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Leukaemia Section

Mini Review

i(17q) in myeloid malignancies

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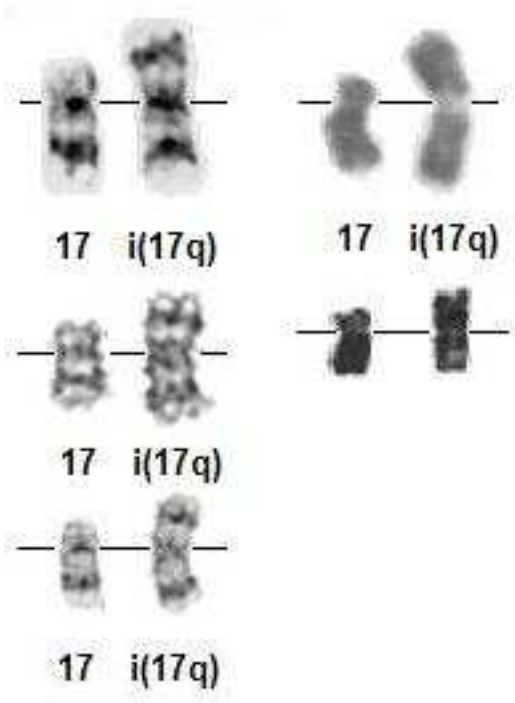

i(17q) G- banding (left) - Courtesy Jean-Luc Lai (top) and Diane H. Norback, Eric B. Johnson, and Sara Morrison-Delap, UW Cytogenetic Services (middle and bottom); and R- banding (right) - top: Editor, bottom: Courtesy Jacques Boyer.

Note: An isochromosome 17 results in a loss of the short arm (17p) and duplication of the long arm (17q) leading to a single copy of 17p and three copies of 17q.

An i(17q), usually observed in a complex karyotype, has been reported in solid tumors and in various types of hematological diseases: acute and chronic myeloid leukemias, acute lymphoid leukemiasand chronic lymphoid leukemias, and Hodgkin and non-Hodgkin lymphomas.

In chronic myeloid leukemia, i(17q) is a frequent and well known secondary anomaly, either solely in 10% of cases, or with other additional anomalies, in at least another 10% of cases, in particular with +8.

Clinics and pathology

Disease

Myeloproliferative / myelodysplastic diseases (MPD/MDS).

Phenotype/cell stem origin

Previous studies on isolated i(17q) have suggested this aberration was associated with chronic myeloid abnormalities with a high rate of progression to ANLL; a new clinico-pathological entity in which i(17q) is the sole abnormality has been reported in a mixed myeloproliferative disorder / myelodysplastic syndrome with an aggressive course; if teen patients were included in this study classified as chronic myeloid malignancy at initial presentation: these features were not confirmed after a negative molecular BCR-ABL analysis in all cases studied (eleven patients).

Etiology

i(17q) as sole cytogenetic aberration represents only 1% of cases in myeloid malignancies.

Cytology

A severe hyposegmentation of neutrophil nuclei (pseudo-Pelger Huet neutrophils (PHH)) and a prominence of the monocyte/macrophage lineage has been noted; other studies have identified an association between hyposegmented neutrophils and loss of 17p

(called 17p- syndrome), always included in complex karyotypes; the i(17q) appeared to be a part of the malignant clone as demonstrated in cases available for a FISH analysis: all myeloid cell lines observed contained the abnormal i(17q), whereas none of the lymphocytes were affected.

Prognosis

By standard Kaplan-Meier analysis, the median survival was 2.5 years (range 0.85-5.25 years).

Genes involved and proteins

Note

The underlying molecular defect that produces the isolated i(17q) is unknown: breakage of the proximal p arm (17p11.2) with rejoining of both centromere-containing chromatids and subsequent inactivation of one centromere; breakpoints could involve important genetic material whose disruption could result in oncogene or tumor suppression gene deregulation.

In understanding the specific i(17q) phenotype, loss of genes localized on 17p were suggested as p53 (17p13.1); a direct correlation between p53 loss and PHH neutrophils was found in a series of MDS and ANLL with 17p- syndrome.

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