

Solid Tumour Section

Short Communication

t(19;22)(q13;q12) in myoepithelial carcinoma

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Clinics and pathology

Disease

Myoepithelioma tumours of soft tissue cover a wide range of tumours of various behaviour. While most are of intermediate aggressivity, some metastasize. There is no sex ratio predominance. Mean age at diagnosis is 38 years; with a range of 3-83 years. Of a hundred of cases reviewed by Hornick and Fletcher (2003), 60% were benign and classified as myoepitheliomas or mixed tumors, and 40% were classified as myoepithelial carcinomas or malignant mixed tumours.

Amongst cases with benign or low-grade cytology, with a mean follow-up of 3 years, 20% recurred locally and none metastasized. Amongst cytologically malignant cases, with a mean follow-up of 4 years, 40% recurred locally, 1/3 metastasized, and 4 out of 31 patients died. Tumours are positive for epithelial markers, and for S100 or GFAP, or myogenic markers (Gleason and Fletcher, 2007).

Epidemiology

One case to date, a 40-year-old female patient. After surgical removal, recurrences occurred during 2 years, and metastases appeared 3 years later. The patient finally died 9.5 years after initial diagnosis (Brandal et al., 2009).

Cytogenetics

Cytogenetics Morphological

The t(19;22)(q13;q12) was found within a complex karyotype.

Genes involved and proteins

ZNF444

Location

19q13

Protein

Possess a SCAN domain and 4 C2H2-type zinc fingers. 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' ZNF444; fuses EWSR1 exon 8 to the very near end of ZNF444 (at nucleotide 967, while the full transcript of ZNF444 is 984 nt long!).

Fusion Protein

Description

Truncated EWSR1 with 6 amino acids added from

ZNF444. This does not fit with the usual model of carcinogenesis found with other EWSR1 translocations, were there is fusion of the N terminal transactivation domain of EWSR1 to the DNA binding domain of the partner (e.g. FLI1).

References

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