t(6;22)(p21;q12) in hidradenoma of the skin

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Clinics and pathology

**Disease**
Hidradenoma or eccrine/apocrine acrospiroma, is a benign adnexal tumour developing most often in adults.

**Epidemiology**
Three cases to date, 3 female patients aged 24, 63, and 85 years (Möller et al., 2008).

**Pathology**
There was one atypical, one poroid, and one solid hidradenoma.

**Prognosis**
Prognosis is good in this benign disease.

Cytogenetics

**Cytogenetics Morphological**
The t(6;22)(p21;q12) was the sole anomaly in the only case with karyotypic studies; the 2 other cases were detected by the presence of the fusion transcript.

Genes involved and proteins

**POU5F1**

**Location**
6p21

**Protein**
Homeobox protein (homeodomain in amino acids 230-289 in the 360 aa isoform) with a POU domain (in aa 138-212). Binds the sequence 5'-ATTTGCAT-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' POU5F1. EWSR1 exon 6 is fused in frame to POU5F1 exon 2.

**Fusion Protein**

**Description**
Fusion of the N terminal transactivation domain of EWSR1 to the POU and the homebox (DNA binding domain) of POU5F1.

References

Möller E, Stenman G, Mandahl N, Hamberg H, et al. POU5F1, encoding a key regulator of stem cell pluripotency, is fused to EWSR1 in hidradenoma of the skin and mucoepidermoid carcinoma of the salivary glands. J Pathol. 2008 May;215(1):78-86

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