

# Online Mendelian Inheritance in Animals (OMIA): a comparative knowledgebase of genetic disorders and other familial traits in non-laboratory animals

Frank W. Nicholas\*

Centre for Advanced Technologies in Animal Genetics and Reproduction (Reprogen), Faculty of Veterinary Science, University of Sydney, NSW 2006, Australia

Received August 24, 2002; Revised and Accepted September 25, 2002

## ABSTRACT

**Online Mendelian Inheritance in Animals (OMIA) provides up-to-date information on inherited disorders and other familial traits in non-laboratory animals. It is freely available online at <http://www.angis.org.au/omia>. With a strong emphasis on comparative biology, OMIA is modelled on, and reciprocally hyperlinked with, Online Mendelian Inheritance in Man (OMIM). It provides a comprehensive catalog of animal models of human inherited disorders, and also provides comprehensive access to information on potential human homologues of inherited disorders and traits in animals. Because its whole structure is based on comparative biology, it provides phenotypic information in a format that is complementary to all the relevant mapping and sequence databases now existing or being created across the animal kingdom.**

## INTRODUCTION

Over many years, veterinarians and others have published a wealth of information on the occurrence and inheritance of disorders and other familial traits in a wide range of non-laboratory animal species. When the present author commenced working as a geneticist in the Faculty of Veterinary Science in 1974, it soon became evident that this wealth of information needed to be assembled in one place that is electronically accessible throughout the world, and in a form that can be regularly updated.

An excellent model was already available, namely Dr Victor McKusick's *Mendelian Inheritance in Man* (MIM) (1), and its electronic version, Online MIM (OMIM) (2)—a comprehensive annotated catalogue of inherited disorders and other familial traits in humans. In 1978, the present author consulted Dr McKusick about his intention to create an animal equivalent of OMIM, and Dr McKusick responded with constructive enthusiasm, providing useful

pointers as to how he would proceed if he were commencing such a task at that time, when computing technology was far advanced from that which was available when MIM was first computerised in 1966.

Being modelled on McKusick's catalogue, the present author's catalogue is called *Mendelian Inheritance in Animals* (MIA). The catalogue and its annotations have been assembled in a relational database, which contains a number of related tables in a hierarchical structure. At the top is a table of disorders and other familial traits. As in MIM, a 6-digit number is allocated to each trait. Unlike in MIM, but consistent with modern practice in database development, this number is completely uninformative; it serves solely as a unique identifier. Wherever the evidence suggests that the animal trait is homologous to a human trait, the relevant trait name from MIM is used for the animal trait, and the relevant MIM 6-digit number is recorded. This approach has resulted in a powerful comparative catalog. Next is a table recording each species in which each trait occurs (containing brief summaries of current knowledge about that trait in that species), and finally there is a table of bibliographic details of papers indexed to relevant traits and species.

Through the sterling efforts of the staff of the Australian National Genomic Information Service (ANGIS), in May 1995 the contents of the relational database became accessible online at <http://www.angis.org.au/omia>, using the same search engine as then used for OMIM, kindly provided by Dennis Benson and Randy Huntzinger at the National Center for Biotechnology Information (NCBI). Direct access to potential human homologues of animal traits was automatically provided by hyperlinking to OMIM via the relevant MIM number. In 1996, in collaboration with Dr David Lipman from NCBI, an automatic means of creating hyperlinks in the reverse direction (from OMIA to OMIM) was established, by providing NCBI with regular access to a list of hyperlinks (mediated via the 6-digit MIA number) for traits in animals for which there is strong evidence of homology (based on molecular information). This two-way hyperlinking provides Online Mendelian Inheritance in Animals (OMIA) users with access to potential human homologues of animal traits, and provides information to OMIM users on potential animal models of human disorders.

\*Tel: +61 293512184; Fax: +61 293522114; Email: [frankn@vetsci.usyd.edu.au](mailto:frankn@vetsci.usyd.edu.au)

**Table 1.** General structure of MIA entries

Field	Description
Trait name	The most standard name for each trait, using (wherever relevant) terms that reflect homology with other species
MIA number	A unique non-informative 6-digit identifier
References	Arranged chronologically (so as to provide a brief 'history' of knowledge of each trait)
MIM number	If it appears that the animal trait has a human homologue, the unique 6-digit identifier of the homologous human trait is included here, providing a hotlink to OMIM
Across-species synonyms	Other terms by which the trait is known across species
Across-species summary	A brief summary of the features of the trait that are common across species
Species synonyms	Terms by which the trait is known in a particular species
Species summary	A brief summary of the features of the trait that are peculiar to a particular species
History	A brief summary of the history of the trait
Clinical summary	A brief summary of clinical signs
Pathology summary	A brief summary of pathology
Prevalence	A brief summary of prevalence
Inheritance	The mode of inheritance
Molecular genetics	A brief summary of molecular knowledge relating to the trait
Mapping summary	A brief summary of map location
Genotype test	Details of genotyping tests for the detection of alleles at the locus
Control	A brief summary of the means of controlling the trait, if it is a disorder

**Table 2.** Summary of the number of disorders/traits grouped according to various categories, in the major domesticated animal species and across all species in the database

Category	Cattle	Chicken	Goat	Horse	Pig	Sheep	Total <sup>a</sup>
Disorders/traits	356	173	65	182	202	177	2498
Single-locus disorders/traits	55	63	8	24	33	57	455
Disorder/trait for which the causative mutation has been identified at the DNA level	23	8	5	7	9	7	115
Disorder/trait with an identified linked marker	9	1	1	1	2	4	31
Potential animal model for a human disorder	115	34	24	84	65	62	352
References	3286	849	241	860	1897	1561	16 243

<sup>a</sup>Total across all 207 species in the database, including those detailed in this table.

Since it was first mounted on the Web, accesses to OMIA have been steadily increasing, with approximately even use from North America, Europe and Australia.

## SEARCHING OMIA

A search of OMIA can be conducted either via one of the lists provided on the home page, or via a text-based search engine towards the bottom of the home page. If one wants to see all of the disorders/traits that have been reported in a particular species, one simply chooses that particular species from the species list. If one wishes to see in which species a particular type of inherited disorder occurs, one can choose among the lists of disorders shown on the home page. If one requires information on a particular disorder/trait, one can enter that particular disorder/trait in the text-based search engine.

## THE STRUCTURE OF ENTRIES IN OMIA

Table 1 presents the general structure of each entry in OMIA. Due to limitations that have so far existed on the time available to the present author for curation of the database, not all fields contain information for all entries.

## CURRENT STATE OF INFORMATION IN OMIA

Table 2 summarises the numbers of disorders/traits in the various categories across the major animal species, together with comparable statistics for the entire database, at the time of writing (August 2002). The differences between the major species are primarily a reflection of the extent of research effort directed to each species. It can be seen that a wealth of useful information has already been gleaned from non-laboratory animals.

## FUTURE DIRECTIONS

Among the many improvements that can be made to the web version are the provision of summary tables and tables showing the details of mutations that have been characterised at the molecular level. Reciprocal hyperlinks need to be established with the relevant mapping databases, and with relevant sequence databases as they come into existence. A recent development that will assist this process has been the establishment of an ANGIS mirror of ArkDB (3)—the main mapping database for non-laboratory animals. In collaboration with Professor John Edwards, the present author is also developing a web-based form of the Oxford Grid (4), which will provide a powerful electronic scaffold for a comprehensive

comparative database, linking all relevant mapping and sequence databases to the information provided on relevant phenotypes in OMIA. Most importantly, in order to give OMIA a life independent of its creator, it is planned to establish an international team of curators and editors who will take responsibility for sections of the database in which they have particular expertise. Finally, a hard-cover version of the database is planned, with much the same format as the hard-cover version of MIM (1).

## ACKNOWLEDGEMENTS

Steve Brown wrote most of the database code that is still in use today, and contributed mightily to the establishment of the database. Paul Le Tissier was an enthusiastic and insightful contributor of ideas during his years in the laboratory in the early 1990s. Jan Graham chased up many of the early references and entered much of the early information, during three years on the project in the early 1980s. Past and present staff of ANGIS, including Alex Reisner, Carolyn Bucholtz,

Camson Huynh, Tim Littlejohn, Mike Poidinger, Dao Mai and Suzanne Payne, have been particularly helpful. As indicated in the text, staff at NCBI, namely Dennis Benson, Randy Huntzinger and David Lipman, have also been very generous in providing assistance. Finally, a special thanks is due to Dr McKusick, for providing so much encouragement from the very beginning, and for helping at several stages along the way.

## REFERENCES

1. McKusick, V.A. (1998) *Mendelian Inheritance in Man. A Catalog of Human Genes and Genetic Disorders*, 12th Edn. The Johns Hopkins University Press, Baltimore, MD.
2. Hamosh, A., Scott, A.F., Amberger, J., Bocchini, C., Valle, D. and McKusick, V.A. (2002) Online Mendelian Inheritance in Man (OMIM), a knowledgebase of human genes and genetic disorders. *Nucleic Acids Res.*, **30**, 52–55.
3. Hu, J., Mungall, C., Law, A., Papworth, R., Nelson, J.P., Brown, A., Simpson, I., Leckie, S., Burt, D.W., Hillyard, A.L. and Archibald, A.L. (2001) The ARKdb: genome databases for farmed and other animals. *Nucleic Acids Res.*, **29**, 106–110.
4. Edwards, J.H. (1991) The Oxford Grid. *Ann. Human Genet.*, **55**, 17–31.