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Two Siblings with Microcephalic Osteodysplastic Primordial Dwarfism Type II (MOPD II)

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We aimed to identify the genetic cause of severe short stature and microcephaly in two siblings.

A ten-year-old boy presented with short stature. He was born with a birth weight of 1300 grams (-2.7 SDS) at 34 weeks of gestational age. On physical examination, height was 86 cm (-8.9 SDS), weight 8.4 kg (-7.9 SDS), BMI 11.4 kg/m² (-5.6 SDS), and head circumference was 37.7 cm (-9.1 SDS). Development was delayed. He started walking at 3 years, speaking with single words at 2 years, and he cannot make sentences yet. He had prominent nose, microcephaly, micrognathia, and microdontia. There were areas of hypo- and hyperpigmentation, cutis marmorata, and few cafe au lait spots on skin. Parents were first-degree cousins. Height of father was 177.5 cm (0.2 SDS) and height of mother was 159 cm (-0.7 SDS); target height was 161.8 cm (-0.2 SDS). His sister presented at 9 months of age and her height was 50.1 cm (-7.1 SDS), weight 3.8 kg (-6.3 SDS), and head circumference was 33.5 cm (-8.1 SDS). She was born with a birth weight of 1135 grams (-2.5 SDS) at 33 weeks of gestational age. Routine laboratory tests, serum levels of free triiodothyronine, free thyroxine, thyroid-stimulating hormone, insulin-like growth factor 1, and insulin-like growth factor binding protein 3 were normal.

Genetic analysis showed homozygous pericentrin mutation c.3109G > T, p. Glu1037 in both siblings. Parents were heterozygous for this mutation.

MPOD II is characterized by severe intrauterine and postnatal growth retardation, microcephaly, and distinctive face; patients with this disorder should be screened for cerebrovascular abnormalities.

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H syndrome (OMIM # 602782), first described in 2008, is a rare autosomal recessive genodermatosis which is multisystemic and is primarily characterized by cutaneous hyperpigmentation, hypertrichosis, hepatosplenomegaly, hearing loss, heart anomalies, hypogonadism, short stature, hyperglycemia (insulin-dependent diabetes mellitus), and hallux valgus/flexion contractures. It is caused by mutations in the solute carrier family 29 (SLC29A3) gene. A 23-year-old female patient who had the characteristic clinical features of H syndrome was referred to our medical genetics outpatient clinic to confirm the clinical diagnosis through molecular testing, to arrange the clinical follow-up and treatment support programme, and to provide the patient with suitable genetic counselling.

Clinical examination was performed. Related tests and imaging methods were planned. All coding exons of SLC29A3 gene were sequenced.

Physical examination revealed cutaneous hyperpigmentation on the body/on lower and upper extremity skin except knees and elbows, bilateral hypertrichosis on lower extremity (proximal), hepatosplenomegaly (splenomegaly), bilateral sensorineural hearing loss, heart anomalies, hyperglycemia (insulin-dependent diabetes mellitus), hallux valgus/flexion contractures (flexion contractures on bilateral hands/feet). Homozygote nonsense mutation causing premature stop codon (p.Y428*) in SLC29A3 gene exon sequencing was detected.

H syndrome is a rare genetic disease which requires multidisciplinary treatment because of its multisystemic involvement. Molecular genetic testing is important to confirm the diagnosis, to provide appropriate genetic counselling and to estimate prenatal diagnosis possibilities for the following pregnancies. As far as we are concerned, there are two more cases reported except ours in Turkey until now. More than 100 patients and 20 mutations in SLC29A3 gene have been described in the world. We find it convenient to present this rarely observed case in order to discuss the clinical findings and the mutation found in our case.

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A Rare Genodermatosis: H Syndrome

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Non-Genetic Factors Altering Birth and Fertility Rates

Dilara Çelebi

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The purpose of this project was to understand how Bosnian war affected the fertility and birth rates.

I selected two countries, namely Bosnia and Herzegovina and Turkey and researched and analyzed the results of the national statistical institute of the countries.