Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Leukaemia Section

Mini Review

t(2;7)(p11;q21), t(7;14)(q21;q32) CDK6/IgH, t(7;22)(q21;q11)

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Identity

Note
The translocation t(7;14)(q21;q32) herein described must not be confused with the t(7;14)(q21;q32) with ERVWE1/IgH involvement, seen in chronic lymphocytic leukaemia (Wahbi et al., 1997). There is also a case of acute myeloid leukaemia with t(7;14)(q21;q32) in a female patient, with no other data (Stephenson et al., 1995).

Clinics and pathology

Disease

Chronic lymphocytic leukaemia (CLL) and splenic marginal zone lymphoma (SMZL) (Corcoran et al., 1999; Hayette et al., 2003).

Phenotype/cell stem origin

Six patients, aged 52-79 years, 5 male / 1 female cases.

Prognosis

Survival data was: 3 years+, 6 years+, and a case dead 16 years after diagnosis in the CLL cases; 4 years+, 5 years+, and 8 years+ in the SMZL and variants cases.

Cytogenetics

Cytogenetics morphological

Sole anomaly in two of six cases.

Additional anomalies

Accompanied with +12 in two CLL cases, del(13q) in one CLL, del(7q) in one SMZL case.

Genes involved and proteins

CDK6

Location
7q21.2

Protein
G1 phase kinase; phosphorylates RB1 but with a distinct activity from CDK4. Control of the cell cycle; blockage of differenciation (Grossel and Hinds, 2006).

IGH

Location
14q32.33

Note
IGH was involved in the one case with a t(7;14), while IGK (2p11) was involved in four cases showing a t(2;7), and IGL (22q11) in the case with a t(7;22).

Protein
Immunoglobulin heavy chain (see Lefranc, 2003).

Result of the chromosomal anomaly

Hybrid gene

Description
Translocation of CDK6 close to enhancers constitutively active in B- cells.

Fusion protein

Note
The case elsewere reported with t(7;14)(q21;q32) ERVWE1/IgH (Wahbi et al., 1997) may also involve

CDK6 instead of ERVWE1, as it is known that IG enhancers may act at long distance. However, the breakpoint in this latter case was found 94 bases upstream ERVWE1. ERVWE1 is 127 kb upstream CDK6 on minus strand of the DNA (ERVWE1 is from 91 935 631 to 91 945 186, and CDK6 from 92 072 173 to 92 303 877), while the breakpoints herein reported are situated within 2 to 66 kb near CDK6, and they appear to be telomeric to CDK6.

Oncogenesis

Overexpression of CDK6 was demonstrated (Hayette et al., 2003).

References

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