CASE 059

CŌRE DIAGNOSTICS™

AUTHOR: Deepak Sharma, Dr. Sanjeev Sharma, Aditi Aggarwal, Dr. Rahul Katara.

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CASE DETAILS:

We present a case of 53 year old male patient who presented with pancytopenia and massive splenomegaly. The patient was JAK2 negative. We detected a rare CALR (Calreticulin) mutation that was identified by Next generation sequencing. We emphasize on the importance of detection of CALR mutation in the diagnosis and classification of myeloproliferative neoplasms in JAK2 and MPL mutation negative patients.

INTRODUCTION:

CALR mutations have been detected rarely in chronic myelomonocytic leukaemia (CMML), myelodysplastic syndrome/myeloproliferative neoplasm (MDS/MPN), and a few myelodysplastic syndrome (MDS) patients, mainly with refractory anaemia with ring sideroblasts, refractory anaemia, and refractory anaemia with excess blasts. The presence of a CALR mutation has been described as an exceptionally rare finding in Janus kinase 2 (JAK2) negative polycythaemia vera (PV) cases [1, 2]. The identified CALR mutation has been proven to be helpful to the pathologist in those bone marrow cases with ambiguous etiology of thrombocytosis, equivocal bone marrow morphologic findings of MPN, and unexplained reticulin fibrosis. Additionally, the identified CALR mutation makes the patient a potential candidate for CALR inhibitors such as ruxolitinib [3].