Introduction:

Diamond-Blackfan anemia (DBA) is a rare, genetically determined disorder which is manifested with selective deficiency of erythrocyte line in bone marrow. Patients with DBA are diagnosed with a hypocellular bone marrow with a significant absence of erythroid precursors typically in early childhood. They require chronic treatment with corticosteroids and frequent blood transfusions, stem cell transplantation is also possible. For those, who don’t respond to corticosteroids therapy, survival depends on blood transfusions. The most common complications of transfusional hemochromatosis are hepatic cirrhosis, cardiomiopathy and endocrinopathies such as hypopituitarism, hypogonadism, hypothyreosis or diabetes mellitus. DBA patients, therefore, require long-term chelation therapy to avoid these complications.

CASE REPORT: A 31-year old male patient with DBA diagnosed at the age of 8, maintained on chronic blood transfusions repeated every 4 weeks since the age of 18 years and iron chelation therapy (deferoxamine) with poor adherence, was admitted to the Department of Endocrinology with suspected primary adrenal insufficiency. Since several months the patient complained about weakness, fatigue, orthostatic hypotension and hyperpigmentation of the skin. The clinical observation and the morning plasma ACTH and cortisol levels excluded primary adrenal failure: 32 pg/ml (7.2-63) and 15.4 ug/dl (4.3-22.4), respectively. Further hormonal assessments suggested hypogonadotropic hypogonadism: LH: 1.8 mIU/ml (1.5-9.3); FSH: 2.8 mIU/ml (1.4-18.1); testosterone: 26 ng/dl (241-824) and the growth hormone deficiency: GH: 0.44 ng/ml (0.03-2.47); IGF-1: 38 ng/ml (113-202), probably in the course of secondary hemochromatosis: plasma Fe 211 ug/dl (65-175), ferritin 5442 ng/ml (22-322), following blood transfusions. MRI of the sella turcica revealed a small pituitary gland with poor enhancement after gadolinium injection, what might confirm iron deposits in this region.

Conclusions:

Pituitary dysfunction following secondary hemochromatosis should be considered in patients maintained on chronic blood therapy. Accurate evaluation and long-term follow-up of all patients with iron over-load are necessary in order to detect the occurrence of hypopituitarism, regardless of clinical evidence for pituitary dysfunction. An adequate replacement therapy is necessary in order to improve quality of life and outcomes in this group of patients.