First Familial Occurrence of Prader-Willi Syndrome in China: Two Cases and Family Studies

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Introduction

• To present the first reported two cases of familial PWS in China.

• To carry out familial studies to analyze different underlying cytogenetics and molecular genetics mechanisms and to formulate a comprehensive summary of this rare condition.

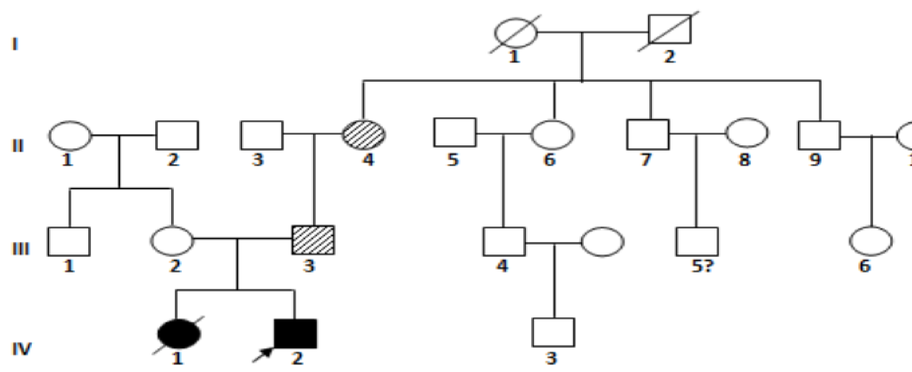
Methods

• Detailed clinical features of the probands and the clinical history of other affected family members were observed and described.

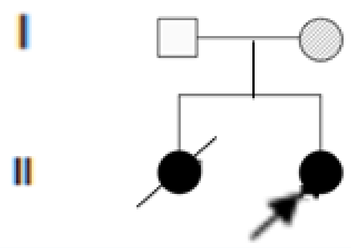
• The genomic DNA isolation and purification from whole blood was applied to the cytogenetic studies and molecular genetics investigations-high-resolution microarray analysis-Single Nucleotide Polymorphism (SNP) Array and Methylation Specific-Multiplex Ligation Probe Amplification(MS-MLPA) analysis.

Results

1、Case 1: The proband was a 1-day-old boy. He and his sister presented the characteristic features of PWS. They have one suspicious paternal cousin with some autistic type behaviours. SNP array and MS-MLPA detected a paternally transmitted submicrodeletion , 417 kilobase pairs (kbp) in size, at 15q11.2-q13 region: array15q11.2(24,963,375-25,380,656)x1, which was verified to pass through the paternal line from the patients' father and paternal grandmother.



2、Case 2：The affected girl is 32 months old, and she had one sister-both of whom fulfilled the diagnostic criteria for PWS. Cytogenetic studies of karyotype analysis revealed that the patients and their unaffected mother shared the same chromosome abnormality-45,XX,rob(15;15)(q10;q10) due to a translocation involving maternal chromosome 15 and hence effective maternal-origin uniparental disomy for the PWS region.



Conclusions

• Molecular genetics investigations are the gold standards for the molecular diagnosis of PWS.

•The familial occurrence of PWS suggests a wide clinical variability of severity within an affected family and a recessive mode of inheritance. The awareness of familial PWS is of great value for early and accurate diagnosis and administration of appropriate therapy.

• Conducting relevant family researches is necessary for understanding the mechanism of familial inheritance, estimating the recurrence risks to provide more corresponding genetic counseling on birth guidance and prenatal diagnosis.

References

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