Endocrine challenges in patients with thalassaemia

Tanja Christa Haamberg1, Schneider Christine2, Jochen Rössler2, Christa Emma Flück1

1 Department of Pediatrics (Division of Pediatric Endocrinology and Diabetology), Inselspital, Bern University Hospital, University of Bern, Bern, Switzerland
2 Department of Pediatrics (Division of Pediatric Hematology and Oncology), Inselspital, Bern University Hospital, University of Bern, Bern, Switzerland

The authors have nothing to disclose / corresponding author tanja.haamberg@insel.ch

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
<table>
<thead>
<tr>
<th>Patient 1</th>
<th>Patient 2</th>
<th>Patient 3</th>
</tr>
</thead>
<tbody>
<tr>
<td>General information</td>
<td></td>
<td></td>
</tr>
<tr>
<td>14 years old female, from middle east</td>
<td>17 years old female (sister of patient 1)</td>
<td>15 years old male, from middle east</td>
</tr>
<tr>
<td>Auxology</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Weight 26.3 kg (-3.75 SDS), height 120cm (-6.37 SDS)</td>
<td>Weight 42.3kg (-1.89 SDS), height 139.7cm (-3.56 SDS)</td>
<td>Weight 31.2kg (-3.65SDS), height 133cm (-4.37SDS)</td>
</tr>
<tr>
<td>Ferritin (7–140 μg/l)</td>
<td>19790 μg/l</td>
<td>10422 μg/l</td>
</tr>
<tr>
<td></td>
<td></td>
<td>8054 μg/l</td>
</tr>
</tbody>
</table>

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

Primary hypothyroidism
- Thyroid hormone replacement

Normal thyroid function

Diabetes mellitus
- Functional insulin treatment started at age 14
- 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.