AN UNUSUAL CAUSE OF SHORT STATURE

Sare Betul Kaygusuz[1], Zeynep Atay[2], Tarık Kırkgoz[1], Tulay Guran[1], Abdullah Bereket[1], Serap Turan[1]

[1]Marmara University, Department of Pediatrics, Division of Pediatric Endocrinology, Istanbul, Turkey

[2]Medipol University, Department of Pediatrics, Division of Pediatric Endocrinology, Istanbul, Turkey

Objectives

Spondyloenchondrodysplasia with immune dysregulation (SPENCDI) is an immunoosseous dysplasia combining the typical metaphyseal and vertebral bone lesions with immune dysfunction and neurologic involvement which is caused by mutations in *ACP5* gene encoding tartrate resistant acid phosphatase 5.

Here, we report a three year old girl presented with primary hypothyroidism, developmental delay and thrombocytopenia and diagnosed as SPENCDI.

Case Report

A 3-year-old girl was referred to our department for primary hypothyroidism which was diagnosed during evaluation for developmental delay at the age of 1 year with mildly elevated TSH and exaggerated response at TRH test. Brain MRI and tests for metabolic diseases, which were done because of inability to talk and walk at two years of age, were unremarkable. She was a product of consanguineous marriage of healthy parents with normal stature. She was born at 40 weeks of gestation and her birth weight was 3300 gr. Family history revealed treatment with rGH for short stature of her 8 year old brother. On initial examination height SDS was -3,5 (85cm) and weight SDS -1,15 (13,3kg). She was noted to have spastic paraparesis, blue sclera and hepatosplenomegaly. Laboratory tests showed thrombocytopenia and autoimmune hemolytic anemia at the age of 4 year and fairly response to steroid treatment. The spine X-rays showed generalized platyspondyly with posterior vertebral bodies were irregularly ossified. A subsequent skeletal survey revealed generalized metaphyseal dysplasia with enchondromatous lesions. Findings were considered consistent with SPENCDI. Laboratory data showed normal levels of serum calcium, phosphate, ALP, intact PTH. A novel homozygous 19 bp deletion in the *ACP5* gene (c. [772-790del19]; [(772-790del19)]) was detected. Tests for immune dysfunction and neurological involvement revealed intense calcification in the bilateral basal ganglia and lateral ventricular frontal horns and a decrease in the number of CD4 + cells.

The older brother also had skeletal dysplasia with 160 cm final height and without any neurological abnormality and immune dysfunction.

Conclusions

SPENCDI is a rare skeletal dysplasia characterized by enchondroma-enchondroma-like metaphyseal lesions in long bones and spondylar dysplasia with immune dysregulation and neurological symptoms. Here we desribed a case with full clinical picture of SPENDI and her less severely effected older brother with only short short stature and bone dysplasia. SPENCD should be kept in mind in children with short stature and typical skeletal findings. Recognition of the disease is important for timely diagnosis and treatment of extraskeletal manifestations and for genetic counseling

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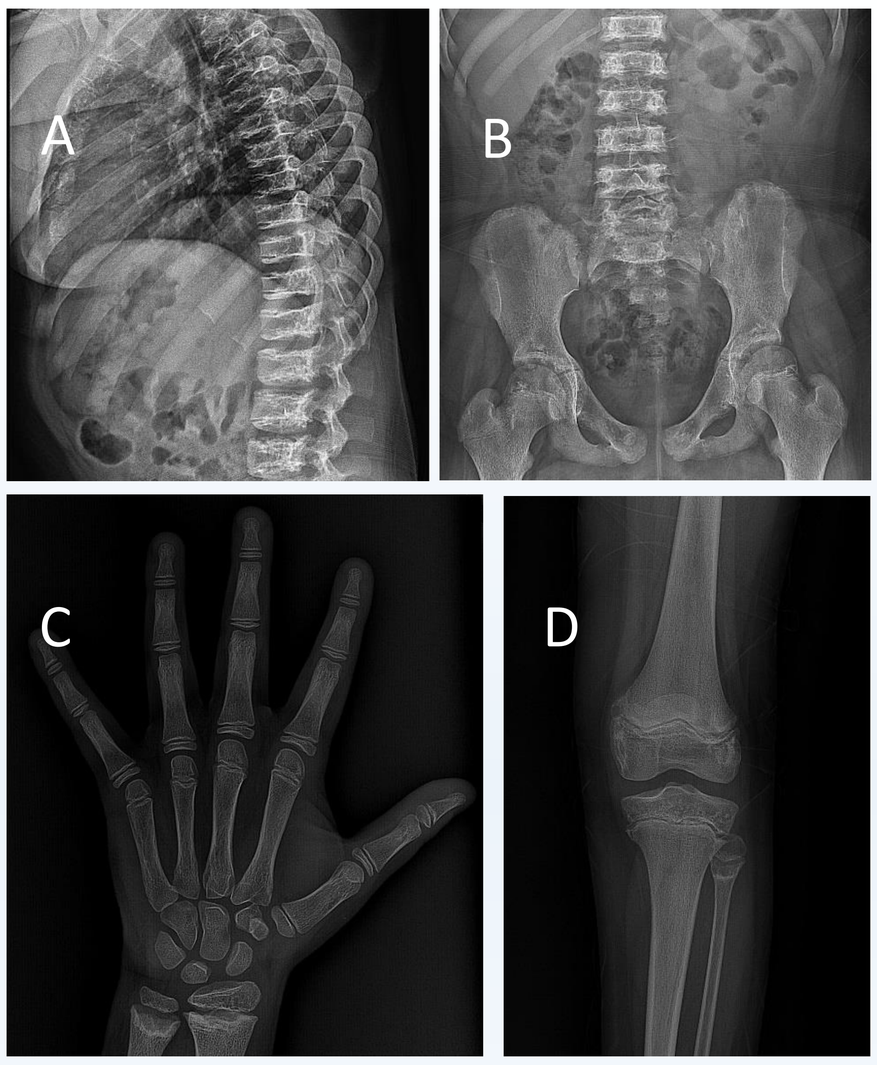

Figure 1: *Lateral spine radiograph showing platyspondyly and irregular ossifications in posterior vertebral bodies (A). Metaphyseal dysplasia with enchondromatous lesions in distal radius and ulna,distal femur, proximal tibia and fibula and the hip (B,C,D)*

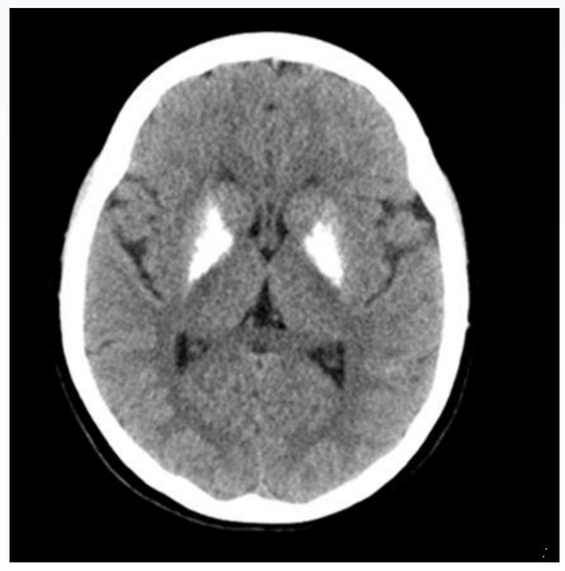

Figure 2: Cranial CT demonstrating bilateral calcification of the basal ganglia

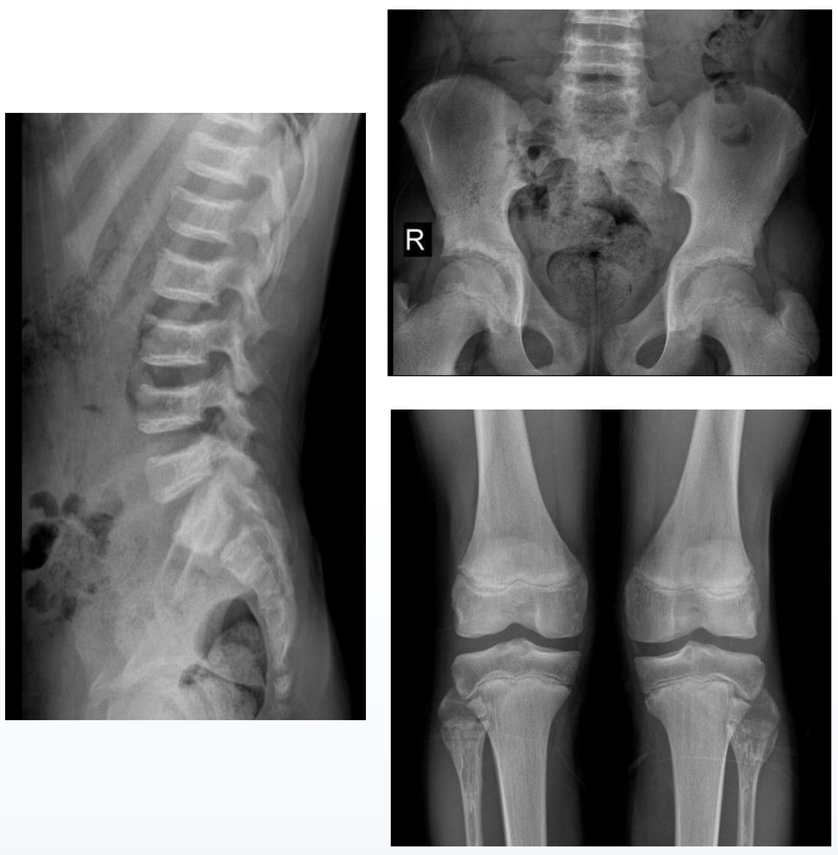

Figure 3: Spinal radiograph and radiograph of the knee and hip of her older brother showing similar metaphyseal and vertebral body changes as the patient