

# Congenital generalized lipodystrophy type 4 - New mutation in the CAVIN1 gene

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## Introduction

Congenital generalized lipodystrophies (CGL); autosomal recessive disorders characterized by dyslipidemia and almost complete absence of body fat associated with insulin resistance. It develops due to mutations in AGPAT2, BSCL2, CAV1, PTRF, PCYT1A and PPAR, genes. CGL type 4 results from PTRF-CAVIN gene mutation. Unlike classical CGL, myopathy, flat and skeletal muscle hypertrophy, heart rhythm disorders (sudden death) and skeletal abnormalities are seen.



## Case

9 years and 4 months old male patient. She presented with weakness, muscle weakness, difficulty in walking, and elevation of blood fat. The patient's history revealed a 34-weeks age of 2800 g, birth, pyloric stenosis at 40 days of age and a 5-years-old undescended testis surgery. The mother and father were cousins of the 1st degree and he received LT4 treatment in the other center from 7 months to 6 years of age due to hypothyroidism. Physical examination weight: 31.7 kg (50-75p), Height: 143.5 cm (75-90p), dysmorphic appearance, whole body subcutaneous fat tissue deficiency, axial muscle weakness, lumbar lordosis, hypertrophy appearance in muscles, hip, knee and ankle joints had limitation of movement and inability to walk. Fasting serum glucose: 89 mg / dl, insulin: 11.3 IU / mL, HbA1c: 5%, AST: 29 IU / L, ALT: 44 IU / L, Total cholesterol: 150 mg / dL, HDL: 25 mg / dL, LDL: 110 mg / dL, Triglyceride: 455 mg / dL, CK: 746 IU / L. The ECG showed an incomplete right bundle branch block, a frequent atrial ectopic beat in the holter, ventricular tachycardia, and a mild hypertrophy of the left ventricle in the ECO. In the genetic analysis, homozygous mutation of c.A406T (p.K136X) was detected in the 1st exon of the CAVIN1 gene.

## Sonuç

Early diagnosis and treatment of arrhythmias is important because of arrhythmia and sudden death in CGL type 4. Also; In addition to myopathy and cardiac disorders, atlanto axial instability, hepatomegaly, high serum triglyceride level, hyperinsulinism or diabetes, low leptin and adiponectin levels, immunoglobulin A deficiency, and low growth hormone levels are secondary features. A few patients with CGL type 4 have been reported in the literature. The c.A406T (p.K136X) mutation found in our patient is a new mutation. In patients with lipodystrophy, the presence of myopathy and skeletal findings should be investigated and CGL type 4 should be kept in mind. There are fetal rhythm disorders with CGL type 4. So, early diagnosis and treatment should be provided with careful investigation of rhythm disorders.