Polycythemia vera (PV)
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Clinics and pathology

Disease
Chronic myeloproliferative syndrome

Phenotype / cell stem origin
Pluripotent - non lymphoid - stem cell is involved.

Epidemiology
Annual incidence: 10/10^6; sex ratio: 1M/1F; median age 60 yrs.

Clinics
Asymptomatic for a long time, revealed by symptoms related to blood hyperviscosity (headache, vertigo...), or by asthenia, pruritus, skin erythrosis, or various other symptoms; splenomegaly is frequent: 70%; hepatomegaly in 40%; blood data: red cell mass of > 36 ml/kg in males, > 32 ml/kg in females; arterial oxygen saturation > 92%; high haemoglobin; WBC and platelets counts may be high.

Prognosis
Chronic disease, with, however, risks of thrombosis and haemorrhages in various tissues, including central nervous system; bone marrow evolution towards: 1- myelofibrosis with myeloid metaplasia (MMM) in 20% of cases; 2- acute leukaemia in 10%, either as an acute transformation, or as a therapy related ANLL; prognosis: median survival is 14 yrs with blood-letting, 12 yrs with 32P, less than 10 yrs with standard chemotherapy.

Cytogenetics

Cytogenetics, morphological
Normal karyotype is found in > 80% of cases at diagnosis, abnormal karyotype occurs with evolution, but the appearance of a clonal anomaly does not indicate progression of the disease, and may also occur during evolution to MMM; finally, up to 100% of cases with acute transformation have chromosome anomalies; these are: del(20q), +8, +9 may be seen solely or simultaneously in 20% of cases with chromosome anomalies, del(13q) and a partial duplication dup(1q)(sometimes in the form of t(1;7)(q10;p10) in 10%, other anomalies in 30%; none of them has prognostic significance; del(5q) and del(7q), hypodiploidy are seen in cases evolving towards therapy related ANLL: they confirm the diagnosis and indicate an adverse prognosis.

Genes involved and Proteins

Note: genes involved are unknown.

References

This article should be referenced as such: