

t(1;12)(p36;p13)

Clinics and Pathology

Disease Myeloid disorders: one [chronic myelogenous leukemia](#) with t(9;22) and one [refractory anemia with excess of blasts in transformation](#)

Epidemiology only 2 cases so far: 1 male and 1 female patient, aged 50 and 66 yrs

Prognosis unknown so far

Genes involved and Proteins

Gene Name [MDS2](#)

Location 1p36

Dna / Rna 7 exons; alternate splicing

Gene Name [ETV6](#)

Location 12p13

Dna / Rna 9 exons; alternate splicing

Protein contains a Helix-Loop-Helix and ETS DNA binding domains; wide expression; nuclear localisation; ETS-related transcription factor

Result of the chromosomal anomaly

Hybrid gene Description 5' ETV6 - 3' MDS2; exons 1 and 2 of ETV6 are fused to exons 6 and 7 of MDS2; fusion is not in frame; the reciprocal fusion gene is not expressed

Fusion Protein Description truncated ETV6 lacking the PTN domain and the DNA binding domain

External links

Other database [t\(1;12\)\(p36;p13\)](#) [Mitelman database \(CGAP - NCBI\)](#)

Other database [t\(1;12\)\(p36;p13\)](#) [CancerChromosomes \(NCBI\)](#)

To be noted

Additional cases are needed to delineate the epidemiology of this rare entity:

you are welcome to submit a paper to our new [Case Report section](#).

Bibliography

Granulocytic sarcoma of the larynx preceding chronic myeloid leukemia.

Vassallo J, Altemani AM, Cardinalli IA, Crespo AN, Passos Lima CSP, Eid KA, Souza CA.

Pathol Res Pract 1993; 189: 1084-1086.

A novel gene, MDS2, is fused to ETV6/TEL in a t(1;12)(p36.1;p13) in a patient with myelodysplastic syndrome.

Odero MD, Vizmanos JL, Roman JP, Lahortiga I, Panizo C, Calasanz MJ, Zeleznik-Le NJ, Rowley JD, Novo FJ.

Genes Chromosomes Cancer 2002; 35: 11-19.

Medline [12203785](#)

Contributor(s)

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<http://www.infobiogen.fr/services/chromcancer/Anomalies/t0112p36p13ID1170.html>

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