A Boy with Adrenal Hypoplasia Congenita without External Genital Abnormalities

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[Introduction]
Adrenal hypoplasia congenita (AHC) is a rare disorder with an estimated frequency of 1 case per 12,500 live births. AHC causes 46,XY disorders in sex development (DSD) due to adrenal androgen deficiency.

[Objective]
Case report on a male AHC patient without external genital abnormalities.

[Case Report]
The baby was born at 37 weeks' gestation with a height of 46.5 cm (~0.49SD), a weight of 2,175 g (~1.57SD), and a head circumference of 30.0 cm (~1.93SD). He had generalized hyperpigmentation at birth. He was discharged without any problems five days after birth and required only phototherapy for a day. However, he was referred to a hospital at the age of 11 days due to poor feeding and poor weight gain. He was not thriving and had peripheral edema. The patient presented generalized hyperpigmentation at birth but no external genital abnormalities.

[Discussion]
Importance of Early Diagnosis of AHC

• AHC often causes adrenal insufficiency during the neonatal period.1)
• However, AHC cannot be detected by neonatal mass screenings because 17OH-progesterone does not increase with AHC.

We suspected that this case was a DAX-1 abnormality with high testosterone in early infancy and has been reported.2) We suspected that this case was a DAX-1 abnormality, but we found no NR0B1 mutation.

[Conclusions]
We encountered a case of AHC with no external genital abnormalities. Hyper-pigmentation of the skin is an important sign of AHC even when there is no abnormality in the external genitalia.


Clinical Characteristics upon Hospitalization

Height: 46.0 cm; Body weight: 2,060 g
Body temperature: 36.9°C
Pulse rate: 140 beats/min; Systolic blood pressure: 72 mmHg
Anterior fontanelle: flat
Respiratory sound: clear
Skin: hyperpigmentation on whole body, especially on lips, areolas, external genitalia
External genitalia: complete male
Periphery: cold

Adrenocorticosteroid Data (at 11 days old)

X-ray (at 5 months old)

Low serum and metabolites of steroids + small adrenal glands ➔ diagnosis of AHC

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Clinic Course During Hospitalization

Growth Curve to Date

Metabolites of pregnenolone

Metabolites of 17αOHP

Metabolites of 17αOHP

Metabolites of DOC

Metabolites of Androgen

Metabolites of Androstenedione

An: high ➔ Δ5 steroid decreased and androstenedione was maintained.

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