

EP07.30 Prenatal Diagnosis of TUBB3-Relate Tubulinopathy

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Abstract

Tubulinopathies, rare prenatal disorders caused by tubulin gene mutations, are characterized by severe cerebral malformations. This case report presents a prenatal diagnosis of TUBB3-associated tubulinopathy to enhance awareness among prenatal practitioners. A 33-year-old woman underwent fetal ultrasound at 23⁺⁵ weeks' gestation at Peking University First Hospital Ningxia Women's and Children's Hospital. The scan revealed midline brain curvature, a distorted cavum septum pellucidum, an oblique platform-like left sylvian fissure, left lateral ventriculomegaly, a congenital pulmonary airway malformation (CPAM), and a right ventricular echogenic focus. Whole-exome sequencing (WES) identified a heterozygous missense mutation in the TUBB3 gene (chr16:89935094; exon 4; NM_006086.4:c.643C>G, p.Leu215Val), resulting in the substitution of leucine with valine at amino acid position 215. This variant was absent from public and population-specific genomic databases. The pregnancy was terminated at 26+6 weeks' gestation. This case highlights the utility of prenatal ultrasound in detecting tubulinopathy-associated anomalies and underscores the necessity of integrating genetic testing, such as WES, for accurate diagnosis and prognostic evaluation. Early multidisciplinary assessment is critical for guiding clinical decisions in such rare fetal disorders.



1.Ultrasound Imaging of Fetal Cranium and Thorax at 23+5 Weeks Gestation: A-C: Transabdominal transverse views of the fetal brain. D-H: Transvaginal (TV) coronal views of the fetal brain. I: Transverse thoracic view demonstrating a Congenital Pulmonary Airway Malformation (CPAM Type II) within the left fetal thoracic cavity.

2.Fetal cranial MRI performed at 25+6 weeks' gestation revealed: Altered sulcation in the left cerebral hemisphere, Mild dilatation of the left lateral ventricle, Curvature of the cerebral midline.

3.Single Nucleotide Variant and Small Insertion/Deletion (InDel) Analysis Depicting the Fetal TUBB3 Mutation(24Y18690: Proband/Fetus; 24Y18690FU0: Father; 24Y18690MU0: Mother)

