A Case with Infantile-Onset Pancytopenia and Hyperglycemia Associated with *SLC19A2* Mutation

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Objective: Homozygous mutations in *SLC19A2* gene result in alterations in cellular transport of thiamine and consequently megaloblastic anemia, deafness, and diabetes mellitus, also named as Roger's syndrome. We present such a patient, who has been followed in our clinic, given that the disease is rarely encountered and significant improvement was achieved especially in hematologic variables with thiamine treatment.

Methods: DNA was extracted from the peripheral blood sample of a female subject who was suspected to have thiamine responsive megaloblastic anemia depending on clinical and laboratory data and *SLC19A2* sequence analysis was performed.

Results: The 4-year-6-month-old girl was first evaluated due to paleness at the age of 3 months. Pancytopenia, megaloblastic anemia, hyperglycemia, sensorineural hearing loss, increased sideroblasts (12%) in the bone marrow aspiration were found. Autoantibodies related with diabetes were negative. Thiamine was started (100 mg/d) and hemoglobin levels increased up to 13 g/dL. During follow-up, maximum insulin requirement and hemoglobin A1c values were 0.6 unit/kg/day and 8.4%, respectively. Her parents were first-degree relatives. Physical examination revealed a weight of 18 kg (0.87 SDS), height 100 cm (-0.65 SDS) and normal systemic findings. Ophthalmologic and cardiac evaluation was normal. Hemoglobin was 9.9 g/dL, mean corpuscular volume 104 fL, platelets 371,000/mm3, leukocyte 7390/mm3, and hemoglobin A1c 4.8%. Sequencing analysis of *SLC19A2* revealed a homozygous mutation (p.Y81*) which was previously reported in the literature. Genetic counselling was provided.

Conclusions: This syndrome, which has various phenotypical features and is autosomal recessively inherited, should be kept in mind because, in particular, hematological parameters improve with thiamine therapy and genetic counselling can be given.

Key words: Infantile-onset pancytopenia and hyperglycemia, *SLC19A2* mutation

Four Cases of SCD (Jarcho-Levin Syndrome) Presenting with Short Stature

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Spondylocostal dysostosis (SCD), in other words Jarcho-Levin syndrome, is a hereditary syndrome presented clinically as short stature. It is characterized by vertebral and costal anomalies. Axial skeletal malformation is detected radiologically and clinically. While sporadic inheritance is seen more often with the multiple organ anomalies in spondylocostal dysostosis, autosomal recessive and dominant inheritance patterns are also known. 4 gene mutations are described for OR-SCD: *DLL3, MESP2, LFNG*, and *HES7*. In our study, we evaluated 4 patients who applied to our clinic with curvature of the back, disproportioned short stature, and multiple hospitalizations for respiratory problems. No similar case was found in their family history. Physical examination and radiological diagnostics showed similar vertebral and costal anomalies. The cases were as follows: a 5-month-old girl, 2-year-old boy, 4.5-year-old girl, and a 8-year-old boy. SCD cases presented with short neck, respiratory problems, kyphosis, scoliosis, pectus deformities, and short stature when cervical vertebras were affected. The patients should be further investigated for other possible system anomalies. Genetic consultation should be given to the family and periodical follow-up visits should be done.

Key words: Short stature, kyphosis, scoliosis, spondylocostal dysostosis