Introduction: Lipid storage myopathy (LSM) is characterized by increased lipid droplets in muscle fibers. Primary carnitine deficiency is the most frequent cause of LSM, clinical presentation ranging from asymptomatic to progressive muscle weakness or cardiomyopathy, carnitine supplementation being effective with remission of symptoms.

Case report: In February 2007 R.A. born in 1996 presented progressive muscle weakness with elevated muscular enzymes (LDH=1155 UI/ml, n=125–234, and CPK=1082 UI/ml, n=25–195). Starting from a muscular biopsy, which suggested polymyositis, a glucocorticoid trial was initiated with partial amelioration followed by clinical relapse. Development of Cushingoid syndrome and growth retardation (height 123 cm, −3 s.d.; weight 22 kg, −2.5 s.d.; normal parental heights) determined endocrinological evaluation which revealed normal IGF1 (172 ng/ml, n=111–551), and basal GH (3.3 µUI/ml) with response at stimulation (stimulated GH 125 µUI/ml) and delayed bone age of 8.5 years. Functional pituitary evaluation was normal. Reevaluation by muscular biopsy revealed presence of increased lipid droplets in type 1 muscle fibers suggesting LSM. Carnitine supplementation was started in December 2008 and continued without pause (1 g/day) with progressive clinical improvement and normalization of muscular enzymes.

Absence of GH deficiency justified expectative but the stagnant height after 6 months was an argument for hGH therapy, which was started in October 2008 followed by a satisfactory growth rate (~0.5 cm/month). Reevaluation in July 2013 revealed Tanner pubertal stage IV, height of 153 cm (−1.95 s.d.) and delayed bone age of 14 years.

Conclusion: We report a patient with short stature and LSM who responded well to hGH therapy and carnitine supplementation. To our knowledge, children with LSM do not usually associate short stature or GH deficiency. Impact of hGH therapy on body composition and muscle structure in LSM cases needs to be further evaluated.
Short stature and carnitine deficiency: the hidden connection

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Systemic primary carnitine deficiency (CDSP) is an autosomal recessive disorder of carnitine transportation. The clinical manifestations of CDSP can vary widely with respect to age of onset, organ involvement, and severity of symptoms, but are typically characterized by episodes of hypoketotic hypoglycemia, hepatomegaly, elevated transaminases, and hyperammonemia in infants; skeletal myopathy, elevated creatine kinase (CK), and cardiomyopathy in childhood; or cardiomyopathy, arrhythmias, or fatigability in adulthood. CDSP should be differentiated from secondary causes of carnitine deficiency such as various organic acidemias and fatty acid oxidation defects.

Systemic primary carnitine deficiency (SPCD) is an inborn error of fatty acid transport caused by a defect in the transporter responsible for moving carnitine across the plasma membrane. Carnitine is an important amino acid for fatty acid metabolism. When carnitine cannot be transported into tissues, fatty acid oxidation is impaired, leading to a variety of symptoms such as chronic muscle weakness, cardiomyopathy, hypoglycemia and liver dysfunction. The specific transporter involved with SPCD is OCTN2. Carnitine deficiency occurs in a wide variety of other conditions and is considered secondary to those conditions. Many inborn errors of metabolism cause secondary carnitine deficiency; the identification of low carnitine levels in a patient should stimulate an intensive evaluation for metabolic disease. The most common causes are genetically determined defects in fatty acid oxidation and amino acid oxidation. Other causes include defects in mitochondrial function, which may reflect genetic abnormalities in mitochondrial DNA.