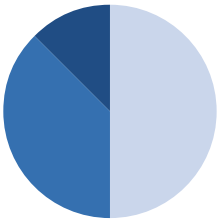
Gene View: Havcr1 (Mammalian)

General View

Report Date: 3:26PM, 2010-06-02
IPA Version: 8.6 (Release Date: 2010-05-28)
Content Version: 3002 (Release Date: 2010-04-22)

General Information

Mutation Information



Mutation to Disease Association

- Asthma 50% Findings (4)
- Rheumatoid Arthritis 37.5% Findings (3)
- Atopic Dermatitis 12.5% Findings (1)

Mutation	Mutation Type	Disease Association	Supporting Evidence
Unspecified	Gene Deletion	rheumatoid arthritis	Show Findings (2)
Unspecified	Gene Deletion	asthma	Show Findings (1)
Unspecified	Gene Deletion	atopic dermatitis	Show Findings (1)
A-1637G	Gene Substitution	rheumatoid arthritis	Show Findings (1)
A-2281G	Gene Substitution	asthma	Show Findings (1)
C2496	Gene Substitution	asthma	Show Findings (1)
Unspecified	Gene Substitution	asthma	Show Findings (1)

Gene Summary

Entrez Gene Name: hepatitis A virus cellular receptor

Synonyms: AI503787, HAVCR, HKIM-1, Kidney Injury Molecule1, KIM-1, TIM-1, TIMD1

Entrez Gene Description: The protein encoded by this gene is a membrane receptor for both human hepatitis A virus (HHAV) and TIMD4. The encoded protein may be involved in the moderation of asthma and allergic diseases. The reference genome represents an allele that retains a MTTVP amino acid segment that confers protection against atopy in HHAV seropositive individuals. Three transcript variants encoding the same protein have been found for this gene. [provided by RefSeq]

Chromosomal Location Information

Homologs of the Havcr1 gene: http://www.ncbi.nlm.nih.gov/mapview/maps.cgi?taxid=9606&chr=5&query=uid%2834583376,16327100,34589047%29&QSTR=26762[gene_id]&maps=gene_set&cmd=focus

Homology Information

The Havcr1 gene is conserved in human, chimpanzee, dog, cow, mouse, and zebrafish.

Protein Family, Domain

Protein Domain	Supporting Evidence
ectodomain	Findings (2)
extracellular domain	Findings (1)
DNA binding	Findings (1)

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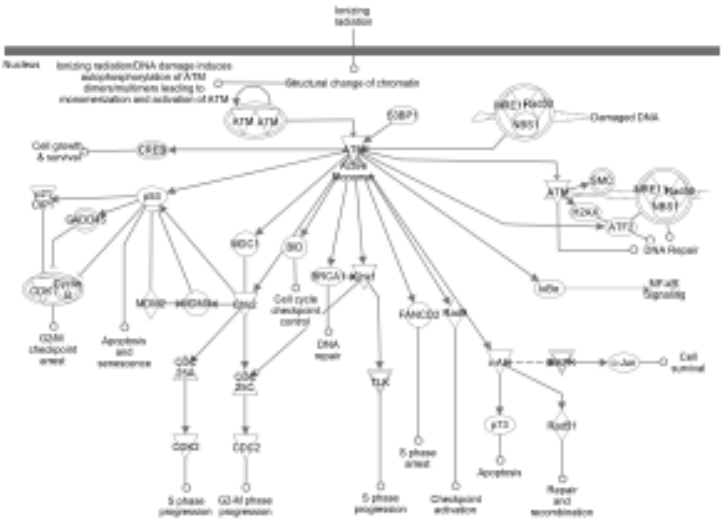
Pathway/Network Information

Canonical Pathways



Location Information

Pathway	Group	Pathway Category	Top Functions and Diseases
ATM Signaling	Signaling Pathway	Cell Cycle Regulation, Cellular Stress and Injury	Cell Cycle; DNA Replication, Recombination, and Repair; Cell Death
Cell Cycle: G2/M DNA Damage Checkpoint Regulation	Signaling Pathway	Cancer, Disease-Specific Pathways	Cell Cycle; Cell Death; Cancer
Hereditary Breast Cancer Signaling	Signaling Pathway	Cancer, Disease-Specific Pathways	DNA Replication, Recombination, and Repair; Cell Cycle; Cancer
Ovarian Cancer Signaling	Signaling Pathway	Cancer, Disease-Specific Pathways	Cell Cycle; Cancer; Neurological Disease
Protein Ubiquitination Pathway	Signaling Pathway	Intracellular and Second Messenger Signaling	Cell-mediated Immune Response; Cellular Development; Cellular Function and maintanance
p53 Signaling	Tox List		Cell Cycle; Cell Death; Cellular Growth and Proliferation
p53 Signaling	Signaling Pathway	Cancer, Ingenuity Toxicity List Pathways	Cell Cycle; Cell Death; Cell-mediated Immune Response
Molecular Mechanisms of Cancer	Signaling Pathway	Cancer, Disease-Specific Pathways	Cell Cycle; Cell Death; Cellular Development
Role of BRCA1 in DNA Damage Response	Signaling Pathway	Cancer, Cellular Stress and Injury	DNA Replication, Recombination, and Repair; Cell Cycle; Cell Death
Role of CHK Proteins in Cell Cycle Checkpoint Control	Signaling Pathway	Cell Cycle Regulation, Cellular Stress and Injury	Cell Cycle; DNA Replication, Recombination, and Repair; Gene Expression
Role of Oct4 in Mammalian Embryonic Stem Cell Pluripotency	Signaling Pathway	Cellular Growth, Proliferation and Development, Organismal Growth and Development	Gene Expression; Organismal Development; Organ Development

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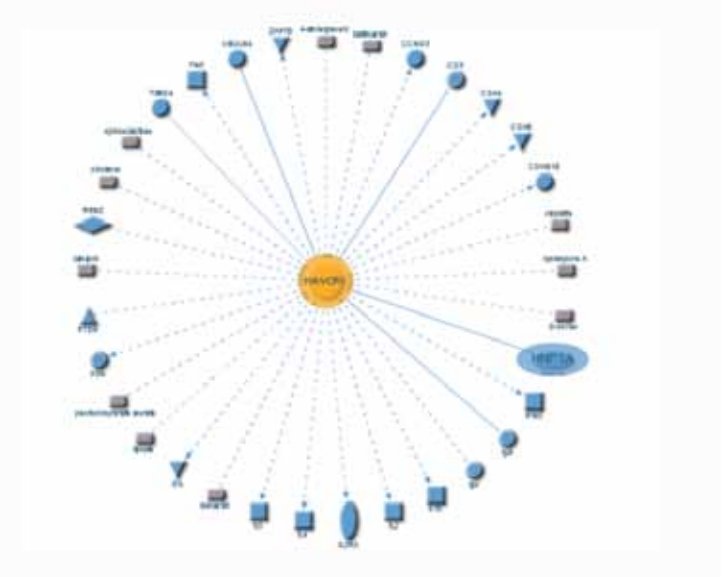
Pathway/Network Information (Continued)

Local Neighborhood



Bibliography

Top Reading List for Havcr1

Rank	Title	Date	Publication	Author	Findings	PMID
1.	Urinary biomarkers for the detection of renal injury	2009	Adv Clin Chem.	Rosner MH.	15	19947356
2.	Kidney injury molecule-1 in renal disease	2010	J Pathol.	Waanders F, van Timmeren MM, Stegeman CA, Bakker SJ, van Goor H.	15	19921716
3.	Kidney injury Molecule-1 (KIM-1): a specific and sensitive biomarker of kidney injury	2008	Scand J Clin Lab Invest Suppl.	Bonventre JV.	9	18569971
4.	Proteomics for the investigation of acute kidney injury	2008	Contrib Nephrol.	Devarajan P.	7	18401157
5.	Monitoring kidney safety in drug development: emerging technologies and their implications	2008	Curr Opin Drug Discov Devel.	Dietrle F, Marrer E, Suzuki E, Grenet O, Cordier A, Vondersher J.	3	1817568
6.	Biomarkers for the diagnosis and risk stratification of acute kidney injury: a systematic review	2008	Kidney Int.	Cocoa SG, Yalavarthy R, Concato J, Parikh CR.	1	18094679

Supporting Literature

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Bibliography (Continued)

Supporting Literature (Continued)

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Recently Added Findings

Findings
Rheumatoid arthritis in humans human is associated with mutant human TIM-1 [HAVCR1] gene (A-1637G with its Promoter [DNA endogenous promoter] mutated).
Atopic dermatitis in humans human is associated with mutant human TIM-1 [HAVCR1] gene (deletion 5383-5397 with its mucin domain mutated)
Asthma in humans human is not associated with mutant human TIM-1 [HAVCR1] gene (C5365T with its mucin domain mutated).
Asthma in humans human is not associated with mutant human TIM-1 [HAVCR1] gene (C2496A).
Rheumatoid arthritis in humans human is associated with mutant human TIM-1 [HAVCR1] gene (deletion 5509-5511CAA with its mucin domain mutated).
Rheumatoid arthritis in humans human is associated with mutant human TIM-1 [HAVCR1] gene (deletion 5383-5397 with its mucin domain mutated).
Asthma in humans human is associated with mutant human TIM-1 [HAVCR1] gene (deletion 5509-5511CAA with its mucin domain mutated).
Asthma in humans human is associated with mutant human TIM-1 [HAVCR1] gene (deletion 5509-5511CAA with its mucin domain mutated).

Date	Publication	Author	Title	PMID
2009, Aug	Trends Mol Med	Meyers JH, Sabatos CA, Chakravarti S, Kuchroo VK	The TIM Gene Family Regulates Autoimmune and Allergic Diseases	16002337