t(2;22)(q31;q12) in a small round cell tumour

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Disease

A case of undifferentiated small round cell tumour of the frontal region, with multiple metastases at diagnosis (in long bones, lungs, kidney), was described in a 16-year-old male patient. The patient died 20 months after diagnosis (Wang et al., 2007).

Cytogenetics

Cytogenetics Morphological
The t(2;22)(q31;q12) was the sole anomaly.

Genes involved and proteins

**SP3**

Location
2q31

Protein
From N-term to C-term: a transactivation domain, a repressor domain, and 3 C2H2 zinc finger domains. Transcription factor.

**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’ SP3. Fusion of EWSR1 exon 7 to exon 6 of SP3.

**Fusion Protein**

Description
The N terminal transactivation domain of EWSR1 was fused to the zinc fingers (DNA binding domain) of SP3.

References


This article should be referenced as such: