

t(5;17)(q33;p11.2)

Clinics and Pathology

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|---------------------|---|
| Disease | Juvenile myelomonocytic leukemia . |
| Epidemiology | Only one case available to date: a 18-month-old boy. |
| Clinics | Marked hepatosplenomegaly, trombocytopenia. |
| Cytology | Peripheral blood leukocytosis with eosinophilia, bone marrow fibrosis (grade III), dry tap. |
| Treatment | Allogeneic BMT. |
| Evolution | Alive and free of disease after 4 years. |

Cytogenetics

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| Probes | LSI CSF1R probe (Vysis) for PDGFRB (5q33); RP11-7904 (telomeric) and RP11-81D5 (centromeric) for HCMOGT-1 (p11.2). |
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Genes involved and Proteins

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|------------------|---|
| Gene Name | PDGFRB |
| Location | 5q33 |
| Protein | Ig like, transmembrane and tyrosine kinase domains. |

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|------------------|--------------------------|
| Gene Name | HCMOGT-1 |
|------------------|--------------------------|

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|-----------------|---------|
| Location | 17p11.2 |
|-----------------|---------|

Result of the chromosomal anomaly

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|--------------------|---|
| Hybrid gene | 5'HCMOGT-1/3'PDGFRB The HCMOGT1 cDNA sequence was fused in-frame to PDGFRB at nucleotide 1936 and situated 5' to the PDGFRB |
| Description | portion, as in previous studies of PDGFRB fusion partners. |
| Transcript | HCMOGT-1/PDGFRB chimeric RNA present. |

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|-----------------------|--|
| Fusion Protein | Coiled-coil domain from HCMOGT-1 fused to the transmembrane domain and the Tyr kinase domain of PDGFRB; the reciprocal |
| Description | PDGFRB/HCMOGT-1 transcript is not expressed. |

External links

| | | |
|----------------|------------------------------------|---|
| Other database | t(5;17)(q33;p11.2) | Mitelman database (CGAP - NCBI) |
| Other database | t(5;17)(q33;p11.2) | CancerChromosomes (NCBI) |

To be noted

Bibliography

HCMOGT-1 Is a Novel Fusion Partner to PDGFRB in Juvenile Myelomonocytic Leukemia with t(5;17)(q33;p11.2).

Morerio C, Aquila M, Rosanda C, Rapella A, Dufour C, Locatelli F, Maserati E, Pasquali F, Panarello C.

Cancer Research 2004; 64 (8): 2643-2648.
Medline [15087372](#)

Contributor(s)

Written 09-2004 Claudio Panarello.

Citation

This paper should be referenced as such :

Panarello C . t(5;17)(q33;p11.2). Atlas Genet Cytogenet Oncol Haematol. September 2004 .

URL :

<http://www.infobiogen.fr/services/chromcancer/Anomalies/t0517q33p11ID1327.html>

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