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Leukaemia Section
Mini Review

t(3;4)(p21;q34)

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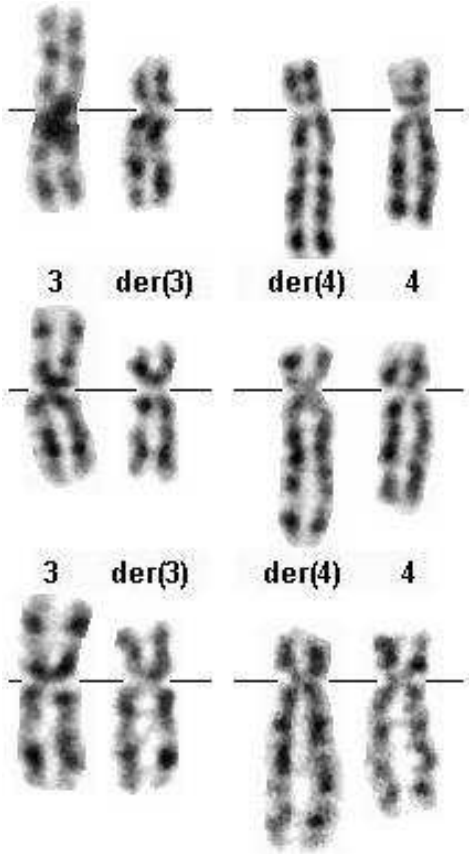


t(3;4)(p21;q34) G-banding

Clinics and pathology

Disease

Myeloid lineage, found in 1 myelodysplastic syndrome (MDS) and 1 Acute Myeloid Leukemia (AML).

Phenotype / cell stem origin

MDS-RA and M1 AML by FAB criteria, a primitive myeloid progenitor is likely to be involved.

Etiology

No known prior exposure.

Epidemiology

Only 2 cases to date, a 69 year old female and a 31 year old male, sex ratio 1M/1F.

Clinics

Elevated WBC ($68x10^9$l), 93% blasts in blood, lymphadenopaty, hepatosplenomegaly, high LDH in AML patient.

Cytology

Positive for CD 34, HLDR, CD33, CD68, MPO in AML.

Treatment

Chemotherapy followed by bone marrow transplantation in AML.

Evolution

After the first cycle of therapy, persistent bone marrow infiltration with 11% blasts.

Prognosis

Survival 6 month in MDS, 15 month+ in AML.

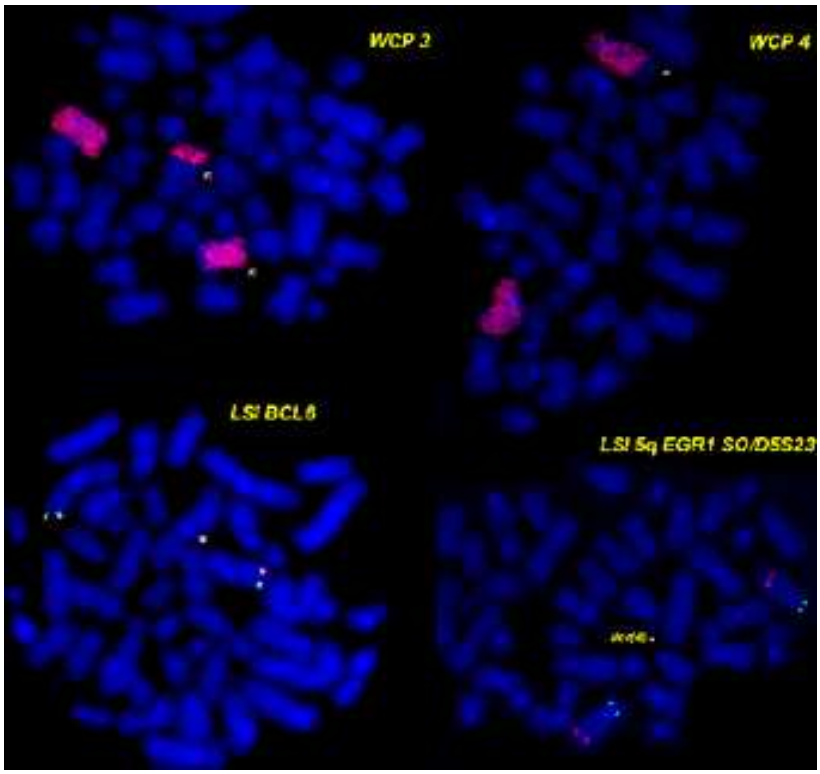
Cytogenetics

Cytogenetics morphological

May be misinterpreted as t(3;5) in suboptimal preparations.

Cytogenetics molecular

FISH analysis is recommended to exclude the more frequent t(3;5).



FISH with WCP 3 and 4 and LSI BCL6 and 5q EGR1 probes.

Probes

WCP 3 and 4 probes, locus specific BCl6 and 5q probes.

Additional anomalies

t(3;4)(p21;q34) is part of a complex karyotype in MDS case associated with del(20q), sole abnormality in AML case.

Genes involved and Proteins

Note: 3p21 is a recurrent breakpoint in MDS/AML and t-MDS/t-AML suggesting, 3p21 site is likely to contain a gene (genes) involved in the pathogenesis of t(3;4)(p21;q34). Frequent deletion or allelic loss of band 3p21 is common in solid tumors, indicating the presence of tumor suppressor genes on this chromosome arm. The association among structural chromosome 3 aberrations and fragile sites on 3p may indicate the importance of previous mutagen exposure in the etiology of these diseases.

Although several cancer-related genes have been located to 3p21, no gene has yet been identified to be related with hematological malignancies. One of the candidate genes may be the AF3p21 gene, a novel fusion partner of the MLL gene described in a patient who had developed therapy-related leukemia with t(3;11)(p21;q23). AF3p21 encodes a protein localized exclusively in the cell nucleus, suggesting the possibility that AF3p21 protein plays a role in signal transduction in the nucleus.

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