

Prenatal sonographic findings of Neu-Laxova syndrome :a genetically confirmed case

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Background :

Neu-Laxova syndrome (NLS) is a rare lethal autosomal recessive disorder with characteristic craniofacial abnormalities, severe fetal growth restriction, limb contractures, and ichthyosis. Prenatal diagnosis is challenging because of its rarity.

Clinical presentation:

28-year-old primigravida. Referred at 30 weeks because of severe fetal growth restriction. Ultrasound showed microcephaly, facial dysmorphism, limb contractures and persistent open mouth. 3D ultrasound confirmed the characteristic facial appearance.

Genetic Findings :

Whole-exome sequencing confirmed Neu-Laxova syndrome

Discussion Combined 2D/3D ultrasound with genetic testing enabled accurate prenatal diagnosis. Early diagnosis improves parental counselling and pregnancy management.

Conclusion :

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microcephaly



Contracture limb



Persistent open mouth and eyes



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