Thyroid Hemiagenesis: A Case Report

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Thyroid hemiagenesis is a rare congenital anomaly. It is characterized by the formation of one lobe of the thyroid. The true incidence is unknown. There is no left lobe in 80% of the cases. It is believed that a defect in the downward movement is responsible for the altered lobulation in thyroid hemiagenesis. The general condition of the patient was good and vital signs were stable. Free triiodothyronine was 4.3 pg/mL, free thyroxine 0.9 ng/dL, thyroid-stimulating hormone 2.6 µU/mL, and thyroglobulin was 10.6 pg/mL. Anti-microsomal and anti-thyroglobulin antibodies were negative. Routine hematological and biochemical parameters were normal. The left lobe of the thyroid could not be monitored by ultrasonography. Isthmus thickness was 1.5 mm and the right lobe was 11x9x19 mm. Thyroid scintigraphy was performed but left lobe of the thyroid was not visualized. Genetic analysis was planned for the patient. The patient is being monitored regularly without treatment.

Key words: Thyroid, hemiagenesis, ultrasonography, scintigraphy

Hypoparathyroidism-Deafness-Renal Disease Syndrome: The First Case Report from Turkey

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Introduction: Hypoparathyroidism-Deafness-Renal Disease (HDR) syndrome is a rare autosomal dominant genetic disorder that is characterized by a triad of conditions, namely hypoparathyroidism, sensorineural deafness, and renal disease. We, here, present the first Turkish case with HDR syndrome.

Case: A two-month-old boy was referred with convulsion related to hypocalcaemia. The patient was born at 39 gestational weeks by vaginal delivery after unremarkable pregnancy. There was no family history of deafness or renal insufficiency. Physical examination findings were unremarkable. His body weight, length, and head circumference were 4470 g (25th p), 59 cm (50th p), and 39 cm (50th p), respectively. Laboratory studies showed BUN 17.7 mg/dL; creatinine (Cr) 1.3 mg/dL; sodium 140 mmol/L; potassium 4.6 mmol/L; calcium (Ca) 6.4 mg/dL; phosphorus 7.2 mg/dL; alkaline phosphatase 408 U/L; magnesium 1.8 mg/dL; parathyroid hormone 7 pg/mL, and 25-hydroxy vitamin D 28.3 ng/mL. Urogram revealed urine density of 1007; pH 6.5; protein (+2); glucose (-), and ketone (-). Spot urine Ca/Cr was 0.01. Hemogram and blood gas analysis were unremarkable. There was thymus tissue on the neck ultrasonography (USG). Urinary USG and DTPA were normal. The brainstem-evoked response audiometry work was normal. The convulsion related to hypocalcaemia was controlled with intravenous Ca gluconate therapy followed by 50 mg/kg/day elementary Ca lactate and 0.25 mg/day calcitriol. Serum Cr level and proteinuria on urogram persisted as 0.8-1.2 mg/dL and trace, respectively. A nonsense mutation (R367X; CGA>TGA) in the *GATA3* gene was found. The patient is followed for renal and hearing functions.

Key words: Hypoparathyroidism-deafness-renal disease syndrome, infant, p.R367X mutation