

TFEB (transcription factor EB)

Identity

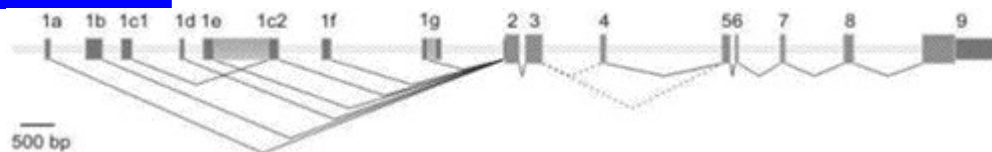
Note Member of the basic-helix-loop-helix leucine-zipper transcription factor MiTF/TFE family (also known as the MiT family), which also contains MiTF, TFEC, and [TFE3](#). The four members form homo- and/or heterodimers to bind the Ebox core sequence CAYGTG.

Other names **T-cell Transcription Factor EB, TCFEB**

Hugo [TFEB](#)

Location 6p21

DNA/RNA



Genomic organization of TFEB gene.

Description TFEB gene contains 8 coding exons and 7 alternative first exons that are differentially expressed.

Transcription Alternative first exon usage points towards the existence of up to seven alternative promoters. Alternative splicing of exon 3 (encoding an acidic activation domain) similar to the closely related TFE3, TFEC, and MiTF genes.

Protein



Functional domains in the TFEB protein.

Description 490 amino acids; 65 kDa; N-terminal Gln-rich stretch (Gln, exon 2), N-term acidic transcriptional activation domain (AAD, exon 3), basic helix-loop-helix region (bHLH, exon 6-8), leucine zipper (LZ, exon 8), proline-rich activation domain (ProAD, exon 9), Ser-rich stretch (Ser)

Expression Wide in fetal and adult tissues, although the various TFEB transcript variants are expressed in a tissue-restricted manner: TFEB-A is enriched in placenta, TFEB-F in spleen, and TFEB-E and TFEB-G in brain.

Localisation Nucleus

Function Transcription factor; member of the basic-helix-loop-helix leucine-zipper transcription factor MiTF/TFE family (also known as the MiT family), which also contains MiTF, TFEC, and TFE3. The four members form homo- and/or heterodimers to bind the Ebox core sequence CAYGTG; the helix-loop-helix - leucine zipper region is implicated in DNA binding and dimerization (homo and heterodimerizations); mice which lack TFEB die due to defects in placental vascularization.

Homology High homology to the other MiTF/TFE members TFE3, TFEC and MiTF, homologous to [myc](#) family of bHLH transcription factors.

Implicated in

Entity [Renal cell carcinoma](#) with t(6;11)(p21;q13) -> Alpha/TFEB gene fusion

Disease Clear cell renal cell carcinomas

Prognosis Limited follow-up available, prognosis appears to be good; no reports of developed metastases

Cytogenetics t(6;11)(p21;q13), usually as the sole cytogenetic anomaly.

Hybrid/Mutated Gene Alpha/TFEB, both fusion genes are expressed; 5'-Alpha-TFEB-3' fusion transcript contains the entire open reading frame of TFEB.

Abnormal Protein No fusion protein, the Alpha gene is a non-protein-encoding transcript.

Oncogenesis Highly induced expression of full-length TFEB protein due to promoter substitution in the Alpha-TFEB fusion gene.

External links

Nomenclature

[Hugo](#) [TFEB](#)
[GDB](#) [TFEB](#)
[Entrez_Gene](#) [TFEB 7942](#) transcription factor EB

Cards

[Atlas](#) [TFEBID531](#)
[GeneCards](#) [TFEB](#)
[Ensembl](#) [TFEB](#)
[CancerGene](#) [TFEB](#)
[Genatlas](#) [TFEB](#)
[GeneLynx](#) [TFEB](#)
[eGenome](#) [TFEB](#)
[euGene](#) [7942](#)

Genomic and cartography

[GoldenPath](#) [TFEB - 6p21](#) [chr6:41759695-41810776 - 6p21.1](#) (hg17-May_2004)

[Ensembl](#) [TFEB - 6p21.1 \[CytoView\]](#)

[NCBI](#) [Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]

[OMIM](#) [Disease map \[OMIM\]](#)

[HomoloGene](#) [TFEB](#)

Gene and transcription

[Genbank](#) [AJ535461](#) [SRS] [AJ535461](#) [ENTREZ]
[Genbank](#) [AL035588](#) [SRS] [AL035588](#) [ENTREZ]
[Genbank](#) [AJ608786](#) [SRS] [AJ608786](#) [ENTREZ]
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[RefSeq](#) [NM_007162](#) [SRS] [NM_007162](#) [ENTREZ]
[RefSeq](#) [NT_086693](#) [SRS] [NT_086693](#) [ENTREZ]
[AceView](#) [TFEB](#) AceView - NCBI
[TRASER](#) [TFEB](#) Traser - Stanford
[Unigene](#) [Hs.485360](#) [SRS] [Hs.485360](#) [NCBI] [HS485360](#) [spliceNest]

Protein : pattern, domain, 3D structure

[SwissProt](#) [P19484](#) [SRS] [P19484](#) [EXPASY] [P19484](#) [INTERPRO]

[Prosite](#) [PS50888 HLH](#) [SRS] [PS50888 HLH](#) [Expasy]
[Interpro](#) [IPR009065 FERM](#) [SRS] [IPR009065 FERM](#) [EBI]
[Interpro](#) [IPR001092 HLH_basic](#) [SRS] [IPR001092 HLH_basic](#) [EBI]
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[Pfam](#) [PF00010 HLH](#) [SRS] [PF00010 HLH](#) [Sanger] [pfam00010](#) [NCBI-CDD]
[Smart](#) [SM00353 HLH](#) [EMBL]
[Blocks](#) [P19484](#)

Polymorphism : SNP, mutations, diseases

[OMIM](#) [600744](#) [[map](#)]
[GENECLINICS](#) [600744](#)
[SNP](#) [TFEB](#) [dbSNP-NCBI]
[SNP](#) [NM_007162](#) [SNP-NCI]
[SNP](#) [TFEB](#) [GeneSNPs - Utah] [TFEB](#) [SNP - CSHL] [TFEB](#) [HGBASE - SRS]

General knowledge

[Family Browser](#) [TFEB](#) [UCSC Family Browser]
[SOURCE](#) [NM_007162](#)
[SMD](#) [Hs.485360](#)
[SAGE](#) [Hs.485360](#)
[Amigo](#) [component|nucleus](#)
[Amigo](#) [process|regulation of transcription, DNA-dependent](#)
[Amigo](#) [function|transcription factor activity](#)
[PubGene](#) [TFEB](#)

Other databases

Probes

[Probe](#) [TFEB Related clones \(RZPD - Berlin\)](#)

PubMed

[PubMed](#) [5 Pubmed reference\(s\) in LocusLink](#)

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Mol Cell Biol 1990; 10: 4384-4388.
Medline [2115126](#)

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Medline [11395386](#)

Recent advances in pediatric renal neoplasia.

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Argani P, Ladanyi M.
Pathology 2003; 35: 492-498 (REVIEW).
Medline [14660099](#)

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Hum Mol Genet 2003; 12: 1661-1669.
Medline [12837690](#)

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Contributor(s)

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URL : <http://www.infobiogen.fr/services/chromcancer/Genes/TFEBID531.html>

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