Case Report

Acute Myeloblastic leukemia and Marfan syndrome: A Case Report and Literature Review

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Abstract

A 22 –year-old male visited our center because of acute myeloblastic leukemia (AML) associated with Marfan syndrome (MFS). The diagnosis of MFS was based on clinical findings such as Ectopia lentis, Anurysmal Aoarta and systemic manifestations. Peripheral blood smear and bone marrow examination showed: AML.M3. We started chemotherapy regimen but unfortunately died by DIC in his hospitalization course.

Keywords: Acute myeloblastic leukemia, Marfan syndrome, case report

Introduction:

Marfan Syndrome (MFS) is an autosomal dominant condition caused by a mutation in the fibrillin 1 (FBN 1) gene on chromosome 15 that encodes the protein fibrillin. This defect results in a set of expression of various organs and system, being musculoskeletal, cardiovascular and ophthalmic manifestations the most notorious. It has an estimated incidence of 2 – 3 per 10,000 inhabitants (1). On the other hand, acute myeloblastic leukemia (AML) is a clonal malignant disease of hematopoietic tissue. AML occurs at any age but is more common in adults, with increased frequency as age advances. It is slightly more common in males (2). In our knowledge, we have found 3 case reports of acute leukemia with marfan syndrome before (2-4).

Case Presentation:

The 22 years old male was admitted to the Nemazee hospital, Shiraz, Iran with ecchymosis in both arms since 2 days before admission. He was referred to hematologist and revealed Pancytopenia in his last tests. He has no history of cough, chills, fever, weight loss, dyspnea, joint pain & low back pain: He had no history of exposure to toxin or alcohol. He was smoker (water pipe and opium) and had history of Rheumatic disorder in his mother. On his admission, his general appearance was ill. He was fully oriented. His vital signs are: Blood pressure = 150/60 pulse rate = 100/min regularly, Respiratory rate = 18, temperature = 37.5 c on his physical exam, we showed: tall stature, thin long face, height =189cm, weight = 89 kg, arm span = 20 cm, the length of the lower segment was 112 cm. The ratio of upper/lower segment was 76/112 cm.

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He had thin long fingers (10cm) (arachnodactyly) and hyper extensible. Ophthalmology exam revealed bilateral intraocular lens. Also he had systolic murmur with ΠΙ/VI degree on apex. Chest exam showed: pectus excavatus deformity & mild scoliosis musculoskeletal exam: arm span substantially exceeds height tall stature ration. Spinal exam: abnormally low US/LS, mild Scoliosis neurologic exam was normal, DEEP Tendon reflex: 2/2, muscle power: 5/5. In all extremities, with no flexion contractures, no muscular atrophy, no hemiparesis. The Babinski was bilateral downward.

Laboratory findings showed in table 1. Peripheral blood smear showed hyper cellular marrow with Blast like cells in favour of AML.M3. Bone marrow aspiration and biopsy approved AML.M3. TTE: enlarged LV size with normal contractility. LVEF=55% Mild MR, mild TR, severe AR. Aneurysmal dilatation of ascending Aorta. Aortic root size= 7.6 cm, AV annulus= 3.2 cm. TEE revealed = dilatation of ascending Aorta, with Aortic root effacement due to annul Aortic ectasia, no dissection. Severe AR. The final diagnosis for our Patient was MFS with AML that remission induction chemotherapy regimen started for him but unfortunately he died cause of DIC during his hospitalization.

Discussion:

MFS is a multisystem connective tissue disorder, with primary involvement of the cardio vascular, ocular and skeletal system. This autosomal heritable disease is mainly attributable to defect in the FBN1 gene on chromosome 15. Aortic dilatation and dissection are the most important and life threatening manifestations, but cardiac, ocular, skeletal and neurological involvement may also impose a considerable burden. MFS affects both men and women of any race and ethnic group (2). In our knowledge we have found 3 case reports of acute leukemia with Marfan syndrome (2-4).

The seven new criteria can lead to a diagnosis, being necessary to fulfill just one of the criteria:

In the absence of a family history:

1. Aortic root Z- score ≥ 2 + Ectopia lentis
2. Aortic root Z- score ≥ 2 + FBN 1 mutation
3. Aortic root Z- score ≥ 2 + systemic score > 7 points
4. Ectopia lentis and an FBN 1 mutation with known aortic pathology.

-In the presence of a family history:

1. Ectopia lentis

2. Systemic score ≥ 7

3. Aortic root Z- score ≥ 2

-Points for systemic score:

1. Wrist and thumb sign= 3

2. Pectus carinatum= 2 (pectus excavatum or chest asymmetry= 1)

3. Hind foot deformity= 2 (pes planus= 1)

4. Dural ectasia= 2 (7).

AML (acute myeloid leukemia) is a type of cancer in which the bone marrow makes abnormal myeloblast (a type of white blood cell), red blood cells, or platelets. It is the most common type of acute leukemia in

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adults. So when this happens, infection, anemia, or easy bleeding may occur. The leukemia cells can spread outside the blood to other parts of the body including CNS, skin, and gums. Possible risk factors of AML, include the followings: male, smoking, having had treatment with chemotherapy or radiation therapy in past, having a history of a blood disorder. It can manifest clinically with variable symptoms which includes: fatigue, weakness, increased bleeding tendencies or predisposition to infections, skin, joint, CNS, heart. Skin can be involved in up to 10% of AML cases. Skin lesions can be as mass like nodules or erythematous papules or plaques. In AML, skin manifestations are indicative of aggressive disease and poor prognosis (9). Since MS is a genetically transmitted disease characterized by immunodeficiency, subject with this disease may have a higher risk of cancer. However, there is no knowledge regarding AML association with MS in the literature. So it seems it's important to Follow/Up the connective tissue disorders for leukemia.

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Table 1: Laboratory findings of the patient

Parameter	Value	Parameter	Value
WBC	0.5×10^9/L	Na	139 mEq/L
Platelet	30×10^9/L	K	3.6 mEq/L
Hemoglobin	9.4 g/L	Stool occult blood	Negative
RBC	$3.2\ 10^6$/L	Blood sugar	83 mg/dL
ANA	Negative	Calcium	9.1 mg/dL
INR	1.52	C-Reactive Protein	9 mg/L
BUN	16 mg/dL	Creatinin	0.8 mg/dL

AST: aspartate transaminase; ALT: alanine transaminase; PTT: Partial thromboplastin time; PT: prothrombin time; INR: International Normalised Ratio; BUN: blood urea nitrogen; RBC: red blood cell; WBC: white blood cell; ANA: Antinuclear antibody

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