

TMPRSS3 (transmembrane protease, serine 3)

Identity

Note	Not to be confused with TMPRSS4 (11q23.3), which was originally named TMPRSS3
Other names	DFNB10 (deafness, autosomal recessive 10) DFNB8 (deafness, autosomal recessive 8) TADG12 (Tumor associated differentially-expressed gene-12 protein) ECHOS1
Hugo	TMPRSS3
Location	21q22.3

DNA/RNA

Description	13 exons spanning 24 kb
Transcription	Four alternative splice isoforms have been described, producing transcripts of 1.3 kb, 2.1 kb, 2.4 kb and 2.5 kb, respectively

Protein

Description	Isoform A (full length) is 454 amino acids; isoforms B and C lack 127 aa at the N-terminus due to alternative splicing; isoform D is 344 aa and has a unique C-terminus due to alternative splicing. The full length isoform comprises an LDL-receptor A domain, a Scavenger receptor (Srcr) domain and a peptidase S1 S6 domain,
Expression	Expressed in many fetal and adult tissues
Localisation	Transmembrane
Function	Transmembrane serine protease; exact function unknown

Mutations

Germinal	Insertion, frameshift and missense mutations in the TMPRSS3 gene have been described in familial congenital (DFNB10) and childhood onset (DFNB8) deafness.
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Implicated in

Entity	Autosomal recessive neurosensory deafness; childhood-onset deafness (DFNB8); Autosomal recessive neurosensory deafness; congenital deafness (DFNB10)
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Entity [Ovarian Cancer](#)

Disease Variant D of TMPRSS3 has been reported to be overexpressed in ovarian carcinomas and has been proposed as a novel diagnostic marker.

Prognosis High expression of variant D is correlated with advanced clinical stages of the disease.

Entity Pancreatic Cancer

Disease TMPRSS3 has been reported to be overexpressed in pancreatic cancer. No information on splice variants or prognostic value is available.

External links

Nomenclature

[Hugo](#) [TMPRSS3](#)

[GDB](#) [TMPRSS3](#)

[Entrez Gene](#) [TMPRSS3 64699](#) transmembrane protease, serine 3

Cards

[GeneCards](#) [TMPRSS3](#)

[Ensembl](#) [TMPRSS3](#)

[CancerGene](#) [TMPRSS3](#)

[Genatlas](#) [TMPRSS3](#)

[GeneLynx](#) [TMPRSS3](#)

[eGenome](#) [TMPRSS3](#)

[euGene](#) [64699](#)

Genomic and cartography

[GoldenPath](#) [TMPRSS3 - 21q22.3](#) [chr21:42665069-42689269 - 21q22.3](#) (hg17-May_2004)

[Ensembl](#) [TMPRSS3 - 21q22.3 \[CytoView\]](#)

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

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

[HomoloGene](#) [TMPRSS3](#)

Gene and transcription

[Genbank](#) [AB038157](#) [SRS] [AB038157](#) [ENTREZ]

[Genbank](#) [AB038158](#) [SRS] [AB038158](#) [ENTREZ]

[Genbank](#) [AB038159](#) [SRS] [AB038159](#) [ENTREZ]

[Genbank](#) [AB038160](#) [SRS] [AB038160](#) [ENTREZ]

[Genbank](#) [AF201380](#) [SRS] [AF201380](#) [ENTREZ]

[RefSeq](#) [NM_024022](#) [SRS] [NM_024022](#) [ENTREZ]

[RefSeq](#) [NM_032401](#) [SRS] [NM_032401](#) [ENTREZ]

[RefSeq](#) [NM_032404](#) [SRS] [NM_032404](#) [ENTREZ]
[RefSeq](#) [NM_032405](#) [SRS] [NM_032405](#) [ENTREZ]
[RefSeq](#) [NT_086913](#) [SRS] [NT_086913](#) [ENTREZ]
[AceView](#) [TMPRSS3](#) AceView - NCBI
[TRASER](#) [TMPRSS3](#) Traser - Stanford
[Unigene](#) [Hs.208600](#) [SRS] [Hs.208600](#) [NCBI] [HS208600](#) [spliceNest]

Protein : pattern, domain, 3D structure

[SwissProt](#) [P57727](#) [SRS] [P57727](#) [EXPASY] [P57727](#) [INTERPRO]
[Prosite](#) [PS01209 LDLRA_1](#) [SRS] [PS01209 LDLRA_1](#) [Expasy]
[Prosite](#) [PS50068 LDLRA_2](#) [SRS] [PS50068 LDLRA_2](#) [Expasy]
[Prosite](#) [PS00420 SRCR_1](#) [SRS] [PS00420 SRCR_1](#) [Expasy]
[Prosite](#) [PS50287 SRCR_2](#) [SRS] [PS50287 SRCR_2](#) [Expasy]
[Prosite](#) [PS50240 TRYPSIN_DOM](#) [SRS] [PS50240 TRYPSIN_DOM](#) [Expasy]
[Prosite](#) [PS00134 TRYPSIN_HIS](#) [SRS] [PS00134 TRYPSIN_HIS](#) [Expasy]
[Prosite](#) [PS00135 TRYPSIN_SER](#) [SRS] [PS00135 TRYPSIN_SER](#) [Expasy]
[Interpro](#) [IPR002172 LDL_receptor_A](#) [SRS] [IPR002172 LDL_receptor_A](#) [EBI]
[Interpro](#) [IPR009003 Pept_Ser_Cys](#) [SRS] [IPR009003 Pept_Ser_Cys](#) [EBI]
[Interpro](#) [IPR001254 Peptidase_S1](#) [SRS] [IPR001254 Peptidase_S1](#) [EBI]
[Interpro](#) [IPR001314 Peptidase_S1A](#) [SRS] [IPR001314 Peptidase_S1A](#) [EBI]
[Interpro](#) [IPR001190 Srcr_receptor](#) [SRS] [IPR001190 Srcr_receptor](#) [EBI]
[CluSTr](#) [P57727](#)
[Pfam](#) [PF00057 Ldl_recept_a](#) [SRS] [PF00057 Ldl_recept_a](#) [Sanger]
] [pfam00057](#) [NCBI-CDD]
[Pfam](#) [PF00089 Trypsin](#) [SRS] [PF00089 Trypsin](#) [Sanger] [pfam00089](#) [NCBI-CDD]
[Blocks](#) [P57727](#)

Polymorphism : SNP, mutations, diseases

[OMIM](#) [605511](#) [map]
[GENECLINICS](#) [605511](#)
[SNP](#) [TMPRSS3](#) [dbSNP-NCBI]
[SNP](#) [NM_024022](#) [SNP-NCI]
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[SNP](#) [TMPRSS3](#) [GeneSNPs - Utah] [TMPRSS3](#) [SNP - CSHL] [TMPRSS3](#) [HGBASE - SRS]

General knowledge

[Family Browser](#) [TMPRSS3](#) [UCSC Family Browser]
[SOURCE](#) [NM_024022](#)

[SOURCE](#) [NM_032401](#)
[SOURCE](#) [NM_032404](#)
[SOURCE](#) [NM_032405](#)
[SMD](#) [Hs.208600](#)
[SAGE](#) [Hs.208600](#)
[Enzyme](#) [3.4.21.-](#) [Enzyme-SRS] [3.4.21.-](#) [Brenda-SRS] [3.4.21.-](#) [KEGG] [3.4.21.-](#) [WIT]
[Amigo](#) [function|chymotrypsin activity](#)
[Amigo](#) [component|endoplasmic reticulum](#)
[Amigo](#) [function|hydrolase activity](#)
[Amigo](#) [component|integral to membrane](#)
[Amigo](#) [component|integral to membrane](#)
[Amigo](#) [function|peptidase activity](#)
[Amigo](#) [process|perception of sound](#)
[Amigo](#) [process|proteolysis and peptidolysis](#)
[Amigo](#) [process|proteolysis and peptidolysis](#)
[Amigo](#) [function|scavenger receptor activity](#)
[Amigo](#) [function|trypsin activity](#)
[PubGene](#) [TMPRSS3](#)

Other databases

Probes

[Probe](#) [TMPRSS3 Related clones \(RZPD - Berlin\)](#)

PubMed

[PubMed](#) [11 Pubmed reference\(s\) in LocusLink](#)

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Medline [16021470](#)

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