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Leukaemia Section
Short Communication

der(4)t(1;4)(q11-32;q34-35)

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Abstract

Unbalanced 1q rearrangements are widely reported in myeloid and lymphoid malignancies. Among unbalanced translocations of 1q, der(4)t(1;4)(q11-32;q34-q35) resulting in complete or partial trisomies of genes located on 1q is a relatively rare anomaly.

Keywords

Unbalanced 1q translocations, chromosome gain, der(4)t(1;4), gene expression.

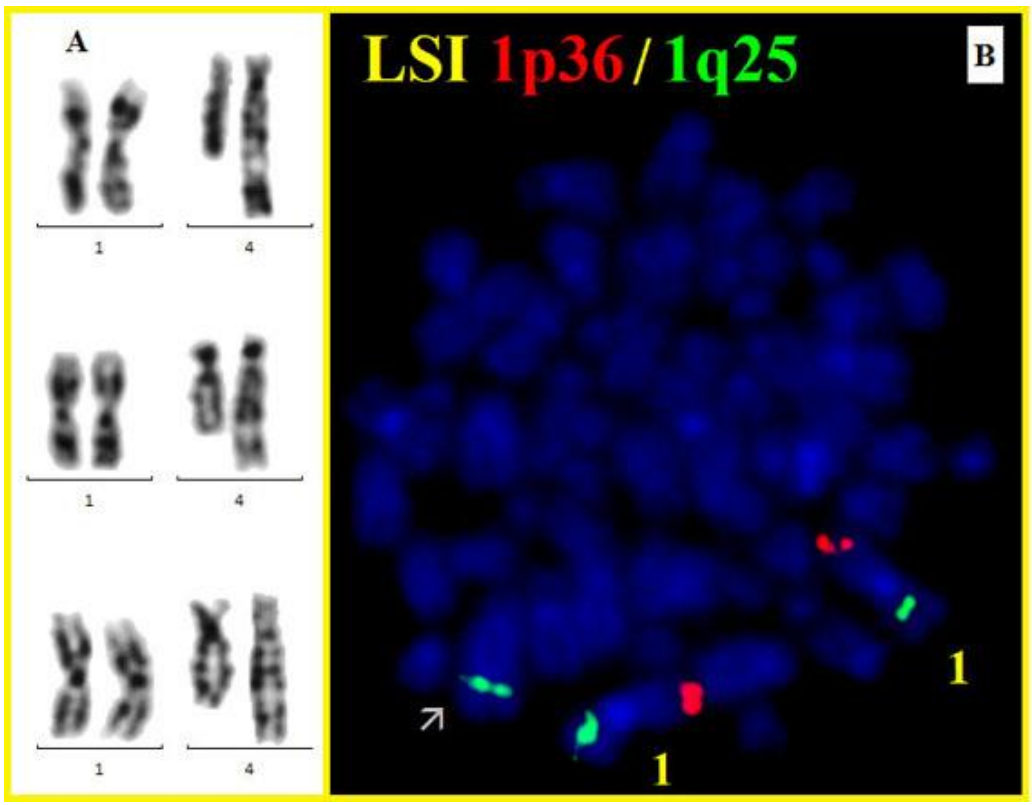

Figure 1. Partial karyotypes with unbalanced translocation between chromosomes 1 and 4 (A). Fluorescence in situ hybridization with LSI 1p36/1q25 dual color probe (Abott Molecular/Vysis, US) showing the extra copy of 1q (green signal) on der(4) chromosome (B).

Clinics and pathology

Disease

Myeloid malignancies, multiple myeloma (MM) and Non-Hodgkin lymphoma.

Myeloid malignancies in 4 (4 males aged 1 to 30 years): 1 refractory anemia with excess blasts-2 (Vundinti et al., 2003), 1 acute myeloblastic leukemia with minimal differentiation (AML-M0) (Creutzig et al., 1996), 1 acute erythroleukemia (AML-M6) (Baumgarten et al., 1993) and 1 acute megakaryoblastic leukemia (AML-M7) (Martinez-Climent et al., 1995). 3 of the AML patients were children with Down syndrome (DS) (aged 1, 2 and 2 years) (Baumgarten et al., 1993; Martinez-Climent et al., 1995; Creutzig et al., 1996).

Multiple myeloma in 7 (4 males and 3 females; ages unknown) (Sawyer et al., 1998; Sawyer et al., 1998; Gutierrez et al., 2000; Lloveras et al.,2004; Wu et al., 2007; Sawyer et al., 2014; Rack et al., 2016).

Lymphoid malignancies 1 acute lymphoblastic leukemia (Lin et al., 1990) (1 female aged 11 years), 1 post-transplant lymphoproliferative disorder (1 male aged 42 years) (Djokic et al., 2006); 10 B-cell lymphomas (6 males and 4 females aged 39 to 74 years), among them 6 patients with follicular lymphoma (Nishida et al., 1989; Bastard et al., 1992; Gray et al., 1997; Itoyama et al., 2002; Aamot et al., 2007; Narayan et al., 2013), 2 with diffuse large B-cell lymphoma (DLBCL) (Le Baccon et al., 2001; Trcic et al., 2010), 2 with mature B-cell neoplasm (Morgan et al., 1999; Veldman et al., 1997) and there was an 14 years old female with T-cell anaplastic large cell lymphoma (Lones et al., 2006).

Epidemiology

15 males and 9 females aged 1 to 74 years (median 42 years).

Prognosis

Reported patients are characterized by complex karyotypes that likely reflects an inherent chromosomal instability correlated with a poor prognosis.

Cytogenetics

Cytogenetics morphological

Various breakpoints on the long arm of chromosome 1; MM and lymphoma patients tend to have more frequently near-centromeric 1q breakpoints (4 out of 7 MM and 7 out of 10 B-cell lymphoma patients).

Additional anomalies

Sole anomaly in 1 patient with DLBCL (Trcic et al., 2010), found in association with +8 in 2 AML patients with Down syndrome (DS) (Baumgarten et al., 1993; Creutzig et al., 1996) and in 1 with i(7)(q10) (Martinez-Climent et al., 1995). Found in a sideline with i(7)(q10) and t(9;22)(q34;q11) in the ALL patient (Lin et al., 1990), t(14;18)(q32;q21), as a part of complex karyotypes in 7 out of 10 B-cell lymphomas (Nishida et al., 1989; Bastard et al., 1992; Morgan et al., 1999; Le Baccon et al., 2001; Itoyama et al., 2002; Aamot et al., 2007; Narayan et al., 2013) and as an additional anomaly to t(2;5)(p23;q35) in patient with anaplastic large cell lymphoma (Lones et al., 2006). Found with del(1)(q21) in 1 (Gutierrez et al., 2000) and as part of highly complex karyotypes in the remaining multiple myeloma patients.

Result of the chromosomal anomaly

Fusion protein

Oncogenesis

1q gains represent nonrandom structural aberrations in hematological malignancies, suggesting the existence of genes in this chromosomal region that are important for disease initiation and/or progression.

Chromosome arm 1q is gene-rich, therefore several genes on 1q may contribute to disease pathogenesis that might cooperate in an additive or synergistic way resulting in their simultaneous downregulation. der(4)t(1;4)(q11-32;q34-35) has been reported as a sole karyotype aberration only in one patient, while it is usually present with additional common abnormalities or along with complex combinations of anomalies in most of the reported cases, indicating that gain of 1q might be relevant for tumor progression and advanced disease.

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