Solid Tumour Section
Short Communication

t(12;22)(q13;q12) in angiomatoid fibrous histiocytoma

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Clinics and pathology

Disease
Angiomatoid fibrous histiocytoma is a rare soft-tissue tumour of low metastatic potential (local recurrence below 15% of cases, and metastases occur in less than 2% of patients), often located in the extremities; it is mostly found in children and young adults. Surgical excision is the treatment of choice.

Epidemiology
Six cases of angiomatoid fibrous histiocytoma with a t(12;22)(q13;q12) have so far been described; there were 4 male and 2 female patients, aged 9, 10, 11, 25, 26, and 79 years (Hallor et al., 2005; Antonescu et al., 2007; Hallor et al., 2007; Rossi et al., 2007; Dunham et al., 2008).

Pathology
Cases seemed to have presented a spindle morphology preferentially, but data is too scarce.

Cytogenetics

Cytogenetics Morphological
The t(12;22)(q13;q12), as well as the t(12;16)(q13;p11) (5' FUS - 3' ATF1), are rarer than the t(2;22)(q34;q12) (5' EWSR1 - 3' CREB1) in angiomatoid fibrous histiocytoma.

Genes involved and proteins

ATF1
Location
12q13

Protein
Contains a KID domain (kinase inductible domain), a basic motif (DNA binding) and a leucine-zipper for dimerization, like CREB1. DNA binding protein, binds the consensus sequence: 5'GTGACGT(A/C)(A/G)-3'. 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' ATF1. EWSR1 exon 7 is fused in frame to ATF1 exon 5 in all but one case, where EWSR1 exon 8 was fused in frame to ATF1 exon 4.

Fusion Protein
Description
Fusion of the N terminal transactivation domain of EWSR1 to the DNA binding domain of ATF1.
References


This article should be referenced as such: