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The authors have nothing to disclose

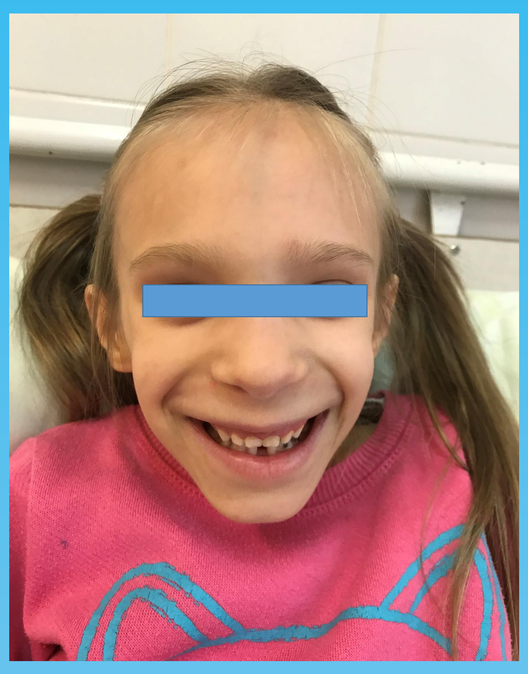
Bloom Syndrome in 7-year-old girl diagnosed with short stature

Hanna Borysewicz-Sańczyk[1], Beata Sawicka[1], Emily Cottrell[2], Tasneem Ladha[2], Helen Storr[2], Artur Bossowski[1]

[1]Department of Paediatrics, Endocrinology, Diabetology with a Cardiology Division, Medical University of Bialystok, Poland,
[2]Centre for Endocrinology, William Harvey Research Institute, Queen Mary University of London, UK

Introduction:

Bloom syndrome (BS) is a rare autosomal recessive disorder caused by mutations in the BLM gene, located on the long arm of the chromosome 15 (15q26.1). The typical symptoms of the disease are short stature, low birth weight, dysmorphic features including long, narrow face (dolichocephaly), micrognathism and prominent nose and ears. Other characteristic features include a rash following sun exposure, hyper-pigmented areas or cafe-au-lait spots on the skin, high-pitched voice, immune deficiency leading to recurrent pneumonia and ear infections, predisposition to the development of cancer and genomic instability. Patients with BS may also have learning disabilities and a predisposition to diabetes.



Case presentation:

We present a 7-year-old girl with hypothyroidism (treated with L-thyroxine 25 ug) referred to the Department of Paediartics Endocrinology, Diabetology with Cardiology, Medical University of Białystok due to short stature. The patient was born at term (39/40) with a birth weight of 1580g (SDS -4.66) with a length of 44cm (small for gestational age, SGA). From birth, she had recurrent infections of upper and lower respiratory tract, frequently requiring antibiotic treatment. Physical examination revealed substantial short stature (SDS -5.25) and low BMI, dysmorphic features with long narrow face, micrognathism and cafe-au-lait spots on her abdomen and right popliteal fossa. General laboratory tests were normal. Further analysis revealed growth hormone (GH) deficiency with a delayed bone age of 4.5 years. Suspecting a genetic abnormality, we referred her to the 'Genetic Research Analysing Short Patients' (GRASP) team at the Centre for Endocrinology in London, UK.
After a positive opinion of National Coordination Team for Growth Hormone Application the treatment with GH was initiated as for GH deficiency patients (initial dose of GH 0.54 U/kg/week). The growth rate of the patient over 9 months of treatment was 5.4 cm/year (5.8 cm/year prior to GH treatment).

The GRASP team identified a homozygous mutation in BLM gene (91306246C>T, c.1933C>T, p.Q645*) which is recognised to cause Bloom syndrome. After we recieved genetic confirmation of BS, the treatment with GH was stopped due to the risk of cancer development.

Conclusions:

Genetic diagnosis in children with short stature and concomitant dysmorphic features is particularly important and some extremely rare syndromes might be a contraindication to GH therapy.