Bone: Subungual exostosis with t(X;6)(q13;q22)

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Identity

Other names: Dupuytren's exostosis

Classification

Note: Benign bone-producing neoplasm of unknown cellular origin.

Clinics and pathology

Disease
Subungual exostosis.

Phenotype stem cell origin
Unknown.

Embryonic origin
Unknown.

Etiology
Unknown.

Epidemiology
Affects children and young adults.

Clinics
Subungual exostosis usually presents as a slowly growing, painful mass localized dorsomedially in the distal phalanx, and in contrast to osteochondroma, there is usually no continuity with the underlying cortex.

Treatment
Surgical excision, but local recurrences are not uncommon.

Prognosis
Excellent.

Cytogenetics

Cytogenetics morphological

\[ \text{t}(X;6)(q22;q13-14). \]

Partial G-banding karyotype showing chromosomes 6 and X in a case of subungual exostosis. The arrows indicate the breakpoints.

Cytogenetics molecular

A Probe specific for COL12A1 (RP11-815E21) identified the breakpoint in 6q14.1, as it showed splitting signals on \text{der}(X) and \text{der}(6). On the same chromosomes, these signals colocalized with the signals of RP11-815E21, encompassing the COL4A5 and IRS4 genes in band Xq22.3.

Probes

RP11-815E21 (COL4A5 and IRS4); RP11-1072D13 (COL12A1).

Variants

The breakpoint on chromosome 6 could be centromeric to COL4A5, in an unknown location.
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FISH experiment revealing the breakpoint regions on both chromosomes 6 and X on a case of subungual exostosis.

Genes involved and Proteins

**COL4A5 (alpha 5 type IV collagen)**

**Location:** Xq22.3  
**Note:** It is currently unknown whether any of these two genes is involved in the pathogenesis of subungual exostosis.

**DNA/RNA**
Genomic (chrX:107,569,810-107,827,431). Three transcript variants: isoform 1 (NM_000495), isoform 2 (NM_033380), isoform 3 (NM_03338).

**Protein**
Three proteins, respectively encoded by the isoform 1 (695 aa), isoform 2 (1691 aa), and isoform 3 (1688 aa).

**COL12A1 (collagen, type XII, alpha 1)**

**Location:** 6q13

**DNA/RNA**
Genomic (chr6:75,850,762-75,972,343). Two transcript variants, a long (NM_004370) and a short isoform (NM_080645).

**Protein**
Two proteins: 1899 amino acids (aa) and 3063 aa, respectively encoded by the short and long transcript isoforms.

Result of the chromosomal anomaly

**Hybride Gene**

**Note:** No detected fusion gene.

To be noted

To elucidate how the transcription of these genes is affected by the translocation, further fresh or fresh frozen samples need to be studied.

References


This article should be referenced as such: