Abstract We present a new case of analbuminemia accompanied by severe lipodystrophy in a young woman. Albumin concentration was below 200 mg/l serum (i.e., less than 0.5% of physiological) as measured by kinetic immunonephelometry, but this was partially compensated by increased circulating levels of both positive and negative acute-phase proteins and of secretory proteins such as cholinesterase. This appears to be the first report describing successful correction of analbuminemia-related lipodystrophy by ultrasound-assisted lipoplasty where, in the first session, around 7800 ml fat was aspirated. The possible relationships between the diagnosed disturbances in lipid metabolism (type IIa hyperlipoproteinemia, increased free fatty acids) and the severe dysregulation in lipid deposition are discussed.

Keywords Analbuminemia · Lipodystrophy · Liposuction

Introduction

Analbuminemia in humans is a rare inherited disease which is transmitted in an autosomal recessive manner. Only 30 cases have been reported worldwide, with no geographical or gender preference [7, 9, 11, 14]. In several patients genetic analyses have been performed, and these reveal point mutations at various regions or a nucleotide insertion in the albumin gene producing a premature stop codon; in one case, a splicing mutation was found resulting in defective ligation of two exons. As a consequence, albumin fragments instead of the complete molecule might be generated, but such fragments have never been detected in serum [11, 13, 18].

In homozygous individuals only 1/50–1/2000 of physiological albumin concentrations is measured, while only slight hypoalbuminemia occurs in heterozygous carriers. Despite the virtual absence of albumin, analbuminemic patients can obviously survive, although there is clear evidence of frequent abortions in the affected families, pointing either to a critical role of albumin itself or to the development of compensatory strategies during fetal life [11, 13, 18].

Edema, fatigue, and low blood pressure are the most frequent symptoms reported in analbuminemic patients [11, 14]. A compensatory increase in other serum proteins such as IgG may maintain the oncotic pressure at a sufficient level. In those cases in which further laboratory analyses have been performed, hypercholesterolemia and hypertriglyceridemia are also reported [14], and metabolic analyses have further benefited from an animal model, the Nagase analbuminemic rat [6, 8, 12]. It is noteworthy that several analbuminemic women are reported to suffer from severe lipodystrophy [7, 11, 14].

We present a recently diagnosed case of analalbuminemia in a woman with severe lipodystrophy. In addition to extensive investigation of serum protein levels and lipid status esthetic correction by lipectomy is described for the first time with regard to analbuminemia. Liposuction in general has been proven as a low-risk curative procedure for removing extensive lipohypertrophic tissue [2, 5]. In this particular case, ultrasound-assisted lipoplasty was used; this gives cosmetically satisfactory results with minimal side effects [19].

Case report

The patient, a woman of Turkish origin, was born in Germany in 1978 and presented for the first time in February 1999 to the plastic Surgery outpatient program of the Florence Nightingale Hospital. She complained of a disproportional fat distribution on her lower body and requested body contouring and liposuction, stating that until the age of 13 years she had normal body propor-
Fig. 1A–D Esthetic correction of lipodystrophy in the analbuminemic patient. A, B Severe disproportion between upper body and the legs and thighs hand prior to liposuction. C, D Improvement noted especially in the waist and hip regions 4 months after surgery. A second liposuction is planned in the near future.

Table 1 Selected serum constituents of the proposita. Value 1 is from a serum specimen obtained in the morning of the day prior to surgery, value 2 is from the 1st postoperative day, value 3 from samples 9 days thereafter (LDL low-density lipoprotein, HDL high-density lipoprotein, AGE-P agarose gel electrophoresis–paragon blue, BTCI butyrylthiocholine iodide, CHOD-POD cholesterol oxidase–peroxidase, ECLIA electrochemiluminescence immunoassay, FPIA fluorescence polarization immunoassay, GC-FID gas chromatography–flame ionization detection, GPPO-POD glycerophosphate oxidase–peroxidase, K-IN kinetic immunonephelometry, MEIA microparticle-enzyme immunoassay, n.d. not detectable)