Prenatal diagnosis and genetic counseling of FGFR3 gene related fetal skeletal dysplasia—with analysis of 13 clinical cases

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Introduction

Objective: To investigate the FGFR3 (fibroblast growth factor receptor 3) related fetal short limb dysplasia, and provide genetic counseling depending on gene diagnosis.

Methods: 13 cases detected with FGFR3 gene heterozygous mutation were enrolled in our study. Samples of fetal umbilical cord blood, amniotic fluid, and/or aborted tissue were collected in all cases. Blood samples were taken from the parents for verification using Sanger sequencing. All cases were screening by ultrasound, X-ray and pathological analyses.

Results: 13 cases with FGFR3 gene heterozygous mutation were distributed in 4 regions, c.742C>T (p.Arg248Cys), c.1144G>A (p.Gly382Arg), c.1124A>G (p.Tyr375Cys) and c.2426G>C (p.X809S, 101); 9 were diagnosed as fatal achondroplasia type I (TD); 4 were diagnosed as achondroplasia (ACH). Sanger sequencing confirmed that none of these mutations were carried by any parent.

Conclusion

The gene detection of FGFR3 mutation can eventually clear classification of fetal short limb dysplasia, so as to provide more accurate prenatal diagnosis and genetic counseling.