Endocrine challenges in patients with thalassaemia

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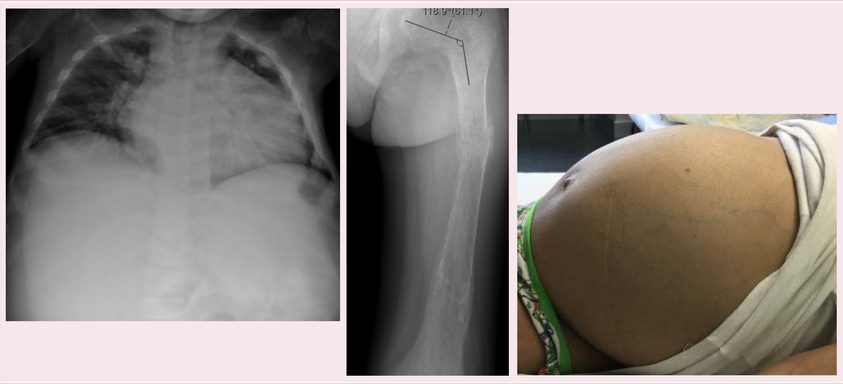
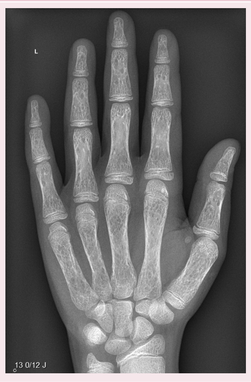
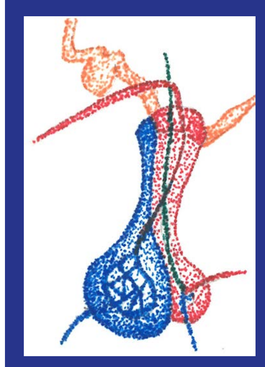
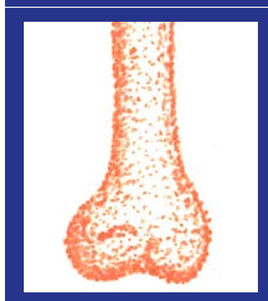
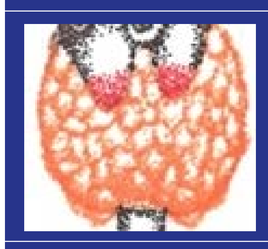
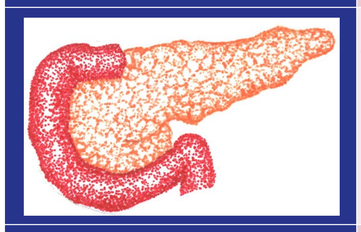
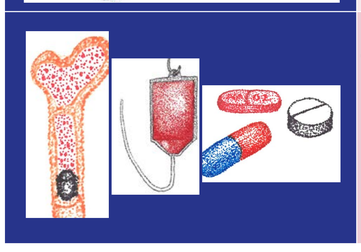
Introduction

Beta-thalassaemia is caused by point mutations leading to decreased production of beta-globin, which results in defective red blood cells and ineffective erythropoiesis. Complications are microcytic hypochromic anaemia, extramedullary haematopoiesis and increased intestinal iron absorption due to compensation mechanisms. The resulting iron overload can be aggravated by recurrent blood transfusions necessary for treatment of anaemia and may cause several endocrine complications such as pituitary dysfunction, diabetes, hypoparathyroidism and hypothyroidism. Beta-thalassaemia intermedia/major may only be cured by a haematopoietic allogenic stem cell transplantation. In developed countries and with optimal treatment possibilities, patients with severe complications are rarely seen. But unfortunately this is not reality for all children.

Case reports

Three refugees from the Middle East came to our institution for treatment late. They all suffered from beta-thalassaemia major and had severe complications due to inadequate therapy in their past.

Results	Patient 1	Patient 2	Patient 3
General information	14 years old female, from middle east	17 years old female (sister of patient 1)	15 years old male, from middle east
Auxology	Weight 26.3 kg (-3.75 SDS), height 120cm (-6.37 SDS)	Weight 42.3kg (-1.89 SDS), height 139.7cm (-3.56 SDS)	Weight 31.2kg (-3.65SDS), height 133cm (-4.37SDS)
Ferritin (7–140µg/l)	19790 µg/l	10422 µg/l	8054 µg/l
Picture			
	Delay of growth and puberty ➔GH-therapy postponed (because of general medical condition)	Delay of growth and puberty ➔Adequate GH secretion in insulin tolerance test at age 18 ➔Spontaneous beginning of puberty	Growth hormone deficiency ➔GH treatment started at age 16 Hypogonadotropic hypogonadism ➔Puberty induction started at age 17
	Severe osteopenia-osteoporosis syndrome Vitamin D deficiency Hypoparathyroidism ➔Calcitriol ➔Calcium	Vitamin D deficiency ➔Vitamin D supplementation	Vitamin D deficiency ➔Vitamin D supplementation
	Primary hypothyroidism ➔Thyroid hormone replacement	Normal thyroid function	Normal thyroid function
	Diabetes mellitus ➔functional insulin treatment started at age 14	Impaired fasting glucose ➔ Oral glucose tolerance test planned	Diabetes mellitus ➔ functional insulin treatment started at age 14
	Hemosiderosis ➔transfusion every 3 weeks ➔oral iron chelators	Hemosiderosis ➔transfusion every 3 weeks ➔oral iron chelators	Hemosiderosis ➔transfusion every 2 weeks ➔oral iron chelators

Conclusion

There are special medical challenges in suboptimal treated thalassaemia patients, which include severe endocrine complications. These are seen in refugee children in our units as a result of war and lack of medical care. An interdisciplinary and individual approach is important to improve the health situation of these patients, in whom some permanent damage is unfortunately irreversible.