

Didem Ozdemir¹, Şefika Burçak Polat¹, Nagihan Beştepe¹, Saliha Ozbek², Reyhan Ersoy¹, Bekir Cakir¹

¹Yildirim Beyazıt University, School of Medicine, Ataturk Education and Research Hospital, Department of Endocrinology and Metabolism, Ankara, Turkiye

²Yildirim Beyazıt University, School of Medicine, Ataturk Education and Research Hospital, Department of Internal Medicine, Ankara, Turkiye

Introduction:

Leri-Weill dyschondrosteosis (LWD) is a rare autosomal dominant dyschondrosteosis characterized by Madelung deformity and mesomelic dwarfism. Majority of cases are associated with haploinsufficiency of the short-stature homeobox-containing (SHOX) gene. Here we report a patient with LWD and diabetes mellitus (DM).

Case Report:

A 31 years old man applied with polyuria and polydipsia. He did not have any chronic disease. In family history, father had type 1 and mother had type 2 DM. His height was 146 cm and weight was 62 kg, with a body mass index of 29.1 kg/m². Laboratory investigation revealed a fasting blood glucose of 298 mg/dl, HbA1c 13.4%, insülin 8 µU/ml (2.6-24.9), c-peptide 1.65 ng/ml (0.1-3.6). AntiGAD, anti-insulin and islet-cell antibodies were negative. Intensive insulin treatment was started. The result of genetic analysis made to determine cause of short stature about 1,5 years ago was 45,X,-Y[5]/46,X,der(Y)t(Y;Y)(p11.2;q11.21). There was deletion in the SHOX gene of Y chromosome. He had bilateral Madelung deformity which is an epiphyseal growth plate disturbance characterized by shortened and bowed radii and ulnae leading to dorsal dislocation of the distal ulna (Figure 1). Thus, he was diagnosed to have LWD.

Figure 1: Bilateral Madelung deformity of wrists



Conclusion:

Haploinsufficiency of the SHOX gene which is located on the pseudoautosomal region of both the X- and Y-chromosomes is a cause of short stature in various clinical conditions including LWD. There are previous reports indicating inverse association between height and diabetes, and showing increased prevalence of gestational DM in patients with short stature. In the literature, clinical features of LWD is limited to skeletal anomalies and short stature. This is the first report of DM in a patient with this syndrome. Although, it is probable that this might be just a coincidence, we think possible association between impaired glucose metabolism and syndromes related with short stature needs further investigation.