

[Prenatal diagnosis of Jacobsen syndrome in a fetus carried by a pregnant woman with intellectual disability]

Abstract

Objective: To assess the value of combined cytogenetic and molecular techniques for the prenatal diagnosis of a pregnant woman with intellectual disability (ID).

Methods: The fetus and its parents were subjected to G-banding karyotyping analysis, single nucleotide polymorphism array (SNP-array) and fluorescence in situ hybridization (FISH) analysis.

Results: G-banding karyotype analysis revealed that the woman has carried a chromosomal microdeletion 46,XX,del(11)(q24), and the fetus was a carrier of 46,XN,del(11)(q24)mat. Subsequent SNP-array and FISH analysis of the pregnant woman indicated that the microdeletion has mapped to 11q24.1-q25. Both the pregnant woman and her fetus were diagnosed with Jacobsen syndrome.

Conclusion: Combined use of cytogenetic and molecular genetic techniques can facilitate diagnosis of patients with intellectual disability.

Guo C, Wang J, Tang Y, Shi H, Liu J, Zhao L. [Prenatal diagnosis of Jacobsen syndrome in a fetus carried by a pregnant woman with intellectual disability]. *Zhonghua Yi Xue Yi Chuan Xue Za Zhi*. 2019 Aug 10;36(8):826-288. Chinese. doi: 10.3760/cma.j.issn.1003-9406.2019.08.018. PMID: 31400138.

