

PMS1 (PMS1 postmeiotic segregation increased 1 (S. cerevisiae))

Identity

Other names **HNPCC3**

PMSL1

Hugo **PMS1**

Location 2q31-33

DNA/RNA

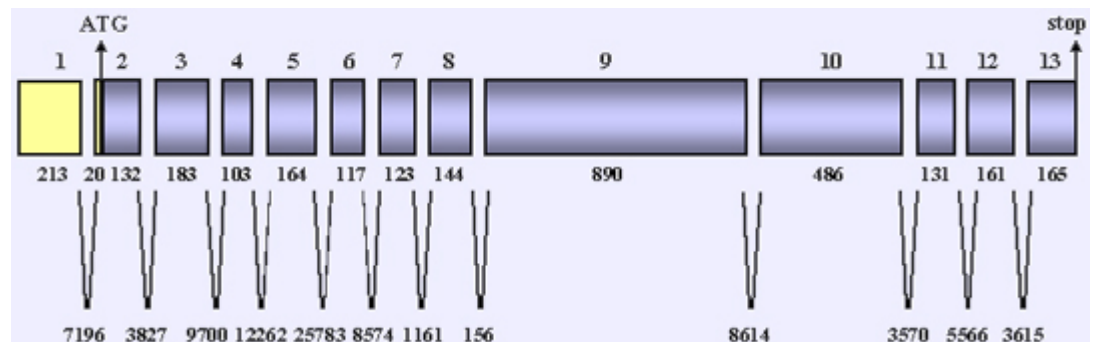


Diagram of the PMS1 gene. Exons are represented by boxes (in scale) transcribed and untranscribed sequences in blue and yellow, with exon numbers on top and number of base pairs at the bottom. Introns are represented by black bars (not in scale) and the number of base pairs indicated. The arrows show the ATG and the stop codons respectively.

Description The PMS1 gene is composed of 13 exons spanning in a region of 93056 bp.

Transcription The transcribed mRNA has 3032 bp

Protein

Description Amino acids: 932. Molecular Weight: 105830 Daltons. PMS1 is a protein involved in the mismatch repair process after DNA replication.

Function PMS1 binds to [MLH1](#) to form a heterodimer, although MLH1 can also bind to [PMS2](#) or [MLH3](#). Although MLH1/PMS2 binds to the heteroduplexes MutSa (composed of [MSH2](#) and [MSH6](#)) or MutSβ (composed of MSH2 and [MSH3](#)), which recognize DNA lesions, it remains to be demonstrated the involvement of the MLH1/PMS1 heterodimer in the mismatch repair process, despite that the heterodimer MLH1/PMS2 is responsible for the recruitment of the

proteins needed for the excision and repair synthesis.

Homology PMS1 is homologue to the bacterial MutL gene and to the Mlh2 gene in yeasts.

Mutations

Germinal A truncating germline mutation of PMS1 was found in one [HNPCC](#) patient. Nevertheless, a MSH2 mutation was found in this family, which was the only one that co-segregated with [colon cancer](#). In addition, no more HNPCC patients have been found with mutations in this gene, and PMS1 -/- mice show no discernible phenotype. So there is no evidence that PMS1 mutations predispose to HNPCC.

External links

Nomenclature

[Hugo](#) [PMS1](#)
[GDB](#) [PMS1](#)
[Entrez_Gene](#) [PMS1_5378](#) PMS1 postmeiotic segregation increased 1 (S. cerevisiae)

Cards

[Atlas](#) [PMS1ID345ch2q31](#)
[GeneCards](#) [PMS1](#)
[Ensembl](#) [PMS1](#)
[Genatlas](#) [PMS1](#)
[GeneLynx](#) [PMS1](#)
[eGenome](#) [PMS1](#)
[euGene](#) [5378](#)

Genomic and cartography

[GoldenPath](#) [PMS1](#) - [chr2:190357355-190450599 + 2q32.2](#) (hg18-Mar_2006)
[Ensembl](#) [PMS1 - 2q32.2 \[CytoView\]](#)
[NCBI](#) [Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]
[OMIM](#) [Disease map \[OMIM\]](#)
[HomoloGene](#) [PMS1](#)

Gene and transcription

[Genbank](#) [AB102869](#) [ENTREZ]
[Genbank](#) [AB102870](#) [ENTREZ]
[Genbank](#) [AB102871](#) [ENTREZ]
[Genbank](#) [AB102872](#) [ENTREZ]
[Genbank](#) [AB102873](#) [ENTREZ]
[RefSeq](#) [NM_000534](#) [SRS] [NM_000534](#) [ENTREZ]
[AceView](#) [PMS1](#) AceView - NCBI

TRASER	PMS1 Traser - Stanford
Unigene	Hs.111749 [SRS] Hs.111749 [NCBI] HS111749 [spliceNest]
Protein : pattern, domain, 3D structure	
SwissProt	P54277 [SRS] P54277 [EXPASY] P54277 [INTERPRO]
Prosit	PS00058 DNA MISMATCH REPAIR 1 [SRS] PS00058 DNA MISMATCH REPAIR 1 [Expasy]
Prosit	PS50118 HMG BOX 2 [SRS] PS50118 HMG BOX 2 [Expasy]
Interpro	IPR003594 ATP bd ATPase [SRS] IPR003594 ATP bd ATPase [EBI]
Interpro	IPR002099 DNA mis repair [SRS] IPR002099 DNA mis repair [EBI]
Interpro	IPR013507 DNA mis repair C [SRS] IPR013507 DNA mis repair C [EBI]
Interpro	IPR009071 HMG-box [SRS] IPR009071 HMG-box [EBI]
Interpro	IPR000910 HMG 1/2 box [SRS] IPR000910 HMG 1/2 box [EBI]
CluSTr	P54277
Pfam	PF01119 DNA mis repair [SRS] PF01119 DNA mis repair [Sanger] pfam01119 [NCBI-CDD]
Pfam	PF02518 HATPase c [SRS] PF02518 HATPase c [Sanger] pfam02518 [NCBI-CDD]
Pfam	PF00505 HMG box [SRS] PF00505 HMG box [Sanger] pfam00505 [NCBI-CDD]
Smart	SM00398 HMG [EMBL]
Blocks	P54277
PDB	2CS1 [SRS] 2CS1 [PdbSum], 2CS1 [IMB] 2CS1 [RSDB]
HPRD	P54277
Protein Interaction databases	
DIP	P54277
IntAct	P54277
Polymorphism : SNP, mutations, diseases	
OMIM	600258 [map]
GENECLINICS	600258
SNP	PMS1 [dbSNP-NCBI]
SNP	NM_000534 [SNP-NCI]
SNP	PMS1 [GeneSNPs - Utah] PMS1 [HGBASE - SRS]
HAPMAP	PMS1 [HAPMAP]
General knowledge	
Family Browser	PMS1 [UCSC Family Browser]
SOURCE	NM_000534
SMD	Hs.111749

SAGE	Hs.111749
Amigo	DNA binding
Amigo	ATP binding
Amigo	nucleus
Amigo	mismatch repair
Amigo	mismatch repair
Amigo	regulation of transcription, DNA-dependent
Amigo	cell cycle
Amigo	negative regulation of progression through cell cycle
PubGene	PMS1

Other databases

Probes

[Probe](#) [PMS1 Related clones \(RZPD - Berlin\)](#)

PubMed

[PubMed](#) [19 Pubmed reference\(s\) in LocusLink](#)

Bibliography

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Medline [11900875](#)

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Fam Cancer 2005; 4: 227-232.
Medline [16136382](#)

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[BiblioGene - INIST](#)

Contributor(s)

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