Online Mendelian Inheritance in Animals (OMIA)

enhancement and curation
by and for the animal* genomics community

* Swine and cattle

FACULTY OF
VETERINARY SCIENCE

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THE UNIVERSITY OF SYDNEY

WHAT IS OMIA?

- An annotated catalogue/compendium of
  - inherited disorders
  - other (single-locus) familial traits in animals*
  - animal genes
- Modelled on, and complementary to, the human catalogue/compendium OMIM
- Available freely on the Internet

* except mice and rats (and humans)

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OMIA - ONLINE MENDELIAN INHERITANCE IN ANIMALS

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WELCOME TO OMIA

Online Mendelian Inheritance in Animals (**OMIA**) is a catalogue/compendium of inherited disorders, other (single-locus) traits, and genes in 186 animal species other than human and mouse and rats, which have their own resources) authored by Professor Frank Nicholas of the University of Sydney, Australia, with help from many people over the years. OMIA information is stored in a database that contains textual information and references, as well as links to relevant PubMed and Gene records at the NCBI and to OMIM and Ensembl.

OMIA is manually curated by a team of specialists. If you see an error or wish to submit an entry, please contact us.

From 1st September 2011 the OMIA number is binomial, with the format OMIAxxxxxx-yyyy, where xxxxxx is the 6-digit number for a trait/disorder, and yyyy is the NCBI species taxonomy id.

Summary

	dog	cattle	cat	pig	sheep	horse	chicken	goat	rabbit	Japanese quail	golden hamster	Other	TOTAL
Total traits/disorders	586	399	304	221	216	208	206	73	58	42	40	467	2820
Mendelian trait/disorder	221	146	77	49	88	40	125	13	28	32	28	151	998
Mendelian trait/disorder; key mutation known	155	80	42	22	34	29	36	9	7	9	3	60	486
Potential models for human disease	299	143	165	78	82	110	42	29	36	11	14	226	1235

RECENT NEWS

Key locomotion mutation identified:
On 29 August, in a paper in Nature, Andersson et al. (2012) reported a nonsense mutation in DMRT3, which encodes a transcription factor, that plays a major role in determining mode of locomotion. For more information, and access to a copy of the paper, see Gaitedness.

Two iconic Mendelian traits resolved in a week! :
One hundred and ten years after they were first described as Mendelian (single-locus) traits, the last two of the six originally-described Mendelian traits have been resolved (or partly so) at the molecular level within a week of each other!

SEARCHING OMIA

Simple Search

Simple searching is available here and also near the top right of every OMIA page. Fields included in the simple search are trait name, species common name, species scientific name and gene symbol. Multiple search terms can be combined with "OR" or "AND". A search with no search term will return all records in the database.

Enter search terms: [ ] [combine search terms with OR] Return a list of [phenes] SEARCH

Exhaustive Search

Fields included in the exhaustive search are trait name, trait species-specific name, trait summary, trait symbol, species common name, species scientific name, gene symbol, gene description, marker, clinical features, genetic testing, inheritance details, molecular genetics, genetic mapping, history, control, pathology, prevalence, article title, article publisher, article author, breed name. Multiple search terms can be combined with "OR" or "AND".

Enter search terms: [ ] [combine search terms with OR] Return a list of [phenes] SEARCH

Advanced Search

The advanced search function can be used to refine your search, or to search with keywords, author names, disease categories, or data within other fields. The advanced search also allows multiple search terms to be combined with AND logic. You may search specific text fields with key words or phrases. You may also enter just a portion of the key word, for a broader search. Wild-card symbols are not required.

Searching for an author name will retrieve all of the traits or diseases that have been linked to papers by that author.

The OMIA database has also classified some diseases or traits into categories. These categories can be searched for using the "category" option below.

Trait name:	[ ]	*Type in a term that is part of a trait name; e.g. "myopathy"*
Trait id (OMIA id):	[ ]	*Type in one or more comma-separated numbers each of which is a trait record identifer (OMIA id); e.g. "001081" or "001081,001199"*
Species-specific trait name:	[ ]	*Type in a term that is part of a species-specific trait name; e.g. "Alport syndrome"*
Species-specific trait summary:	[ ]	*Type in a term that is part of a species-specific trait summary; e.g. "causative mutation"*
Species-specific trait symbol:	[ ]	*Type in a term that is part of a species-specific trait symbol; e.g. "HFMD"*
Species NCBI taxonomy id:	[ ]	*Type in one or more comma-separated numbers each of which is a record identifer in the NCBI taxonomy database; e.g. "9913" or "9913,9615"*

Species scientific name:

Species common name:

Marker:

Clinical features:

Genetic testing:

Mode of inheritance: ---

Inheritance details:

Molecular genetics:

Genetic mapping:

History:

Control:

Pathology:

Prevalence:

Model of human disease: ---

MIM id:

Considered a defect: ---

Mendelian trait/disorder: ---

Key mutation known: ---

Gene id:

Gene symbol:

Gene synonym: ☑

Gene description:

Breeds:

Article pubmed id:

Article author:

Article title:

Article keyword:

Category: ---

Created on or before:

Created on or after:

Last modified on or before:

Last modified on or after:

Inborn error of metabolism
Dwarfism
Congenital heart disease
Inherited bleeding disorder
Lysosomal storage disease
Colour
Progressive retinal atrophy (PRA)
Cone-rod dystrophy (CRD)
Stationary retinal disorder
Developmental retinal disorder
Retinal disorder
Disorder of Sexual Development (DSD)

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phene is to gene
as
phenotype is to genotype

McKusick??

22 phene records found [show instead gene records]

- OMIA 001089-9825 **Blood group system ABO** in *Sus scrofa domesticus* (domestic pig) Gene: GGTA1
- OMIA 001249-9825 **Coat colour, brown** in *Sus scrofa domesticus* (domestic pig) Gene: TYRP1
- OMIA 000209-9825 **Coat colour, dominant white** in *Sus scrofa domesticus* (domestic pig) Gene: KIT
- OMIA 001199-9825 **Coat colour, extension** in *Sus scrofa domesticus* (domestic pig) Gene: MC1R
- OMIA 001743-9825 **Coat colour, patch** in *Sus scrofa domesticus* (domestic pig) Gene: KIT
- OMIA 001216-9825 **Coat colour, roan** in *Sus scrofa domesticus* (domestic pig) Gene: KIT
- OMIA 001745-9825 **Coat colour, white belt, due to KIT** in *Sus scrofa domesticus* (domestic pig) Gene: KIT
- OMIA 000259-9825 **Deafness** in *Sus scrofa domesticus* (domestic pig) Gene: MITF
- OMIA 001718-9825 **Dwarfism, Schmid metaphyseal chondrodysplasia** in *Sus scrofa domesticus* (domestic pig) Gene: COL10A1
- OMIA 001579-9825 **Ear size** in *Sus scrofa domesticus* (domestic pig) Gene: PPARD
- OMIA 000499-9825 **Hypercholesterolaemia** in *Sus scrofa domesticus* (domestic pig) Gene: LDLR
- OMIA 000621-9825 **Malignant hyperthermia** in *Sus scrofa domesticus* (domestic pig) Gene: RYR1
- OMIA 001085-9825 **Meat quality (Rendement Napole)** in *Sus scrofa domesticus* (domestic pig) Gene: PRKAG3

OMIA 000621-9825 : Malignant hyperthermia in *Sus scrofa domesticus*

See the equivalent entry at NCBI

In other species: turkey , dog , domestic cat , horse , deer , cattle , rabbit

Possible human homologue (MIM number): 145600

Mendelian trait/disorder: yes

Mode of inheritance: Autosomal Recessive

Considered a defect: yes

Key mutation known: yes

Year key mutation first reported: 1991

Cross-species summary: A progressive increase in body temperature, muscle rigidity and metabolic acidosis, leading to rapid death.

Species-specific name: Porcine Stress Syndrome

Species-specific symbol: PSS

Species-specific description: In pigs, malignant hyperthermia (MH) leads to rapid post-mortem changes in muscle, resulting in pale soft exudative (PSE) meat. MH can be triggered by a minor stress, such as loading, transport, sexual intercourse, high ambient temperature, or exposure to the anaesthetic halothane. Susceptibility to halothane-induced MH is an autosomal recessive trait in pigs. Together, sudden death syndrome and PSE constitute porcine stress syndrome (PSS), which became a major economic problem in many countries in the 1970s, as indicated by the number of references in the list below. In part, the increasing problem of PSS was due to strong selection for increased leanness, which is associated with susceptibility to PSS.

Molecular basis: The molecular basis of MH in pigs was discovered via identification of a strong candidate gene, namely RYR1, that encodes a calcium release channel of skeletal muscle sarcoplasmic reticulum. When it was shown that this candidate gene mapped very closely to MH in pigs and in humans, the race was on to clone and sequence the RYR1 gene. The race was won by a Canadian research team led by David MacLennan (Fujii et al., 1991) who showed that MH is due to a base substitution (C-T) in the 1843rd nucleotide of the RYR1 gene. The base substitution causes an amino-acid substitution (arginine - cysteine) in the 615th position of the calcium release channel, resulting in altered calcium flow. It is remarkable that the smallest possible change (a single base-substitution) leading to a single amino-acid-substitution in a very large molecule (comprising 5,035 amino acids) can have caused a disorder that was a major financial burden for the global pig industry for several decades.

Interestingly, Bates et al. (2012) reported that "A proportion of pigs normal for RYR1 did exhibit limb rigidity during halothane gas challenge, and subsequently tended to have lower 45 min pH and greater longissimus muscle fluid loss post harvest." This suggests that the RYR1 locus is not the only factor determining reaction to halothane.

Genetic testing: Various PCR genotyping tests have been devised, all based on detection of an RFLP resulting from the causative base substitution. Over a roughly ten-year period, these tests enabled the harmful allele to be removed from most pig populations throughout the world.

Associated gene:

Symbol	Description	Species	Chr acc	Chr name	Start	Stop	OMIA gene details page	Other Links
RYR1	ryanodine receptor 1 (skeletal)	*Sus scrofa*	NC_010448.3	6	42840239	42960105	RYR1	Homologene, Ensembl, NCBI gene

Gene RYR1 : ryanodine receptor 1 (skeletal) in *Sus scrofa*

See the equivalent entry at NCBI

In other species: dog, horse

Symbol: RYR1

Synonyms: CRC, RYR

Description: ryanodine receptor 1 (skeletal)

Type of gene: protein-coding

NCBI gene id: 396718

Other designations: RYR-1|calcium release channel|halothane|porcine stress syndrome|ryanodine receptor 1|skeletal muscle calcium release channel|skeletal muscle ryanodine receptor|skeletal muscle-type ryanodine receptor|type 1 ryanodine receptor

Links: Homologene, Ensembl

Genomic location: 6:42840239..42960105 [Chromosome accession NC_010448.3]

Related phenes:

OMIA 000621-9825 : Malignant hyperthermia in *Sus scrofa domesticus*

References

Note: the references are listed in reverse chronological order (from the most recent year to the earliest year), and alphabetically by first author within a year.

2002 Martins-Wess, F., Voss-Nemitz, R., Drogemuller, C., Brenig, B., Leeb, T. :
Construction of a 1.2-Mb BAC/PAC contig of the porcine gene RYR1 region on SSC 6q1.2 and comparative analysis with HSA 19q13.13 *Genomics* 80:416-422, 2002. Pubmed reference: 12376096.

2000 Giese, A., Deppe, A., Brenig, B., Leeb, T. :
Genomic structure of the 5 ' end of the porcine ryanodine receptor 3 gene (RYR3) *DNA Sequence* 11:175-179, 2000. Pubmed reference: 10902927.

1999 Leeb, T., Giese, A., Pfeiffer, I., Brenig, B. :
Two highly polymorphic microsatellites within the porcine ryanodine receptor 3 gene (RYR3) *Animal Genetics* 30:321-322, 1999. Pubmed reference: 10467714.

1998 Leeb, T., Giese, A., Al-Bayati, H., Rettenberger, G., Brenig, B. :
Assignment of the porcine ryanodine receptor 3 gene (RYR3) to chromosome 7q22-->q23. *Cytogenet Cell Genet* 83:244-5, 1998. Pubmed reference: 10072592. DOI: 15193.

1996 Brenig, B., Leeb, T. :
Identification of a G/C transversion polymorphism in intron 38 of the porcine skeletal muscle ryanodine receptor gene *Animal Genetics* 27:128, 1996. Pubmed reference: 8856914.

References OMIA 000621-9825 : Malignant hyperthermia in *Sus scrofa domesticus*

Note: the references are listed in reverse chronological order (from the most recent year to the earliest year), and alphabetically by first author within a year.

Around 860 in total, back to 1964

2012 Bates, R.O., Doumit, M.E., Raney, N.E., Helman, E.E., Ernst, C.W. :
Association of halothane sensitivity with growth and meat quality in pigs. *Animal* 6:1537-42, 2012. Pubmed reference: 23031527. DOI: 10.1017/S1751731112000134.

Pastoret, S., Ameels, H., Bossiroy, F., Decreux, A., De Longueville, F., Thomas, A., Desmecht, D. :
Detection of disease resistance and susceptibility alleles in pigs using oligonucleotide microarray hybridization. *J Vet Diagn Invest* 24:479-88, 2012. Pubmed reference: 22529114. DOI: 10.1177/1040638712442878.

Schütte, J.K., Schäfer, U., Becker, S., Oldewurtel, C., Starosse, A., Singler, P., Richard, A., Wappler, F., Gerbershagen, M.U. :
3,4-Methylenedioxymethamphetamine induces a hyperthermic and hypermetabolic crisis in pigs with and without a genetic disposition for malignant hyperthermia. *Eur J Anaesthesiol* :, 2012. Pubmed reference: 23138574. DOI: 10.1097/EJA.0b013e32835a1127.

Vandenhaute, E., Culot, M., Gosselet, F., Dehouck, L., Godfraind, C., Franck, M., Plouët, J., Cecchelli, R., Dehouck, M.P., Ruchoux, M.M. :
Brain pericytes from stress-susceptible pigs increase blood-brain barrier permeability in vitro. *Fluids Barriers CNS* 9:11, 2012. Pubmed reference: 22569151. DOI: 10.1186/2045-8118-9-11.

Weschenfelder, A.V., Torrey, S., Devillers, N., Crowe, T., Bassols, A., Saco, Y., Piñeiro, M., Saucier, L., Faucitano, L. :
Effects of trailer design on animal welfare parameters and carcass and meat quality of three Pietrain crosses being transported over a long distance. *J Anim Sci* 90:3220-31, 2012. Pubmed reference: 22966081. DOI: 10.2527/jas.2012-4676.

2011 Cherel, P., Pires, J., Glenisson, J., Milan, D., Iannuccelli, N., Herault, F., Damon, M., Le Roy, P. :
Joint analysis of quantitative trait loci and major-effect causative mutations affecting meat quality and carcass composition traits in pigs. *BMC Genet* 12:76, 2011. Pubmed reference: 21875434. DOI: 10.1186/1471-2156-12-76.

Fiege, M., Weisshorn, R., Kolodzie, K., Wappler, F., Gerbershagen, M.U. :
Effects of theophylline on anesthetized malignant hyperthermia-susceptible pigs. *J Biomed Biotechnol* 2011:937479, 2011. Pubmed reference: 22131820. DOI: 10.1155/2011/937479.

Krischek, C., Natter, R., Wigger, R., Wicke, M. :
Adenine nucleotide concentrations and glycolytic enzyme activities in longissimus muscle samples of different pig genotypes collected before and after slaughter. *Meat Sci* 89:217-20, 2011. Pubmed reference: 21592677. DOI: 10.1016/j.meatsci.2011.04.022.

Metterlein, T., Schuster, F., Kranke, P., Hager, M., Roewer, N., Anetseder, M. :
Magnesium does not influence the clinical course of succinylcholine-induced malignant hyperthermia. *Anesth Analg* 112:1174-8, 2011. Pubmed reference: 21474662. DOI: 10.1213/ANE.0b013e31821263d6.

Overall total number of references in OMIA: 20,660

most hyperlinked to PubMed

many have a link to actual paper via doi

OMIA 001718-9825 : Dwarfism, Schmid metaphyseal chondrodysplasia in *Sus scrofa domesticus*

See the equivalent entry at NCBI

Possible human homologue (MIM number): 156500

Mendelian trait/disorder: yes

Mode of inheritance: Autosomal Dominant

Considered a defect: yes

Key mutation known: yes

Year key mutation first reported: 2000

Species-specific description: In a single paper, Nielsen et al. (2000) reported a new form of dwarfism in pigs, and its causative mutation.

Inheritance: Nielsen et al. (2000) reported autosomal dominant inheritance.

Mapping: An initial genome scan with 70 microsatellite markers implicated chromosome SSC1. Mapping with additional SSC1 markers mapped the disorder to 8.3cM from marker Sw781. The authors noted that this region is homologous to human chromosome HSA6q21-22.3, which harbours the gene COL10A1, mutations in which cause Schmid metaphyseal chondrodysplasia, a disorder very similar to the pig disorder. Thus the authors had identified a comparative positional candidate gene.

Molecular basis: Following a comparative positional candidate gene approach (described above in the Mapping section), Nielsen et al. (2000) cloned and sequenced the porcine COL10A1 gene and identified a causative missense mutation, namely "a single G to A transition in exon 3 that results in a Gly-to-Arg substitution, G590R, in the carboxyl terminus of the protein".

Clinical features: The disorder is characterised by "Metaphyseal chondrodysplasia in the long bones" (Nielsen et al., 2000).

Breed: Yorkshire.

Associated gene:

Symbol	Description	Species	Chr acc	Chr name	Start	Stop	OMIA gene details page	Other Links
COL10A1	collagen, type X, alpha 1	*Sus scrofa*	NC_010443.4	1	91889387	91882389	COL10A1	Homologene, Ensembl, NCBI gene

Reference

2000 Nielsen, V.H., Bendixen, C., Arnbjerg, J., Sørensen, C.M., Jensen, H.E., Shukri, N.M., Thomsen, B. :
Abnormal growth plate function in pigs carrying a dominant mutation in type X collagen. *Mamm Genome* 11:1087-92, 2000. Pubmed reference: 11130976. DOI: 10.1007/s003350010212

OMIA 000483-9913 : Horns/Polled in *Bos taurus*

See the equivalent entry at NCBI

In other species: goat , sheep , water buffalo , bighorn sheep , kouprey

Possible human homologue (MIM number): 110100

Mendelian trait/disorder: yes

Mode of inheritance: Autosomal

Considered a defect: no

Key mutation known: yes

Year key mutation first reported: 2012

Cross-species summary: There is substantial variation in the extent of horn growth, making classification difficult. However, in general, the presence or absence of horns can be attributed to the action of two alleles at an autosomal locus, with the polled condition being dominant to horned.

Species-specific description: The absence of horns (polledness) is of substantial benefit in cattle, from an economic and welfare point of view: bruising due to horns is eliminated, and the stress associated with de-horning is avoided. (Information complied by Ulrika Tjälldén and Vanja Kinch, Uppsala, March 1998)

History: In cattle, one of the first Mendelian traits to attract attention was the presence/absence of horns. The inherited nature of this trait was well recognised (but not understood) long before the rediscovery of Mendelism (see, e.g., Darwin 1859 [p. 14]; Darwin 1868 [vol ii, p. 316]). In 1902, polledness was one of the first six animal traits to be shown to have Mendelian inheritance (Bateson and Saunders, 1902). In 1906, the American agricultural polymath W.J. Spillman (who is not only regarded as a founding father of agricultural economics, but also independently rediscovered Mendelism while crossing strains of wheat!) published a paper in Science (Spillman 1906a) and another in the newly-founded Journal of Heredity (Spillman 1906b), providing convincing evidence that the presence/absence of horns is a Mendelian trait, with polled being dominant to horned. This trait soon became a classic Mendelian trait, cited in many textbooks. Indeed, as delightfully recorded by Crow (1992), this trait even attracted the attention of the Nobel-prize winning physicist Erwin Schrödinger, who wrote two letters to J.B.S Haldane in 1945, in relation to "the hornless cattle problem". In these letters, Schrödinger derived an equation that predicts the frequency of horned offspring in a closed herd after any number of generations of complete selection against horned bulls, but with no selection on cows.

Nothing much was added to our knowledge of this trait until the first wave of genomics tools provided sufficient microsatellite markers to enable Georges et al. (1993) to map the presence/absence of horns to within a recombination fraction of 13% with two markers on chromosome BTA1 (see Mapping section). To present readers, such "loose" linkage might seem to be not worthy of much celebration. At the time, however, this result was sufficiently important and novel to warrant publication in Nature Genetics. Subsequent progress in mapping is summarised in the Mapping section. (Most of the wording under this heading is from Nicholas, F.W. (2012; Mendelian Inheritance in Cattle, chap 2 [pp. 11-19] in Bovine Genomics [ed. J. Womack], Wiley-Blackwell, Ames, Iowa.)

Medugorac, I., Seichter, D., Graf, A., Russ, I., Blum, H., Göpel, K.H., Rothammer, S., Förster, M., Krebs, S. :
Bovine polledness - an autosomal dominant trait with allelic heterogeneity. *PLoS One* 7:e39477, 2012. Pubmed reference: 22737241. DOI: 10.1371/journal.pone.0039477.

Associated gene:

Symbol	Description	Species	Chr acc	Chr name	Start	Stop	OMIA gene details page	Other Links
POLLED		*Bos taurus*	no genomic information	-	-	-	POLLED	Ensembl

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OMIA 000151-9913 : Brachyspina in *Bos taurus*

See the equivalent entry at NCBI

Mendelian trait/disorder: yes

Mode of inheritance: Autosomal Recessive

Considered a defect: yes

Key mutation known: yes

Year key mutation first reported: 2012

PAG XXI 2013 !!

Molecular basis: Charlier et al. (2012) reported the causal mutation for brachyspina in Holstein cattle as a deletion in the FANCI gene. Noting that the carrier frequency is far too high (up to 7.4%) to be consistent with a relatively rare autosomal recessive disorder, Charlier et al. (2012) also showed that a large proportion of affected calves die in utero. Thus this causal mutation also contributes to natural abortions and hence to reduced fertility.

Breed: Holstein.

Associated gene:

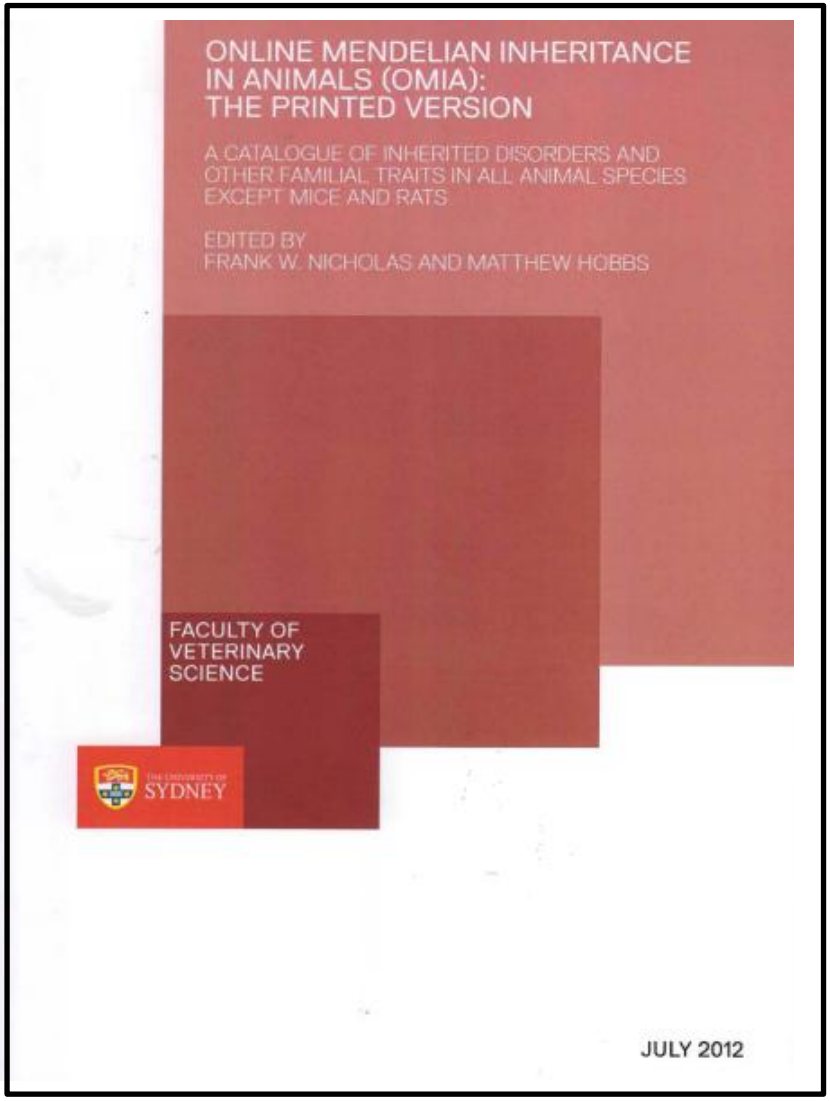
Symbol	Description	Species	Chr acc	Chr name	Start	Stop	OMIA gene details page	Other Links
FANCI	Fanconi anemia, complementation group I	*Bos taurus*	AC_000178.1	21	21137917	21198617	FANCI	Homologene, Ensembl, NCBI gene

References

Note: the references are listed in reverse chronological order (from the most recent year to the earliest year), and alphabetically by first author within a year.

2012 Charlier, C., Agerholm, J.S., Coppieters, W., Karlskov-Mortensen, P., Li, W., de Jong, G., Fasquelle, C., Karim, L., Cirera, S., Cambisano, N., Ahariz, N., Mullaart, E., Georges, M., Fredholm, M. :
A Deletion in the Bovine FANCI Gene Compromises Fertility by Causing Fetal Death and Brachyspina. *PLoS One* 7(8):e43085, 2012. Pubmed reference: 22952632. DOI: 10.1371/journal.pone.0043085.

2013 Akiyama, K., Hirano, T., Masoudi, A.A., Uchida, K., Tsuji, T., Kumagai, T., Onwada, K., Sugimoto, Y., Kunieda, T. :
A Mutation of the GFRF1 Gene is Responsible for Forelimb-Girdle Muscular Anomaly (FMA) of Japanese Black Cattle *Plant and Animal Genome XXI* Abstract P0555:, 2013.



Breeds: Mongrel
Associated Gene: RPGR: retinitis pigmentosa GTPase regulator [chromosome X:38105005-38156370]

Beltran, W.A., Cideciyan, A.V., Lewis, A.S., Iwabe, S., Khanna, H., Sumaroka, A., Chiodo, V.A., Fajardo, D.S., Roman, A.J., Deng, W.T., Swider, M., Alemán, T.S., Boye, S.L., Genini, S., Swaroop, A., Hauswirth, W.W., Jacobson, S.G. and Aguirre, G.D. : Gene therapy rescues photoreceptor blindness in dogs and paves the way for treating human X-linked retinitis pigmentosa. *Proc Natl Acad Sci U S A* 109:2132-7, 2012.

Miyadera, K., Acland, G.M. and Aguirre, G.D. : Genetic and phenotypic variations of inherited retinal diseases in dogs: the power of within- and across-breed studies. *Mamm Genome* :, 2011.

Genini, S., Zangerl, B., Slavik, J., Acland, GM., Beltran, WA. and Aguirre, GD. : Transcriptional Profile Analysis of RPGRORF15 Frameshift Mutation Identifies Novel Genes Associated with Retinal Degeneration. *Invest Ophthalmol Vis Sci* 51:6180-90, 2010.

Zangerl, B., Johnson, J.L., Acland, G.M. and Aguirre, G.D. : Independent origin and restricted distribution of RPGR deletions causing XLPRA. *J Hered* 98:526-30, 2007.

Beltran, WA., Hammond, P., Acland, GM. and Aguirre, GD. : A frameshift mutation in RPGR exon ORF15 causes photoreceptor degeneration and inner retina remodeling in a model of X-linked retinitis pigmentosa. *Invest Ophthalmol Vis Sci* 47:1602-11, 2006.

Zhang, Q., Acland, GM., Wu, WX., Johnson, JL., Pearce-Kelling, S., Tulloch, B., Vervoort, R., Wright, AF. and Aguirre, GD. : Different RPGR exon ORF15 mutations in Canids provide insights into photoreceptor cell degeneration. *Hum Mol Genet* 11:993-1003, 2002.

Xanthinuria
MIM: 278300

Canis lupus familiaris [dog]
Single locus: yes
Mode of inheritance: Autosomal Recessive
Considered a defect: no
Causative mutation known: no

Flegel, T., Freistadt, R. and Haider, W. : Xanthine urolithiasis in a dachshund. *Veterinary Record* 143:620-623, 1998.

Kucera, J., Balkova, T., Pychla, R. and Jahn, P. : Bilateral xanthine nephrolithiasis in a dog. *Journal of Small Animal Practice* 38:302-305, 1997.

Vanmuilen, C.D., Nickel, R.F., Vandijk, T.H. and Reijngoud, D.J. : Xanthinuria in a family of Cavalier King Charles spaniels. *Veterinary Quarterly* 19:172-174, 1997.

Vanmuilen, C.D., Nickel, R.F. and Reijngoud, D.J. : Xanthinuria (xanthine oxidase deficiency) in two Cavalier King Charles spaniels. *Veterinary Quarterly* 18:5 24-5 25, 1996.

Felis catus [domestic cat]
Single locus: unknown
Considered a defect: yes
Causative mutation known: no

Tachida, S., Kagi, A., Koyama, H. and Tagawa, M. : Xanthine urolithiasis in a cat: a case report and evaluation of a candidate gene for xanthine dehydrogenase. *J Feline Med Surg* 9:503-8, 2007.

White, R.N., Tick, N.T. and White, H.L. : Naturally occurring xanthine urolithiasis in a domestic shorthair cat. *Journal of Small Animal Practice* 38:299-302, 1997.

Yellow fat

Bos taurus [cattle]
Single locus: yes
Considered a defect: no
Causative mutation known: yes
Year causative mutation first reported: 2009
Associated Gene: BCO2: beta-carotene oxygenase 2 [chromosome 15:22808239-22885964]

Berry, SD., Davis, SR., Beattie, EM., Thomas, NL., Burnett, AK., Ward, HE., Stanfield, AM., Biswas, M., Ankersmit-Udy, AE., Oxley, PE., Barnett, JL., Pearson, JF., van der Linen, Y., Macgibbon, AH., Spelman, RJ., Lehnert, K. and Snell, RG. : Mutation in bovine beta-carotene oxygenase 2 affects milk color. *Genetics* 182:923-6, 2009.

Equus caballus [horse]
Single locus: unknown
Considered a defect: yes
Causative mutation known: no

Suárez-Bonnet, A., Espinosa de los Monteros, A., Herráez, P., Rodríguez, F., Andrada, M. and Caballero, M.J. : Fat embolism secondary to yellow fat disease in an Appaloosa horse. *J Vet Diagn Invest* 20:694-7, 2008.

Sus scrofa [pig]
Single locus: unknown
Considered a defect: yes
Causative mutation known: no

Danse, L.H. and Steenbergen-Bonenkamp, W.A. : Enzyme histochemical studies of adipose tissue in porcine yellow fat disease. *Vet Pathol* 11:465-76, 1974.

[Yellow fat disease in piglets and fattening pigs (author's transl)]. *Tijdschr Diergeneeskd* 98:1118-32, 1973.

DAVIS, C.L. and GORHAM, J.R. : The pathology of experimental and natural cases of yellow fat disease in swine. *Am J Vet Res* 15:55-0, 1954.

Oryctolagus cuniculus [rabbit]
Single locus: yes
Mode of inheritance: Autosomal Recessive
Considered a defect: no
Causative mutation known: no

Jones, D., Gresham, G.A., Lloyd, H.G. and Howard, A.N. : "Yellow fat" in the wild rabbit. *Nature* 207:205-6, 1965.

Castle, WE. : The Linkage Relations of Yellow Fat in Rabbits. *Proc Natl Acad Sci U S A* 19:947-50, 1933.

Ovis aries [sheep]
Single locus: yes
Mode of inheritance: Autosomal Recessive
Considered a defect: yes
Causative mutation known: yes
Year causative mutation first reported: 2010
Description: The presence of three xanthophylls (lutein, lutein-5-6-epoxide, and flavoxanthin) in sheep fat, presumably due to the inability to oxidise xanthophylls. Inherited as a single-locus autosomal recessive trait.
Associated Gene: BCO2: beta-carotene oxygenase 2

Vage, DI. and Boman, IA. : A nonsense mutation in the beta-carotene oxygenase 2 (BCO2) gene is tightly associated with accumulation of carotenoids in adipose tissue in sheep (Ovis aries). *BMC Genet* 11:10, 2010.

Baker, R.L., Steine, T., Vaboen, A.W. and Breimen, D. : The inheritance and incidence of yellow fat in Norwegian sheep. *Acta Agriculturae Scandinavica* 35:389-397, 1985.

Geddie-Dahl, T.W. : [Reduction of the yellow fat incidence by different breeding plans for sheep]. *Nord Vet Med* 24:620-30, 1972.

Hill, E. : Xanthophyll pigmentation in sheep fat. *Nature* 194:865-6, 1962.

Castle, W.E. : Yellow fat in sheep. *Journal of Heredity* 25:246-247, 1934.

Yellow-vernen syndrome

Meleagris gallopavo [turkey]
Single locus: unknown
Considered a defect: yes
Causative mutation known: no

Thurston, R.J. and Korn, N. : Semen quality in the domestic turkey - the yellow-vernen-syndrome. *Poultry & Avian Biology Reviews* 8:109-121, 1997.

~750,000 words =
661 A4 pages of 8-point double-column text

OMIA home | Browse | Search | Landmarks, Reviews, Maps | **Download** | Curate | Contact | Ci

You are here: OMIA / Download

DOWNLOAD DATA

To download a current MySQL dump of OMIA please click on the appropriate link below.

	Zip	gzip
sql	omia.sql.zip	omia.sql.gz
xml	omia.xml.zip	omia.xml.gz

A table of phenes for which there is a causal mutation in a known gene is available here.

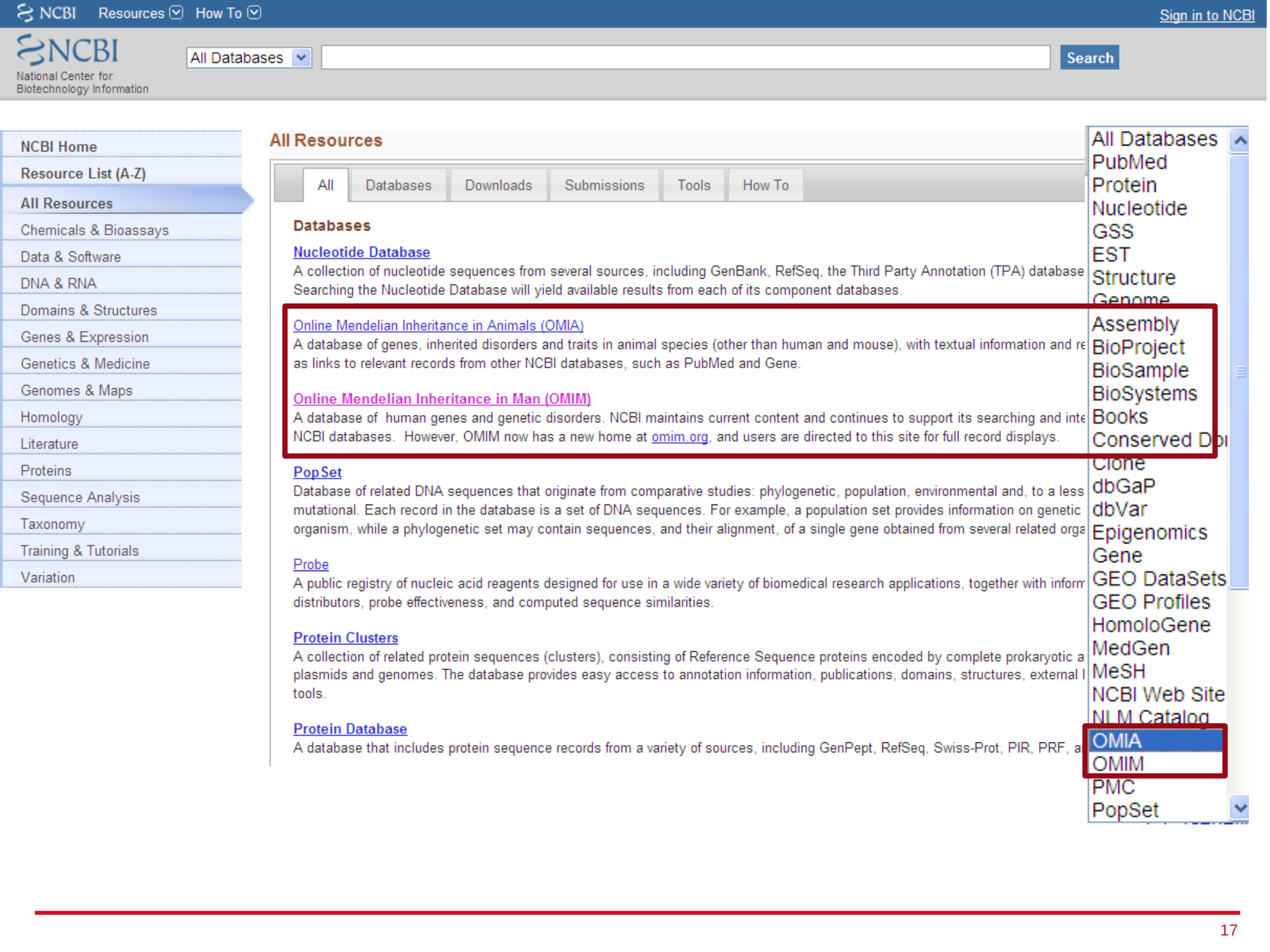
There is also a print version of the OMIA database (with all records up to 2nd July 2012) available here.

OMIA: a brief history

1974	Lecturer in animal genetics → many queries → many trips to library
	Became aware of Mendelian Inheritance in Man (MIM) flat file on mainframe at Johns Hopkins print editions (printouts): 1971 3rd edn
1978	Accosted Victor McKusick at Genetics Congress in Moscow animal equivalent of MIM? Sure, go ahead
	Key features: >1 species → extra dimension → strong comparative emphasis modelled on MIM; hence Mendelian Inheritance in Animals (MIA)
1980	Small grant → create MIA relational database on mainframe cf MIM flatfile
1980s	Gradual entry of backlog and manual servicing of queries

OMIA: a brief history

1991	Birth of WorldWideWeb
1995	both MIM and MIA launched on the web → Online MIM (OMIM) and Online MIA (OMIA) both using NCBI's birx search engine
	OMIA: regularly-updated flat file from database on laptop published via Australian National Genomic Information Service (ANGIS) http://omia.angis.org.au
1997	Reciprocal hyperlinks between OMIM and OMIA highlight animal models of human disorders
2005	OMIA transferred to MySQL database on server Interactive web page !! **Instant updating by curators anywhere in the world!!**
2005	NCBI asked for an OMIA mirror integrated in their Entrez system weekly dump → OMIA alongside OMIM !!


NCBI Resources How To
Sign in to NCBI
NCBI
National Center for Biotechnology Information
All Databases
Search
NCBI Home
Resource List (A-Z)
All Resources
Chemicals & Bioassays
Data & Software
DNA & RNA
Domains & Structures
Genes & Expression
Genetics & Medicine
Genomes & Maps
Homology
Literature
Proteins
Sequence Analysis
Taxonomy
Training & Tutorials
Variation
All Resources
All
Databases
Downloads
Submissions
Tools
How To
Databases
Nucleotide Database
A collection of nucleotide sequences from several sources, including GenBank, RefSeq, the Third Party Annotation (TPA) database
Searching the Nucleotide Database will yield available results from each of its component databases.
Online Mendelian Inheritance in Animals (OMIA)
A database of genes, inherited disorders and traits in animal species (other than human and mouse), with textual information and re
as links to relevant records from other NCBI databases, such as PubMed and Gene.
Online Mendelian Inheritance in Man (OMIM)
A database of human genes and genetic disorders. NCBI maintains current content and continues to support its searching and inte
NCBI databases. However, OMIM now has a new home at omim.org, and users are directed to this site for full record displays.
PopSet
Database of related DNA sequences that originate from comparative studies: phylogenetic, population, environmental and, to a less
mutational. Each record in the database is a set of DNA sequences. For example, a population set provides information on genetic
organism, while a phylogenetic set may contain sequences, and their alignment, of a single gene obtained from several related orga
Probe
A public registry of nucleic acid reagents designed for use in a wide variety of biomedical research applications, together with inform
distributors, probe effectiveness, and computed sequence similarities.
Protein Clusters
A collection of related protein sequences (clusters), consisting of Reference Sequence proteins encoded by complete prokaryotic a
plasmids and genomes. The database provides easy access to annotation information, publications, domains, structures, external l
tools.
Protein Database
A database that includes protein sequence records from a variety of sources, including GenPept, RefSeq, Swiss-Prot, PIR, PRF, a
All Databases
PubMed
Protein
Nucleotide
GSS
EST
Structure
Genome
Assembly
BioProject
BioSample
BioSystems
Books
Conserved Do
Clone
dbGaP
dbVar
Epigenomics
Gene
GEO DataSets
GEO Profiles
HomoloGene
MedGen
MeSH
NCBI Web Site
NLM Catalog
OMIA
OMIM
PMC
PopSet

OMIA: a brief history

2011	Revised web site with improved curation tools based on a django framework (Matthew Hobbs)
	Vicki Meyers-Wallen (Cornell): dogs and cats
2012	Zhiliang Hu: reciprocal links with AnimalGenome.org
	Fiona Cunningham: reciprocal links with Ensembl (thanks to Dave Burt)
	Thomas Peterson & Maricel Kann, University of Maryland, Baltimore County systematic catalogue of all ORF causal mutations in HGVS notation protein domain hotspots of disease mutations

Pig Genome Assembly 10.2

Showing 119.9 kbp from Chr.6, positions 42,840,240 to 42,960,106

Instructions

Click to start on a chromosome: Chr.1, Chr.2, Chr.3, Chr.4, Chr.5, Chr.6, Chr.7, Chr.8, Chr.9, Chr.10, Chr.11, Chr.12, Chr.13, Chr.14, Chr.15, Chr.16, Chr.17, Chr.18, Chr.X; Or search your region of interests using a query string formatted like: Chr.1:1000000-2000000

Navigation: Click one of the rulers to center on a location, or cl and dra region. Use the Scroll/Zoom buttons to ge magnification and position.

[Bookmark this] [Upload your own data] [Hide banner] [Share thes racks] [Hi-res Image] [Help] Reset

Search

Landmark or Region:

ryr1 Search

Data Source

Pig Genome Assembly 10.2

Overview

Chr.6

0M 10M 20M 30M 40M 120M 130M 140M 150M

Details

42850k 42930k 42940k 42950k 42960k

NCBI Genes

RYR1

OMIA Genes

"RYR1"

OMIA Gene

Name:	RYR1
OMIA ID:	000621
Symbol:	RYR1
Phene:	Malignant hyperthermia
Gene ID:	396718
Map Location:	Chr.6:42840239-42960105
Description:	RYR-1|calcium release channel|halothane|porcine stress syndrome|ryanodine receptor 1|skeletal muscle calcium release channel|skeletal muscle ryanodine receptor|skeletal muscle-type ryanodine receptor|type 1 ryanodine receptor

Flip

Zhiliang's handywork!!!

Cattle Genome UMD3.1 Track -- QTL, SNPs, Genes, etc.

Showing 60.7 kbp from Chr.21, positions 21,137,917 to 21,198,617

Instructions

Click to start on a chromosome: Chr.1, Chr.2, Chr.3, Chr.4, Chr.5, Chr.6, Chr.7, Chr.8, Chr.9, Chr.10, Chr.11, Chr.12, Chr.13, Chr.14, Chr.15, Chr.16, Chr.17, Chr.18, Chr.19, Chr.20, Chr.21, Chr.22, Chr.23, Chr.24, Chr.25, Chr.26, Chr.27, Chr.28, Chr.29, Chr.X, Or search your region of interests using a query string for matted like: **Chr.1:1000000-2000000**

Once you get a chromosome view in the "Overview" window, highlight a region under 100MB in length for details. Highlight select in "Details" window to narrow your scope, and use "Scroll/Zoom" to fine tune your focus.

[Bookmark this] [Upload your own data] [Hide banner] [Share these tracks] [Link to Image] [High-res Image] [Help] [Reset]

Search

Landmark or Region:

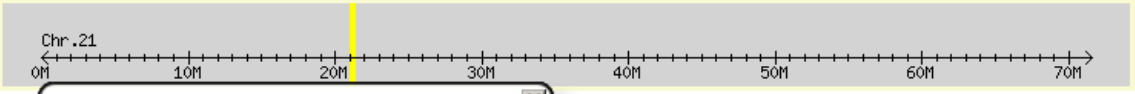
FANCI Search

Data Source

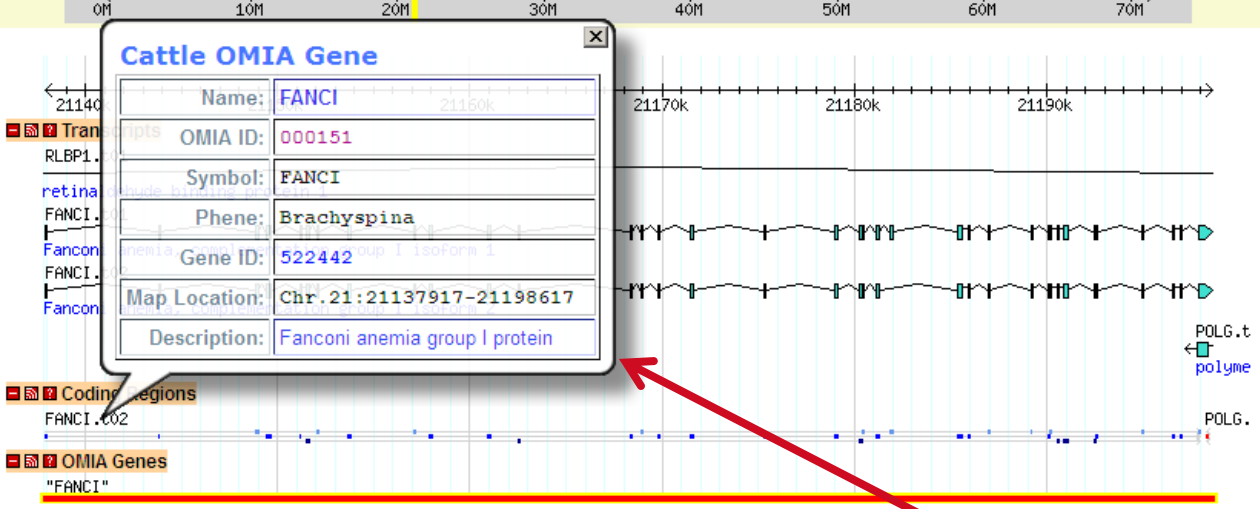
Cattle Genome Sequence (UMD Data)

Scroll/Zoom: Show 60.7 kbp Flip

Overview



Details



Clear highlighting

Update Image

Zhiliang's handywork!!!

Research and applications

Incorporating molecular and functional context into the analysis and prioritization of human variants associated with cancer

Thomas A Peterson,[1] Nathan L Nehrt,[1,2] DoHwan Park,[1] Maricel G Kann[1]

Nehrt *et al. BMC Genomics* 2012, **13**(Suppl 4):S9
http://www.biomedcentral.com/1471-2164/13/S4/S9

BMC Genomics

PROCEEDINGS Open Access

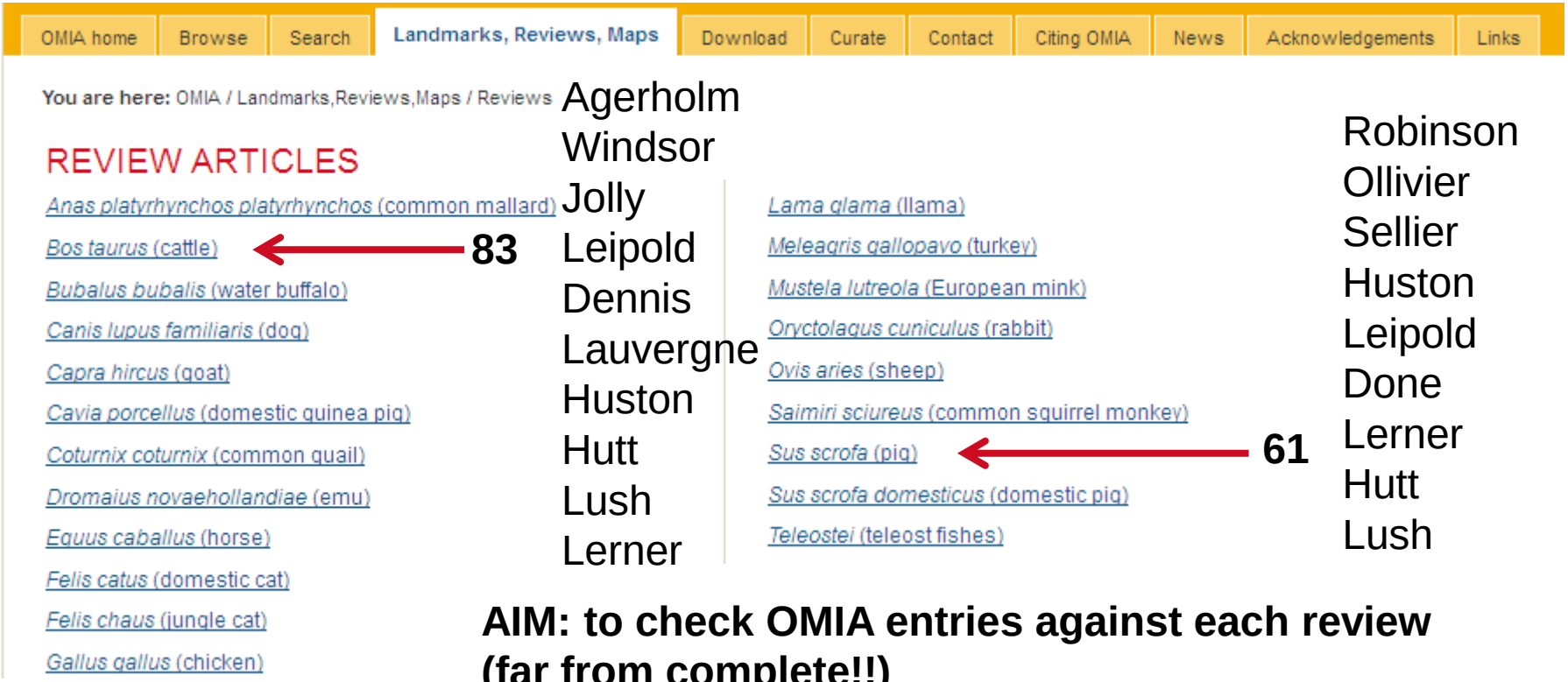
Domain landscapes of somatic mutations in cancer

Nathan L Nehrt[1,2†], Thomas A Peterson[1†], DoHwan Park[3], Maricel G Kann[1*]

From SNP-SIG 2011: Identification and annotation of SNPs in the context of structure, function and disease
Vienna, Austria. 15 July 2011

OMIA now

- Alternatives?
  - several dog web catalogues
  - *Mendelian Inheritance in Cattle*: http://dga.jouy.inra.fr/lgbc/mic2000/
    (COGNOSAG: Keith Huston, Paul Millar, JJ Lauvergne and S. Dolling)
  - many reviews



AIM: to check OMIA entries against each review (far from complete!!)

OMIA now

Obvious problem with “static” reviews:

- rapidly out of date
- no functional links to other information
- but still valuable, from time to time

Given that OMIA

1. exists (albeit in an incomplete state)
2. is freely available
3. is/can be kept up to date
4. is increasingly hyperlinked to other relevant databases

There is no point in anyone starting from scratch to collect information and references for a review

→ **OMIA: a one-stop global resource for animal genetics/genomics ??**

CURATION

Already have a team of volunteers

- Frank Nicholas
- Imke Tammen
- Mohammad Shariflou
- Bethany Wilson
- Matthew Hobbs
- Vicki Meyers-Wallen
- Martha MaloneyHuss
- Paul McGreevy
- Mark Haskins
- Tosso Leeb
- Hamutal Mazrier
- Marilyn Menotti-Raymond
- Peter Windsor
- Jerry Wei
- Carole Charlier
- Michel Georges
- Bianca Hasse
- Ben Dorshorst
- Mario Van Poucke
- Emily Piper
- Zhiliang Hu
- Zena Wolf

and a handbook

OMIA CURATION GUIDE
by Frank Nicholas
Version 120129

and a practice site:

http://sg-web-prd-1.ucc.usyd.edu.au/

and curation tools:

CURATION TOOLS

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OMIA ADMINISTRATION

OMIA application		
Articles	Add	Change
Breeds	Add	Change
Cross-species phenes	Add	Change
Genes	Add	Change
HTML fragments	Add	Change
Inheritance types	Add	Change
Mutation types	Add	Change
Mutations	Add	Change
News items	Add	Change
Phene categories	Add	Change
Species	Add	Change
Species-specific phenes	Add	Change

Auto-fill:

- PubMed refs
- Species names
- Phene names
- Gene symbols
- Breed names

CURATION TOOLS

ADD SPECIES-SPECIFIC PHENE

Phene:  **Auto-complete**

Species: **Auto-complete**

Type in part of a species common name, or the beginning of a species scientific (binomial) name, or an NCBI taxonomy database species identifier, and make a selection from the suggested list.

Breeds: **Auto-complete**

Type in part of a breed name and make a selection from the suggested list. Multiple selections can be accrued. Clicking on the green [+] icon opens a separate window allowing the creation of a new breed record.

Species-specific phene name:

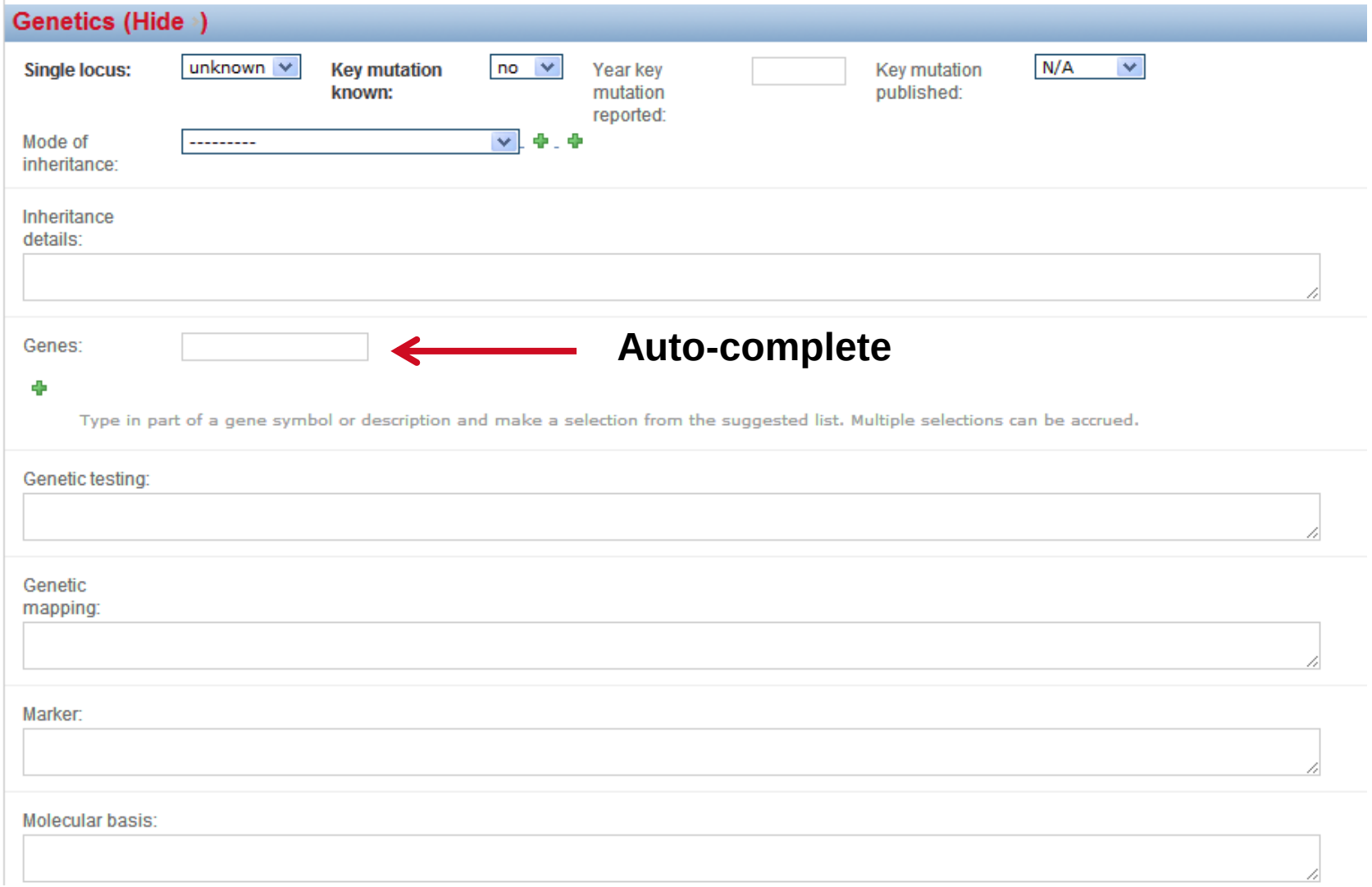
Symbol:

Summary:

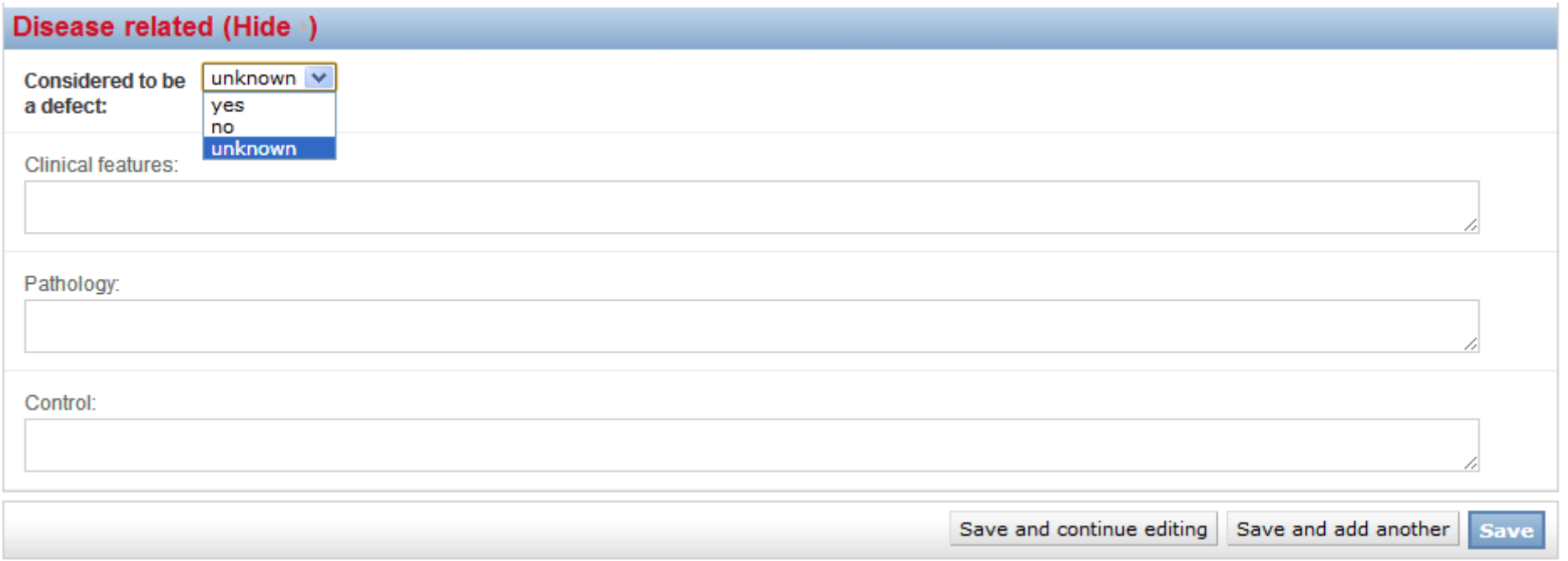
History:

Prevalence:

CURATION TOOLS



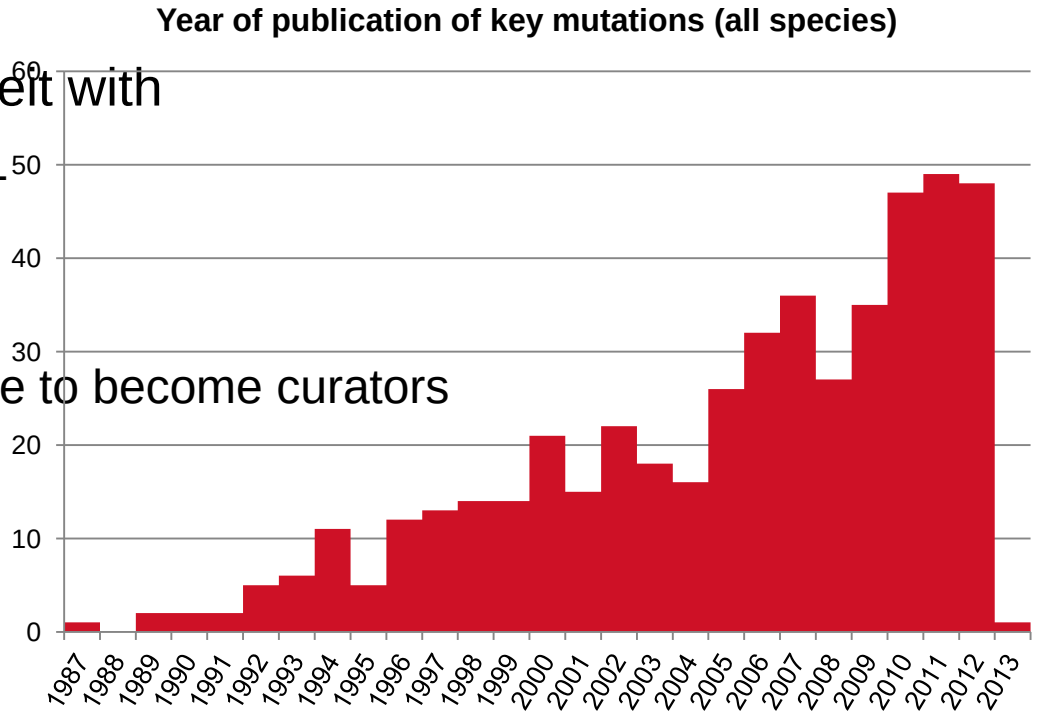
CURATION TOOLS



Most fields in most entries currently empty !!
More fields can be added in any section !!

CURATION

- Essentially OMIA has been a one-person operation til now
- Retired “early” at age 60, primarily to work on OMIA
- But can’t do it complete justice
- Now 65, and still fit and able, albeit with
  - one pre-cancer operation in 2011
  - contemporaries having strokes !!
- Serious need for Plan B
  - colleagues with motivation and time to become curators
    - species
    - traits (across species)



A flood of KOs in non-laboratory animals?

OMIA 000499-9825 : Hypercholesterolaemia in *Sus scrofa domesticus*

See the equivalent entry at NCBI

In other species: rabbit , dog

Possible human homologue (MIM number): 143890

Mendelian trait/disorder: yes

Mode of inheritance: Autosomal Recessive

Considered a defect: yes

Key mutation known: yes

Year key mutation first reported: 1998

Species-specific name: familial hypercholesterolemia, recessive

Species-specific symbol: FH-r

History: This trait was the first in non-laboratory animals to be investigated via the use of Transcription Activator-Like Effector Nucleases (TALENs) (Carlson et al., 2012) to create knockouts of the key gene (in this case, LDLR) (Carlson et al., 2012).

Molecular basis: A genome scan conducted by Haslerrapacz et al. (1998) showed that the gene for this disorder in pigs maps near to the centromere of chromosome 2, which is homologous to the region of human chromosome 19 containing the gene for low-density lipoprotein receptor (LDLR), a strong candidate for involvement in this disorder. Sequence analysis of the LDLR gene from homozygous normal and affected pigs showed that the disorder is due to a single missense mutation (resulting in the amino-acid substitution Arg84Cys). The causal mutation was thus identified via the comparative positional candidate gene approach.

In a proof-of-principle study, Carlson et al. (2012) used Transcription Activator-Like Effector Nucleases (TALENs) to create cloned pigs with a range of mutations in the porcine LDLR gene, namely 289_290ins34, 285_287delATG, 211_292del128, 289_290del10 and 289_290insA. The phenotypes of these mutant pigs were not reported in this paper.

Associated gene:

Symbol	Description	Species	Chr acc	Chr name	Start	Stop	OMIA gene details page	Other Links
LDLR	low density lipoprotein receptor	*Sus scrofa*	NC_010444.3	2	70206817	70193425	LDLR	Homologene, Ensembl, NCBI gene

References

Note: the references are listed in reverse chronological order (from the most recent year to the earliest year), and alphabetically by first author within a year.

2012 Carlson, D.F., Tan, W., Lillico, S.G., Stverakova, D., Proudfoot, C., Christian, M., Voytas, D.F., Long, C.R., Whitelaw, C.B., Fahrenkrug, S.C. : **Efficient TALEN-mediated gene knockout in livestock.** *Proc Natl Acad Sci U S A* 109:17382-7, 2012. Pubmed reference: 23027955. DOI: 10.1073/pnas.1211446109.

CURATION

- Potential curators:
  - authors of recent species reviews
  - postgrads/postdocs, as a part of their training (Ernie Bailey):
    - to work through reviews critically is a really useful exercise
      - gives a feeling for the history of discovery
      - is there sufficient data to justify the Mendelian claim?
      - is there sufficient evidence to justify the claim of a causal mutation?
    - check OMIM links (is this a valid model of a human disorder?)
    - sort out confusing nomenclature/terminology
      - rename/merge/split entries
  - updating entries as an assessment task
  - tools for curators to check and release new/revised text (Matthew)

CURATION

American Journal of Medical Genetics 24:505–511 (1986)

Updating McKusick: An Educational Exercise for Medical Students

Joann N. Bodurtha, J. Ives Townsend, Virginia K. Proud, and Walter E. Nance

Department of Human Genetics, Medical College of Virginia, Richmond, Virginia

Am J Hum Genet. 1987 August; 41(2): 304–305.

II. RECENT INNOVATIONS IN HUMAN GENETICS EDUCATION

The Curricularization of McKusick

JOANN N. BODURTHA,* SANDI VERBIN,* KLARA K. PAPP, † AND WALTER E. NANCE*

*Department of Human Genetics and †Center for Educational Development and Faculty Resources, Medical College of Virginia, Richmond

LANDMARK PAPERS

LANDMARK ARTICLES

Links

1902 Bateson, W. :
Experiments with poultry *Reports to the Evolutionary Committee of the Royal Society* 1:87-124, 1902.

Why is this an OMIA landmark paper? It is first of two adjacent papers (the other being Bateson and Saunders, 1902) that first reported Mendelian inheritance in animals. This paper reported five Mendelian poultry traits, namely Pea comb, Rose comb, polydactyly, shank colour, and white plumage (dominant white).

Bateson, W., Saunders, E.R. :
The facts of heredity in the light of Mendel's discovery *Reports to the Evolution Committee of the Royal Society* 1:125-160, 1902.

Why is this an OMIA landmark paper? It is the second of two adjacent papers (the other being Bateson, 1902) containing the very first reports of Mendelian inheritance in domesticated animals. In addition to the five Mendelian poultry traits reported in the preceding paper (Bateson, 1902), this paper also reported polled in cattle as being a Mendelian trait.

1908 Bateson, W., Punnett, R.C. :
Experimental studies in the physiology of heredity. Poultry *Reports of the Evolution Committee of the Royal Society* 4:18-35, 1908.

Why is this an OMIA landmark paper? It was the first paper to describe a phenotype resulting from the interaction of two genes, i.e. epistasis. The two genes were Rose-comb and Pea-comb in chickens. Birds with mutant alleles at both loci have a "walnut" comb, which is markedly different from either Rose-comb or Pea-comb. Another landmark paper (Imsland et al., 2012) has provided a molecular explanation for this pleiotropy.

1928 Serebrovsky, A.S., Petrov, S.G. :
A case of close autosomal linkage in the fowl *Journal of Heredity* 19:305-306, 1928.

Why is this an OMIA Landmark paper? It presents the first-ever linkage map for any domesticated animal species.

1987 Ricketts, M.H., Simons, M.J., Parma, J., Mercken, L., Dong, Q., Vassart, G. :
A nonsense mutation causes hereditary goitre in the Afrikander cattle and unmasks alternative splicing of thyroglobulin transcripts *Proceedings of the National Academy of Sciences of the United States of America* 84:3181-3184, 1987. Pubmed reference: 3472203.

Why is this an OMIA landmark paper? It is the very first report of a causal mutation in domesticated non-laboratory animals. The discovery was made possible by the specific clinical signs, which suggested only one possible candidate gene, namely the TG gene, encoding thyroglobulin.

1991 Fujii, J., Otsu, K., Zorzato, F., Deleon, S., Khanna, V.K., Weiler, J.E., Obrien, P.J., Maclennan, D.H. :
Identification of a Mutation in Porcine Ryanodine Receptor Associated with Malignant Hyperthermia *Science* 253:448-451, 1991. Pubmed reference: 1862346.

Why is this an OMIA landmark paper? It was the first report of the causal mutation of one of the most-investigated and economically-important disorders to have occurred in domesticated animals. Extensive comparative mapping between humans and pigs eventually suggested the RYR1 gene encoding the ryanodine receptor as a very likely candidate gene. It turned out to be a huge gene (120 kb), the sequencing of which was a mammoth task at that time (late 1980s, early 1990s). These authors were the first to show that the smallest possible mutation (a single-base missense mutation) that changed just one amino acid in a very large molecule comprising 5,035 amino acids, was the cause of a disorder that had been a major financial burden for the global pig industry for several decades.

With thanks to Leif Andersson

CURATION

- INCENTIVES FOR CURATORS ?
- Acknowledgement:
  - at bottom of each edited page
  - at end of a section of text
  - but these not much help on CV
- Publications:
  - publish static OMIA reviews from time to time (highly cited ☺)
- Create specific OMIA roles, e.g. OMIA Chief Swine editor
- Become regarded as the “guardian/authority” of information for a species or for a set of traits

ENHANCEMENTS

- Co-author Matthew Hobbs has done an amazing job
  - 500 hours in 2012 ≈ $50,000
  - the last of my leftover funds
- But still a long shopping list of enhancements required for OMIA
- On the web in **Roundup Issue Tracker**

List of issues

Search

Your Queries (edit)

Issues
Create New
Show Unassigned
Show All
Search
Show issue:

Keywords
Create New

Administration
User List

Hello, frankn
Your Issues
Your Details
Logout

Help
Roundup docs

ID	Activity	Title	Status	Creator	Assigned To
		critical			
183	2 weeks ago	Can't add a reference from Pubmed	chatting	frankn	matthew
176	1 month ago	Tool for superuser to check changes before they go live	chatting	frankn	matthew
182	1 month ago	Facilitate reciprocal links with Ensembl	chatting	frankn	matthew
166	1 month ago	AND/OR search term combinations in advanced search	testing	matthew	matthew
		urgent			
177	1 month ago	Check contents of dump to NCBI	chatting	frankn	matthew
167	1 month ago	technical documention	in-progress	matthew	matthew
168	1 month ago	system housekeeping	in-progress	matthew	matthew
		bug			
184	2 weeks ago	automatic update of local copies of NCBI DTD documents	chatting	matthew	
173	1 month ago	Create an email "newsletter" tool to advise subscribers of additions to OMIA	chatting	frankn	matthew
181	1 month ago	Enable curators to enter links to photos and/or movies of phenes?	chatting	frankn	matthew
85	18 months ago	Off-site backup?	unread	frankn	matthew
		feature			
143	1 month ago	better use of MPO	in-progress	matthew	matthew
158	5 months ago	improve curation history page	chatting	matthew	matthew

ENHANCEMENTS

- Two major enhancement issues:
- 1. ONTOLOGIES
  - Matthew: entire MPO (Jackson lab) as auto-complete on test site, but very slow
  - Very useful collaboration with Zhiliang Hu
  - Still a long way to go
- 2. AUTOMATIC TEXT MINING
  - Now: use myNCBI daily searches for phenes or authors
    - Many irrelevant refs
  - Automatic addition of new refs for each entry
    - e.g. Miotto et al. (2005) **Supporting the curation of biological databases with reusable text mining.** Genome Inform.16(2):32-44
  - Curator check; then go live

OMIA SUMMARY

- Developed over the last 35 years
- Freely available at http://omia/angis.org.au
- Covers (incompletely) 186 non-human animal species
- 2,820 phene-species entries
  - Nearly 1,000 Mendelian phene-species entries
    - Including 486 with known key mutation
- Hyperlinks with NCBI, OMIM, Ensembl, AnimalGenome
- 20,000 references
  - Most hyperlinked to PubMed
  - Many with doi access to full paper
- = Groundwork for others to build-on/develop
- Curation tools and development/testing/learning version

OMIA SUMMARY

- OMIA has the potential to be the global one-stop shop for up-to-date information on
  - inherited disorders
  - single-locus traits
- Of course, the world does not owe a living to OMIA or to me
- Aim of this talk
  - make people aware that if I am knocked out, OMIA is dead !
- If sufficient people feel OMIA should be maintained and improved, then
  - need a Plan B for curation
  - need (modest) funding for enhancement
- Here at PAG, I am willing to
  - help anyone work through curation tools
  - correct/update/create entries

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