t(11;22)(q24;q12) in giant cell tumour of bone

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Disease

Giant cell tumour of bone is a locally destructive tumor, usually seen in patients over 20 years of age, a borderline lesion between benign and malignant tumours, with a good prognosis, despite of recurrences and, more rarely, pulmonary metastases. The most frequent genetic finding is telomeric associations.

Genetics

Note

In a study by Scotlandi et al., 2000, was found that a minor population of cells from giant cell tumour of bone samples had an EWSR1/FLI1 transcript, but this was found in a high percentage of samples (13/15).

Genes involved and proteins

FLI1

Location

11q24

Protein

From N-term to C-term: a transactivation domain (TAD) containing multiple degenerate hexapeptide repeats, 3 arginine/glycine rich domains (RGG regions), a RNA recognition motif, and a RanBP2 type Zinc finger. Role in transcriptional regulation for specific genes and in mRNA splicing.

EWSR1

Location

22q12

Protein

5’ EWSR1 - 3’ FLI1. EWSR1 exon 7 is fused in frame to FLI1 exon 6 and/or 5 (type 1 and type 2 fusions respectively), indicating, when both transcripts were produced in a given sample, genetic heterogeneity within the tumour.

Fusion Protein

Fusion of the N terminal transactivation domain of EWSR1 to the ETS type DNA binding domain of FLI1.

References


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