Bone: t(1;17)(p34;p13) in aneurysmal bone cyst

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Clinics and pathology

**Disease**
Aneurysmal bone cysts

**Note**
Benign but locally aggressive tumor.

**Phenotype / cell stem origin**
Occurs mainly in vertebrae and flat bones. Multiple involvement is frequent.

**Etiology**
May involve the arrest of maturation of the osteoblasts caused by USP6 overexpression and dysregulation of autocrine BMP (bone morphology protein) signaling (Lau et al., 2010).

**Epidemiology**
Usually seen in patients aged 10-20 years; represents about 5% of primary bone tumours; slightly more frequent in female patients.

**Clinics**
Forms a spongy hemorrhagic mass; symptoms are pain, swelling, pathological fractures. One case to date was found with a t(1;17)(p34;p13), a 7-year-old boy with a tumor located in the tibia (Althof et al., 2004; Oliveira et al., 2005).

**Treatment**
Surgical curetage.

**Prognosis**
Recurrence occurs in one fourth of cases.

Cytogenetics

**Cytogenetics Morphological**
The t(1;17)(p34;p13) was the sole anomaly.

**Genes involved and proteins**

**THRAP3**

**Location**
1p34

**Protein**
THRAP3, also called TRAP150, is made of an arginine/serine-rich sequence in the N-terminal region and domains with similarity with BCLAF1 and with CASC3/MLN51 in the C-terminal region. It is part of the transcription regulatory complex TRAP/Mediator, and a component of the spliceosome. It both activates pre-mRNA splicing and induces mRNA degradation. The arginine/serine-rich N-term of TRAP3 is responsible for its splicing activity, and the C-term part for its mRNA degradation activity (Lee et al., 2010).

**USP6**

**Location**
17p13

**Protein**
USP6, also called TRE17/ubiquitin-specific protease 6 (USP6), is a deubiquitinase. It is the first de-ubiquitinating enzyme to activate NF-KB, and requires both catalytic subunits of IKK (IKKalpha and IKKbeta) (Pringle et al., 2011).
Result of the chromosomal anomaly

Hybrid Gene

Description
5' THRAP3 - 3' USP6

Fusion Protein

Description
Fusion of the noncoding exon 1 of THRAP3 to a splicing variant of USP exon 1 (i.e. to the entire coding sequence of USP6, resulting in upregulation of USP6).

References


This article should be referenced as such: