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Leukaemia Section
Short Communication

i(22)(q10) in myeloid malignancies

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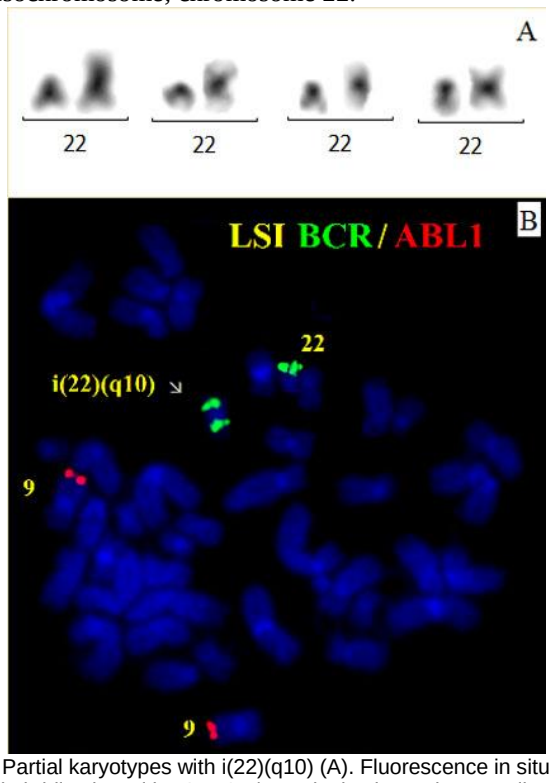
Abstract

Isochromosome i(22)(q10) is a rare but non-random karyotypic change in hematologic malignancies, often associated with complex karyotypes and with partial or complete loss of chromosomes 5 and/or 7.

Keywords
Isochromosome; chromosome 22.



Partial karyotypes with i(22)(q10) (A). Fluorescence in situ hybridization with LSI BCR/ABL dual color probe revealing extra copy of the BCR gene on 22q11 as a result of isochromosome formation (Abott Molecular/Vysis, US) (B).

Clinics and pathology

Disease

Chronic and acute myeloid malignancies

Phenotype/cell stem origin

Chronic myeloid malignancies in 7 (4 males and 3 females aged 59 to 73 years, median 68 years): refractory anemia with excess of blasts in 5 (Musilova & Michalova 1988; Geddes et al., 1990; Padua et al., 1998; Martinez-Ramirez et al., 2004; Lessard et al., 2007), 1 myelodysplastic syndrome (Andersen et al., 2005) and 1 chronic myeloid leukemia (Werner et al., 1991).
Acute myeloid leukemia in 16 (10 males and 6 females; median age 61 years; range 2-76 years): 5 with M2 (Berger et al., 1987; Van Limbergen et al., 2002; Rucker et al., 2006; Xu et al., 2008;), 2 with M4 (Shurtleff et al., 1995; Johansson et al., 1997; Asou et al., 2009;), 1 with M5 (Gervais et al., 2008), 1 with M6 (Bitter et al., 1985) and 7 with unspecified AML (Michels et al., 1989; Pedersen & Jensen; GFCH 1996; Morrison et al., 2002; Lugthart et al., 2010; Haferlach et al., 2012; Lavallee et al., 2015).

Epidemiology

23 patients (14 males, 9 females aged 2 to 76 years; median 63 years).

Prognosis

The presence of i(22)(q10) in association with chromosome 5 and/or chromosome 7 anomalies in complex karyotypes is indicator of poor prognosis, representing a therapeutic challenge.

Cytogenetics

To exclude the possibility that der(22) chromosome contain two centromeres, use of centromere 22-specific FISH probes is recommended.

Cytogenetics morphological

Sole anomaly in 1 patient, loss of 5 and/or 7 chromosomal material seemed the most common event, and their losses in combination were observed in many cases: found with del(5q)/-5 in 4, with simultaneous del(5q)/del(7q) or del(7q)/-7 in 8 and with monosomy of both chromosomes in 2. Found as an additional anomaly to inv(16)(p13q22) in 2 and with miscellaneous or highly complex anomalies in the remaining patients.

Result of the chromosomal anomaly

Fusion protein

Oncogenesis

While trisomy 22 is a well know anomaly in myeloid leukemia, in particular, acute myelomonocytic leukemia with eosinophilia, i(22)(q10) is less common. The i(22q) formation leads to the gain of the entire long arm of chromosome 22 suggesting its possible role in oncogenesis. Gain of the whole long arm leads to overexpression of genes located on 22q due to a dosage effect. i(22)(q10) is mainly found as part of complex karyotypes, therefore may not be the primary disease determining aberration but rather a secondary event associated with disease evolution.

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