

# Analysis of genetic mutations in a Chinese pedigree affected with idiopathic hypogonadotropic hypogonadism Syndrome

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## Objectives:

The aim of this study was to detect potential gene mutation of idiopathic hypogonadotropic hypogonadism Syndrome (IHH) 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. A Disorder of Sexual Development (DSD) gene panel was applied to identify the pathogenic mutation responsible for the DSD and verified by Sanger.

## Results:

A novel mutation c.533G>C (p.W178S) of the PROKR2 gene was found in the patient and his father. The same mutations were not found among 100 healthy controls.

## Conclusions:

A novel mutation c.533G>C (p.W178S) of the PROKR2 gene mutation can be a cause of IHH in Chinese. We think that genetic studies to may assist in making IHH diagnosis and providing the consultant for their families. The novel mutations have enriched the mutation spectrum of the IHH gene.

