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# Online Mendelian Inheritance in Man 'OMIM'

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The skin is one of the commonest organ systems involved in genetic or inherited diseases. Genodermatoses, genetic syndromes, inherited skin diseases and also inflammatory or immunological disorders with a genetic basis occupy an important part of the dermatology curriculum. The decoding of the human genome has opened up exciting possibilities for a student of dermatology. One of the best websites for detailed and updated information of genetic diseases is the OMIM.

#### WHAT IS OMIM?

Victor McKusick and his colleagues at the Johns Hopkins University, Baltimore, USA authored a book titled 'Mendelian inheritance in man' which is in its 12th edition today.<sup>1</sup> Keeping in mind the swiftly changing developments in the field of genetic diseases, an online version – the OMIM or Online Mendelian Inheritance in Man – was set up in 1995 by the National Centre for Biotechnology Information (NCBI). This is a comprehensive computerized database of human genes and genetic disorders that is updated daily.<sup>2</sup>

#### WHICH DISORDERS ARE DEALT WITH IN OMIM?

OMIM contains disorders which are inherited in a Mendelian fashion. It does not include conditions resulting from chromosomal aberrations. The dermatologist can get current information on virtually

all the genetic skin diseases and syndromes such as the ichthyoses, epidermolysis bullosa, neurofibromatosis and lipoid proteinosis. Immunological or inflammatory disorders with a polygenic inheritance such as atopic dermatitis, psoriasis, alopecia areata and lichen sclerosis are also included.

#### WHAT ARE OMIM NUMBERS?

Each genetic disorder has been allotted a unique six digit OMIM number.<sup>2</sup> The first digit indicates the mode of inheritance of the disorder, viz.

1. (100000- ) Autosomal dominant loci or phenotypes (entries created before May 15, 1994)
2. (200000- ) Autosomal recessive loci or phenotypes (entries created before May 15, 1994)
3. (300000- ) X-linked loci or phenotypes
4. (400000- ) Y-linked loci or phenotypes
5. (500000- ) Mitochondrial loci or phenotypes
6. (600000- ) Autosomal loci or phenotypes (entries created after May 15, 1994)

A number of symbols are also used in conjunction with the OMIM number, and an explanation of these is available in the OMIM FAQs section on the website.

Examples of OMIM numbers include:

Neurofibromatosis 1	166220
Epidermolysis bullosa letalis	226700

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Incontinentia pigmentii	308300
Retinitis pigmentosa, Y linked	400004
Psoriasis susceptibility 1(psors1)	177900,
	Psors2 – 602723, etc.
Atopic dermatitis	603165,
	605803 – 605805, etc.
Systemic lupus erythematosus	152700
Lichen sclerosus	151590

**HOW IS A DISEASE ACCESSED ON OMIM?**

Searching OMIM is very similar to searching Pubmed. The website allows different levels of search - basic, advanced and complex Boolean.<sup>2</sup>

The basic search involves typing in the name of a disease, a chromosome number or a gene symbol, and clicking on 'Go'. In fact, almost 1900 entries show up for the word 'skin', giving one some idea of how many genetic disorders involve the skin. For example, a search for 'epidermolysis bullosa' shows about 71 results, which include various subtypes of epidermolysis bullosa and related genetic conditions. Clicking on the OMIM number of any of these gives details about that condition – synonyms, gene map locus to a single or multiple genes, clinical and genetic details, references and an update log for that disorder. Additionally, where the diagnosis is uncertain, typing in a few of the key features, e.g. photosensitivity, ichthyosis or bullae, gives a list of diseases which can be considered in the differential diagnosis.

**WHAT ARE THE OMIM GENE MAP AND THE OMIM MORBID MAP?**

There are two main systems which can be searched, the OMIM gene map, and the OMIM morbid map.<sup>2</sup> The OMIM gene map lists disorders gene wise, starting with chromosome 1p and ending with 22q, along with the X and Y chromosomes. Diseases which have been linked to specific genes on these chromosomes are available in this map. This map can be searched using

the chromosome number, gene symbol or name of disorder. The OMIM morbid map is an alphabetical list of disorders and their cytogenetic locations. It can be searched in the same fashion as the OMIM gene map.

**WHAT OTHER FEATURES ARE AVAILABLE IN OMIM?**

A click on OMIM statistics lets one know the total number of entries available on that day, e.g. the total number of entries on 3 March 2004 was 15,196, with 14,240 autosomal, 848 X-linked, 48 Y-linked and 60 mitochondrial. The website also provides details of changes, additions and deletions on a monthly basis since its inception in 1995. OMIM also provides links to Pubmed, the Human Genome websites and other related genetic research websites.

**WHAT DOES OMIM NOT PROVIDE?**

OMIM does not have a complete list of all genetic disorders contained in it; these have to be searched by name or symbol. It also does not provide facilities for the clinical evaluation and genetic testing of patients of suspected genetic diseases. For these types of queries and information, other sources such as Genetic Alliance<sup>3</sup>, NORD<sup>4</sup> and Gene Tests<sup>5</sup> may be useful.

**REFERENCES**

1. McKusick VA. Mendelian inheritance in man. 12th ed. Baltimore: Johns Hopkins University Press; 1998.
2. Online Mendelian Inheritance in Man, OMIM (TM). McKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University (Baltimore, MD) and National Center for Biotechnology Information, National Library of Medicine (Bethesda, MD), 2000. Available from: URL: <http://www.ncbi.nlm.nih.gov/omim/>
3. Genetic Alliance [cited 2003 Dec 6] Available from: URL: <http://www.geneticalliance.org>
4. National Organization for Rare Diseases [cited 2003 Dec 7] Available from: URL: <http://www.NORD-rdb.com/~orphan>
5. Gene Tests [cited 2003 Dec 8] Available from: URL: <http://www.genetests.org>

