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# 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 sequencher software for sequence alignment.

## Results

Result 1　Analysis of SF1 gene variations in 45 children with micropenis and 50 controls

| variation | case | Control | Amino acid change | SNP number |
|---|---|---|---|---|
| 437 ( G/C ) ( exon4 ) | 10 | 17 | G146A | rs1110061 |
| 565 ( C/T ) ( exon4 ) | 2 | / | P189S | this study |
| 1056(G/T) ( exon6 ) | 1 | / | Q352H | this study |
| 1062(G/A) ( exon6 ) | 1 | / | 354L/ | this study |

Result 2　Hardy-Weinberg population genetic equilibrium test of SNP in case group and normal control group

| | case | | | | H-W | | | α=0.05 | |
|---|---|---|---|---|---|---|---|---|---|
| | genetype | | | total | genetype | | | $X^2$ | P |
| | GG | GC | CC | | GG | GC | CC | | |
| case | 35 | 8 | 2 | 45 | 33.83 | 10.38 | 0.79 | 2.396 | 0.122 |
| control | 33 | 13 | 4 | 50 | 31.21 | 16.59 | 2.21 | 2.341 | 0.126 |

Result 3　Frequencies of allele G/C of SNP rs1110061 and genotype(GG/GC/CC) between case group and normal control

| | | case | control | $X^2$ | P |
|---|---|---|---|---|---|
| genetype | GG | 35(77.8%) | 33 ( 66% ) | | |
| | GC | 8(17.8%) | 13 ( 26% ) | 1.66 | > 0.05 |
| | CC | 2(4.4%) | 4 ( 8% ) | | |
| allele | G | 78(86.7%) | 79(79%) | 1.94 | > 0.05 |

Result 4 Results of GnRH and HCG test in 3 SF1 missense mutations in children with micropenis

| variations | Case number | age | GnRH and HCG test | | | | |
|---|---|---|---|---|---|---|---|
| | | | Base T nmol/L | Base DHT pg/ml | After HCG T | After HCG DHT | LH peak /FSH peak |
| 565(C/T ) | 41 | 10y10m | <0.69 | 258.47 | 9.5 | 350.39 | 2.8/4.44 |
| (p. 189S) | 43 | 9y4m | <0.69 | 124.68 | 5.86 | 205.34 | 2.24/3.73 |
| 1056(G/T) (p. 352H) | 14 | 9y5m | <0.69 | 48.41 | 14.8 | 207.04 | 11.5/14.8 |

## Conclusion

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:

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