X-linked adrenal hypoplasia congenita with a novel DAX1 missense mutation

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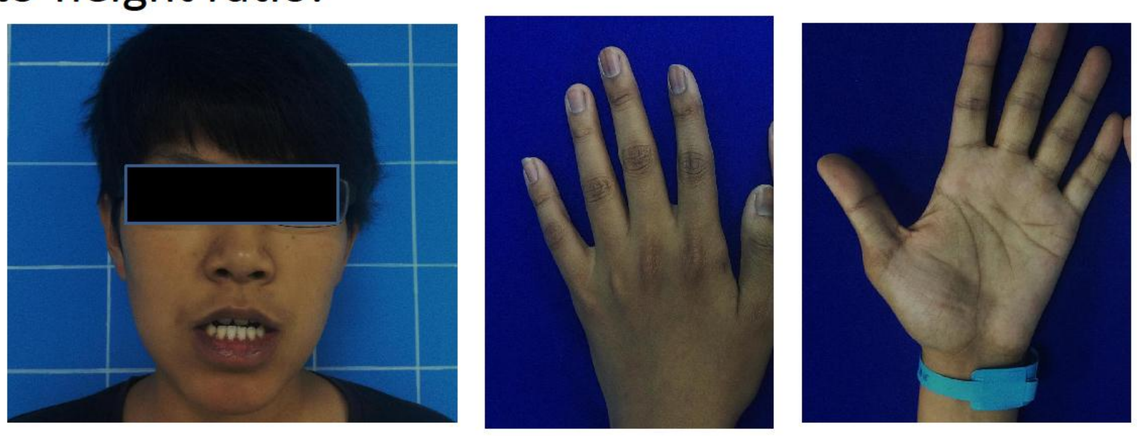
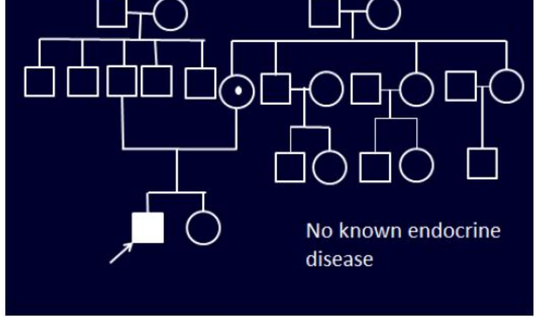
Introduction:

X-linked adrenal hypoplasia congenita (AHC) is a rare developmental disorder of the adrenal cortex. Adrenal insufficiency typically begins in infancy or in childhood. A few individuals present adrenal insufficiency in adulthood.

Case report:

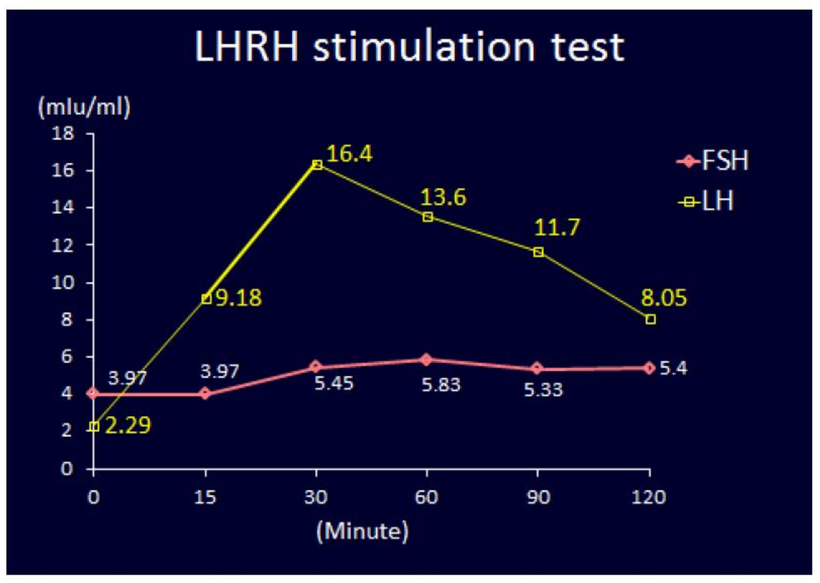
The 21-year-old man had underdeveloped secondary sexual characteristics. Physical examination revealed pigmentation over mouth, palmar creases, and joints, small bilateral testes, sparse pubic, axillary hair, and a large arm span-to-height ratio.





Endocrine function test:

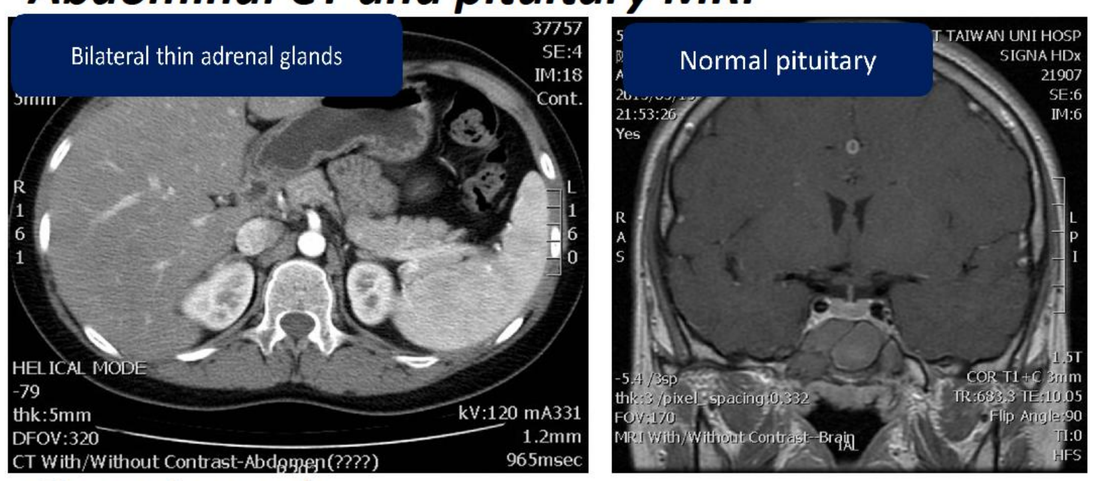
Hormone	Level	Reference
IGF-1 (ng/mL)	256.2	116~358
hGH (ng/mL)	0.537	0~3
Prolactin (ng/mL)	9.79	2.5~17
FSH (mIu/ml)	4.07	0.9~15
LH (mIu/ml)	2.19	1.3~13
Testosterone (ng/ml)	0.451	1.3~8.1
DHEA-S (µmol/L)	< 0.41	4.6~13.4
Free T4 (ng/dL)	1.02	0.6~1.75
hsTSH (µIU/mL)	1.74	0.10~4.50
ACTH (pg/mL) 8 am	>1250	10~65
ACTH (pg/mL) 4 pm	>1250	10~65
Cortisol (µg/dL) 8 am	5.37	8~24
Cortisol (µg/dL) 4 pm	4.07	2.5~12.5



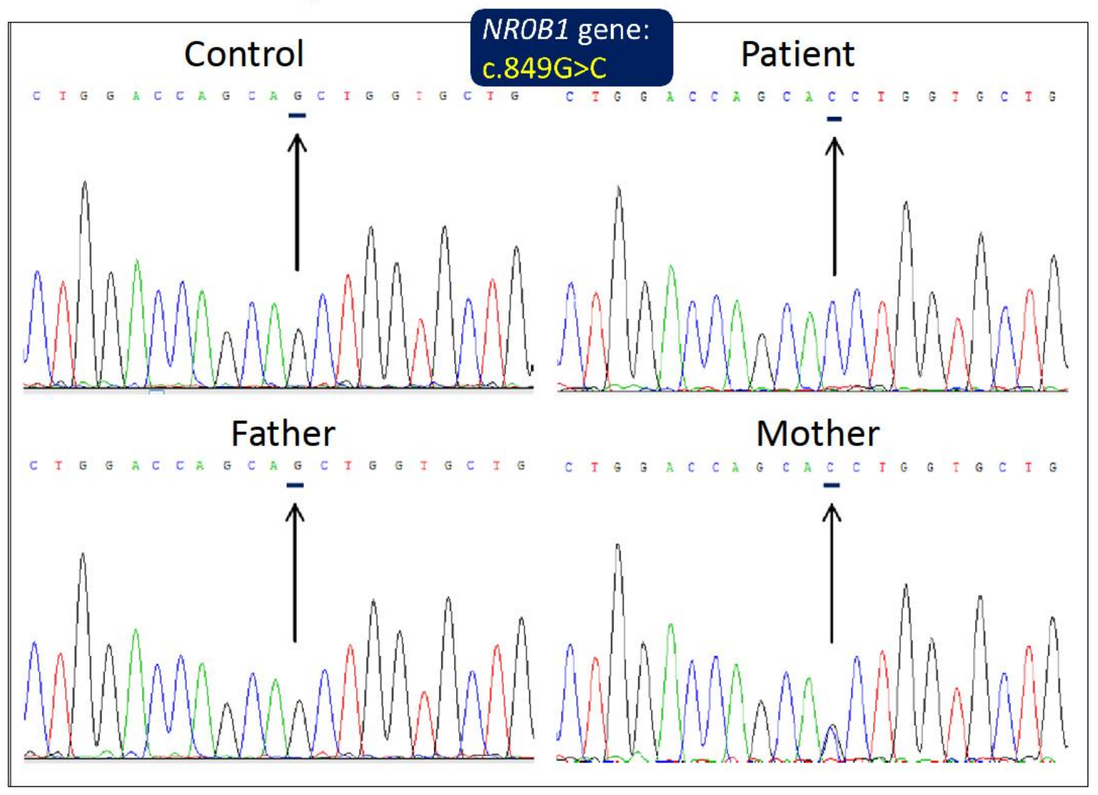
Clomiphene test (50mg BID PO)

	Day 1	Day 7
FSH (mIU/ml)	3.97	5.98
LH (mIU/ml)	2.29	1.84
Testosterone (ng/ml)	0.45	< 0.2

Abdominal CT and pituitary MRI



Genetic study



Conclusion:

DAX-1 gene deletions or mutations have been reported to be responsible for AHC. We report a case of AHC with adult-onset adrenal insufficiency, hypogonadotropic hypogonadism, and a novel missense mutation in the DAX-1 gene.

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