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Leukaemia Section
Mini Review

-Y / Y loss in leukemia

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Identity

Note: Indeed, it is a normal elderly phenomenon in men over 60 years, to loose the Y chromosome.

Clinics and pathology

Disease

-Y is frenquently observed in myeloproliferative diseases (MPD), myelodysplasic syndromes (MDS), and acute non lymphocytic leukemias (ANLL), but rarely in lymphoproliferation.

Epidemiology

Frequency of Y loss cases in the control and patient populations:
- In the control population: 10.5% of the males as a whole, 0 to 2% before 60 years;
- In myeloid diseases: 9.5% of MDS male patients, 3.2% of ANLL male patients, and up to 10% of chronic myelogenous leukemia (CML) male patients.
Percentage of -Y cells:
- loss of the Y chromosome in 100% of the cells appears in about 20% of control cases who have lost their Y and about 30% of myeloid disease patients with a -Y karyotype; controls and MDS populations show comparable % values of missing Y cells;
- In ANLL and in MPD the percentages are significantly higher than in the control population; this may partly be due to the swiftness of the monoclonal proliferation.
Age:
- In CML and in ANLL with a t(8;21), Y loss occurs at a younger age than in the normal population.
- No significant difference was demonstrated between MDS patients and the control group in the unique study with statistical analysis.

Clinics

The functional significance of Y loss is at present unknown:
- Partial or complete reappearance of the Y chromosome has been described in 8 cases of leukemia remissions showing that this abnormality may be a neoplastic event.
- In CML, the occurrence of the Y loss does not indicate progression of the disease; it may be a sporadic event in both normal and CML populations in patients younger than 60 years.
- In ANLL with t(8;21), Y is lost in 61% of men; in some of these cases, Y loss is in association with other well known secondary abnormalities; Y loss mosaicism in t(8;21) cells is observated in about 18% of males; X chromosome is lost in 41% of females; loss of sex chromosome is not elderly related in this disease.

Cytology

No particular association.

Prognosis

No clear relation between Y loss and the prognosis; as the age and the disease have a specific prognostic value affecting the clinical course, it will be difficult to determine the prognositic significance of the Y loss per se.

Cytogenetics

Cytogenetics morphological

The constitutive heterochromatin of the Y chromosome is the "bodyguard" of the genome (1975).
Y loss may prevent other marrow cells from being involved in further karyotypic progression (1980).
The gene for the receptor of GM-CSF is located on the pseudoautosomal region of the Y chromosome (1992).
The Y chromosome contains a tumor suppressor gene (1994).

Probes

All available probes for the Y chromosome.

Additional anomalies

Y loss is generally considered as a secondary event, most often in association with t(9;22) in CML or with t(8;21) in M2 AML.

Variants

As a sole event, the loss of an X chromosome in females, -X, is much less frequent than the loss of the Y chromosome in males, but it may also occur.

Genes involved and proteins

Note

Genes involved, if any, are unknown.

To be noted

Note

All data herein evaluated were extracted from bone marrow studies; there is a difference between the active marrow progenitor cells and the stimulated phytohemagglutinin lymphocyte cell population; in one study on one individual, the Y loss rate had been found lower in peripherical cells in culture than in the bone marrow cells.

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