A Case with Acrodysostosis and Hormone Resistance

Selma Tunç[1], Korcan Demir[1], Filiz Hazan[2], Özgür Kırbıyık[3], Eren Soyaltın[1], Özlem Nalbantoğlu Elmas[1], Melek Yıldız[1], Hüseyin Anıl Korkmaz[1], Behzat Özkan[1]

[1]Dr. Behçet Uz Children's Hospital, Clinic of Pediatric Endocrinology, İzmir, Turkey

[2]Dr. Behçet Uz Children's Hospital, Clinic of Medical Genetics, İzmir, Turkey

[3]Ministry of Health Tepecik Training and Research Hospital, Clinic of Medical Genetics, İzmir, Turkey

Aim: Acrodysostosis is a rare genetic syndrome characterized by small hands and feet with short, stubby fingers and toes, cone shaped epiphyses, broad nasal root, and various abnormalities of mandible, skull and vertebra, short stature, and mental retardation. Because of the hormone resistance that would accompany, acrodysostosis can be confused with pseudohypoparathyroidism. Mutations of PRKAR1A and PDE4D are reported to be responsible for the disease in less than 50 cases.

Methods: A case considered to have acrodysostosis was discussed regarding clinical and laboratory findings.

Results: A 12-year-old male patient was referred due to short hands and feet. These complaints were present since birth without a history of regular drug use or major disease. Motor and mental development was delayed compared to his peers. His parents were not relatives but from the same village. Physical examination disclosed the following: weight 45 kg [0.50 standard deviation score (SDS)], height 143.7 cm (-0.83 SDS), synophrys, arched eyebrows, low-set ears, and small squared hands. Upper/lower segment ratio was normal (0.95). Optic atrophy was bilateral, but the left side was predominantly affected. Skeletal survey was normal except short tubular bones in hands and feet and cone-shaped epiphyses. Calcium was 9.5 mg/dL, phosphorus 6 mg/dL, alkaline phosphatase 304 IU/L, parathormone 441 pg/mL, 25-hydroxy vitamin D 12.4 ng/mL, thyroid-stimulating hormone 11.5 mIU/mL, free thyroxine 1.02 ng/dL, free triiodothyronine 4.7 pg/mL, and anti-thyroid peroxidase and anti-thyroglobulin antibodies were negative. Thyroid ultrasonography revealed a volume of 2.79 mL (-1.52 SDS) and low echogenicity but no nodule. Bone age was compatible with chronological age. Vitamin D 2000 U/day and L-thyroxin 50 µg/day were administered. Genetic analysis for PRKAR1A and PDE4D was planned.

Conclusion: Acrodysostosis should be kept in mind and appropriately evaluated when hormone resistance is detected in cases of patients who present with small hands and feet as well as pseudohypoparathyroidism.

Key words: Acrodysostosis, hormone resistance, pseudohypoparathyroidism, short stature, mental retardation

A Case of Thyroid Hormone Resistance

Ruken Yıldırım[1], Yusuf Kenan Haspolat[1], Handan Deniz[2], Fırat Can[3]

[1]Dicle University Faculty of Medicine, Department of Pediatric Endocrinology, Diyarbakır, Turkey

[2]Bismil State Hospital, Diyarbakır, Turkey

[3]Dicle University Faculty of Medicine, Clinic of Pediatrics, Diyarbakır, Turkey

Thyroid hormone resistance is a hereditary disease characterized by high triiodothyronine, thyroxine, and non-suppressed thyroid-stimulating hormone. It usually shows an autosomal dominant inheritance feature. The most common is due to mutations in the thyroid hormone receptor beta (*THRB*) gene. Patients with thyroid hormone resistance are generally clinically euthyroid. However, although rare, signs and symptoms of hypothyroidism or thyrotoxicosis may be present. In this report, we discuss a case of thyroid hormone resistance and tachycardia; the patient had positive family history, normal intelligence level, and no goiter.

Key words: Thyroid hormone resistance, family survey