A Case of Pycnodysostosis with Bilateral Choanal Atresia

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Objectives: Pycnodysostosis is an uncommon, autosomal recessive condition characterized by short stature, bone fragility with osteosclerosis, acroosteolysis of the distal phalanges, delayed suture closure, obtuse mandibular angle, prominent eyes, and brachydactyly. Here, we describe a case of pycnodysostosis with bilateral choanal atresia in addition to previously described features which have not been reported in literature before.

Method: A 23-year-old woman consulted to us for genetic assessment before an operation for bilateral choanal atresia. Detailed physical examination was performed and pedigree was analyzed. The patient was seen by several departments including neurosurgery, endocrinology, and radiology.

Results: The clinical and laboratory examinations revealed short stature, dysmorphic facial features, metopic craniosynostosis, unilateral conductive hearing loss, open cranial sutures, and sandal gap deformity. Her mental status seemed appropriate for her age. Pedigree analysis showed autosomal recessive pattern with two similar individuals. Radiologic findings were metopic craniosynostosis, open cranial suture, acroosteolysis, sclerosis of vertebral bodies, and Madelung deformity of the forearm, all of which were supported by the diagnosis pycnodysostosis.

Conclusion: The patient was diagnosed with pycnodysostosis both clinically and radiologically. Genetic counseling was performed. Although its incidence is low, pycnodysostosis is relatively common in Turkey because of the high consanguinity rate. Hence, clinicians who work in countries with a high consanguinity mating rate should consider pycnodysostosis as a differential diagnosis in patients with short stature and skeletal findings.

Key words: Pycnodysostosis, ostesclerosis, choanal atresia, open cranial sutures, short stature

H Syndrome: A Rare Monogenic Cause of Insulin-Dependent Diabetes Mellitus

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Introduction: H syndrome (HS) is an autosomal recessive multisystemic disorder, named after the first letter of the characteristic clinical features of hyperpigmentation, hypertrichosis, hepatosplenomegaly, hearing loss, heart anomalies, hypogonadism, low height (short stature), hyperglycemia/diabetes mellitus (DM), and hallux valgus/flexion contractures. Causative mutations have been identified in the *SLC29A3* gene encoding the human equilibrative nucleoside transporter 3 (hENT3), which is thought to serve as a transporter of nucleosides from the inside of the late endosome/lysosomes to the cytoplasm and also across the inner mitochondrial membrane.

Case: A seven-year-old boy, with insulin-dependent DM (IDDM) diagnosed at 18 months of age, developed bilateral cervical masses at the age of 6 years. His height was 120.4 cm [-0.2 standard deviation (SD)] and weight was 25 kg (0.5 SD). He had coarse face, depressed nasal bridge, hypermetropia, right esotropia, cervical lymphadenopathy (LAP), and multiple nevi at various sites of the body. The laboratory evaluation for LAP revealed normal results with regard to hematologic and serologic tests except the presence of AntiSLC70 antibodies. Excisional lymph node biopsy demonstrated massive sinus dilatation, histiocyte proliferation which was stained with S100 and CD68, capsular and pericapsular fibrosis in the adipose tissue and emperipolesis, consistent with Rosai-Dorfman disease. Both LAP and DM suggested the presence of HS. Because of the combination of two known but relatively rare manifestations of HS genetic analysis of the *SLC29A3* gene was carried out and revealed a novel homozygous mutation of p.367A>T; (c.1099G>A).

Discussion: HS is a genetic dermatologic disease with different manifestations. Early-onset IDDM may even be the first manifestation of H syndrome as in our case, presenting a diagnostic challenge. Diabetic patients with cutaneous signs or histiocytic LAP should be investigated for HS.

Key words: H syndrome, monogenic diabetes, SLC29A3, Rosai-Dorfman disease