http://jmscr.igmpublication.org/home/
ISSN (e)-2347-176x ISSN (p) 2455-0450
crossref DOI: https://dx.doi.org/10.18535/jmscr/v9i5.25

IGM Publication
Journal Of Medical Science And Clinical Research
An Official Publication Of IGM Publication

Case Report

Chronic Myeloid Leukaemia in a case of Poland Syndrome with cytological features suggestive of myeloid sarcoma in skin: a totally unexpected case diagnosed at the time of fine needle aspiration

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Abstract

Introduction: *Chronic myeloid Leukaemia is a form of Myeloproliferative neoplasm associated with recurrent (9,22) chromosomal translocation and formation of BCR-ABL fusion gene. Here we report a case of CML diagnosed in a 36 year old male with features of Poland syndrome, and skin lesions cytologically suggestive of Myeloid Sarcoma. It was an accidental diagnosis by fine needle aspiration in a case of clinically suspected lipomatosis.*

Case Report: *A 36 year old male presented with few soft nodular lesions of recent onset in the lumbosacral region without any other apparent symptom and a clinical diagnosis of Lipomatosis. Fine needle aspirate revealed myeloblasts and immature myeloid progenitors admixed with necrosis and fat. History and clinical examination showed that basically the patient is a case of Poland syndrome with congenital absence of Right sided pectoralis major and hypoplastic right upper limb. Associated finding was a huge splenomegaly.*

Blood was drawn in the next visit and peripheral blood picture was that of florid Chronic Myeloid Leukaemia. The necrosis intimately mixed with myeloblasts and myeloid precursors; present on the aspirate of the skin lesions, points towards a case of myeloid sarcoma in the background of CML. Otherwise the aspiration findings could be easily explained as mere blood elements drawn from dermal blood vessels in a patient of CML.

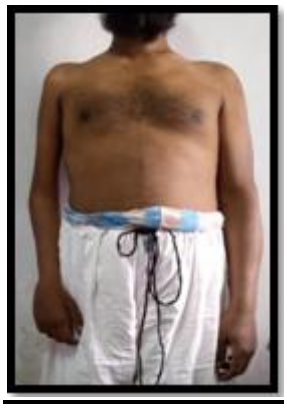
However, before histo-pathological confirmation and other ancillary investigations were done; the patient was referred to higher centre and further follow up was not possible.

Conclusion: *The overall diagnosis was chronic myeloid Leukaemia with skin lesions cytologically suggestive of myeloid Sarcoma in a case of Poland Syndrome. The patient was referred to higher centre dedicated to Hematology for genetic analysis and initiation of therapy.*

Keywords: *Poland syndrome, chronic myeloid leukemia, myeloid sarcoma.*

Introduction

Chronic myeloid Leukaemia is a form of Myeloproliferative neoplasm associated with recurrent (9,22) chromosomal translocation and formation of BCR-ABL fusion gene. By definition, myeloid sarcoma is a tumour mass consisting of myeloid blasts, with or without maturation, occurring at an anatomical site other than the bone marrow, effacing the tissue architecture. Here we report a case of CML diagnosed in a 36 year old male with features of Poland syndrome, and skin lesions cytologically suggestive of Myeloid Sarcoma. It was an unexpected, accidental diagnosis by fine needle aspiration in a case of clinically suspected lipomatosis.



Case History

A 36 year old male presented with few soft nodular lesions of recent onset in the lumbosacral region without any other apparent symptom and a clinical diagnosis of Lipomatosis. Fine needle aspirate revealed myeloblasts along with immature myeloid progenitors admixed with necrosis and fat. History and clinical examination showed that basically the patient is a case of Poland syndrome with congenital absence of Right sided pectoralis major and hypoplastic right upper limb. Associated finding was a huge splenomegaly.

Blood was drawn by venepuncture in the next visit and peripheral blood picture was that of florid Chronic Myeloid Leukaemia showing:

Hemoglobin- 9.3 gm/dl with normocytic normochromic RBCs

Total Leucocyte count: 1,14,960/mm3

Platelet count- 5,36,000/mm3

Differential count: Neutrophil-23% Lymphocyte-09% Eosinophil-8% Monocyte-2% Basophil-4% Myeloblast-5% Promyelocyte-2% Myelocyte-26% Metamyelocyte-21%

Repeated aspirations from the skin lesions revealed similar cytological findings. The necrosis intimately mixed with myeloblasts and myeloid precursors; present on the aspiration of the skin lesions, points towards a case of myeloid sarcoma in the background of CML. Otherwise the aspiration findings could be easily explained as mere blood elements drawn from dermal blood vessels in a patient of CML.

However, before histo-pathological confirmation and other ancillary investigations were done; the patient was referred to higher centre and further follow up was not possible.

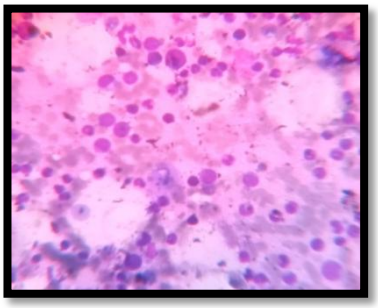


Fig 1: FNAC finding

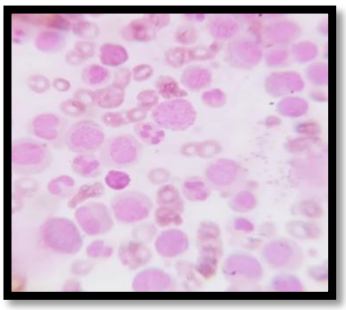


Fig 2: Peripheral smear

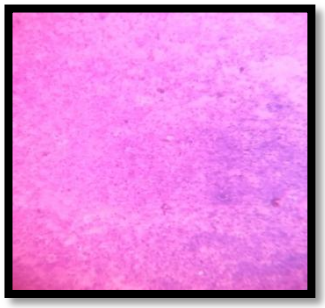


Fig 3: Necrosis on cytology smear

Discussion

The natural history of chronic myeloid Leukaemia is known to evolve through three distinct phases: 1. Chronic phase 2. Accelerated Phase and 3. Phase of Blast crisis, which is defined as presence of more than 20% blasts either in bone marrow or peripheral blood or extramedullary proliferation of blasts[1].

Myeloid sarcoma can occur de novo or in the background of AML,MDS, MPN and MPN/MDS[1].KMT2A rearrangement or the alteration in NPM1 gene is frequently found while investigations of the cases. Chloracetate esterase commonly gives positive staining pattern in this scenario and that lies behind the earlier name of this entity as chloroma. Non effacing nodular proliferations of myeloid precursors in the extramedullary sites are to be differentiated from myeloid sarcoma, as they are commonly found in cases of myeloproliferative neoplasms in advanced stages[1].

Named after Sir Alfred Poland in 1841, the syndrome is characterized by ipsilateral aplasia of chest wall muscles and abnormally short webbed fingers of the hand (symbrachydactyly) of same side (right side in 75% cases.) There is typically absence of pectoralis major and minor muscle and underdevelopment of nipple. This rare congenital anomaly has varieties of presentation, has an incidence rate between 1 in 10000 to 1 in 100000 live births and often associated with another related condition known as Moebius syndrome[2]. No genetic abnormality or familial clustering has been found in most of the cases. It is often referred to as Poland sequence (a sequence is a constellation of symptoms often attributed to be caused by a single insult during embryogenesis) and suggested that it may come under the umbrella term 'subclavian artery supply disruption sequence'[3].

Bangerter M et al[4] described the varied cytological features of myeloid sarcoma on fine needle aspiration. Among the 20 cases of myeloid sarcoma, Kaygusuz et al[5] found evidence of necrosis in 2 cases and it was found to be an indicator of aggressiveness. Vasconselos et al[6] reported a similar case of cutaneous myeloid sarcoma like ours, in the background of peripheral blood picture findings consistent with CML, chronic phase.

Multiple cases of Poland syndromes are reported in the literature, though we have not found any recent case report on coexistence of CML and Poland Syndrome even after our best effort; Though Weaker link between Poland syndrome and CML has been found to be reported in one instance[7].

Conclusion

The overall diagnosis was chronic myeloid Leukaemia with skin lesions cytologically suggestive of myeloid Sarcoma in a case of Poland Syndrome. No cases have been reported with similar clinical scenario till date in the literature available, as far our knowledge goes. The patient was referred to higher centre dedicated to Hematology for genetic analysis and initiation of therapy.

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