Bone: t(9;17)(q22;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 tumors; 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(9;17)(q22;p13), a 10-year-old boy with a tumor located in the clavicle (Oliveira et al., 2005). However, a malignant fibrous histiocytoma has also been described with a t(9;17)(q22;p13), as part of a complex karyotype (Sozzi et al., 1997).

**Treatment**
Surgical curettage.

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

**Cytogenetics**

**Cytogenetics Morphological**
The t(9;17)(q22;p13) was the sole anomaly.

**Genes involved and proteins**

**OMD**

**Location**
9q22

**Protein**
OMD (osteomodulin), also called OSAD (osteoadherin), is a member of the small leucine rich-repete proteoglycan (SLRP) family. It is an extracellular matrix keratan sulfate proteoglycan restricted to mineralized tissues. OMD is a marker for terminally differentiated matrix producing osteoblasts. OMD expression enhances the differentiation and maturation of osteoblasts. It is induced by osteoclast activity (Rehn et al., 2008).

**USP6**

**Location**
17p13

**Protein**
USP6, also called TRE17/ubiquitin-specific protease 6 (USP6), is a deubiquitinase. It is the first deubiquitinating 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’ OMD - 3’ USP6
**Fusion Protein**

**Description**
Fusion of the noncoding exon 1 of OMD to a splicing variant of USP exon 1, resulting in upregulation of USP6.

**References**


Rehn AP, Cerny R, Sugars RV, Kaukua N, Wendel M. Osteoadherin is upregulated by mature osteoblasts and enhances their in vitro differentiation and mineralization. Calcif Tissue Int. 2008 Jun;82(6):454-64


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