P31. AN UNUSUAL INITIAL PRESENTATION OF CML – CASE STUDY.

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Chronic myeloid leukemia (CML) is a myeloproliferative disease arising at the level of a pluripotent stem cell and consistently associated with the BCR-ABL fusion gene.

CML most commonly manifest in a chronic phase of the disease with leukocytosis owing to neutrophils in different stage of maturation, basophilia, thrombocytosis or thrombocytopenia, anemia, splenomegaly, and the demonstration of the Ph chromosome is the ultimate confirmation of the diagnosis.

We describe the case of a female patient presenting with isolated marked thrombocytosis resembling Essential Thrombocytemia (ET). The clinical and morphological initial findings were typical for ET: asymptomatic patient, no splenomegaly, no leukocytosis, no immature myeloid cells on smear, no anemia, only persistent raised platelet count (2.200.000x109/L).

We performed the PCR test for mutated JAK2 and the result was negative. Also, the analysis of peripheral blood cells by FISH for t(9,22) came negative. Bone marrow biopsy specimen was suggestive for 5q- syndrome. To confirm this hypothesis we performed the conventional cytogenetic study. Unexpectedly, it showed the presence of the Philadelphia chromosome, and the absence of other genetic anomalies. The diagnosis of CML was confirmed by PCR testing positive for BCR-ABL p210 type b3a2 transcript.

The particularities of this CML patient case are thrombocytemic onset mimicking ET, the unusual finding of negativity for BCR-ABL fusion gene by FISH concomitant with the presence of Ph chromosome by cytogenetic analysis, demonstrating that in some cases the diagnosis of CML may be a challenging one.