous parents. The last prenatal ultrasound imaging revealed severe intrauterine growth restriction and microcephaly without polyhydramnios. The newborn had significant dysmorphic features, such as microcephaly, slanted forehead, protruding eye, flattened nose, micrognathia, cleft palate, ichthyosis skin, edematous hands and feet and flexion contractures of the joints. Moreover, she had the usual female karyotype. Results of plain x-ray imaging demonstrated microcephaly, kyphosis, and arthrogryposis.Conclusion: According to the results of this study, NLS is a severe serine deficiency disorder. Given the confirmation of the possibility of diagnosing NLS early in gestation by several studies, it is suggested that early maternal supplementation with serine and glycine be used in families at risk of this disease or those who are diagnosed in early gestation with NLS in order to decrease the severity and fatality of the disease.

کلمات کلیدی:

Deficiency, Microcephaly, Serine