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Leukaemia Section

Review

t(10;14)(q24;q11) TLX1/TRD

t(7;10)(q34;q24) TRB/HOX11

Tatiana Gindina, Karolien Beel

Cytogenetics Lab, Raisa Gorbacheva Memorial Institute of Children's Oncology, Hematology and Transplantation at First Pavlov St. Petersburg State Medical University, Saint-Petersburg, Russia; tatgindina@gmail.com (TG); Center for Human Genetics, UZ Leuven, Belgium; karolien.beel@uzleuven.be (KB)

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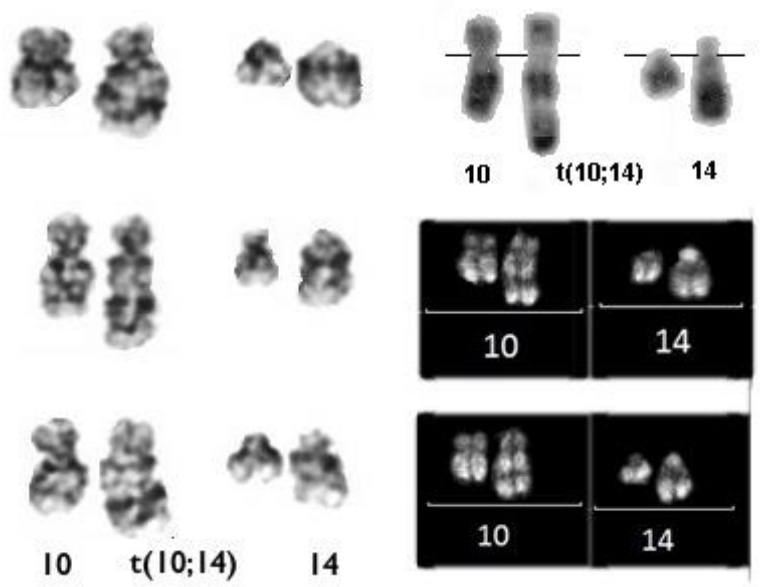
Abstract

Review on t(10;14)(q24;q11) and t(7;10)(q34;q24) translocations, with data on clinics, and the genes involved.

KEYWORDS

chromosome 10; chromosome 14; t(10;14)(q24;q11); t(7;10)(q34;q11); TLX1; TRD; TRB; T-cell Acute lymphoblastic leukemia

Identity



Left: Partial karyograms showing t(10;14)(q24;q11), GTG-banding - Courtesy Tatiana Gindina. Right: top: t(10;14)(q24;q11) R-banding - Courtesy Pascale Cornillet-Lefebvre and Stéphanie Struski; middle and bottom: t(10;14)(q24;q11) R-banding - Courtesy Karolien Beel, Peter Meeus and Lucienne Michaux

Clinics and pathology

Disease

T-cell acute lymphoblastic leukemia (ALL) and non-Hodgkin lymphoma (NHL) with medullary involvement.

A t(10;14)(q24;q11)TLX1/TRD was found in 69 cases of T-ALL (Dube et al., 1986; Raimondi et al., 1988; Kagan et al., 1989; Uckun et al., 1989; Parket al, 1992; Secker-Walker et al., 1992; Martin et al, 1996; Rack et al., 1997; Forestier et al., 1998; Heerema et al., 1998; Lai et al., 2000; Schneider et al., 2000; Kahl et al., 2001; Pedersen et al., 2001; Thomas et al., 2001; Mancini et al., 2002; Nordgren et al., 2002; Kristensen et al., 2003; Barber et al., 2004; Speleman et al., 2005; Stergianou et al., 2005; Cauwelier et al., 2006; Reichard et al., 2006; van Grotel et al., 2006; Strefford et al., 2007; Kwon et al., 2009; Le Noir et al., 2012; Setoodeh & Zhang, 2012; Grossmann et al., 2013; Park et al., 2014; Safavi et al., 2015; Gindina T. three personal unpublished cases).

A t(7;14)(q34;q24) TLX1/TRB was found in 21 cases of T-ALL (Kahl et al., 2001; Cauwelier et al., 2006; Le Noir et al., 2012; Raimondi et al., 1988).

In exceptional cases, a t(10;14) or a t(7;10) was found in T-prolymphocytic leukemia and ataxia telangiectasia (Rack et al., 1997), and, more surprisingly, in chronic lymphocytic leukemia, a B-cell disease (Delhomme-Bachy et al., 1992; Lu et al., 2006).

Phenotype/cell stem origin

T lineage. The gene expression pattern of TLX1-expressing lymphoblasts is similar to that of early cortical thymocytes, compatible with a leukemic arrest at the stage of the early cortical thymocyte (virtually all arrested at the early cortical (CD1+) CD4+ CD8+ "double-positive" stage of thymocyte development).

Epidemiology

Found in 5% of pediatric T-ALL (0.3% of all pediatric ALL) and 30% of adult T-ALL cases. Median age was 24 years (range 4-58). Sex ratio was 3 male : 1 female patients.

Clinics

Organomegaly with marked hepatosplenomegaly, lymphadenopathy, mediastinal mass, high WBC count (100 to 200 X 109/l) sometimes with anemia.

Cytology

High leukocyte count, very high circulating and central blast cell count.

Prognosis

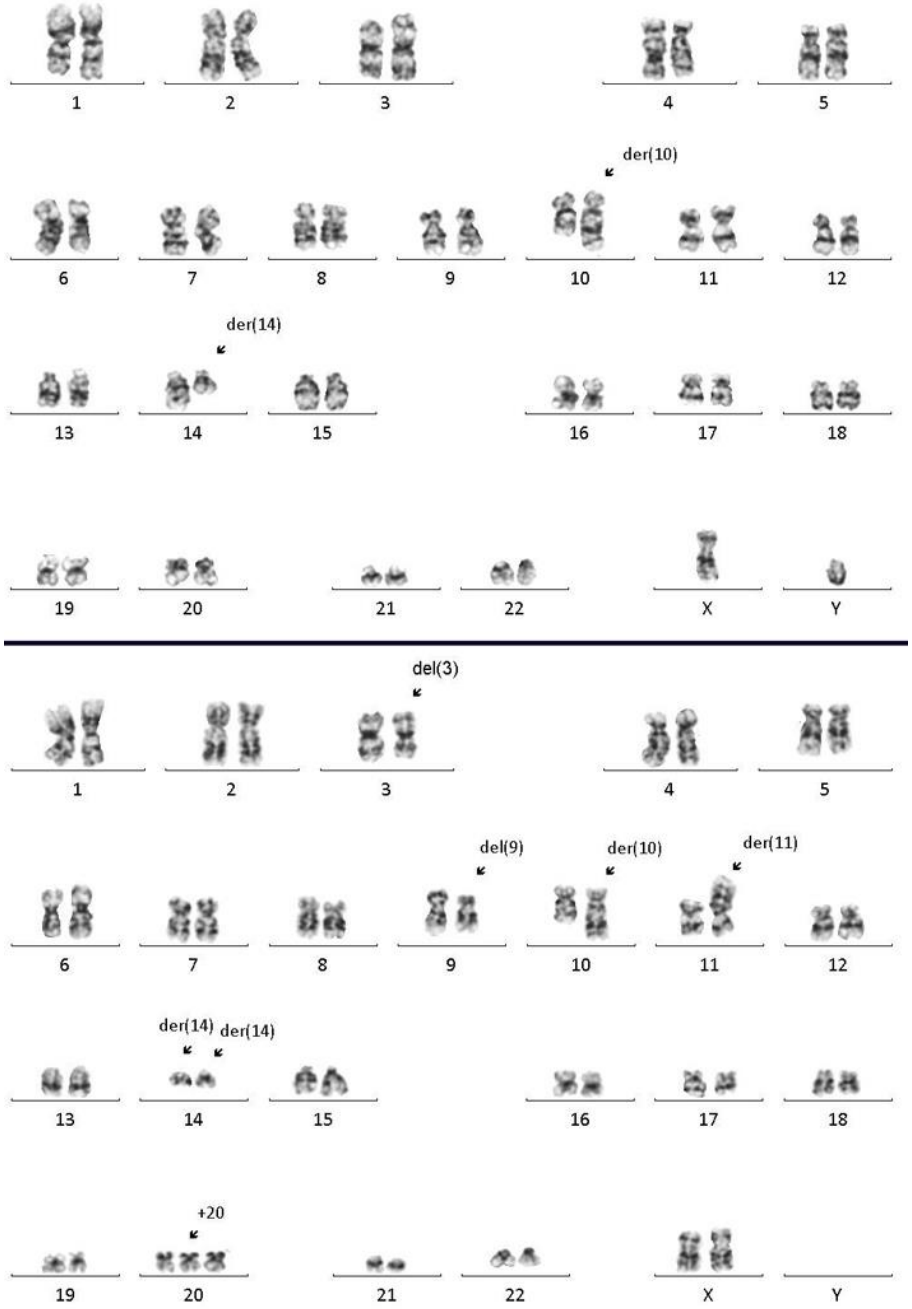
TLX1 expression has been linked with a favorable prognosis and low risk of relapse in children and adults (van Vlierberghe et al., 2012). TLX1+ patients have a 92% probability of survival at 5 years. The lack of expression of anti-apoptotic genes in this stage of thymocyte development (and in TLX1-expressing lymphoblasts) leads to a high responsiveness to drug-induced apoptosis.

Genetics

Note

Both translocations t(10;14)(q24;q11.2) and t(7;10)(q34;q24) are insufficient to initiate malignancy in mice: activation of other mutant genes, including NOTCH1, is found in most TLX1+ T-ALL. This finding suggests that multiple cooperating changes, leading to impaired DNA repair, lead to T-cell differentiation arrest and leukemogenesis.

Cytogenetics



Top: Translocation t(10;14)(q24;q11) in a 32-year-old male with T-ALL and OS 33 months+ - Courtesy Tatiana Gindina. Bottom: Complex karyotype with two translocations t(10;14)(q24;q11) and t(11;14)(p13;q11), deletions of 3q, 9p, trisomy 20 in a 31-year-old female with T-ALL and OS 37 months - Courtesy Tatiana Gindina.

Additional anomalies

Additional chromosome anomalies were observed in about half of the cases (Dube et al., 1986; Raimondi et al., 1988; Kagan et al., 1989; Rack et al., 1997; Forestier et al., 1998; Lai et al., 2000; Schneider et al., 2000; Kahl et al., 2001; Pedersen et al., 2001; Mancini et al., 2002; Nordgren et al., 2002; Kristensen et al., 2003; Barber et al., 2004; Speleman et al., 2005; Stergianou et al., 2005; Cauwelier et al., 2006; Reichard et al., 2006; Strefford et al., 2007; Le Noir et al., 2012; Setoodeh & Zhang, 2012; Grossmann et al., 2013; Safavi et al., 2015; Gindina T., three personal cases). The most common of them

are deletions of 9p (17%), 6q (13%), and 12p (9%). Deletions of 11p, 6p, 3q, 7q, 13q were less common. Trisomy 8 was present in 6 (7%) patients, and trisomy 20 was in 4 (4,5%) patients. Trisomies of other chromosomes were detected very seldom. In 34 (38%) cases the translocations t(10;14) and t(7;10) were a part of a complex karyotype.

Genes involved and proteins

TLX1 (T-cell leukemia homeobox 1)

Location
10q24.31

Note
Alias HOX11 (homeobox 11) alias TCL3 (T-cell leukemia 3).

DNA/RNA
spans over 7 kb, 3 exons, mRNA 7 kb

Protein
The homeobox gene TLX1 (HOX11) encodes for TLX1, a homedomain-containing transcription factor, nuclear transcription factor that belongs to the NK-linked or NK-like subfamily of homeobox genes. TLX1 is the founding member of a family of HOX genes, homeoprotein, 61 amino acids, nuclear localization. It binds to the DNA sequence 5'-GGCGGTAAGTGG-3'. The encoded protein is required and critical for normal development of the spleen during embryogenesis and is also involved in specification of neuronal cell fate. TLX1 is not normally expressed in adult tissues at levels detectable by routine Northern analysis.

TLX-1 leukemias show specific cooperating mutations rarely present in non-TLX-induced leukemias, including the NUP214 / ABL1 fusion oncogene and mutations in the PTPN2, Wilms tumor 1 (WT1), and PHF6 tumor supressors (van Vlierberghe et al., 2012).

TRD (T cell Receptor Delta)

Location
14q11.2

Note
or TRB in the case of a 7q34-36 involvement

Protein
T-cell receptor

Result of the chromosomal anomaly

Fusion protein

Description
Both translocations place TLX1 under the control of strong enhancers in the T-cell receptor loci. The t(10;14) translocation places the TLX1 coding region under the transcriptional control of the TCR delta receptor (TRD)(14q11.2) promoter, the t(7;10) translocation places the TLX1 coding region under the transcriptional control of the TCR beta locus (TRB)(7q34-36), both leading to increased expression of TLX1 in T-cells. The t(10;14) can be detected by PCR, and a dual-color FISH probe is often used to detect HOX11 translocations on 10q24. However, TLX1 overexpression in leukemic blasts has been observed in the absence of 10q24 rearrangement in as many as 50% of T-ALL cases.

Oncogenesis
These genetic aberrations induce aberrant and abundant TLX1 expression in T- lineage cells bearing the translocation. In addition, it was recently proposed that the unique cortical thymic maturation arrest in TLX-induced leukemias may be related to the binding of TLX1- ETS1 complexes to TCRA enhancer sequences, with the consequent down regulation of TCRA gene rearrangement and expression (van Vlierberghe et al., 2012).

TLX1 overexpressing lymphoblasts are arrested at the stage of beta-selection in the thymocyte development. Leukemogenesis results from decreased cell death and increased proliferation of immature TLX1 expressing thymocytes, in the absence of normal DNA repair systems. The lack of anti-apoptotic actors in this stage explains the high responsiveness to chemotherapy, and the associated excellent outcome.

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