

# Short Stature with Lipodystrophy: Reminder of a Forgotten Syndrome

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## BACKGROUND

The combination of various severe manifestations of hypothyroidism with pseudo muscular hypertrophy is called Kocher Debre Semelaigne syndrome (KDS). KDS is very rare in countries where newborn screening for hypothyroidism is in place. Most of the reports of KDS have come from India and developing countries with only a single report from Europe over last five decades (1). Earlier, one case each was reported from Italy and Portugal (2,3) in early 1970's and none from UK till date. We present a 7-year-old boy from UK who had short stature and apparent partial lipodystrophy.

## CASE PRESENTATION

A 7-year-old Caucasian boy was referred with possible partial lipodystrophy. His past history included bilateral uveitis diagnosed at 7 months. On examination, he had a sallow complexion, slightly dry skin and marked loss of fat with athletic physique and prominent musculature in lower limbs. His facial appearance was sallow with sparse eyebrows and chubby cheeks. He had protruding abdomen with small umbilical hernia and hepatomegaly. His height SDS was  $-2.07$ . Investigations demonstrated TSH  $>100$  mU/l, free T<sub>4</sub> 1.2 pmol/l, ALT levels of 175 U/l with normal IGF1 levels, lipids, OGTT, coeliac screen, and leptin levels. Bone age was markedly delayed (2 years and 8 months at 7 years). Anti-TPO antibodies were positive. USG of thyroid was normal and USG of liver suggested fat accumulation. He was started on Levothyroxine replacement. His thyroid function, ALT levels, hepatomegaly, growth, delayed bone age, complexion, and facial appearance have improved gradually over time (current height SDS  $-1.5$ ). The fat loss and pseudomuscular hypertrophy is improving more slowly.

## CONCLUSIONS

Long standing untreated hypothyroidism can cause severe fat loss associated with other systemic effects. Although KDS is very rare in the developed world with newborn screening in place, it can still occur with unrecognized autoimmune hypothyroidism. This association with hypothyroidism is also highlighted in two recent reports from Turkey (4,5). Recognition of this condition is important to facilitate prompt treatment and to prevent further unnecessary investigation.

## REFERENCES

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