Bone: t(16;17)(q22;p13) in aneurysmal bone cyst

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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. About eleven cases to date have been described with a t(16;17)(q22;p13), 7 female patients aged 5, 7, 13, 13, 14, 15, and 17 years, and 4 male patients aged 10, 12, 13, and 30 years (Panoutsakopoulos et al., 1999; Herens et al., 2001; Wyatt-Ashmead et al., 2001; Althof et al., 2004; Oliveira et al., 2004).

Treatment
Surgical curetage.

Prognosis
Recurrence occurs in one fourth of cases.

Cytogenetics

Cytogenetics Morphological
In 8 of the 11 cases, the t(16;17)(q22;p13) was the sole anomaly.

Genes involved and proteins

CDH11
Location
16q22

Protein
Cell-cell adhesion molecule that mediates adhesion by Ca2+-dependent interactions. Its intracellular domain is anchored to the actin cytoskeleton through alpha and beta-catenin.

Role in maintaining tissue architecture and cell polarity, limiting cell movement and proliferation. CDH11 antagonizes Wnt/beta-catenin signaling pathway, induces apoptosis, and regulates epithelial-mesenchymal transition (Li et al., 2011). CDH11 is involved in various cancers. Tumor suppressor function.

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 (IKK alpha and IKK beta) (Pringle et al., 2011).
Result of the chromosomal anomaly

**Hybrid Gene**

**Description**
5' CDH11 - 3' USP6

**Fusion Protein**

**Description**
The promotor of CDH11 is juxtaposed to the entire sequence of USP6.

**References**


This article should be referenced as such: