Leukaemia Section
Mini Review

**der(6)t(1;6)(q21-23;p21)**

Adriana Zamecnikova
Kuwait Cancer Control Center, Laboratory of Cancer Genetics, Department of Hematology, Shuwaikh, 70653, Kuwait (AZ)

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**Identity**

Partial karyotypes showing the chromosomal translocation der(6)t(1;6)(q21-23;p21) identified by G-banding.

**Clinics and pathology**

**Disease**
Most frequently observed in chronic myeloproliferative disorders, occurs with higher frequency in patients with chronic idiopathic myelofibrosis, polycythemia vera and post-polycythemic myelofibrosis; may be present either at diagnosis or during transformation to advanced stages of the disease.

**Epidemiology**
Described in 20 cases (11 males, 9 females): 1 biphenotypic leukemia (16 years old male); 1 B-cell lymphoma (73 years old female); 2 acute myeloid leukemia (AML) patients (1 male 71 years old, 1 female 28 years old); and in 16 patients with myelofibrosis with myeloid metaplasia (9 males; 7 females); eleven patients had myelofibrosis with myeloid metaplasia, three post-polycythemic myeloid metaplasia, and one post-thrombocythemic myeloid metaplasia; one of these patients, a 47 years old male, progressed to AML. From the known data of 14 patients with myelofibrosis, median age was 65.5 years (range, 38-72 years).

**Clinics**
In the largest study, the anomaly was associated with splenomegaly, elevated WBC count, elevated levels of alkaline phosphatase and lactate dehydrogenase; median overall survival was 7.8 years: five patients have died (one transformed to acute myeloid leukemia and the others died because of sepsis or thrombosis).

**Cytogenetics**

**Cytogenetics morphological**
Breakpoints may be controversial and difficult to ascertain in poor quality preparations. Recently, the same breakpoint on 6p21.3 and clustering of breakpoints near the paracentric region 1q21-23 was described in 14 patients with myelofibrosis with myelocytic metaplasia.
**Additional anomalies**

Sole anomaly in 9 cases (2 AML and 7 cases with myelofibrosis); no recurrent additional anomaly observed in patients with complex karyotypes. 4 patients had two or more different clones (1 patient with biphenotypic leukemia and 3 myelofibrosis cases); among them 2 patients had 1q21-23 rearrangements involving the homologous chromosome 1.

**Result of the chromosomal anomaly**

**Fusion protein**

**Oncogenesis**

The presence of the der(6)t(1;6) results in partial trisomy for 1q21-23 to 1qter and in loss of 6p21 to 6pter. The pathogenetic significance may be the consequence of gain of gene(s) on 1q and/or haplo-insufficiency of gene(s) from 6p and alternatively, rearrangements of one or more genes at the breakpoints. The significance of the 6p21 breakpoint is unclear; however a number of published reports of myelofibrosis with chromosome 6p breakpoints in the region raise the possibility of a gene involved in the pathogenesis of this hematologic disorder. The inability to identify common breakpoints on 1q, suggests that an increase in gene copy number is a pathogenetic event. Whether trisomy 1q is a secondary event to a primary (cryptic? e.g. JAK2 V617F mutation) anomaly as well as the roles of methylation, cytotoxic treatments and the underlying molecular consequences of the rearrangement remain to be determined.

**To be noted**

**Case Report**

der(6)t(1;6)(q21;p21) in myelofibrosis following polycythemia vera.

**References**


*This article should be referenced as such:*