Clinical outcomes of fetuses with chromosome 16 short arm copy number variants

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Abstract

Objective: The short arm of chromosome 16 consists of several copy number variants (CNV) that are crucial in neurodevelopmental disorders; however, incomplete penetrance and diverse phenotypes after birth aggravate the difficulty of prenatal genetic counseling. Design: Case series. Setting: This study uses data from National Taiwan University Hospital. Sample: We screened 15,051 pregnant women who underwent prenatal chromosomal microarray analysis between July 2012 and December 2017. Methods: Patients with positive array results were divided into four subgroups based on the type of mutation identified on screening (16p13.3, 16p13.11, 16p12.2, and 16p11.2), and the maternal characteristics, prenatal examinations, and postnatal outcomes of different cases were reviewed. Main outcome measured: Postnatal prognosis. Results: Chromosome 16 CNVs were identified in 35 fetuses, including four with 16p13.3 CNVs, 22 with 16p13.11 CNVs, two with 16p12.2 microdeletions, and seven with 16p11.2 CNVs. Of the 35 fetuses, 17 delivered without early childhood neurodevelopmental disorders, three developed neurodevelopmental disorders during childhood, and 11 were terminated. Conclusion: Incomplete penetrance and variable expressivity make prenatal counseling challenging. We report a few cases of de novo 16p CNVs without further neurodevelopmental disorders.

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