# Long-term follow-up in a Chinese child with congenital lipoid adrenal hyperplasia due to a *StAR* gene mutation

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## *Objectives:*

To show some special character and Long-term follow-up in a Chinese child with congenital lipoid adrenal hyperplasia due to a *StAR* gene mutation

## *Methods:*

We report a case of CLAH caused by mutations in the steroidogenic acute regulatory protein (*StAR*) gene.

## *Results:*

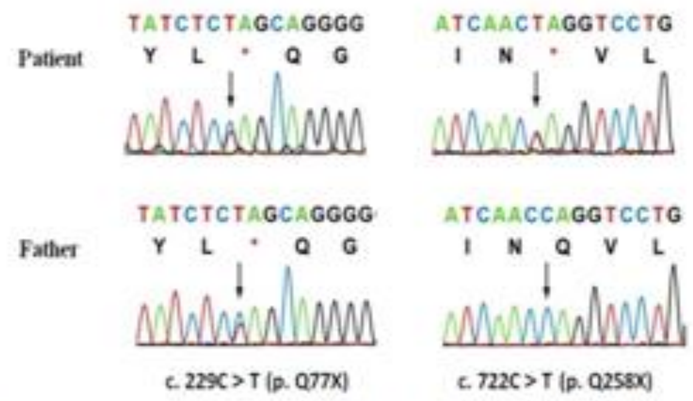


**Fig 1.** Sequence electropherograms showing the *STAR* gene mutations in the patient and her father. Sequence analysis of the *STAR* gene revealed two hemizygous nonsense mutations at c. 229C>T (p. Q77X) and c. 722C>T (p. Q258X). The heterozygous mutation of c. 229C>T (p. Q77X) was found in the patient's father. Because patient's mother was died due to a traffic accident, the mother's sequences could not be tested. The black arrows indicate the

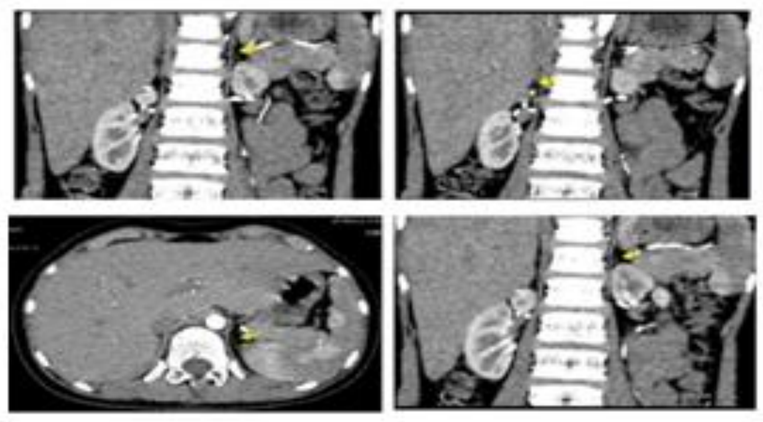

**Fig 2.** Adrenal computed tomography (CT) scans revealed that the adrenal glands on both sides were much smaller than normal. The yellow arrows indicate the adrenal gland.

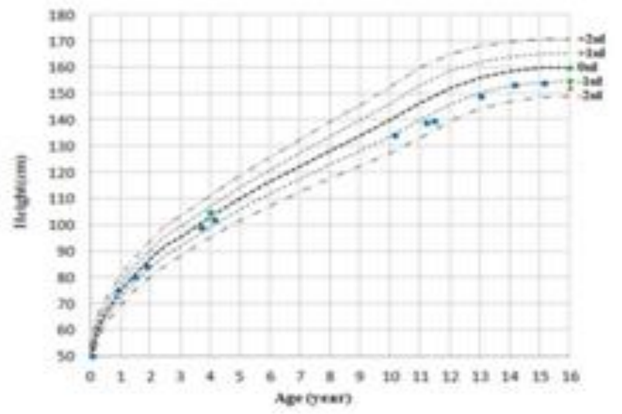


**Fig 3.** Chart showing the progression of the patient's height during the 15-year follow-up. Blue dots indicate the patient's height. Green dots indicate the height of the patients' sisters. The red triangle represents the midparent height. The green square represents the height of the patient reported in Fu's paper.

The patient had typical early-onset adrenal crisis at 2 months of age. She had normal-appearing female genitalia and a karyotype of 46, XY. The serum cortisol and adrenal steroids levels were always nearly undetectable, but the adrenocorticotropic hormone levels were extremely high. Genetic analysis revealed compound heterozygous mutations at c. 229C>T (p.Q77X) in exon 3 and c. 722C>T (p.Q258X) in exon 7 of the *StAR* gene. The former mutation was previously detected in only two other Chinese CLAH patients. Both mutations cause truncation of the StAR protein. The case reported here seemed to be a classic instance of CLAH with very small adrenal glands and is thus far the second reported CLAH case with small adrenal glands. In a 15-year follow-up, her height was approximately average for females before age 4 and fell to -1 SDS at 10 years of age. Her bone age was similar to her chronological age from age 4 to age 15.

## *Conclusions:*

In conclusion, this is a classic case of CLAH with exceptionally small adrenal glands. Q77X mutation seems to be more common in the Chinese CLAHs. Additionally, this is the first report of the growth pattern of CLAH after a 15-year follow-up.

## *References:*

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