

Solid Tumour Section

Short Communication

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

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Clinics and pathology

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 ATTTGCAT.

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

Yamaguchi S, Yamazaki Y, Ishikawa Y, Kawaguchi N, Mukai H, Nakamura T. EWSR1 is fused to POU5F1 in a bone tumor with translocation t(6;22)(p21;q12). *Genes Chromosomes Cancer*. 2005 Jun;43(2):217-22

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