Leukaemia Section
Mini Review

**i(6)(p10)**

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**Identity**

Partial GTG banded karyotype showing i(6)(p10). Courtesy of Cytogenetics Laboratory, Garrahan Pediatrics Hospital, Buenos Aires, Argentina.Clinics and pathology.

**Disease**
Acute lymphoblastic leukemia (ALL)

**Phenotype/cell stem origin**
B-cell precursor (pro-B, common, pre-B) and exceptionally T ALL.

**Etiology**
It has been suggested that, although the isochromosomes are rarely the sole abnormality in the majority of cases, they do occur early in the leukemic process. Martineau et al. proposed that isochromosomes are mechanical indicators of genetic events central to the etiology of the leukemia.

**Epidemiology**
Isochromosomes are a nonrandom chromosomal anomaly in ALL. The incidence of i(6)(p10) in ALL is 0.07%. Only sixteen cases have been reported in ALL and only one in immunoblastic lymphoma. All the patients, except for one adult, were children with a median age of 5 years; sex ratio: 12M/5F.

**Clinics**
The clinical characteristics are virtually unknown.

**Prognosis**
Remains to be determined. The median survival of the reported cases varies from 7 to 40 months.

**Genetics**

**Note**
It has been suggested that a central part of the short arm of chromosome 6p harbours one or more oncogenes directly involved in tumour progression. On the other hand, despite accumulating evidence those deletions of chromosomal bands 6q16-q21 are a critical event in ALL, no suppressor genes have been identified in this region. Recently, it has been described that a minimal deleted interval in 6q21 encompasses the FOXO3A, PRDM1 and HACE1 candidate genes.

**Cytogenetics**

**Note**
The isochromosome 6p was found in the stemline in 12 cases and in a sideline in 5 cases. It was associated with...
pseudo, hyper and hypodiploidies and high ploidies. It occurred more frequently at diagnosis than at relapse.

**Cytogenetics molecular**
Comparative genomic hybridisation studies have detected copy-number increases affecting chromosome 6p in several types of cancer.

**Additional anomalies**
The isochromosome 6p was not observed as a sole anomaly. The majority of cases are part of a complex karyotype and some cases occur with established abnormalities such as der(19)(t(1;19), t(12;21)(p13;q22) and t(14;18)(q32;q21).

**Genes involved and proteins**

**Note**
Not yet defined.

**Result of the chromosomal anomaly**

**Hybrid gene**

**Note**
As a result of the formation of the isochromosome, the structural abnormality results in monosomy for the genes on the long arm, and trisomy for the genes in the short arm. It is not known whether the overexpression of a proto-oncogene or the deletion of a tumour-suppressor gene from the isochromosome contributes to development of proliferation of leukaemia.

**References**


**This article should be referenced as such:**