Bone: t(17;17)(p13;q21) 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. Two cases to date were found with a t(17;17)(p13;q21), an 8-year-old boy with a tumor of the soft tissues, and a 13-year-old boy with a tumor located in the tibia (Oliveira et al., 2005; Panagopoulos et al., 2008).

Treatment
Surgical curetage.

Prognosis
Recurrence occurs in one fourth of cases.

Cytogenetics

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

Genes involved and proteins

COL1A1
Location
17q21

Protein
Two pro a1(I) chain associate in trimers with one pro a2(I) chain to form the type I collagen fibrils after proteolysis. Constituent of the extra cellular matrix in connective tissue of bone, skin, tendon, ligament, teeth.

Germinall mutations
COL1A1 has been found mutated in osteoporosis, osteogenesis imperfecta types I-IV, Ehlers-Danlos types I and VIIA, and Caffey disease (Stover and Verrelli, 2011).

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' COL1A1 - 3' USP6

Fusion Protein

Description
Fusion of the exon 1 of COL1A1 to a splicing variant of USP exon 1 in the case reported in Oliveira et al., 2005.

References


This article should be referenced as such: