Solid Tumour Section
Short Communication

t(6;22)(p21;q12) in undifferentiated sarcoma

Jean-Loup Huret

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

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Disease
A case of undifferentiated sarcoma of the pubic bone, with multiple pulmonary metastases at diagnosis, was described in a 39-year-old female patient. The patient died 6 months after diagnosis (Yamaguchi et al., 2005).

Cytogenetics

Cytogenetics Morphological
The t(6;22)(p21;q12) was accompanied with a marker chromosome.

Genes involved and proteins

POU5F1
Location
6p21
Protein
360 amino acids in the longest isoform; contains a bipartite DNA-binding domain, composed of a POU-specific domain (amino acids 138-212), and a homeobox (aa 230-289). Transcription factor with a major role during embryogenesis. Binds specifically ATTTGCA.

EWSR1
Location
22q12
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.

Result of the chromosomal anomaly

Hybrid Gene
Description
5’ EWSR1 - 3’ POU5F1. Fusion of exons 1-6 of EWSR1 to part of exon 1, and exons 2-5 of POU5F1.

Fusion Protein
Description
The N terminal transactivation domain of EWSR1 was fused to the DNA binding domains of POU5F1.

References

This article should be referenced as such: