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Pancreatic cancer in bloom syndrome

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Abstract

Bloom syndrome is a rare autosomal recessive disorder characterized by distinct physical features, such as short stature, genomic instability, and predisposition to numerous cancers. The *BLM* gene encodes for the RecQ helicase that plays an important role in genome editing, maintenance, and stability. Mutations in the *BLM* gene cause genomic instability that exposes the carriers to a variety of cancers, and in particular hematological and gastrointestinal cancers. Herein, we report the first case of pancreatic cancer in a 32-year-old patient with bloom syndrome.

Keywords

Cancer prevention, cancer screening, oncology

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Introduction

Bloom's syndrome (BS) was first described in 1954,[1] but fewer than 300 cases were documented.[2] It is a rare autosomal recessive genetic disorder characterized by defects in the *BLM* gene, which codes for the DNA repair enzyme RecQL3 helicase, resulting in genetic instability characterized by elevated levels of spontaneous sister chromatid exchanges (SCEs) and chromosomal radial formation.[3] The Ashkenazi Jewish population is particularly susceptible, as ~1% of this population is heterozygous carriers of the *BLM* gene.[3] This mutation is the most common BLM mutation, designated as blm^{Ash}.[4]

The clinical picture frequently includes short stature, a facial rash, and recurrent infections due to severe immunodeficiency. The genomic instability is also manifested by an increased susceptibility to a wide range of cancers, in particular, hemato-oncology and gastrointestinal tumors; thus, frequent screenings and awareness by physicians are imperative[1–4]. DNA sequencing can be used to diagnose carriers, and high-risk populations should undergo genetic testing.

Cancer is the leading cause of death for subjects with BS. No patients have been reported to reach the age of 50 (median lifespan is <30 years.). Herein, we report the first case of pancreatic cancer (PC) in BS.

Case

A 32-year-old male with BS and a past medical history significant for well-differentiated pancreatic adenocarcinoma, diabetes mellitus type II, and restrictive lung disease presented for a consultation and annual checkup at the Integrated Cancer Prevention Center (ICPC) at Tel Aviv Medical Center.

At presentation, he was 147 cm and 33.4 kg (body mass index (BMI) 15.5). He was presented with no complaints and his physical examination was unremarkable, except for severe cachexia. He was examined, as all patients are at the ICPC, by specialists in internal medicine, surgery, plastic surgery, urology, oral surgery, and gastroenterology.[5] He also underwent routine imaging (US abdomen) and blood tests.

A well-differentiated pancreatic adenocarcinoma (T2N0M0) was diagnosed on March 2018 by abdominal computed tomography (CT) and increased blood levels of carcinoembryonic antigen (CEA) and CA19-9. He underwent near total pancreatectomy. Histology was positive for CA19-9 and CEA, and negative for P53 and SMAD4. No tumor-based genetic testing was done.

Discussion

BS is a rare human autosomal recessive disorder with the highest prevalence in Ashkenazi Jews. It is associated with a

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vastly increased predisposition to a wide range of cancers. BS poses a risk for a number of cancers, including leukemias, lymphomas, and carcinomas—all of which present at an early age. The mean age at diagnosis for acute myelocytic leukemia (AML) was age 18 years (range 2–47) and for acute lymphocytic leukemia (ALL), it was age 20 years (range 5–40).[6]

Patients with BS have an excess not only of common cancers but also of rare cancers, for example, Wilms tumor. Surprisingly, there are no reports in the literature of PC in this setting. It is odd, as PC is common in other syndromes with DNA replication errors like lynch syndrome, BRCA mutations, retinitis pigmentosa, and so on.[1–4] Ashkenazi Jews have an increased incidence of BS and thus have an increased risk of developing PC.

Conclusion

Herein, we report the first case of PC in the setting of BS. Given the wide range of cancers associated with BS, we suggest that BS patients should have an extensive annual checkup focusing on early cancer detection, and it should be conducted at a tertiary care facility such as the Integrated Cancer Prevention Center in the Tel Aviv Medical Center.

Declaration of conflicting interests

The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Ethical approval

Ethical approval to report this case was obtained from the Institutional Review board at Tel Aviv Sourasky Medical Center.

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Informed consent

Written informed consent was obtained from the patient for their anonymized information to be published in this article.

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