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Leukaemia Section

Mini Review

i(5)(p10) in acute myeloid leukemia

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Identity

i(5)(p10) G-banding - Claudia Schoch (left), and R-banding - Nathalie Douet-Guilbert (right).

Note

In literature, two types of i(5)(p10) are observed:
Type 1: i(5)(p10) inducing a loss of the long arm of chromosome 5 (5q) and a trisomy of the short arm of the chromosome 5 (5p);
Type 2: +i(5)(p10) (or supernumerary i(5)(p10) or gain of i(5)(p10)) inducing a tetrasomy of the short arm of chromosome 5 (5p). The i(5)(p10) occurred in addition to two normal chromosomes 5.
The isochromosome of the short arm of chromosome 5 - i(5)(p10) - has only been described in a few cases of myeloid leukemia.

Clinics and pathology

Phenotype/cell stem origin

Type 1: classified as myelodysplastic syndrome (4 cases), acute myeloid leukemia (4 cases) predominantly AML M2;
Type 2: classified as acute myeloid leukemia (5 cases), predominantly AML M5a.

Etiology

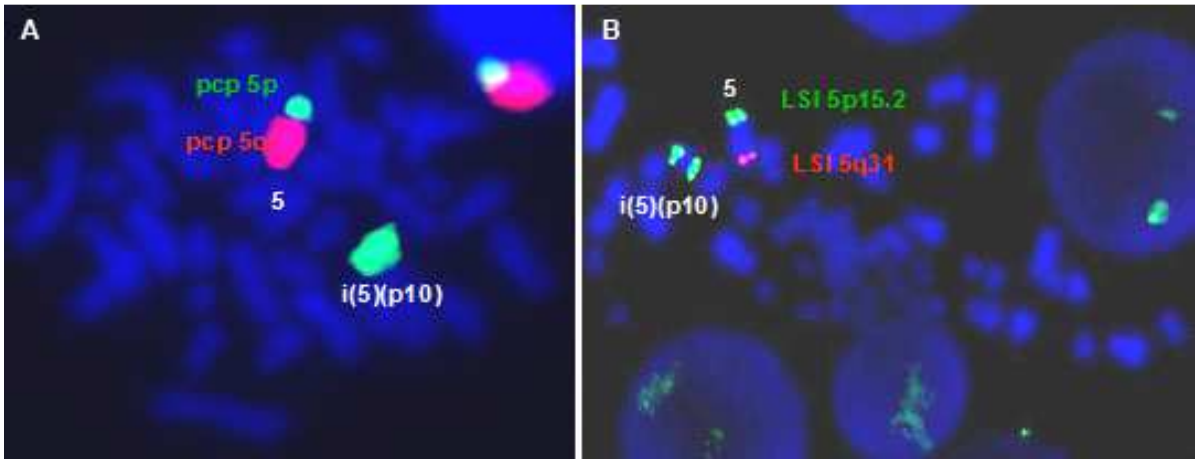
Unclear

Epidemiology

Type 1: it is found in young adults in MDS (average age: 35 years; range: 19-67) and in older patients in AML (average age: 66 years; range: 50-85).
Type 2: the +i(5)(p10) is found in patients with an average age of 48.5 years (range : 24-78).

Prognosis

Prognosis of patients with i(5)(p10) seems to be poor compared to patients with del(5q), but it is unclear due to the very small number of cases and the usually associated complex chromosomal abnormalities.



A - FISH with partial chromosome painting 5p (pcp 5p) (Green signal) and 5q (pcp 5q) (Red signal). B - FISH with LSI 5p15.2 (Green signal) / 5q31 (Red signal). Nathalie Douet-Guilbert.

Cytogenetics

Cytogenetics morphological

The formation of i(5p) results from the loss of the long arm of chromosome 5 and duplication of its short arm inducing trisomy 5p and monosomy 5q in type 1 and tetrasomy 5p in type 2.

A metacentric del(5q) could be an isochromosome of the short arm of chromosome 5. FISH technique with specific probes of chromosome 5p/5q used as a complement of conventional karyotype is necessary to identify i(5)(p10). The i(5p) is a variant of del(5q). The i(5p) is monocentric or dicentric.

Additional anomalies

In one case, i(5)(p10) was the sole anomaly but rapidly evolved into a complex karyotype. Complex karyotypes were present in the other cases: -12/del12p (3 cases), -17/del17p (2 cases), del9q (2 cases).

Supernumerary +i(5)(p10) was accompanied by several additional anomalies, especially trisomy 8

Genes involved and proteins

Note

Type 1: to explain the specific phenotype of i(5)(p10), loss of tumor suppressor genes in the deleted region (5q) associated with gene dosage effect of genes located on 5p is suggested.

Type 2: gene dosage effect of genes located on the short arm of chromosome 5.

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