УДК: 61:615.1(043.2)
ББК: 5:52.8
ISBN: 978-985-21-0532-3

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CASE OF 7-YEAR-OLD GIRL WITH BLOOM SYNDROME

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Bloom syndrome is a rare (1/48 000 births) disorder with significant chromosome fragility caused by mutation in the BLM gene located on the long arm of the 15th chromosome (15q26.1) that encodes helicase RecQ13. It is an autosomal recessive disease. In patients with this syndrome short stature, low birth weight, dysmorphic features including long, narrow face (dolichocephaly), micrognathism and prominent nose and ears are observed. Moreover frequent respiratory and digestive tract infections are linked to immunodeficiency occurring in this disorder. Very characteristic symptoms are also a rash on the skin that develops after the exposition to the sun, hyper-pigmented areas or cafe-au-lait spots and high-pitc hed voice. Due to mutation in gene encoding helicase RecQ13 patients with Bloom Syndrome are more prone to develop cancer in early age, especially leukemia or lymphoma, and adenocarcinoma in adulthood comparing to the rest of the population. Patients with BS may also manifest some learning disabilities and have predisposition to diabetes.

7-year old girl was admitted to the Department of Paediatrics, Endocrinology, Diabetology with Cardiology Division, Medical University of Białystok due to short stature. Medical history of the patient revealed hypothyroidism treated with L-thyroxine 25 ug. She was born at therm (39 weeks of pregnancy) with body mass 1580g (<5 pc) and length 44cm (small for gestational age, SGA) and since then the patients suffered from recurrent infections of upper and lower respiratory tract, frequently treated with antibiotics. Performing physical examination of the girl we observed substantial short stature (-5,25 SD) and body mass deficiency, dysmorphic features with long narrow face, micrognathism, cafe-au-lait spots on the skin of abdomen and right popliteal fossa. Laboratory tests revealed growth hormone (GH) deficiency while general parameters remained on a normal level. Thyroid hormones concentration with supply of L-thyroxine was normal. Adrenal gland dysfunction was excluded. We assessed bone age for 4 years and 6 months. As genetic abnormality was suspected, the blood sample for genetic tests was collected and sent to laboratory. 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 after 9 months of the treatment was 5.4 cm/year (before treatment 5.8 cm/year). Nevertheless the treatment with GH had to be stopped due to genetic confirmation of Bloom syndrome and possible increased risk of cancer development.

Some rare genetic disorders such as Bloom syndrome should be taken into consideration while diagnosing children with short stature and concomitant dysmorphic features. Due to possible increased risk of cancer development in such patients standard GH therapy may be contraindicated.