The Research about SF1 gene abnormality in 45 children with micropenis

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Background

Micropenis are the most common signs of incomplete masculinisation, but do not receive enough attention. The etiology is very complex, including endocrine factors, genetic factors and environmental endocrine disruptors.

Objective

To explore 45 cases of micropenis children steroidogenesis factor 1 genetic abnormalities and to research the influence of the mutation on sex gland function.

Method

45 micropenis boys were collected from endocrinology department in October 2011 to February 2013 and 50 healthy children as control, and blood DNA was extracted, then PCR amplification products and SF1 gene sequencing were analysed. Sequencing results using sequencer software for sequence alignment.

Results

SF1 genetic abnormality is an infrequent cause in children with micropenis, only one sample of c.1056G>T (p.Q352H) may be one of the pathogenic mutations in children with micropenis.

Reference: