Bone: t(3;17)(q21;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(3;17)(q21;p13), a 7-year-old girl with a tumor located in the tibia (Oliveira et al., 2005).

**Treatment**
Surgical curettage.

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

**Cytogenetics**

**Cytogenetics Morphological**
The t(3;17)(q21;p13) was the sole anomaly.

**Genes involved and proteins**

**CNBP**

**Location**
3q21

**Protein**
CNBP, also called ZNF9, is made of 7 CCHC-type Zn fingers. Nucleic acid binding protein; binds single stranded DNA and RNA; act as a regulator of transcription and translation of many genes, including MYC. CNBP may regulate gene expression by catalyzing the formation of G4s (G-quadruplexes, formed by intramolecular four-stranded DNA structures).

**Germinal mutations**
Myotonic dystrophy DM2 is caused by expansion of a (CCTG)(n) in CNBP. CNBP has also been implicated in sporadic inclusion body myositis (review in Calcacerta 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’ CNBP - 3’ USP6
**Fusion Protein**

**Description**
Fusion of the noncoding exon 1 of CNBP to USP exon 2 (i.e. to the entire coding sequence of USP6, resulting in upregulation of USP6).

**References**


This article should be referenced as such: