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Leukaemia Section
Short Communication

ider(20q) in Myeloid Malignancies

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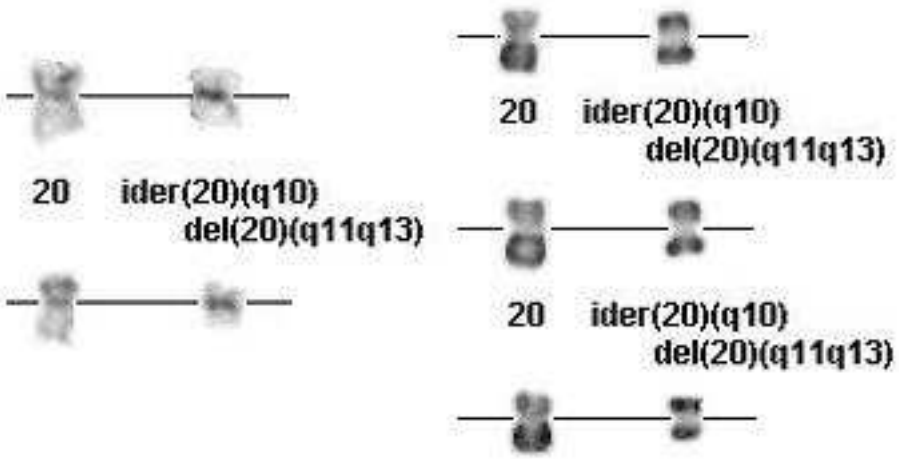
Identity



Partial karyotypes for ider(20q) in G-banding (left) and R-banding (right).

Clinics and pathology

Disease

Myelodysplastic syndrome (21 cases), Acute Myeloid Leukemia (5 cases), Chronic Myelomonocytic Leukemia (1 case).

Phenotype/cell stem origin

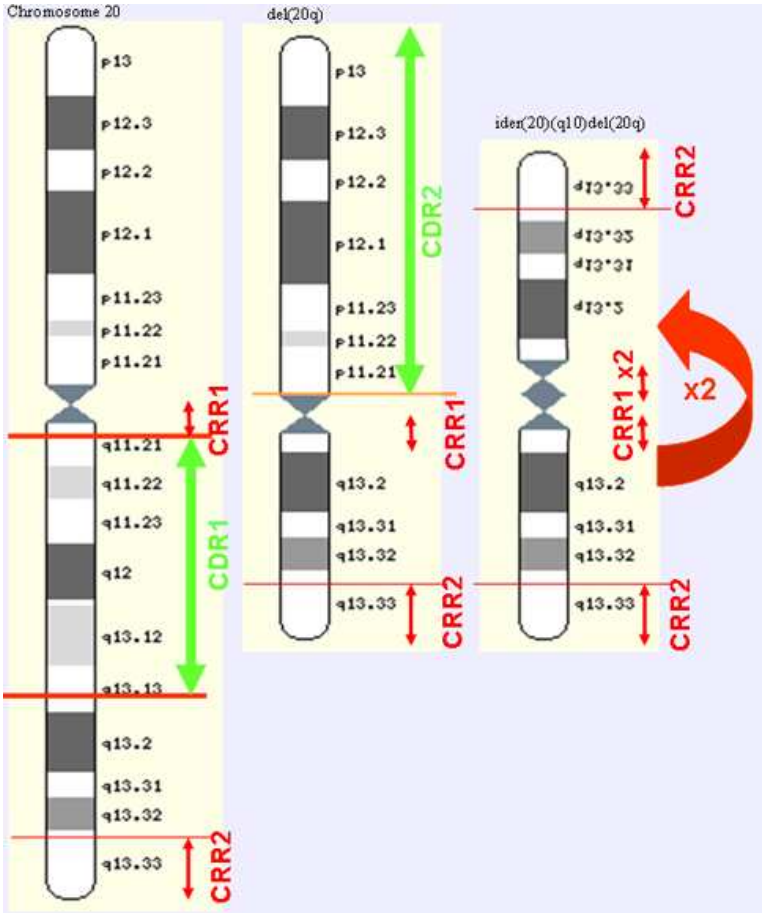
Thrombopenia (90%) with anemia (60%).
Dysplastic changes in bone marrow: dyserythropoeisis associated with dysgranulopoieisis and/or dysmegakaryocytopoeisis.

Epidemiology

The frequency of ider(20q) is estimated at 0.49% in myelodysplastic syndrome and 0.26% in acute myeloid leukemia according to one study.
They are found in older patients (average age: 68 years; range: 38-91).

Prognosis

Prognosis of patients with ider(20q) seems to be poor compared to patients with del(20q), but it is unclear due to the small number of cases.



Ideograms of normal chromosome 20, of del(20q), of ider(20q) and commonly deleted regions (CDR1, CDR2) and commonly retained regions (CRR1 et CRR2).
The ider(20q) is described as a secondary event, signing a clonal evolution of deletion 20q positive cells.
The formation of ider(20q) results from loss of the short arm of chromosome 20 and duplication of the deleted long arm of chromosome 20.

Cytogenetics

Cytogenetics morphological

A monosomy of chromosome 20 with small metacentric marker chromosome: 46,XX or XY,-20,+mar is most likely an isoderivative of chromosome 20.
The ider(20)(q10)del(20)(q11q13) is a variant of del(20)(q11q13).

Cytogenetics molecular

The ider(20q) is monocentric or dicentric.
The proximal breakpoints are consistently located in 20q11.21 band. The distal breakpoints span from band 20q13.13 to band 20q13.33.
The commonly deleted region include the short arm of chromosome 20 and a large region on the long arm of chromosome 20 spanning from 20q11.21 to 20q13.13.
A commonly proximal retained region (from centromere to 20q11.21) and commonly distal retained region (from 20q13.33 to telomere 20qter) of the long arm of chromosome 20 were determined. These retained regions are duplicated.

FISH with subtelomeric probes 20p (Green signal) and 20q (Red signals). The ider(20q) contains two red signals and no green signal.

Additional anomalies

Additional anomalies in decreasing frequency:
- del(20q) detected by conventional cytogenetics and/or by FISH,

- 2 copies of ider(20q),
- monosomy 7,
- complex karyotypes in acute myeloid leukemia.

Genes involved and proteins

Note

To explain specific phenotype, loss of tumor suppressor genes in deleted region (ADA, L3MBTL) and gene dosage effect of genes located on the retained region of chromosome 20 are suggested.

References

Li T, Xue Y, Wu Y, Pan J. Clinical and molecular cytogenetic studies in seven patients with myeloid diseases characterized by i(20q-). Br J Haematol. 2004 May;125(3):337-42

Ligon AH, DeAngelo DJ, Atkins L, Dal Cin P. Isochromosome of a deleted 20q may be a relatively common abnormality in myeloid malignancies. Cancer Genet Cytogenet. 2005 Oct 1;162(1):89-91

MacKinnon RN, Campbell LJ. A comparison of two contrasting recurrent isochromosomes 20 found in myelodysplastic syndromes suggests that retention of proximal 20q is a significant factor in myeloid malignancies. Cancer Genet Cytogenet. 2005 Dec;163(2):176-9

Saunders K, Czepulkowski B, Sivalingam R, Hayes JP, Aldouri M, Sekhar M, Cummins M, Ho A, Mufti GJ. Isochromosome of a deleted 20q: a rare but recurrent chromosome abnormality in myelodysplastic syndromes. Cancer Genet Cytogenet. 2005 Jan 15;156(2):154-7

Li T, Xue Y, Wu Y, Pan J. FISH studies identify the i(20q-) anomaly as a der(20)del(20)(q11q13)idic(20)(p11). Genes Chromosomes Cancer. 2006 Jun;45(6):536-9

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