Molecular genetic analysis of 3 patients featuring mental retardation and developmental delay

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Objective

To explore the clinical application value in genetic diagnosis for children featuring developmental delay and mental retardation applying single nucleotide polymorphisms array.

Methods

The karyotype of each child was analyzed with G-banded and SNP array. The validation of parents was continued in order to make clear sources of variations when necessary.

Results

Case 1 and case 2 karyotype analysis results were 46, XX. Karyotype analysis of case 3 was 46, XY, del(7)(pter→q36.1). SNP array has detected a 7.6 Mb deletion at 15q11.2-q13.2 in case 1, but no deletion or duplication was detected in her parents. SNP signal linkage analysis found that the deletion was inherited from his father. SNP array has detected a 1.7 Mb microdeletion at 1q21.1-q21.2 in the case 2 and a 9.9Mb deletion at 7q36.1-q36.3 in the case 3.

Conclusion

SNP Array combined with chromosome karyotype analysis technology provides a detailed diagnostic information of chromosomal abnormality. SNP array can be the preferred method of genetic testing for children featuring agnogenic developmental delay and mental retardation.