

POLH (polymerase (DNA direct), eta)

Identity

Note see also the deep insight on: [END_IDENTITY_GENE](#)
 Other names **XP-V**
RAD30A
 Hugo **[POLH](#)**
 Location 6p21.1

DNA/RNA

Description 11 exons.

Of the 11 splice donor/acceptor sites, 10 contained consensus GT/AG dinucleotides; only the splice donor site in exon 11 (sequence CT) varied from the consensus pattern.

The POLH gene lacked a TATA sequence in the region upstream of the transcription-initiation site and the upstream region was GC rich (76% in the sequence between +1 and +270).

The first ATG codon for initiation of translation was included in the second exon. Exon 11 contained the termination codon followed by 661 bp of 3'-untranslated sequence

Protein

Description 713 amino acids

Function The POLH gene encodes DNA polymerase h, which catalyzes the translesion synthesis past a cis-syn T-T pyrimidine dimer, one of the major DNA photoproducts induced by UV light

Homology mXPV: 80.3% amino acids identity and 86.9% similarity

Mutations

Germinal 12 mutated sites involved in the XP variant syndrome

Implicated in

Entity [xeroderma pigmentosum](#), XP group V
 Disease predisposition to skin cancer

External links

[Hugo](#)

[GDB](#)

[Entrez Gene](#)

Nomenclature

[POLH](#)

[POLH](#)

[POLH 5429](#) polymerase (DNA directed), eta

Cards

[Atlas](#)

[GeneCards](#)

[Ensembl](#)

[CancerGene](#)

[Genatlas](#)

[GeneLynx](#)

[eGenome](#)

[euGene](#)

[XPVID303](#)

[POLH](#)

[POLH](#)

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[POLH](#)

[5429](#)

	Genomic and cartography
GoldenPath	POLH - 6p21.1 chr6:43651923-43691357 + 6p21.1 (hg17-May_2004)
Ensembl	POLH - 6p21.1 [CytoView]
NCBI	Genes Cyto Gene Seq [Map View - NCBI]
OMIM	Disease map [OMIM]
HomoloGene	POLH
	Gene and transcription
Genbank	AB038008 [SRS] AB038008 [ENTREZ]
Genbank	AL353602 [SRS] AL353602 [ENTREZ]
Genbank	AY388614 [SRS] AY388614 [ENTREZ]
Genbank	AB024313 [SRS] AB024313 [ENTREZ]
Genbank	AF158185 [SRS] AF158185 [ENTREZ]
RefSeq	NM_006502 [SRS] NM_006502 [ENTREZ]
RefSeq	NT_086693 [SRS] NT_086693 [ENTREZ]
AceView	POLH AceView - NCBI
TRASER	POLH Traser - Stanford
Unigene	Hs.439153 [SRS] Hs.439153 [NCBI] HS439153 [spliceNest]
	Protein : pattern, domain, 3D structure
SwissProt	Q9Y253 [SRS] Q9Y253 [EXPASY] Q9Y253 [INTERPRO]
Prosite	PS50173 UMUC [SRS] PS50173 UMUC [Expasy]
Interpro	IPR001126 UMUC_like [SRS] IPR001126 UMUC_like [EBI]
CluSTr	Q9Y253
Pfam	PF00817 IMS [SRS] PF00817 IMS [Sanger] pfam00817 [NCBI-CDD]
Blocks	Q9Y253
	Polymorphism : SNP, mutations, diseases
OMIM	603968 [map]
GENECLINICS	603968
SNP	POLH [dbSNP-NCBI]
SNP	NM_006502 [SNP-NCI]
SNP	POLH [GeneSNPs - Utah] POLH [SNP - CSHL] POLH [HGBASE - SRS]
	General knowledge
Family Browser	POLH [UCSC Family Browser]
SOURCE	NM_006502
SMD	Hs.439153
SAGE	Hs.439153
Amigo	process DNA replication
Amigo	function damaged DNA binding
Amigo	function eta DNA polymerase activity
Amigo	function magnesium ion binding
Amigo	component nucleoplasm
Amigo	process regulation of DNA repair
Amigo	function transferase activity
KEGG	Purine Metabolism
KEGG	Pyrimidine Metabolism
KEGG	DNA Polymerase
PubGene	POLH
	Other databases
	Probes
Probe	POLH Related clones (RZPD - Berlin)

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