

Solid Tumour Section

Short Communication

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 was found with a t(17;17)(p13;q21), a 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 $\alpha 1(I)$ chain associate in trimers with one pro $\alpha 2(I)$ 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.

Germinal 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

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