Atlas of Genetics and Cytogenetics
in Oncology and Haematology

OPEN ACCESS JOURNAL AT INIST-CNRS

# Gene Section
**Review**

# ZNF384 (zinc finger protein 384)

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Published in Atlas Database: November 2007

Online updated version: http://AtlasGeneticsOncology.org/Genes/ZNF384ID42881ch12p13.html
DOI: 10.4267/2042/38549

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## Identity

**Hugo:** ZNF384
**Other names:** CAGH1; CAGH1A; CIZ; ERDA2; NMP4; NP; TNRC1
**Location:** 12p13.31
**Local order:** centromere 5'-ZNF384- 3' telomere.

## DNA/RNA

**Note:** GeneLoc location for GC12M006646: Start: 6,645,904 bp from pter; End: 6,668,930 bp from pter; Size: 23,026 bases (23 kb); Orientation: minus strand

### Transcription

Transcript Variant: different alternative splicing isoforms are described.

## Protein

**Note:** Similarity: belongs to the Kruppel C2H2-type zinc-finger protein family; contains 8 C2H2-type zinc fingers.

### Description

Nucleocytoplasmic shuttling protein and transcription factor which appear to bind and regulate the promoter of MMP1, MMP3, MMP7 and COL1A1. Multiple transcript variants encoding several protein isoforms have been found.

### Localisation

Nucleus.


The diagram shows all genes (including ZNF384), with their orientation from centromere to telomere, which are localized in a region going from 6,590 Kbp to 6,720 Kbp at 12p13.


Schematic representation of CIZ protein. LZ: leucine-rich domain SR: serine rich domain PR: Proline rich domain NLS: Nuclear Localization signal ZFs: Kruppel-type C2H2 zinc finger domains QA: Gln-Ala repeat (See also Martini et al., Cancer Research 2002).

# Implicated in

## *Acute lymphoblastic leukemia with t(12;17)(p13;q11) ⟶ TAF15/ZNF384*

**Disease**

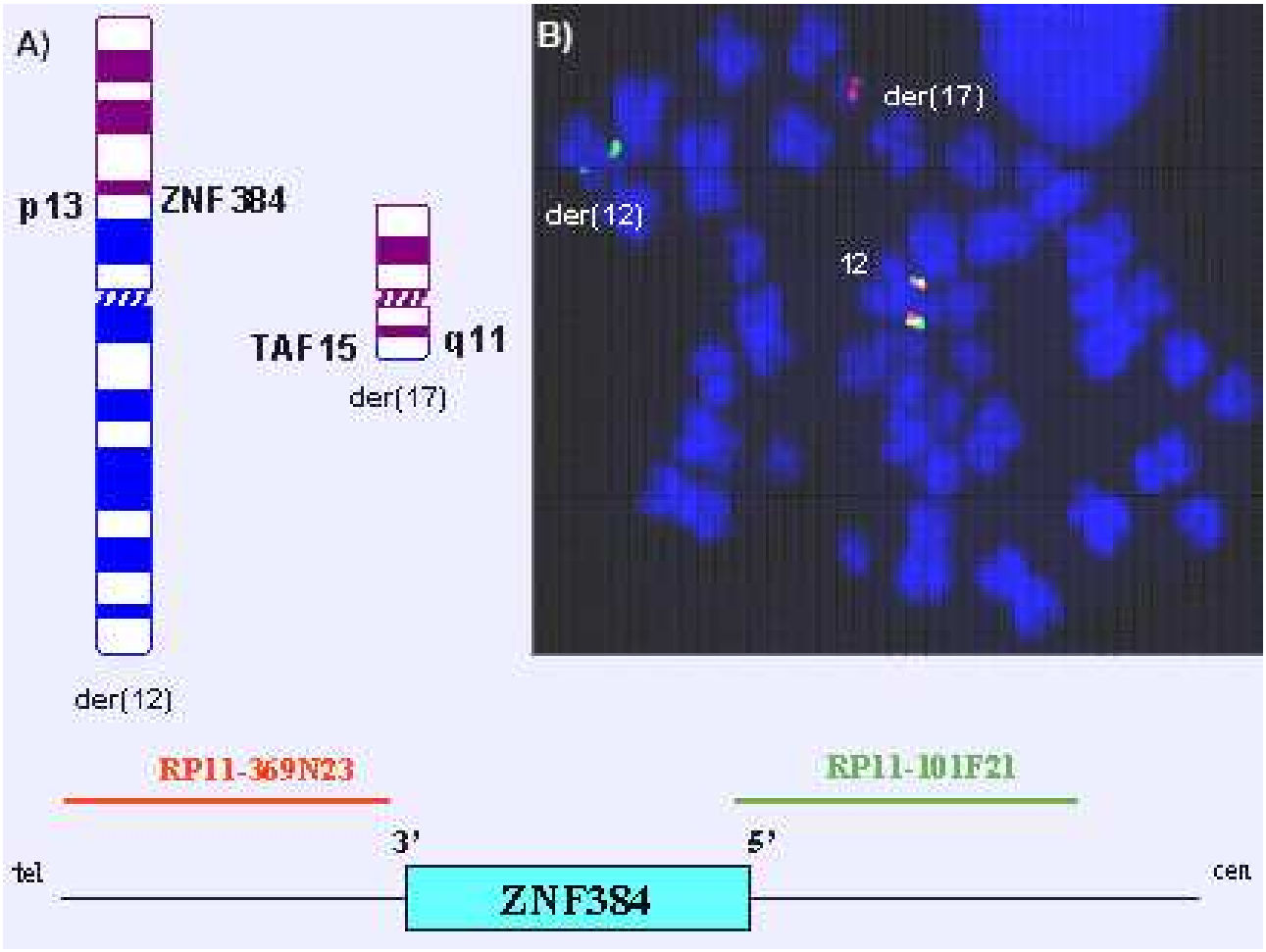
pro-B Acute lymphoblastic leukemia with expression of myeloid antigens (ANPEP/CD13 and/or CD33, and less frequently FUT4/CD15); acute myeloid leukemia.

**Prognosis**

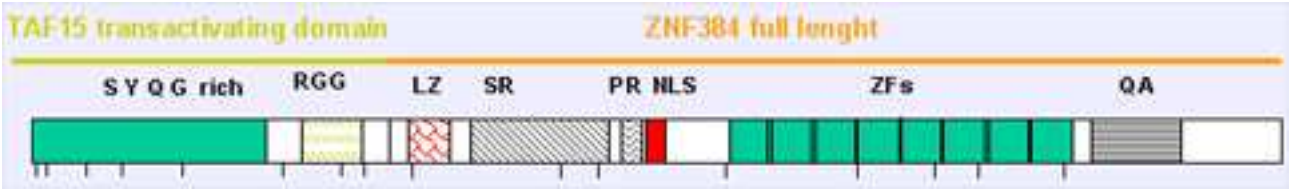
Relatively good prognosis.

**Abnormal Protein**

TAF15-ZNF384




A) schematic representation of the reciprocal t(12;17)(p13;q11) translocation; B) Break-a-part FISH: RP11-369N23 maps telomeric to the 3' ZNF384 while RP11-101F21 partially overlaps with the 5' end of ZNF384 (RP11 clones belong to the Peter De Jong library and were kindly provided by M Rocchi).




Schematic representation of the TAF15-ZNF384 fusion protein. SYQG, Ser-Tyr-Gln-Gly transactivating domain; RGG, Arg-Gly-Gly rich region, (RNA binding domain); LZ, leucine-rich domain; SR, serine rich domain; PR, Proline rich domain; NLS, Nuclear Localization signal; ZFs, Kruppel-type C2H2 zinc finger domains QA: Gln-Ala repeat (see also Martini et al., Cancer Res 2002).

### *Acute lymphoblastic leukemia with t(12;19)(p13;p13) ⟶ E2A/ZNF384*

**Disease**

pro-B Acute Lymphoblastic Leukemia with expression of myeloid antigens.

**Prognosis**

Relatively good prognosis.

**Cytogenetics**

The t(12;19)(p13;p13) is cryptic.

**Abnormal Protein**

ZNF384-E2A

### *Acute lymphoblastic leukemia with t(12;22)(p13;q12) ⟶ EWSR1/ZNF384*

**Disease**

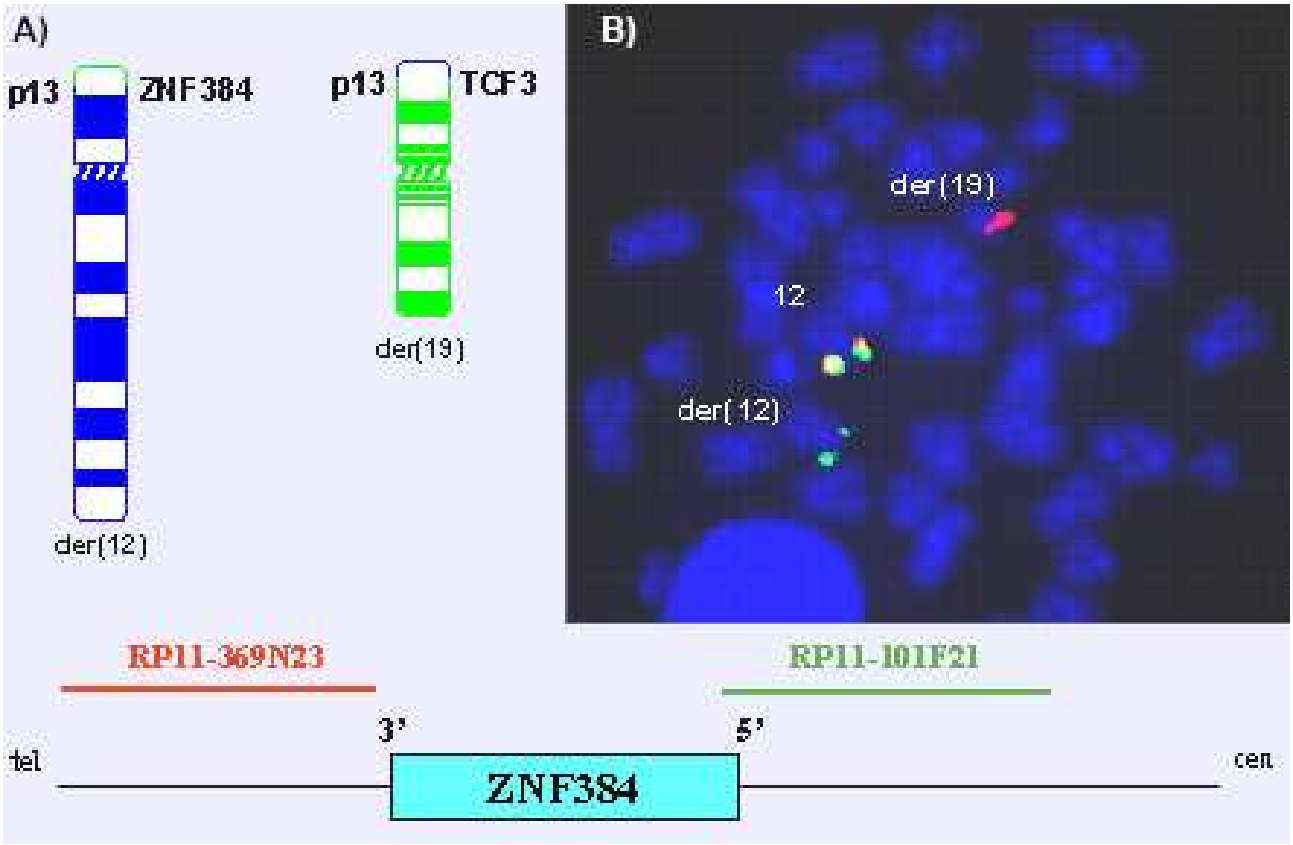
pro-B Acute Lymphoblastic Leukemia with expression of myeloid antigens; biphenotypic leukemia.

**Prognosis**

Relatively good prognosis.

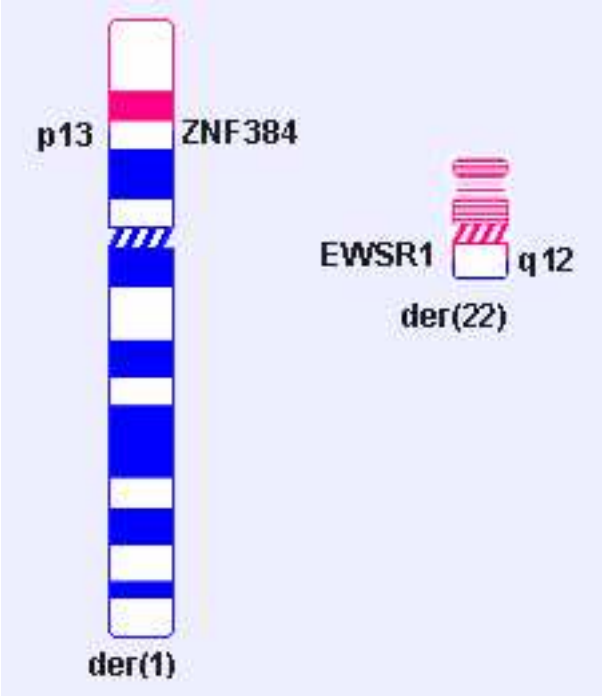
**Abnormal Protein**

EWSR1-ZNF384



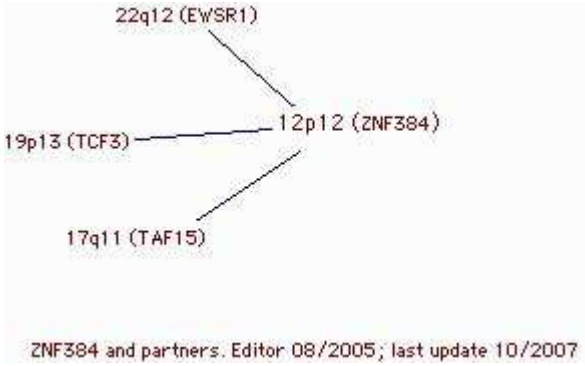

A) schematic representation of the reciprocal t(12;19)(p13;p13) translocation; B) Break-a-part FISH: RP11-369N23 maps telomeric to the 3'ZNF384 while RP11-101F21 partially overlaps with the 5' end of ZNF384 (RP11 clones belong to the Peter De Jong library and were kindly provided by M Rocchi).


Schematic representation of the EWSR1-ZNF384 fusion protein. SYQG, Ser-Tyr-Gln-Gly transactivating domain; LZ, leucine-rich domain; SR, serine rich domain; PR, Proline rich domain; NLS, Nuclear Localization signal; ZFs, Kruppel-type C2H2 zinc finger domains; QA, Gln-Ala repeat (see also Martini et al., Cancer Res 2002).




Schematic representation of the reciprocal t(12;22)(p13;q12) translocation producing the EWSR1-ZNF384 fusion gene.

## Breakpoints

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*This article should be referenced as such:*

Gorello P, La Starza R, Mecucci C. ZNF384 (zinc finger protein 384). Atlas Genet Cytogenet Oncol Haematol.2008;12(4):299-302.