Acta Scientific MEDICAL SCIENCES (ISSN: 2582-0931)

Volume 5 Issue 9 September 2021

Case Report

Bloom Syndrome: Case Report

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Received: July 07, 2021

Published: August 04, 2021

Abstract

Bloom syndrome (BS) is characterized by chromosomal instability and increased risk of malignancy at an early age. We report the case of 38 years old man having two brothers suffering from bloom syndrome. Our patient developed anaplasic T lymphoma revealed by multiple cervical adenopathies and complicated with pulmonary embolism. Patient died before beginning the chemotherapy.

Keywords: Bloom Syndrome; Lymphoma; Chromosomal Instability; Cytogenetically Analysis

Introduction

Bloom syndrome is an extremely rare, inherited autosomal recessive genetic disorder caused by a mutation in the BLM gene. It is evoked in patient with short stature, a photosensitive rash, telangiectasis and pigmentary disorders. Other features can exist such include learning disabilities, diabetes, chronic obstructive pulmonary disease and recurrent infections of the upper respiratory tract and ears [1]. Patient with Bloom syndrome have also an increased risk of cancer. They can develop any type of cancer, but the cancers arise earlier in life compared to the general population, and affected individuals often develop more than one type of cancer [2]. In this report we present the clinical data of a patient suffering from anaplastic T lymphoma complicating a bloom syndrome.

Case Presentation

An 38 years old man presented with multiple peripheral adenopathies, fever and weight loss since 2 months. His family history showed two brothers followed up in dermatology department for achromic lesions related to a bloom syndrome. At admission, physical examination showed multiple dysmorphic manifestations including a long, narrow face a small lower jaw and prominent nose and ears (Photo 1). There was also right jugular adenopathies and the biggest was 5cm in diameter. Abdominal examination showed hepatomegaly at 15 cm and moderate ascites. Blood tests revealed anemia at 8.1 g/dl, lymphopenia at 264 element, hypoalbuminemia at 22.5 g/L, and hypogammaglobulinemia at 5.4 g/L. There are also hepatic cytolysis and cholestasis with a negative serology B and C hepatitis. Ascites punction showed exudative liquid. Cervical ultrasound revealed multiple jugular right adenopathies. Body computed tomograghy scan showed cervical, axillar, mediastinal adenopathies, nodular hepatomegaly and heterogenous splenomegaly with intraperitoneal effusion. Biopsy of cervical adenopathy was in favor of anaplastic T lymphoma. During hospitalization, the patient presented an acute respiratory failure related to distal pulmonary embolism. Family decided to stop taking him charge and patient died 7 days after at home. The diagnosis of bloom syndrome complicated with lymphoma is very likely in our patient but cytogenetic analysis could not be performed.

Photo 1

Discussion

Bloom syndrome is an autosomal recessive disease resulting from a defect of the cell's DNA repair system, which incriminates a gene located on 15q26.1. This gene is responsible for encoding a protein known as BLM. The chromosomal breakage sites have been correlated with locations of oncogenes and breakpoints in chromosomal rearrangements, which are linked to cancers [2]. Thus, patients with Bloom syndrome have an overall 150- to 300-fold increased risk of malignancy compared with the general population [4]. The most frequent malignancies are acute leukemia, lymphoma, gastrointestinal adenocarcinoma, and squamous cell cancer of the skin. The mean age cancer onset is 24.7 years and over one-third die at a mean age of 24 years. The diagnosis of BS should therefore be considered in all patients with a malignancy of unusually early onset, short stature and a photosensitive rash [4]. Cytogenetic analysis looking for increased sister chromatid exchanges can then confirm or refute the diagnosis the identification of the syndrome is mandatory to follow-up and diagnose malignant lesions earlier in our case report two brothers of our patient were cytogenetically diagnosed with bloom syndrome, screening early symptoms and regular check up is mandatory for early diagnosis of any type of cancer and therefore better prognosis after treatment.

Prenatal diagnosis of Bloom syndrome is available by cytogenetic and molecular analysis of fetal cells obtained by either amniocentesis or chorionic villus sampling. Molecular DNA testing is now available. Genetic counseling is to be offered to the parents as there is an autosomal recessive transmission mode in Bloom syndrome; siblings of heterozygous carriers are at 25% risk of having Bloom syndrome and 50% risk of being a carrier [5].

The other notable trend to come out of the case report is the incidence of other complications of disease and treatment in this patient and other case reports mentioned in literature. Even those who are successfully treated for malignancy frequently succumb to pneumonia or other pulmonary complications, hepatic disease or sepsis. This is not altogether surprising as Bloom syndrome causes diabetes, lung and liver problems and immunodeficiency independently of any malignancy or its treatment. A number of the patients discussed above have had stormy in-patient courses and have finally opted for palliative treatment [6].

Conclusion

Bloom's syndrome is a rare autosomal recessive disorder that characteristically shows a telangiectatic, sun-sensitive facial rash in conjunction with stunted growth. These patients are prone to respiratory and gastrointestinal infections along with immunologic abnormalities. To the best of our knowledge this is the first Tunisian case to be reported.

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Volume 5 Issue 9 September 2021