

Leukaemia Section

Short Communication

t(5;12)(q13;p13) ?/ETV6

Jean-Loup Huret

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

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Abstract

Short Communication on t(5;12)(q13;p13) ?/ETV6, with data on clinics, and the genes implicated.

Clinics and pathology

Disease

Acute myeloid leukemia (AML) and myelodysplastic syndrome (MDS), as well as acute lymphoblastic leukemia (ALL)

Phenotype/cell stem origin

Five cases are available to date: 3 myeloid cases: an AML not otherwise specified, a M2-AML, and a RAEB transforming into M2-AML (Kobayashi et al., 1994; Yamamoto et al., 2000; Park et al., 2007); and 2 lymphoid cases: 2 ALLs (Heinonen et al., 1988; Sato et al., 1997).

Is not taken into account in this review a case of translocation between 5q13, 12p13, 22q11, and 3q12 with no possible fusion transcript on any derivative chromosome, although ETV6 (12p13) and MN1 (22q12), with also sequences on 5q and on 3q are implicated in the rearrangements (Belloni et al., 2004).

One case occurred after exposure to mutagenic agents: a patient experienced a renal carcinoma 3 years before onset of the secondary MDS (Yamamoto et al., 2000).

Epidemiology

The myeloid cases were a 58-year old female patient and two 65-year old male patients; the ALL cases were a 1-year old girl and a 12-year old girl.

Prognosis

Scarce data available: a M2-AML case was still alive but in relapse 9 months after diagnosis (Park et al., 2007), and the MDS case evolved towards a M2-AML and the patient died 4 months after diagnosis of the MDS (Yamamoto et al., 2000).

Cytogenetics

Cytogenetics morphological

The t(5;12) was the sole anomaly in two myeloid cases and part of a complex karyotype in the treatment-related myeloid case.

The t(5;12) was accompanied with a marker chromosome in the infant case, and with numerical anomalies in the other lymphoid case.

Genes involved and proteins

Note

The partner(s) of ETV6 in 5q13 remain(s) unknown.

Both one myeloid and one lymphoid cases were tested, and it appears that the breakpoint in ETV6 is not identical: the breakpoint was located in intron 1, and exon 1 was deleted, in the treatment-related myeloid case (Yamamoto et al., 2000), whereas it was located 3' of the ETV6 coding sequence in an ALL case (Sato et al., 1997).

ETV6

Location

12p13

Protein

452 amino acids. ETV6 is composed of a HLH domain responsible for hetero- and homodimerization in N-term, and an ETS domain responsible for sequence specific DNA-binding in C-term (binds to the DNA sequence 5'-CCGGAAGT-3'). Transcriptional regulator; tumor suppressor. Involved in bone marrow hematopoiesis.

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