

## Solid Tumour Section

### Mini Review

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

Clelia Tiziana Storlazzi, Fredrik Mertens

Department of Genetics and Microbiology, University of Bari, Bari, Italy (CTS); Department of Clinical Genetics, Lund University Hospital, Lund, Sweden (FM)

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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.

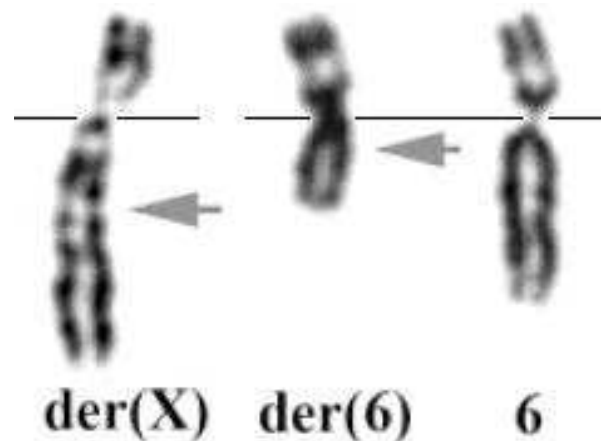
#### Prognosis

Excellent.

### Cytogenetics

#### Cytogenetics morphological

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 der(X) and 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.



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

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