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Clinical and molecular genetic analysis of a Chinese family with congenital X-linked adrenal hypoplasia caused by novel mutation 1268delA in the *DAX-1* gene

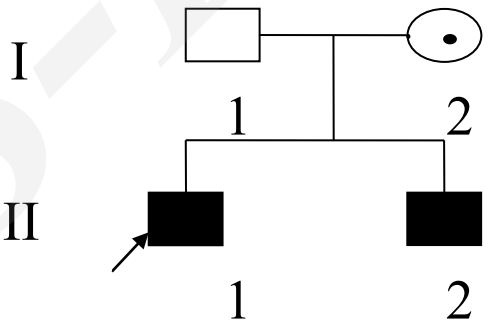
Key words: Congenital adrenal hypoplasia, *DAX-1* gene, Frameshift mutation

Case Report

Congenital X-linked adrenal hypoplasia (AHC) is a rare disorder. In this paper, we present a Chinese kindred with AHC with following aspects:

- **Clinical characteristics**
- **Direct sequencing of the DAX-1 gene**

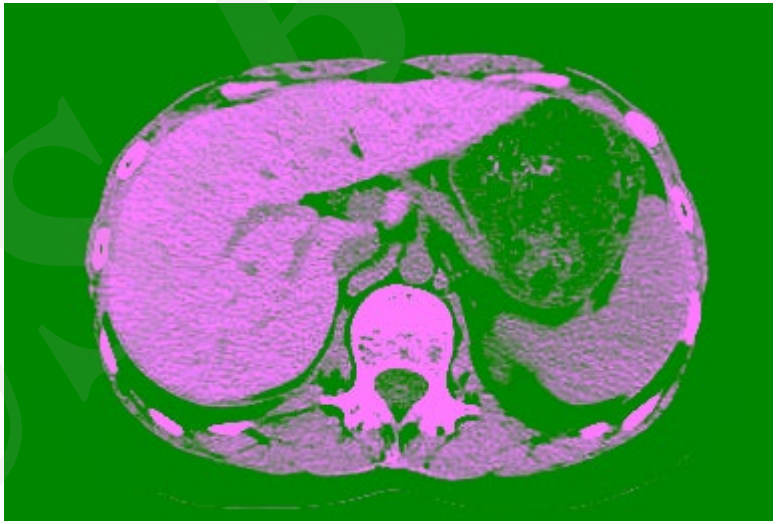
Fig.1



The proband, II-1, had the phenotypic spectrum of X-linked AHC. II-2 was the younger brother of the proband and had symptoms similar to those of his brother. The two patients' father (I-1) and mother (I-2) had both undergone normal puberty without any symptoms of adrenal insufficiency.

Clinical characteristics

- **primary adrenal insufficiency**
- **delayed puberty**
- **hypogonadotropic hypogonadism**
- **testicular defect**
- **CT: small adrenal glands**
- **MRI: pituitary gland hasn't any definite abnormality**



CT revealed small adrenal glands .it is useful to differentiat from congenital adrenal hyperplasia or Addison's disease secondary to multiple etiologies.

Direct sequencing of DAX-1 gene

A

B:

C

D

A, proband's father - normal;
B. proband's mother- heterozygous carrier;
C. proband,
D.proband's brother.

Direct sequencing of exon 2 shows a deleted peak (red arrow) at nucleotide position 1268 in the proband and his brother, overlapping peaks due to a heterozygous single base pair deletion in his mother (red arrow).

This nucleotide 1268 deletion results in a frameshift and a premature stop codon at position 436 (c.1268delA p.His423Leufs*14). The mutated gene encodes a truncated protein missing a large portion of the terminal region corresponding to the ligand binding domain.