A Young Diabetic Case with Bloom Syndrome

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Bloom syndrome (BS) is a rare autosomal recessive disorder characterized by short stature, telangiectasias, and a rash on the face triggered by the sun. Other clinical features include immune deficiency and predisposition to the development of cancer and diabetes. We present this case as the diabetic BS is rare.

Case: A 20-year-old male patient who had diabetes for 3 years was admitted. The patient had complaints of rash on sun-exposed parts of the body such as cheeks and arms since he was 2 years old. In his medical history, he was found to be followed up for growth failure. His parents had a cousin marriage. The diepoxybutane test was performed as BS was suspected due to sun-induced rashes, growth failure, and telangiectasias. The chromosome analysis showed that our case had 40 breakages per 100 metaphases, whereas there were 2 breaks in control. Immunoglobulin levels were low. BS was diagnosed with phenotypic findings. The physical examination revealed rashes and telangiectasias on the face. He was 152 cm and 49 kg. In the oral glucose tolerance test (OGTT) of 3 years earlier, fasting plasma glucose (FPG) was found to be 125 mg/dL and in the 2nd hour of the test, plasma glucose was 295 mg/dL and fasting insulin was 57.73 IU/mL. He was diagnosed with diabetes according to OGTT. The laboratory findings of our patient who was on metformin were: FPG 91 mg/dL, hemoglobin A1c 6%, and basal insulin 29.3 IU/mL. In the follow-ups, metformin was observed to regulate his blood glucose levels.

Results: Diabetes is diagnosed in 10% of patients with BS, most often type 2 diabetes. BS is caused by mutations in the *BLM* gene on chromosome 15q26.1. The genomic instability characterized by mutations and the elevated rate of sister chromatid exchange lead to tendency to malignancy. These patients should be closely followed for malignancy and development of diabetes.

Key words: Bloom syndrome, type 2 diabetes, genomic instability, malignancy, diepoxybutane test

A Case of Dyskeratosis Congenita Associated with Hypothyroidism and Hypogonadism

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Dyskeratosis congenital (DC) is a rare multisystemic disease characterized by skin atrophy, pigmentation, nail dystrophy, leukoplakia in mucous membrane, bone marrow failure, and tendency to malignancy. We present a rare case of DC associated with hypothyroidism and hypogonadism.

Case: A 30-year-old male patient was referred to us with the findings of micropenis and atrophic testicles. His parents had a cousin marriage. His physical examination results were as follows: blood pressure 130/80 mmHg, BMI 28 kg/m^2. There were hypopigmented skin lesions on the whole body. The skin was dry and the nails were dystrophic. Axillary hair and pubic hair were normal, but facial hair was scarce. He had alopecia in 1/3 of outer eyebrow (Omnibus sign) and saddle nose. Micropenis was present and testicles were found to be hypoplasic. During the auscultation, roncus was common and expiration was long. Laboratory findings were as follows: WBC 3140/mm^3, neutrophils 1740/mm^3, Hb 12.5 gr/dL, plt 187000/mm^3, free testosterone 1.4 pg/mL, total testosterone 0.70 ng/mL, follicle-stimulating hormone (FSH) 77.28 mIU/mL, luteinizing hormone (LH) 15.38 mIU/mL, estradiol <20 pg/mL, prolactin 6.77 ng/mL, free triiodothyronine 3.35 pg/mL, free thyroxine 0.98 ng/dL, thyroid-stimulating hormone 10.88 µU/mL, anti-thyroglobulin 203 IU/mL, adrenocorticotropic hormone 24.1 pg/mL, and cortisol 13.34 µg/dL. Hypergonadotropic hypogonadism was suspected as testosterone was low and FSH and LH were high. Hyperkeratosis keratoderma was found in the skin biopsy. In the biopsy of cervical lymph node from the right side, we found disseminated histiocytic proliferation changing the normal appearance of the lymph node and granulomas that were characterized by rare giant cell formation without necrosis.

Conclusion: DC generally shows X-linked recessive inheritance along with autosomal dominant and recessive forms. Although the pathogenesis of the disease is still unknown, *DKC1* gene localized to Xq28 is thought to be responsible for the X-linked DC. Our patient is still genetically studied. In this syndrome, the association of hypothyroidism and hypogonadism should be kept in mind.

Key words: Dyskeratosis congenital, nail dystrophy, hypothyroidism, hypogonadism, bone marrow failure