-7/del(7q) in childhood

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Identity

Note

-7/del(7q) is a more common entity of blood malignancies in the adults.

Epidemiology

The most frequent abnormality in childhood myeloid disorders; found in 30% of the MDS and in 4% of the ANLL; sex/age: 90% of the children with this anomaly are younger than 5 years; before 5 years, there is a majority of boys (3M/2F), with -7 as the sole cytogenetic abnormality; after 5 years, girls are in majority, and the -7/del(7q) is then often associated with additional anomalies.

Clinics

Several clinical forms: the most frequents are JCML and the monosomy 7 syndrome; these disorders have some common features:
- JCML is defined by clinical and cytological observations; 6 to 24% of JCML children show monosomy 7 in the bone marrow;
- monosomy 7 syndrome is a cytogenetic-defined entity.

The therapy related cases of monosomy 7 had been exposed to alkylating agents, they have a myelodysplastic phase preceding acute leukemia with multilineage bone marrow dysplasia. In opposite, therapy including anti-topoisomerase drug induce myelodysplastic syndromes and leukemias with 11q23 abnormalities.

Cytology

Before 5 years, the disease presents as a specific myeloid leukemia characterized by leucocytosis with monocytosis but thrombopenia, anemia in blood and hyperplasia of the bone marrow; for some authors, the diagnosis of monosomy 7 syndrome should be made in any FAB class (principally CMML), whereas the diagnosis of JCML applies to cases of CMML with fetal hemoglobin > 10 %,and with no monosomy 7; the remaining CMML are diagnosed as CMML; for others,
a - 7 does not exclude the diagnosis of JCML; however, cases of JCML without visible monosomy 7 appear to have no loss of heterozygosity on chromosome 7. After 5 years, the disease presents as a MDS with cytopenia in blood and hypodysplasia of bone marrow, like in adults.

**Prognosis**

Slow evolution of the ANLL in infants before 6 month; for children older than 1 year, the survival is less than 2 years; the European Working Group on MDS in Childhood noted a superior survival for children with MDS having a - 7 alone than for those with other anomalies (3 year survival of 56% vs 24%); but this was the reverse in children with ANLL.

**Cytogenetics**

**Cytogenetics morphological**

Deletion (7q): cluster of breakpoints in 7q11 to 7q36, is a with two common minimal zones in q22 and in q32-34.

**Cytogenetics molecular**

Using loss of heterozygocity (LOH) studies and YAC libraries, a 2 to 3 Mb segment in 7q22 has been designated as the proximal common deleted area; the 7q33-34 zone is the consensual area for the distal deletion; LOH studies suggest that a specific mechanism, such as mitotic recombination in bone marrow stem cell leading to homozygosity in both granulocytes and lymphocytes, may be implicated.

**Probes**

Chromosomal band 7q221: marker D7S658, through D7S2494, YAC HSC7E441 to HSC7E572; 7q33-34: D7S498 to D7S505; near-centromeric probes of chromosome 7 and 7q31 probes are produced by commercial companies.

**Additional anomalies**

7 alone is observed in 75% of MDS cases and in 32% of ANLL; the specific additional anomalies are - 5/del(5q), and trisomy 8.

**Variants**

The balanced translocation t(1;7)(q10;p10), and many unbalanced translocation, having for consequence a partial monosomy 7 of the 7q22 to 7q34 bands may, in a way, be considered as variants.

**Genes involved and proteins**

**Note**

-7/del(7q) is frequent in secondary MDS or ANLL, and also in leukemias occurring in individuals with constitutional syndromes including predisposition to myeloid disorders; these findings suggest the presence of a putative myeloid leukemia suppressor gene in the commonly deleted genomic segment 7q22 and even multiple genes in 7q22 -31.1 that are playing a role in leukemogenesis; candidate genes are : ASNS (asparagin synthetase gene) in 7q21.3-q22.1; ACHE (acetyl cholinesterase), EPO (erythropoietin), PLANH1 (plasminogen activator inhibitor 1) in 7q22; and MET in 7q31.2-31.3.

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