

PAX9 (Paired box gene 9)

Identity

Other names	Paired box homeotic gene 9
Hugo	<u>PAX9</u>
Location	14q12

DNA/RNA

Description	4 exons
Transcription	3 alternative splicing isoforms

Protein

Description	341 amino acids; 36.3kD
Expression	PAX9 is expressed in developing somites, specifically in the posterior ventrolateral region. These cells undergo an epithelial-mesenchymal transition, gaining increased motile capability as a consequence. Subsequent migration of this population generates the lateral sclerotome, which in turn gives rise to the ribs and neural arches. PAX9 contains a DNA binding paired domain, an octapeptide region and a carboxyl-terminal transactivation domain.
Localisation	Nuclear.
Function	PAX9 is a transcription factor that regulates the expression of genes involved in mediating cell proliferation, resistance to apoptosis, and cell migration. Mice homozygous for Pax9 mutations die shortly after birth, lacking the thymus, parathyroid glands and ultimobranchial bodies derived from the third and fourth pharyngeal pouches, presenting aberrant head and visceral skeleton development and a complete absence of teeth.
Homology	PAX9 shares homology through the conserved paired box domain with the other members of the nine strong PAX gene family.

Mutations

Germinal	PAX9 mutations are associated with oligodontia.
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Implicated in

Entity	Oligodontia
Disease	Caused by missense and frameshift PAX9 mutations. Patients present normal primary dentition but lack most permanent molars.

Entity	Jarcho-Levin syndrome
Disease	In humans, a reduction in expression levels of Pax9, together with those of the related Pax1 gene, have been reported in vertebral column chondrocytes of autopsied foeti presenting Jarcho-Levin syndrome, a segmentation anomaly affecting thoracic and vertebral skeletal development. This syndrome shares similarities with the phenotype of the Pax9/Pax1 double mutant mouse.

External links

	Nomenclature	
Hugo	PAX9	
GDB	PAX9	
Entrez_Gene	PAX9_5083 paired box gene 9	
	Cards	
GeneCards	PAX9	
Ensembl	PAX9	
CancerGene	PAX9	
Genatlas	PAX9	
GeneLynx	PAX9	
eGenome	PAX9	
euGene	5083	
	Genomic and cartography	
GoldenPath	PAX9 - 14q12 chr14:36200656-36215621 + 14q13.3 (hg17-May_2004)	
Ensembl	PAX9 - 14q13.3 [CytoView]	
NCBI	Genes Cyto Gene Seq [Map View - NCBI]	
OMIM	Disease map [OMIM]	
HomoloGene	PAX9	
	Gene and transcription	
Genbank	AJ238381 [SRS]	AJ238381 [ENTREZ]
Genbank	AJ238382 [SRS]	AJ238382 [ENTREZ]
Genbank	AJ238383 [SRS]	AJ238383 [ENTREZ]
Genbank	AY338688 [SRS]	AY338688 [ENTREZ]
Genbank	L09745 [SRS]	L09745 [ENTREZ]
RefSeq	NM_006194 [SRS]	NM_006194 [ENTREZ]
RefSeq	NT_086806 [SRS]	NT_086806 [ENTREZ]
AceView	PAX9 AceView - NCBI	
TRASER	PAX9 Traser - Stanford	
Unigene	Hs.132576 [SRS]	Hs.132576 [NCBI] HS132576 [spliceNest]

Protein : pattern, domain, 3D structure

SwissProt	P55771 [SRS] P55771 [EXPASY] P55771 [INTERPRO]
Prosite	PS00034 PAIRED_BOX [SRS] PS00034 PAIRED_BOX [Expasy]
Interpro	IPR009057 Homeodomain_like [SRS] IPR009057 Homeodomain_like [EBI]
Interpro	IPR001523 Paired_box_N [SRS] IPR001523 Paired_box_N [EBI]
CluSTr	P55771
Pfam	PF00292 PAX [SRS] PF00292 PAX [Sanger] pfam00292 [NCBI-CDD]
Smart	SM00351 PAX [EMBL]
Blocks	P55771

Polymorphism : SNP, mutations, diseases

OMIM	167416 [map]
GENECLINICS	167416
SNP	PAX9 [dbSNP-NCBI]
SNP	NM_006194 [SNP-NCI]
SNP	PAX9 [GeneSNPs - Utah] PAX9 [SNP - CSHL] PAX9 [HGBASE - SRS]

General knowledge

Family Browser	PAX9 [UCSC Family Browser]
SOURCE	NM_006194
SMD	Hs.132576
SAGE	Hs.132576
Amigo	function DNA binding
Amigo	process development
Amigo	component nucleus
Amigo	process regulation of transcription, DNA-dependent
PubGene	PAX9

Other databases

Probes

Probe	PAX9 Related clones (RZPD - Berlin)
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PubMed

PubMed	12 Pubmed reference(s) in LocusLink
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Bibliography

The paired box encodes a second DNA-binding domain in the paired homeo domain protein.

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Genes Dev 1991; 5: 594-604.

Medline [1672661](#)

Pax genes and organogenesis.

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Medline [14747376](#)

Pax9-deficient mice lack pharyngeal pouch derivatives and teeth and exhibit craniofacial and limb abnormalities.

Peters H, Neubuser A, Kratochwil K, Balling R.
Genes Dev 1998; 12: 2735-2747.
Medline [9732271](#)

Pax1 and Pax9 synergistically regulate vertebral column development.

Peters H, Wilm B, Sakai N, Imai K, Maas R, Balling R.
Development 1999; 126: 5399-5408.
Medline [10556064](#)

Mutation of PAX9 is associated with oligodontia.

Stockton DW, Das P, Goldenberg M, D'Souza RN, Patel PI.
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Medline [10615120](#)

Haploinsufficiency of PAX9 is associated with autosomal dominant hypodontia.

Das P, Stockton DW, Bauer C, Shaffer LG, D'Souza RN, Wright T, Patel PI.
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Aberrant Pax1 and Pax9 expression in Jarcho-Levin syndrome: report of two Caucasian siblings and literature review.

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A missense mutation in PAX9 in a family with distinct phenotype of oligodontia.

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Pax1 and Pax9 activate Bapx1 to induce chondrogenic differentiation in the sclerotome.

Rodrigo I, Hill RE, Balling R, Munsterberg A, Imai K.
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Novel mutation of the initiation codon of PAX9 causes oligodontia.

Klein ML, Nieminen P, Lammi L, Niebuhr E, Kreiborg S.
J Dent Res 2005; 84: 43-47.
Medline [15615874](#)

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[BiblioGene - INIST](#)

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Citation

This paper should be referenced as such :

Robson E, Whall J, Eccles M . PAX9 (Paired box gene 9). Atlas Genet Cytogenet Oncol Haematol. August 2005 .

URL :

<http://www.infobiogen.fr/services/chromcancer/Genes/PAX9ID41644ch14q12.html>

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