

Solid Tumour Section

Short Communication

Bone: t(16;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 tumours; 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- κ B, and requires both catalytic subunits of IKK (IKK α and IKK β) (Pringle et al., 2011).

Result of the chromosomal anomaly

Hybrid Gene

Description

5' CDH11 - 3' USP6

Fusion Protein

Description

The promoter of CDH11 is juxtaposed to the entire sequence of USP6.

References

Panoutsakopoulos G, Pandis N, Kyriazoglou I, Gustafson P, Mertens F, Mandahl N. Recurrent t(16;17)(q22;p13) in aneurysmal bone cysts. *Genes Chromosomes Cancer*. 1999 Nov;26(3):265-6

Herens C, Thiry A, Dresse MF, Born J, Flagothier C, Vanstraelen G, Allington N, Bex V. Translocation (16;17)(q22;p13) is a recurrent anomaly of aneurysmal bone cysts. *Cancer Genet Cytogenet*. 2001 May;127(1):83-4

Wyatt-Ashmead J, Bao L, Eilert RE, Gibbs P, Glancy G, McGavran L. Primary aneurysmal bone cysts: 16q22 and/or 17p13 chromosome abnormalities. *Pediatr Dev Pathol*. 2001 Jul-Aug;4(4):418-9

Althof PA, Ohmori K, Zhou M, Bailey JM, Bridge RS, Nelson M, Neff JR, Bridge JA. Cytogenetic and molecular cytogenetic findings in 43 aneurysmal bone cysts: aberrations of 17p

mapped to 17p13.2 by fluorescence in situ hybridization. *Mod Pathol*. 2004 May;17(5):518-25

Oliveira AM, Hsi BL, Weremowicz S, Rosenberg AE, Dal Cin P, Joseph N, Bridge JA, Perez-Atayde AR, Fletcher JA. USP6 (Tre2) fusion oncogenes in aneurysmal bone cyst. *Cancer Res*. 2004 Mar 15;64(6):1920-3

Oliveira AM, Perez-Atayde AR, Dal Cin P, Gebhardt MC, Chen CJ, Neff JR, Demetri GD, Rosenberg AE, Bridge JA, Fletcher JA. Aneurysmal bone cyst variant translocations upregulate USP6 transcription by promoter swapping with the ZNF9, COL1A1, TRAP150, and OMD genes. *Oncogene*. 2005 May 12;24(21):3419-26

Lau AW, Pringle LM, Quick L, Riquelme DN, Ye Y, Oliveira AM, Chou MM. TRE17/ubiquitin-specific protease 6 (USP6) oncogene translocated in aneurysmal bone cyst blocks osteoblastic maturation via an autocrine mechanism involving bone morphogenetic protein dysregulation. *J Biol Chem*. 2010 Nov 19;285(47):37111-20

Li L, Ying J, Li H, Zhang Y, Shu X, Fan Y, Tan J, Cao Y, Tsao SW, Srivastava G, Chan AT, Tao Q. The human cadherin 11 is a pro-apoptotic tumor suppressor modulating cell stemness through Wnt/ β -catenin signaling and silenced in common carcinomas. *Oncogene*. 2011 Dec 5;

Pringle LM, Young R, Quick L, Riquelme DN, Oliveira AM, May MJ, Chou MM. Atypical mechanism of NF- κ B activation by TRE17/ubiquitin-specific protease 6 (USP6) oncogene and its requirement in tumorigenesis. *Oncogene*. 2011 Nov 14;

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