

Magnetic Resonance Imaging Characteristics of a Dunnigan-Type Familial Partial Lipodystrophy Patient

Nilufer Özdemir Kutbay¹, Banu Şarer Yürekli¹,
Emin Karaca², Hüseyin Onay², Barış Akıncı³,
Mehmet Erdoğan¹, Şevki Çetinkalp¹,
Mustafa Seçil⁴, A. Gökhan Özgen¹,
L. Füsün Saygılı¹

¹Ege University Faculty of Medicine, Department of
Endocrinology, Izmir, Turkey

²Ege University Faculty of Medicine, Department of Medical
Genetics, Izmir, Turkey

³Dokuz Eylül University Faculty of Medicine, Department of
Endocrinology, Izmir, Turkey

⁴Dokuz Eylül University Faculty of Medicine, Department of
Radiology, Izmir, Turkey

Introduction: The lipodystrophic syndromes are rare congenital or acquired disorders which are characterized by partial or complete lack of adipose tissue. The extent of fat loss correlates with the severity of the metabolic abnormalities. We present the magnetic resonance imaging (MRI) of our patient with familial partial lipodystrophy (FPLD).

Cases: Physical examination of three sisters from the same family revealed absent subcutaneous fat tissue and distinctive muscle structure. The patient was diagnosed with lipodystrophy based on presence of diabetes, insulin resistance,

hypertriglyceridemia, and her physical examination findings. As for the genetic study of the patient, *BSCL2* (Exon 2-11), *AGPAT2* (Exon1-6), *CAV1* (Exon 1-3), *PTRF* (Exon 1a, 1b, 2a, 2b), *LMNA* mutations were not detected. All of the patients had macroalbuminuria although they did not have retinopathy. Renal biopsy was performed in two sisters. Electron microscopic studies have not been completed yet. MRIs of one of the sisters are as follows: Coronal, T1-weighted whole body MRI showed preserved subcutaneous fat tissue in the body and upper extremities and reduced fat tissue especially in the thigh and hip region; Dual-echo, chemical shift gradient-echo T1-weighted MRIs showed hepatic steatosis. Liver signal was lost on the out-of-phase image (b) compared to the in-phase image (a). Single-voxel proton magnetic resonance spectroscopy image revealed spectroscopic peaks derived from hepatic lipids at 0.9-1.1 ppm, 1-1.5 ppm, and 2-2.5 ppm.

Discussion: FPLD type 2 (Dunnigan's syndrome) is associated with fat loss from the extremities, abdomen, and thorax and excess subcutaneous fat in the chin and supraclavicular area. Patients have normal adipose tissue in childhood, but lose subcutaneous adipose tissue from the extremities later, usually with the onset of puberty. In lipodystrophy patients, MRI is helpful to observe the pattern of subcutaneous fat tissue.

Key words: Familial partial lipodystrophy, MR images, insulin resistance, macroalbuminuria, hypertriglyceridemia, diabetes