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Summary

Acute Megakaryoblastic Leukaemia and Retinal Vein Thrombosis!

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Summary

Acute megakaryoblastic leukaemia is characterised by megakaryoblasts. These patients often present with retinal vein thrombosis!
It is associated by 30% or more blasts in the marrow.
Blasts are megakaryocytic in nature and express specific antigens for megakaryocytes and are platelet perfoxidase positive on electron microscopy.
It is associated with GATA 1 and is seen predominantly in Down's syndrome.
Other genes may however be associated with (AMKL; Acute megakaryoblastic leukaemia.)
Another related gene is MKL 1, which is also known as “MAL.”
This gene is a cofactor of serum response factor.

Presentation:

They usually present with pancytopaenia; there may be myelofibrosis, hepatomegaly, lymphadenopathy and poor response to chemotherapy.
In young children, leukocytosis and organomegaly are commonly seen.
The prognosis in children is better.

Diagnosis:

Morphology of AML M7 is characterized by megakaryoblasts on the bone marrow aspirate and trephine biopsy.
Immunophenotype is detected by flow cytometry and immunohistochemistry assay.
Megakaryoblasts have a high nuclear-cytoplasmic ratio and are medium to large-sized cells.
The basophilic cytoplasm may be vacuolated and budding platelets may be seen.
Megakaryoblasts lack myeloperoxidase and stain positively with Sudan Black B.
They are alpha mapthyl butyrate eterase negative, have variable alpha napthyl acetate esterase activity and have variable PAS staining activity!
The marrow aspirate may be difficult to obtain due to the myelofibrosis.
More precise identification by immunophenotyping or with electron microscopy is often of necessity.
Immunophenotyping using MoAb (monoclonal antibodies) to megakaryocyte restricted antigen (CD41 and CD61) may be diagnostic.

Prognosis:

Prognosis depends on the cause.
One third of cases is associated with at (1;22) (p13; q13) mutation in children.
These cases have a poor prognosis.
Another third' predominantly Downs cases have a fair prognosis.
The last third, those of a heterogeneous nature have a somewhat poor prognosis!

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