Analysis of genetic mutations in a Chinese patient affected with Noonan Syndrome

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Objectives:

The aim of this study was to detect potential gene mutation of Noonan Syndrome in a Chinese family.

Methods:

Patient with clinical diagnosis and parents were analyzed in this study. The analysis included medical histories, clinical analysis, and genetic tests. ThePTPN11,KRAS,SOS1,RAF1,NRAS,BRAF,RIT1,SOS2, LZTR1,SHOC2,CBL,NF1 gene was sequenced to identify the pathogenic mutation responsible for the development of Noonan Syndrome by PCR and Sanger.

Results:

A novel mutation c.2600A>G（p.N867S） of the SOS2 gene was found in the patient, but not in his parents. The same mutations were not found among 100 healthy controls.

Conclusions:

A novel SOS2 c.2600A>G（p.N867S）mutation can be a cause of Noonan Syndrome in Chinese. We think that genetic studies to may assist in making Noonan Syndrome diagnosis and providing the consultant for their families.The novel mutations have enriched the mutation spectrum of the SOS2 gene.

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Growth and syndromes (to include Turner syndrome)

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