

Case Report: A Rare Case Presentation and Detection of Acute Promyelocytic Leukemia

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Introduction

Acute Promyelocytic leukemia (APL) is an uncommon subtype of acute myeloid leukemia (AML) characterized by the translocation t (15;17) (q22; q21) with a favorable prognosis upon immediate recognition and treatment.

Case Presentation

- Acute Promyelocytic leukemia , discovered in 1950 with t (15;17) [1,2].It is very rare when
- A case of a 44-year-old female who presented in October 2020 to the Emergency Room with severe fatigue and blood tests showing Hb 6 g/dL, thrombocytopenia, and leukopenia (WBC 2.5 x10⁹/L).
- Bone marrow biopsy and flow cytometry showed a negative result. Initial diagnosis was mainly related to a viral infection ,
- December 15, 2020, the patient presented with leukocytosis (55.31 x10⁹/L),anemia (Hb 7.6 g/dL), thrombocytopenia PLT (67 x10⁹/L), and swelling in her right knee.
- Repeated flow cytometry showed signs of dysplasia with abnormal myeloid lineage.
- Stills disease was diagnosed, treated by steroids 1 mg/kg post a total body scan showed findings mostly related to

- underlying myeloproliferative or lymphoproliferative disease with hepatosplenomegaly, retroperitoneal soft tissue thickening encasing the abdominal aorta, with aortic wall mostly related to associated para-neuroplastic aortitis.
- A repeated Cytogeneteics and molecular analysis showed PML-RARA transcripts t (15;17) , negative for Leukemia , hypercellular aspirates showing left – shifted myeloid hyperplasia and <1% blasts.
- Patient was diagnosed with Promyelocytic Leukemia (APL) .
- Treatment initiated with ATRA + Arsenic per Guidliunes .

Discussion

- This case report describes a rare presentation of Acute Promyelocytic Leukemia ,with no reported cases .
- Usually APL is diagnosed on the bone marrow morphology and confirmed by the presence of the t(15;17) and detection of the PML/RARA fusion transcript via RT-PCR [3].
- Amelioration of the case after introduction of steroids 1 mg /kg as a Still Diseases as diagnosed .
- Repeated Cytogenetics and molecular analysis after the false negative results , showed a diagnosis of PML-RARA transcripts t (15;17).
- Fast intervention of chemotherapy by ATRA +Arsenic per guidelines , improve the patient situation .

Conclusions

- Our case of a 44-year-old female with APL is quite a unique presentation and warrants to be reported ,
- It is important to be aware of the false negative rate for large PML/RARA FISH probes in the setting of masks rearrangements
- There is a need to conduct more observational and experimental studies to understand the mechanisms , by identifying masked PML/RARA fusion
- A rapid Diagnosis and the administration of the chemotherapies are essential to improve the outcome in patrients with APL,

Reference

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