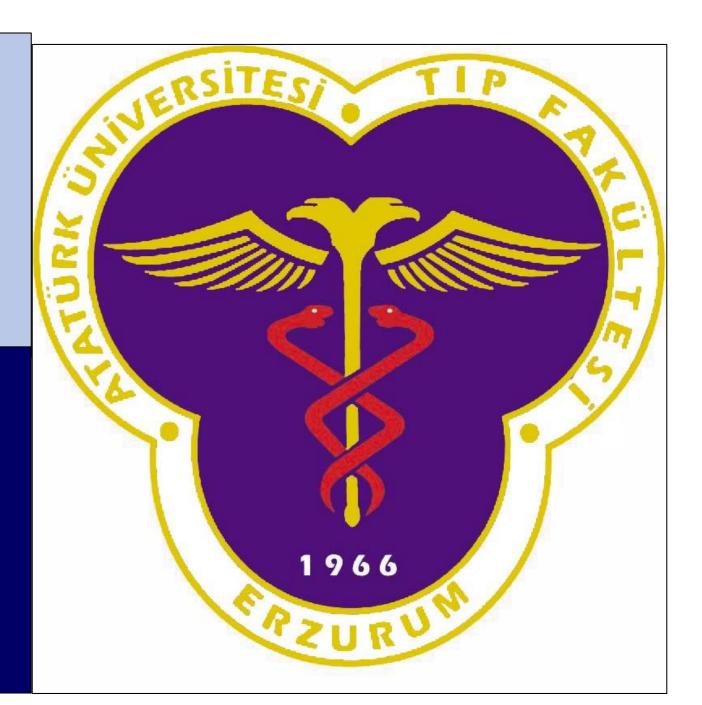# Dopamine Beta-Hydroxylase Deficiency Leading to Growth Hormone Deficiency

Hakan Doneray[1], Ayse Ozden[1], Remziye Seda Yesilcıbik[2],

[1]Ataturk University Faculty of Medicine, Department of Pediatric Endocrinology, Erzurum, Turkey, [2]Ataturk University Faculty of Medicine, Department of Pediatrics, Erzurum, Turkey

BACKRAUND

- Dopamine beta-hydroxylase (DBH) deficiency is characterized by lack of sympathetic noradrenergic function.
- Affected individuals exhibit profound deficits in autonomic regulation of cardiovascular function.
- DBH deficiency is a congenital disorder; however, the diagnosis is not generally recognized until late childhood.
- In this report, we present a case with DBH deficiency leading to growth hormone deficiency.

CASE

- A ten year old girl who has been taking growth hormone therapy was referred with a history of sudden onset malaise and somnolence.
- Her body weight and height were 19 kg (-2.4 SD) and 115 cm (-2.3 SD), respectively.
- Physical examination findings were unremarkable except for irregular heartbeat and blood pressure, somnolence, and weak deep tendon reflexes.
- Ophthalmoscopic eye and cardiovascular examinations were normal.
- Laboratory studies revealed abnormal levels of serum catecholamins and their urine metabolites.
- Serum;

- Epinephrine 11.1 pg/ml (N: 4-83)
- Norepinephrine 78.1 pg/ml (N: 80-498)
- Dopamine 52.9 pg/ml (<30)

- Urine;
- Metanephrine 47 µg/day (N: 26-230)
- Normetanephrine 40.8 µg/day (N: 44-540)

- Other laboratory and radiological studies were unremarkable.

- Oral administration of 50 mg Droxidopa, twice daily, increased blood pressure and reversed all symptoms.

CONLUSION

- This patient with growth hormone deficiency is the first case associated with DBH deficiency.

- Norepinephrine is a major regulator of the release of growth hormone.

- Low norepinephrine synthesis inhibits growth hormone secretion.

- DBH deficiency should keep in mind in case of irregular heartbeat and blood pressure together with growth hormone deficiency.