

# Gene Section

## Mini Review

# TFE3 (transcription factor E3)

Jean-Loup Huret, François Desangles

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH), Laboratoire de Biologie, Hopital du Val de Grace, 75230 Paris, France (FD)

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## Identity

Location: Xp11.2

## DNA/RNA



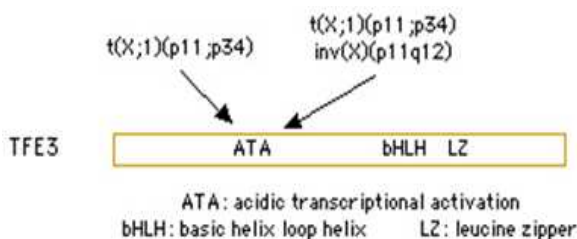
## Description

8 exons.

## Transcription

Differential splicing removing exon 3 (with dominant negative activity of the resulting protein).

## Protein



## Description

743 amino acids; 80 kDa; N-term acidic transcriptional activation domain (domain 260-271, exon 3), helix-loop-helix (344-400), leucine zipper (409-430), and a proline/arginine rich sequence (575-743) C-term.

## Expression

Wide; in fetal and adult tissues.

## Localisation

Nucleus.

## Function

Transcription factor; member of the basic helix-loop-helix family (b-HLH) of transcription factors primarily found to bind to the immunoglobulin enhancer muE3 motif, Ig K enhancers and Ig H variable regions promoters; the helix-loop-helix - leucine zipper region is implicated in DNA binding and dimerization (homo and heterodimerizations); mice which lack TFE3 in their B and T lymphocytes reconstitute the B- and T-cell compartments, but IgM levels are reduced.

## Homology

To other members of the myc family of helix-loop-helix transcription factors.

## Implicated in

***t(X;1)(p11.2;q21.2) in renal cell carcinoma --> PRCC/TFE3***

## Prognosis

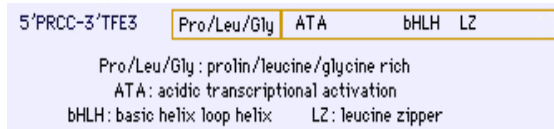
Overall 5-year survival rate around 85%.

## Hybrid/Mutated gene

5' PRCC - 3' TFE3; variable breakpoint in PRCC; breakpoint in the 1st intron of TFE3.

## Abnormal protein

N-term PRCC with the proline rich sequence fused to most of TFE3, including the acidic transcriptional activation domain, the helix-loop-helix, and the leucine zipper; the reciprocal TFE3-PRCC is expressed; it is to be noted that the normal TFE3 transcript is lost in female patients.



### ***t(X;1)(p11.2;p34) in renal cell carcinoma --> PSF/TFE3***

#### **Disease**

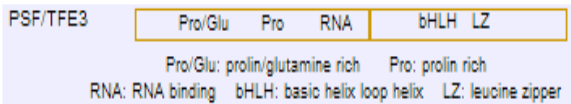
t(X;1)(p11.2;p34) has only been found in a handful cases of papillary renal cell carcinoma.

#### **Hybrid/Mutated gene**

5' PSF - 3' TFE3.

#### **Abnormal protein**

N-term PSF and most of it fused to the DNA binding domains of TFE3 (excluding the acidic transcriptional activation domain, including the C-term helix-loop-helix, and the leucine zipper); no TFE3-PSF reciprocal transcript, as the der(X) t(X;1) is missing; the normal TFE3 transcript is found.



### ***inv(X)(p11.2q12) in renal cell carcinoma --> NONO/TFE3***

#### **Disease**

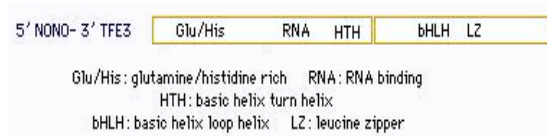
Only one case of papillary renal cell carcinoma.

#### **Hybrid/Mutated gene**

5' NONO - 3' TFE3.

#### **Abnormal protein**

N-term NONO and most of it except the C-term proline rich sequence fused to the DNA binding domains of TFE3 (excluding the acidic transcriptional activation domain, including the C-term helix-loop-helix, and the leucine zipper); the reciprocal transcript is found.



### ***Other Xp11 involvements in renal cell carcinoma (t(X;17)(p11;q25), t(X;10)(p11;q23), etc ...) are likely to implicate TFE3.***

## References

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*This article should be referenced as such:*

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