Endocrinopathies in a 17-years-old girl with Diamond Blackfan anemia and transfusional iron overload

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Objectives:
Diamond Blackfan anemia (DBA) is an inherited bone marrow failure syndrome, which presents with anemia in early infancy. Survival depends on blood transfusions, which in consequence lead to iron overload (IOL). The most common complications of IOL are hepatic cirrhosis, endocrinopathies and cardiomyopathy.

Results:
We present the case of 17 years old girl with DBA and IOL-associated endocrinopathies. Her treatment consists of multiple blood transfusions and corticosteroids. Iron chelation therapy with deferasirox and desferrioxamine was started at the age of 10 years (irregularly). At first examination in our clinic at age of 15 years the girl was severely growth retarded (-4,20 SDS, bone age=15 years, serum IGF-I level were within normal ranges for age) and had excess weight (1,10 SDS BMD). Serum ferritin (SF) level was in the range of 1785-4277 ng/ml (IOL - SF>1000 ng/ml). Hypogonadotropic hypogonadism was diagnosed at the age of 15 (there was no response FSH/LH to LHRH stimulation; AMH, marker for ovarian reserve, was normal), impaired glucose tolerance (75-g oral OGTT was performed), also insulin resistance (ISI Matsuda=2,5 (N>2,6), HOMA-IR=4,6 (N<3,2)), second hyperparathyroidism and osteoporosis (BMD of the lumbar spine was examined, Z-score= -2,7SD) were found. Basal adrenal and thyroid function were normal. We used (3T MRI) to measure pituitary volume and iron concentration. The pituitary height and volume were significantly decreased: V=64-88 mm³ (N 305- 86 mm³). Results of pituitary T2* MRI were in the range of 6,9-7,1 ms (N>20 ms, Pic. 1, 2).

Conclusions:
Pituitary iron overload and volume loss are independently predictive of hypogonadism. MRI (T2*) can be used as a non-invasive tool to recognize pituitary IOL and identification of proper chelation therapy for the prevention of irreversible pituitary tissue damage. Our observations emphasize the importance of periodic, meticulous evaluation of the endocrine function of patients with transfusion-associated IOL.