Leukaemia Section
Mini Review

del(9q) solely
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Identity

del(9q) G-banding - Courtesy Diane H. Norback, Eric B. Johnson, Sara Morrison-Delap Cytogenetics at the Waisman Center (left and middle) and Jean-Luc Lai (right).

Note: del(9q) as the sole abnormality must be distinguished from syndromes where it is associated with other chromosome rearrangements; in particular, there is frequent association with LAM2 expressing t(8;21)(q22;q22), and, also, with t(15;17)(q24;q21); we herein describe del(9q) as the sole anomaly, when not otherwise specified.

Clinics and pathology

Disease
ANLL mainly; rarely observed in myelodysplastic syndroms (MDS) or myeloproliferative disorders; biphenotypic T-lymphoid / myeloid leukemias cases have also been described.

Phenotype / cell stem origin
ANLL: M1, M2, M4, M6 FAB subtypes; pluripotent stem cell probably involved; there is a trilineage myelodysplasia; six patients (4 M1, 1 M2 and 1 T-ALL) from two reports have been described with del(9q) and CD34+, CD7+, T lymphoid / myeloid biphenotypic leukemia.

Epidemiology
0 to 3% of ANLL cases, depending on series; both sexes equally represented; adults and children may be affected.

Cytology
Frequent sideroblasts; leukemic blasts are agranular, with large vacuoles on Giemsa staining and localized positivity for myeloperoxidase (MPO); giant MPO positive granules are described, corresponding to =AB pseudo-Chediak-Higashi =BB granules; most blast cells are CD34 positive.

Prognosis
When del(9q) is the unique chromosome abnormality the prognosis, depending on AML subtype, is variable; (del(9q) as a secondary anomaly in t(8;21) has no prognostic consequence for some workers and is a factor of worse prognosis for others).
**Cytogenetics**

**Cytogenetics, morphological**
Interstitial deletion of chromosome 9 long arm, called del(9q) or 9q-, involving a variable chromosome segment; the region 9q21-22 seems constantly involved.

**Cytogenetics, molecular**
This constantly deleted region has not yet been more precisely defined and it is not known whether deletion of one or more critical gene(s) are involved. Thus there are presently no 9q molecular probes available to assess 9q deletion.

**Probes**
Whole chromosome 9 painting, to exclude 9q translocations.

**Additional anomalies**
On 31 reviewed cases of ANLL with del(9q) as a primary change, none had additional anomalies del(9q) as a secondary anomaly:
- Association with t(8;21) represents the majority of cases; t(8;21) occurs in 5 to 10 % of patients with ANLL, and its association with del(9q) is the second more frequent, after the association with loss of one sex chromosome; it represents approximately 10-15 % of cases.
- Association with t(15;17), in promyelocytic leukaemia, has also seldom (1%) been observed.
- In these two syndromes, del(9q) is usually not present at diagnosis but appears as an additional change at relapse.
- del(9q) has never been described in association with other recurrent primary changes.

**Variants**
Unbalanced translocations involving 9q may, in a way, be considered as del(9q) variants.

**Genes involved and Proteins**

*Note:* genes involved are unknown; there is probable deletion of one or several tumor suppressor gene(s) involved in the progression of the disease.

**References**


Kwong YL, Chan TK, Chan LC. Interstitial deletion of the long arm of chromosome 9 as the sole anomaly in acute myeloid leukaemia is associated with dyserythropoesis. Leukemia 1992 Jan;6(1):64-5.


This article should be referenced as such: