

POU6F2 (POU domain, class 6, transcription factor 2)

Identity

Other names **RPF-1 (Retina-derived POU-domain factor-1)**
 Hugo [POU6F2](#)
 Location 7p14.1
 Cen-CDC2L5-RALA-POU6F2-VPS41-AMPH-Tel

DNA/RNA

Note POU6F2, previously named RPF-1 was isolated from a retina cDNA library

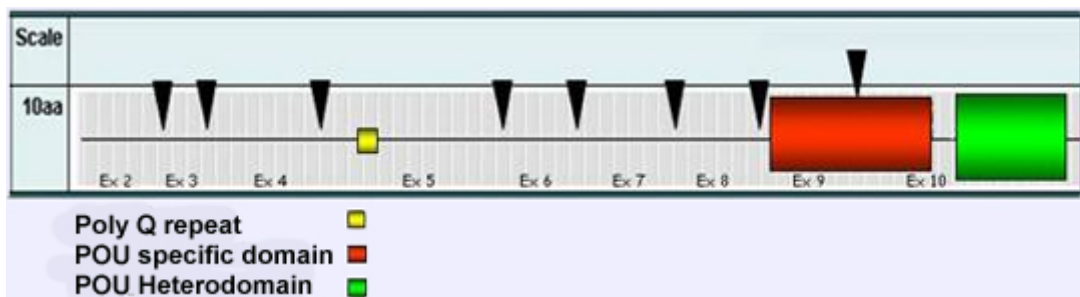


modified from <http://genome.ucsc.edu/>. Exons 1D to 10 of the gene are indicated by the vertical bars.

Description Thirteen exons, including 4 alternative exons 1, encompassing 458 Kb of genomic DNA (exons 1D to 10).

Transcription Representative mRNA: U91935 2159 bases. Alternative splicings: four alternative exons 1; variable skipping of exon 6; variable skipping of both exons 8 and 9; +/- 36 aminoacids at the 5' end of exon 10.

Protein



modified from: <http://www.ebi.ac.uk/>

Description POU6F2 is a member of a gene family whose products are characterized by the presence of a bipartite DNA-binding domain, consisting of a POU-specific domain and a POU heterodomain.

separated by a variable polylinker. Both subdomains contain helix-turn-helix motifs that directly associate with the two components of bipartite DNA-binding sites. In addition, the POU6F2 protein contains a poly-glutamine (poly-Q) domain. Glutamine repeats are evolutionary conserved domains that may act as polar zippers by joining proteins bound to separate DNA segments and thus regulating their activity. POU domain family members are transcriptional regulators, many of which show highly restricted patterns of expression and are known to control cell type-specific differentiation pathways. POU6F2 encodes a deduced 648-amino acid protein. Alternative splicing potentially generates 24 distinct mRNA isoforms coding for proteins with different DNA-binding activity. The most abundant POU6F2 isoforms in human retina have an insertion of an evolutionarily conserved 36-amino acid peptide into the DNA recognition helix of the POU-specific domain. In vitro, the POU domain of POU6F2 lacking the insert binds to a consensus binding site for the product of another gene of the POU family, OCT1, whereas the alternatively spliced POU domain does not.

Expression Immunohistochemical and ribonuclease protection assays showed that in adult mouse *Pou6f2* is expressed within the central nervous system, where its expression is restricted to the medial habenula, to a dispersed population of neurons in the dorsal hypothalamus, and to subsets of ganglion and amacrine cells in the retina. In mouse embryo, *Pou6f2* expression was detected during the earliest stages of retinal differentiation where it appears to be involved in the initial steps of amacrine and ganglion cell commitment. RT-PCR analysis of the mouse *Pou6f2* gene revealed expression in kidney, adrenal gland, heart, stomach, muscle, and eye, but not in lung or skin, of mouse fetuses at embryonic day (E) 18, and in kidney, heart, muscle, spleen, and ovary, but not in lung, of adult mice.

Localisation nuclear (presumptive).

Function POU-domain family transcription factor (presumptive).

Homology Other POU-domain family genes

Mutations

Note The POU6F2 gene is located within an interval on chromosome 7p14 where loss of heterozygosity (LOH) was detected in a fraction of Wilms tumors (WTs), a kidney malignancy of childhood characterized by highly heterogeneous genetic alterations. By sequencing the POU6F2 gene in 12 WTs showing LOH on chromosome 7p14, 2 germline mutations of possible pathogenic significance were identified. The finding of the expression of the POU6F2 mouse homolog in both fetal and adult kidney, together with the demonstration of mutations in WT patients, suggest that the gene is a tumor suppressor and is involved in hereditary predisposition to WT.

Germinal In a patient with WT and LOH at chromosome 7p14, a germline 552G-T transversion in exon 5 of the POU6F2 gene, resulting in a gln184-to-his (Q184H) substitution in a glutamine repeat domain, was identified. The patient showed loss of the constitutionally wildtype allele in tumor DNA.

Neither the mother nor the father carried this mutation. Marker studies indicated that the deletion in tumor DNA was of maternal origin, suggesting that the identified base change most likely occurred as a de novo germline point mutation on the paternal chromosome.

In a patient with WT showing LOH at chromosome 7p14, a germline C-to-G transversion in the untranslated portion of the alternatively spliced exon 1C of the POU6F2 gene was identified. The mutation was inherited from the unaffected mother.

Implicated in

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|------------------|---|
| Entity | Wilms tumor, or nephroblastoma |
| Prognosis | good with treatment according to National Wilms Tumor Study Group (NWTSG) or International Society of Paediatric Oncology (SIOP) or Associazione Italiana Ematologia Oncologia Pediatrica (AIEOP) |

External links

| Nomenclature | |
|-----------------------------|---|
| Hugo | POU6F2 |
| GDB | POU6F2 |
| Entrez_Gene | POU6F2 11281 POU domain, class 6, transcription factor 2 |
| Cards | |
| GeneCards | POU6F2 |
| Ensembl | POU6F2 |
| Genatlas | POU6F2 |
| GeneLynx | POU6F2 |
| eGenome | POU6F2 |
| euGene | 11281 |
| Genomic and cartography | |
| GoldenPath | POU6F2 - 7p14.1 chr7:38819648-39277630 + 7p14.1 (hg17-May_2004) |
| Ensembl | POU6F2 - 7p14.1 [CytoView] |
| NCBI | Genes Cyto Gene Seq [Map View - NCBI] |
| OMIM | Disease map [OMIM] |
| HomoloGene | POU6F2 |
| Gene and transcription | |
| Genbank | AC005483g [SRS] AC005483g [ENTREZ] |
| Genbank | AC073345g [SRS] AC073345g [ENTREZ] |
| Genbank | AC092174g [SRS] AC092174g [ENTREZ] |
| Genbank | U91934g [SRS] U91934g [ENTREZ] |
| Genbank | U91935m [SRS] U91935m [ENTREZ] |
| RefSeq | NM_007252 [SRS] NM_007252 [ENTREZ] |

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|--|--|
| RefSeq | NT_086706 [SRS] NT_086706 [ENTREZ] |
| AceView | POU6F2 AceView - NCBI |
| TRASER | POU6F2 Traser - Stanford |
| Unigene | Hs.137106 [SRS] Hs.137106 [NCBI] HS137106 [spliceNest] |
| Protein : pattern, domain, 3D structure | |
| SwissProt | P78424 [SRS] P78424 [EXPASY] P78424 [INTERPRO] |
| Prosite | PS00027 HOMEobox_1 [SRS] PS00027 HOMEobox_1 [Expasy] |
| Prosite | PS50071 HOMEobox_2 [SRS] PS50071 HOMEobox_2 [Expasy] |
| Prosite | PS00035 POU_1 [SRS] PS00035 POU_1 [Expasy] |
| Prosite | PS00465 POU_2 [SRS] PS00465 POU_2 [Expasy] |
| Interpro | IPR001356 Homeobox [SRS] IPR001356 Homeobox [EBI] |
| Interpro | IPR009057 Homeodomain like [SRS] IPR009057 Homeodomain like [EBI] |
| Interpro | IPR010982 Lambda like DNA [SRS] IPR010982 Lambda like DNA [EBI] |
| Interpro | IPR000327 POU [SRS] IPR000327 POU [EBI] |
| Interpro | IPR007103 POU homeo [SRS] IPR007103 POU homeo [EBI] |
| CluSTr | P78424 |
| Pfam | PF00046 Homeobox [SRS] PF00046 Homeobox [Sanger]] pfam00046 [NCBI-CDD] |
| Pfam | PF00157 Pou SM00389 [SRS] PF00157 Pou SM00389 [Sanger]] pfam00157 [NCBI-CDD] |
| Prodom | PD000010 Homeobox [INRA-Toulouse] |
| Prodom | P78424 PO6F2 HUMAN [Domain structure] P78424 PO6F2 HUMAN [sequences sharing at least 1 domain] |
| Prodom | PD000010 [INRA-Toulouse] |
| Prodom | P78424 PO6F2 HUMAN [Domain structure] P78424 PO6F2 HUMAN [sequences sharing at least 1 domain] |
| Blocks | P78424 |
| Polymorphism : SNP, mutations, diseases | |
| OMIM | 609062 [map] |
| GENECLINICS | 609062 |
| SNP | POU6F2 [dbSNP-NCBI] |
| SNP | NM_007252 [SNP-NCI] |
| SNP | POU6F2 [GeneSNPs - Utah] POU6F2 [SNP - CSHL] POU6F2 [HGBASE - SRS] |
| General knowledge | |
| Family Browser | POU6F2 [UCSC Family Browser] |
| SOURCE | NM_007252 |
| SMD | Hs.137106 |

[SAGE](#) [Hs.137106](#)
[Amigo](#) [process|central nervous system development](#)
[Amigo](#) [process|ganglion mother cell fate determination](#)
[Amigo](#) [component|nucleus](#)
[Amigo](#) [process|regulation of transcription, DNA-dependent](#)
[Amigo](#) [function|transcription factor activity](#)
[Amigo](#) [process|transcription from RNA polymerase II promoter](#)
[Amigo](#) [process|visual perception](#)
[PubGene](#) [POU6F2](#)

Other databases

Probes

[Probe](#) [POU6F2 Related clones \(RZPD - Berlin\)](#)

PubMed

[PubMed](#) [4 Pubmed reference\(s\) in LocusLink](#)

Bibliography

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Medline [8601806](#)

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Genes Chromosomes Cancer 2001 May; 31(1): 42-47.

Medline [11284034](#)

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Hum Mutat 2004 Nov; 24(5): 400-407.

Medline [15459955](#)

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Contributor(s)

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URL :

<http://www.infobiogen.fr/services/chromcancer/Genes/POU6F2ID42963ch7p14.html>

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