t(1;22)(q23;q12) in myoepithelioma

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Clinics and pathology

Disease
Myoepitheliomas are tumours which can occur in the salivary glands, in soft tissues and other organs, with a variable but generally low to intermediate aggressiveness (myoepithelial carcinoma for the most adverse histology).

Epidemiology
One case to date, a 59-year-old female patient with a 10 years long history. The patient was well 7 months after treatment of a myoepithelioma with an uncertain degree of malignancy (Brandal et al., 2008).

Cytogenetics

Cytenogenetics Morphological
The t(1;22)(q23;q12) was the sole anomaly.

Genes involved and proteins

PBX1
Location: 1q23
Protein
Homeobox protein (homeodomain in amino acids 233-295). Binds the sequence 5'-ATCAATCAA-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' FLI1. EWSR1 exon 7 is fused in frame to PBX1 exon 5.

Fusion Protein
Description
Fusion of the N terminal transactivation domain of EWSR1 to the homeobox (DNA binding domain) of PBX1.

References


This article should be referenced as such: