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Partial Lipodystrophy with Renal Disorder

A Case Report

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Lipodystrophy, which is characterised by the symmetrical loss of subcutaneous fat, may be partial or total. Partial lipodystrophy has been identified by various terms: Progressive lipodystrophy, Barraquer Simons Disease, Lipodystrophia Facialis, Progressive cephalothoracic lipodystrophy, Cephalothoracicobrachial form of lipodystrophy.[1-4]

This syndrome is described as a disorder with the loss of facial fat alone or with involvement of the neck, arms, chest and abdomen. There is normal or even increased fat deposition over the legs. In addition to the loss of fat, some other abnormalities may be associated.

We have reported this case because of the rareness of partial lipodystrophy with renal involvement and to the best of our knowledge no similar case has been reported from Turkey.

Case Report

An 8-year-old girl, was admitted to Hacettepe Children's Hospital on October 17, 1969 with complaints of hematuria, loss of weight, and abdominal pain. Her history revealed that she was the product of a normal pregnancy and delivery. About 2 1/2 years prior to admission she developed anorexia, perspiration on the face, and loss of weight. She was diagnosed and treated by a local doctor for tuberculosis for 2 years. In spite of this treatment, her complaints persisted. Family history was not contributory.

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Physical examination: The patient was 123 cm. tall and weighed 26 kg. Her pulse rate was 98 per minute, and blood pressure was 120/80 mm. Hg. Temperature was 36.8°C. Her cheeks were hollow and the skeletal structure of the face was prominent (Figure 1). The subcutaneous tissue was well developed everywhere except in the face (Figure 2). There were no hirsuties and pigmentation. A grade I. systolic murmur was heard over mid-precordium. There was no transmission of this murmur; and the heart sounds were normal. Lungs were clear. Liver was palpated 1 cm. below the right costal margin. Kidneys were not palpable. The remainder of the physical examination was unremarkable. Laboratory findings were as follows: Hb: 7.8 gm/100 ml., Hct: 26 %. Sedimantation rate: 65 mm/h., white blood count 8600/cmm. (with predominance of segmented cells), thrombocytes normal, erythrocytes hypochrom-microcytic, reticulocytes were less than 0.1 %, nonprotein nitrogen 92 mg/100 ml., fasting blood sugar 108 mg/100 ml., CO_2 18.64, mEq/lt., K 3.9 mEq/lt., Cl 106.7, mEq/lt., total proteins 6 gm/100 ml., albumin 4 gm/100 ml., globulin 2 gm./100 ml. serum glutamic-oxalacetic transaminase 24 U., serum glutamicpyruvic trans-

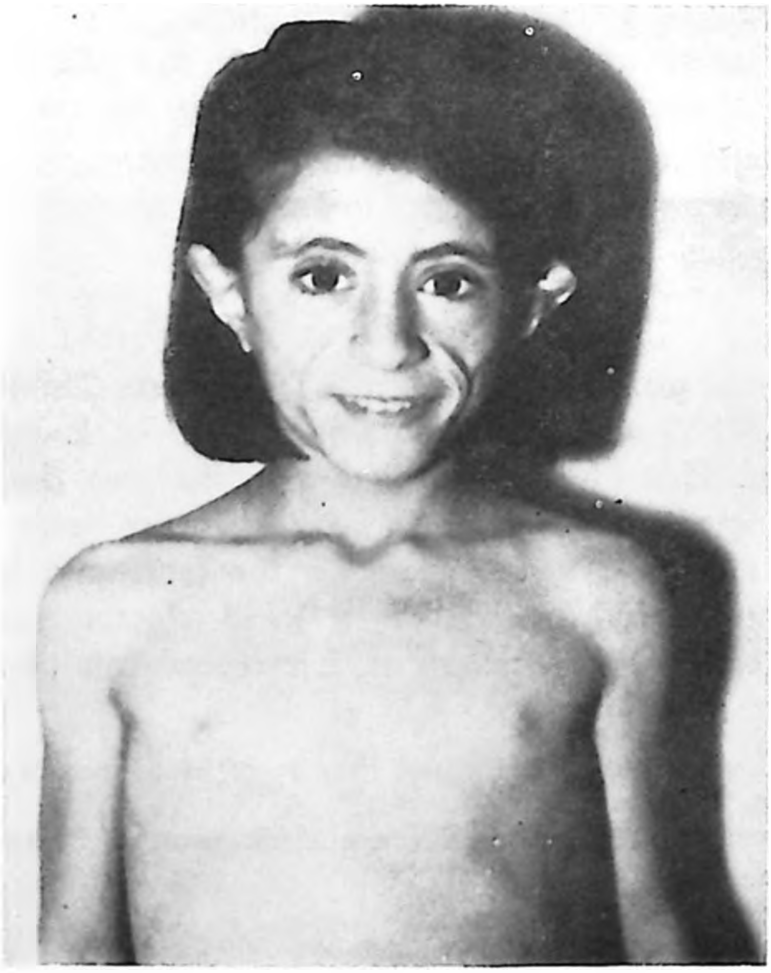

Figure 1

Picture of M. T. showing characteristic appearance of the face for partial lipodystrophy.

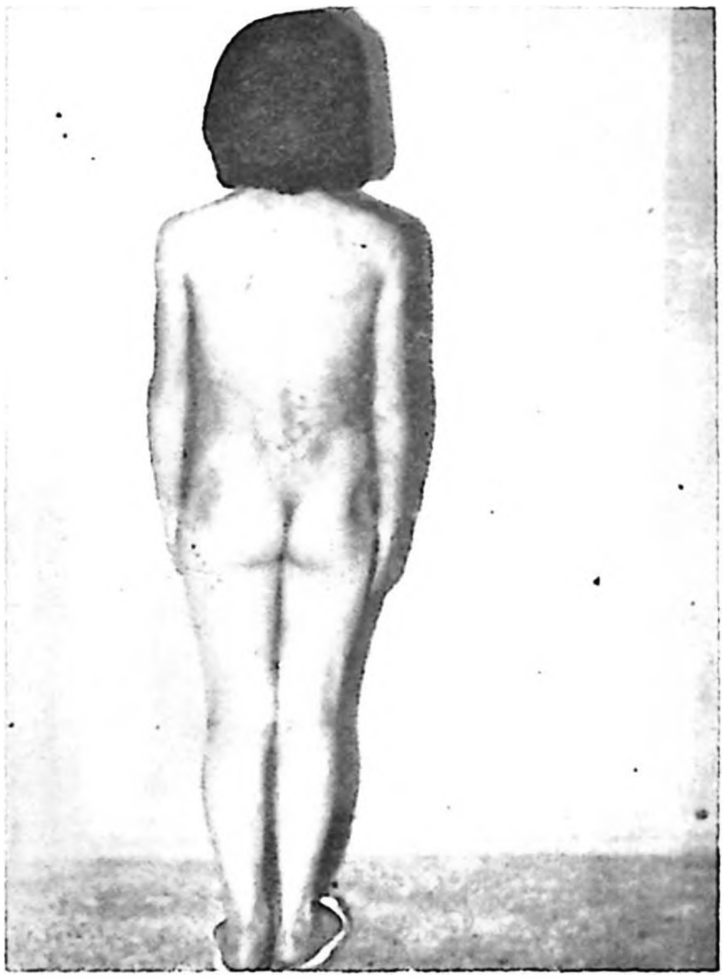

Figure 2
Picture of the case presented, showing normal fat deposits on legs.

aminase 18 U., Ca 8.8 mg/100 ml., P 712 mg/100 ml., alkaline phosphatase 5.7 Bodansky Units, uric acid 4.6 mg/100 ml., creatinin 1.3 mg/100 ml., total cholesterol 182 mg/100 ml., total lipid 900 mg/100 ml., fibrinogen 360 mg/100 ml., serum iron 26 gamma/100 ml., serum iron binding capacity 317 gamma/100 ml., serum electrophoresis: albumin 44.6 %, α_1 globulin: 7.4 %, α_2 globulin: 20.2 %, β globulin: 6.6 %, γ globulin: 21 %. Serum C_3 (Beta 1 C/Beta 1 A) 15 mg./100 ml. (determined with immuno-diffusion plate method, normal ranges 140-150 mg/100 ml.).

Urinalysis revealed a specific gravity of 1004, albumin (+), abundent erythrocytes and leucocytes in sediment. Renal concentrating ability test revealed a 1015 specific gravity. Ascaris and trichuris tricuria eggs were seen in the faces. 100,000 colonies of E. Coli cultured in urine. Several guinea pig inoculations and urine cultures for renal tuberculosis were negative.

L. E. phenomenon and PPD skin test were also negative.

Esophageal and gastric X-ray examinations, intravenous pyelograms and retrograde pyelograms were normal.

Punch biopsy of the skin of the face (Figure 3) showed unremarkable epidermis and dermis with appendiges. No inflammatory cells were seen. The only abnormality noticed was that of the adipose tissue which was markedly decreased, otherwise this was not remarkable. Dermal collagen showed focal areas of homogenization.

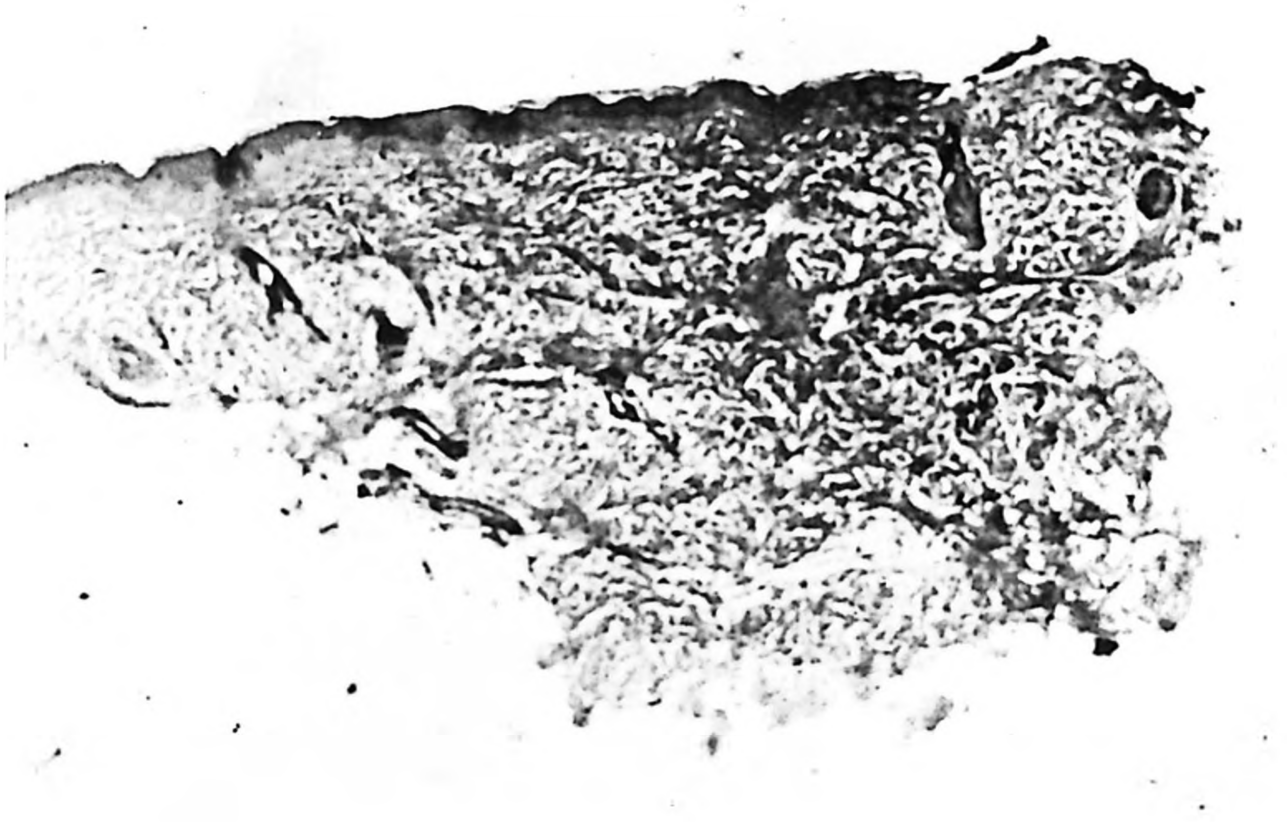

Figure 3

Punch biopsy of the skin of the face shows markedly decreased amount of subcutaneous adipose tissue. Epidermis and dermis with appendeges are not remarkable.

Punch biopsy of the skin on the leg showed slight hyperkeratosis of the epidermis. Dermal collagen showed similar focal areas of homogenization as above, but subcutaneous adipose tissue was not decreased. The impression of the skin biopsies were compatible with the clinical diagnosis of partial lipodystrophy. Needle biopsy of the kidney (Figures 4 and 5) presented 42 glomeruli and an adequate number of renal tubuli. [Sections stained with hematoxylen-eosin, P.A.S. (perodic-acid schiff), P.T.A.H. (prosphotungitic acid hematoxylen), Mason's trichrom and Prussian blue reaction for hemosiderin were examined]. The glomeruli showed various degrees of increased cellularity which was focal in some glomeruli and segmental in others and involving all glomerular taft in still others. Except for a few localized areas, basal membranes appeared not to be thickened. Basal membranes which were thickened in rare instances resembled the wireloop lesions (In

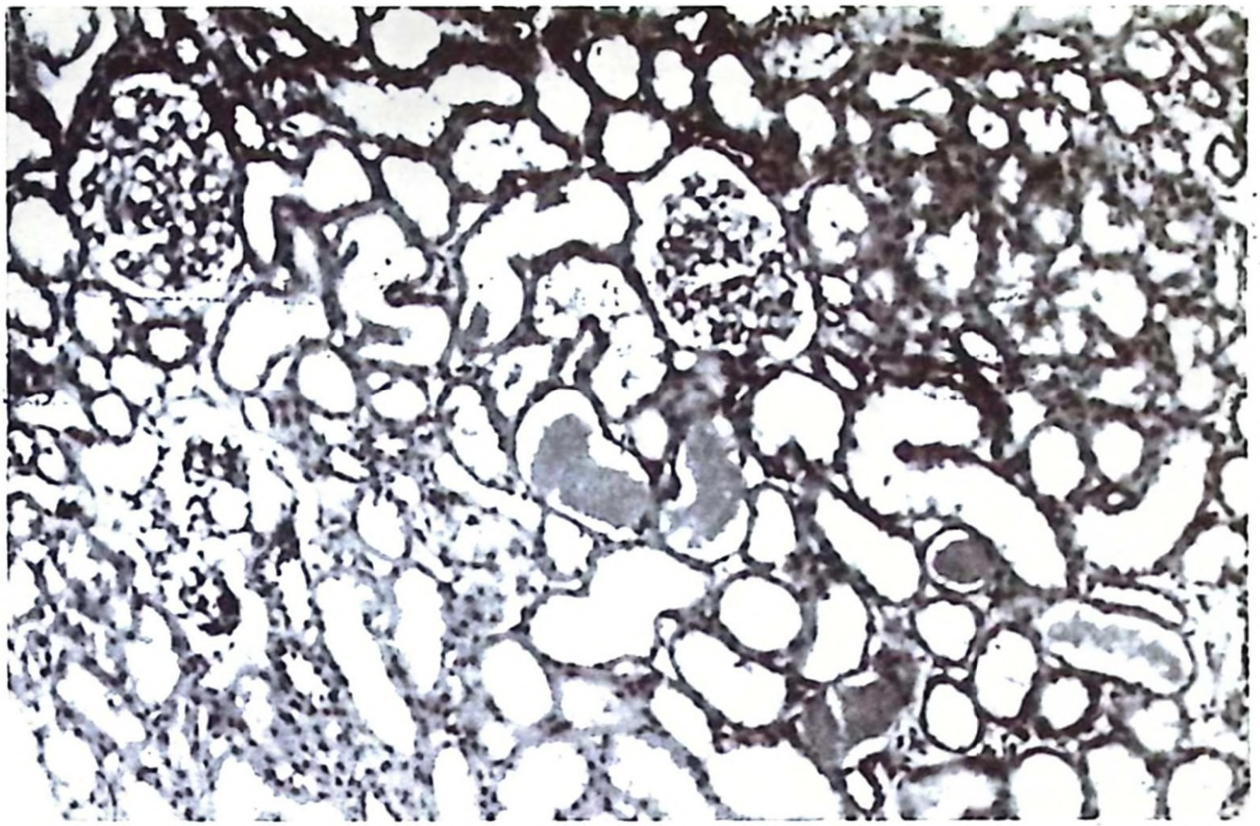

Figure 4

Needle biopsy of the kidney shows focal areas of slight dilatation of the tubuli of which contains erythrocytes and precipitated proteineccous material in the lumina.

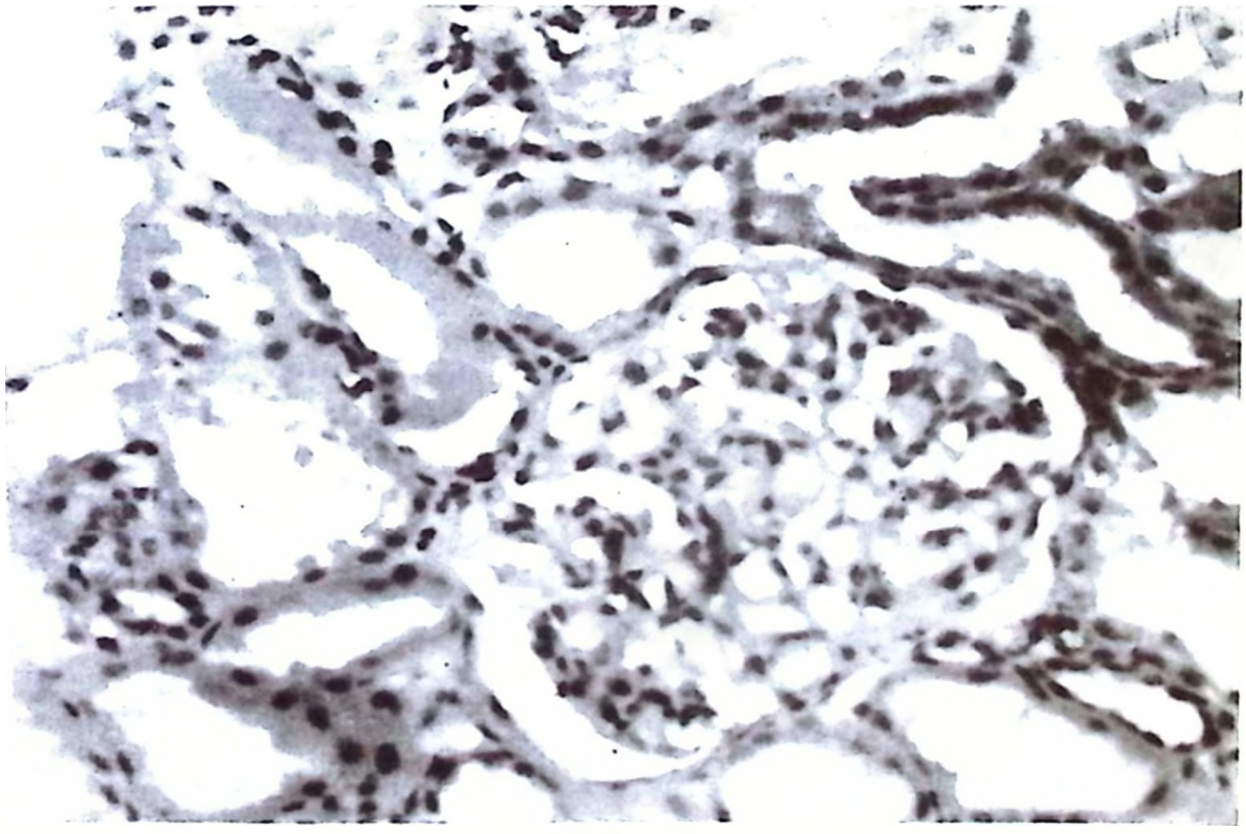

Figure 5

Needle biopsy of the kidney shows glomeruli with focal areas of increased cellularity and thickened basement membranes.

some glomeruli, debris of nuclear material was observed). The Bowman's capsule showed localized thickened areas some of which indicated few inflammatory cell reactions and foci of fibrinoid changes. The Bowman's spaces were narrowed and contained some precipitated proteineceous material and a few erythrocytes. The tubuli appeared slightly dilated, and in a few areas contained erythrocytes and precipitated proteineceous material in the lumina of the interstitial tissue Lymphocytes and plasma cell collections were seen in two focal areas. A local and mild degree of hemosiderin pigment was found in the tubuli, in the interstitium and adjacent areas of the Bowman's capsule.

The impression gathered from the kidney biopsy was that of glomerulonephritis, suggestive of autoimmunization or hypersensitivity reaction. Due to technical reasons, other more conclusive tissue study methods for autoimmune reactions could not be employed.

Clinical Course: Since E. Coli was cultured in urine Furadantin 7 mg/kg and Gantrisin 150 mg/kg were administered. Fersamal was started for iron deficiency anemia. Her hemoglobin level rose to 12.05 gr./100 ml. in three weeks. Prednisolone was started at a dose of 2 mg./kg. for renal disorder. At the end of three weeks treatment, urinalysis was as follows: specific gravity 1011, albumin negative, 3-5 erythrocytes in the sediment. She was discharged from hospital and advised to take prednisolone (30 mg a day) and gantrisin (2 gm a day). Three months later she was seen again at the out-patient clinic and found in good health with the exception of cadaveric facial appearance. At this time laboratory findings revealed: Erythrocyte sedimentation rate 10 mm/h., hemoglobin 11.73 gm./100 ml., white blood count 6000/cmm., fasting blood sugar 87 mg/100 ml., total serum protein 7.8 gm./100 ml., albumin 5.5 gm./100 ml., globulin 2.3 mg./100 ml., alkaline phosphatase 5.6 Bodansky units, total cholesterol 155 mg./100 ml., total lipid 670 mg./100 ml., serum glutamic oxalacetic transaminase 20 U., serum glutamic-pyruvic transaminase 13 U., thymol turbidity 1.3, cephalin-cholesterol flocculation (+), total bilirubin 0.8 mg.100 ml. Urinalysis: albumin negative, reaction acid, specific gravity 1014, sediment normal. There was no growth in the urine culture. Gantrisin was discontinued and prednisolone was decreased gradually.

Discussion

A definition of this syndrome was suggested by Senior and Gellis[3] as symmetrical absence of facial fat with or without disappearance of fat from the arms, chest, abdomen and hips, but with retention of distal addipose depots. They also stated that this definition was not a perfect

one. However they called attention to the need of such a definition to exclude facial hemiatrophy, malnutrition, thyrotoxicosis. Partial lipodystrophy was first mentioned by Mitcehel[9] in 1885. More than 200 cases have subsequently been published.[3] This syndrome affects females 4 times more frequently than males. Clinical onset most often occurs between 5 and 15 years of age. However it may be seen as early as the first year of life.[5]

Although, occurrence of this syndrome among relatives has been reported,[3 4] our patient did not have a history of consanguinity.

Etiology and pathogenesis of lipodystrophy is still obscure. However, various theories such as infection, endocrine disturbances and central nervous system lesions have been proposed.[3 6 7]

Zanghof and Zabel[8] showed that an autotransplant of fatty tissue to an atrophic site resulted in loss of fat and conversly transplanted atrophic tissue to an adipose site accumulated fat. They strongly suggested that in partial lipodystrophy the disorder is based on the environment of the cell rather than on the cell itself.

Clinical diagnosis is easily made in well established cases. Tuberculosis is often erroneously diagnosed as in our reported case. Although our patient had not had any positive criterion for tuberculosis, she was given antituberculous treatment by a local physician.

The only diagnostic histological change for partial lipodystrophy is the absence of subcutaneous fat in affected areas, the dermis and epidermis lie directly on the fascia and muscle.

Previously it was thought that other than the cadaverous appearance these patients usually showed no disability and had a normal life expectancy.[2 3] However hepatomegaly, disorders in glucose metabolism and hyperlipemia, disturbances of the central nervous system have been associated. Our patient had slight liver enlargement, but there were no disturbances in glucose metabolism, liver functions and the central nervous system. Recently, association of renal disorders with this syndrome has been reported.[1 3 4 7] Senior and Gellis[3] reviewed 14 cases of partial lipodystrophy with renal disorder in the literature and they also added their own 14 patients with renal involvement out of 25 partial lipodystrophy cases. The diversity of clinical diagnosis of renal diseases, ranging from pyelonephritis to acute and chronic glomerulonephritis and the nephrotic syndrome were found.

The association of glomerulonephritis with this syndrome leads to the consideration of antibody concept, possibly relating to adipose

tissue as well as the kidney. The presence of glomerulonephritis in our case is contributory for this proposal. The impression of the kidney biopsy of our patient was glomerulonephritis, suggestive of autoimmunization or hypersensitivity reaction. Due to technical reasons more conclusive methods in tissue for autoimmune reaction could not be done. Since hypocomplementamic nephritis was shown in autoimmune glomerulonephritis, the low complement level finding of our patient is considered to be contributory for autoimmune renal disease. There is no known treatment for this disorder.[5] However, treatment for facial deformity in partial lipodystrophy is possible, by means of cosmetic surgery.[2]

We could find only 28 similar cases in the world medical literature, of partial lipodistrophy with renal involvement.

Summary

An 8-year-old girl with partial lipodystrophy with renal involvement is reported. The diagnosis of our patient is confirmed by skin and renal biopsies. The impression of the kidney biopsy was glomerulonephritis, suggestive of autoimmunization or hypersensetivity reaction.

We found only 28 similar cases in the world literature. To our knowledge this is the first report from Turkey.

A possible pathogenesis of the disease is also speculated.

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