Medizininformatik – Initiative
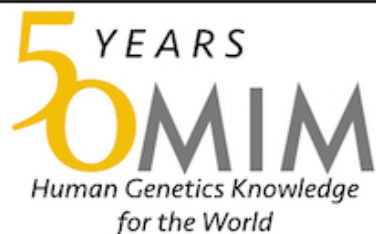# Wokshop Semantische Interoperabilität

TMF – Technologie- und Methodenplattform
für die vernetzte medizinische Forschung
Berlin, Charlottenstrasse 42
Montag, den 22. Mai 2017

OMIM – ein online Kompendium menschlicher Gene und Phänotypen

Thomas F. Wienker (wienker@molgen.mpg.de)

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Institut für Medizinische Informatik und Statistik (IMIS), und
Institut für Klinische Molekularbiologie (IKMB), UKSH/CAU, Kiel

MPIMG

Var Watch

5**0** YEARS
MIM
Human Genetics Knowledge
for the World

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OMIM®

Online Mendelian Inheritance in Man®

An Online Catalog of Human Genes and Genetic Disorders

Updated May 19, 2017

Search OMIM for clinical features, phenotypes, genes, and more...   🔍

Advanced Search : OMIM, Clinical Synopses, Gene Map

Need help? : Example Searches, OMIM Search Help, OMIM Tutorial

Mirror site : mirror.omim.org

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Make a donation!

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Genetic Medicine

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M E D I C I N E

OMIM® - Online Mendelian Inheritance in Man®

Welcome to OMIM®, Online Mendelian Inheritance in Man®. OMIM is a comprehensive, authoritative compendium of human genes and genetic phenotypes that is freely available and updated daily. The full-text, referenced overviews in OMIM contain information on all known mendelian disorders and over 15,000 genes. OMIM focuses on the relationship between phenotype and genotype. It is updated daily, and the entries contain copious links to other genetics resources.

This database was initiated in the early 1960s by Dr. Victor A. McKusick as a catalog of mendelian traits and disorders, entitled Mendelian Inheritance in Man (MIM). Twelve book editions of MIM were published between 1966 and 1998. The online version, OMIM, was created in 1985 by a collaboration between the National Library of Medicine and the William H. Welch Medical Library at Johns Hopkins. It was made generally available on the internet starting in 1987. In 1995, OMIM was developed for the World Wide Web by NCBI, the National Center for Biotechnology Information.

OMIM is authored and edited at the McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine, under the direction of Dr. Ada Hamosh.

NLM's Profiles in Science -- The McKusick Papers

PERSPECTIVES IN HUMAN GENETICS

Mendelian Inheritance in Man and Its Online Version, OMIM

Victor A. McKusick

Last year marked the 40th anniversary of the publication of the first print edition of *Mendelian Inheritance in Man* (MIM). This seems an appropriate juncture at which to review its origins, evolution, and present status, including and particularly those of its online version, OMIM (Online Mendelian Inheritance in Man).
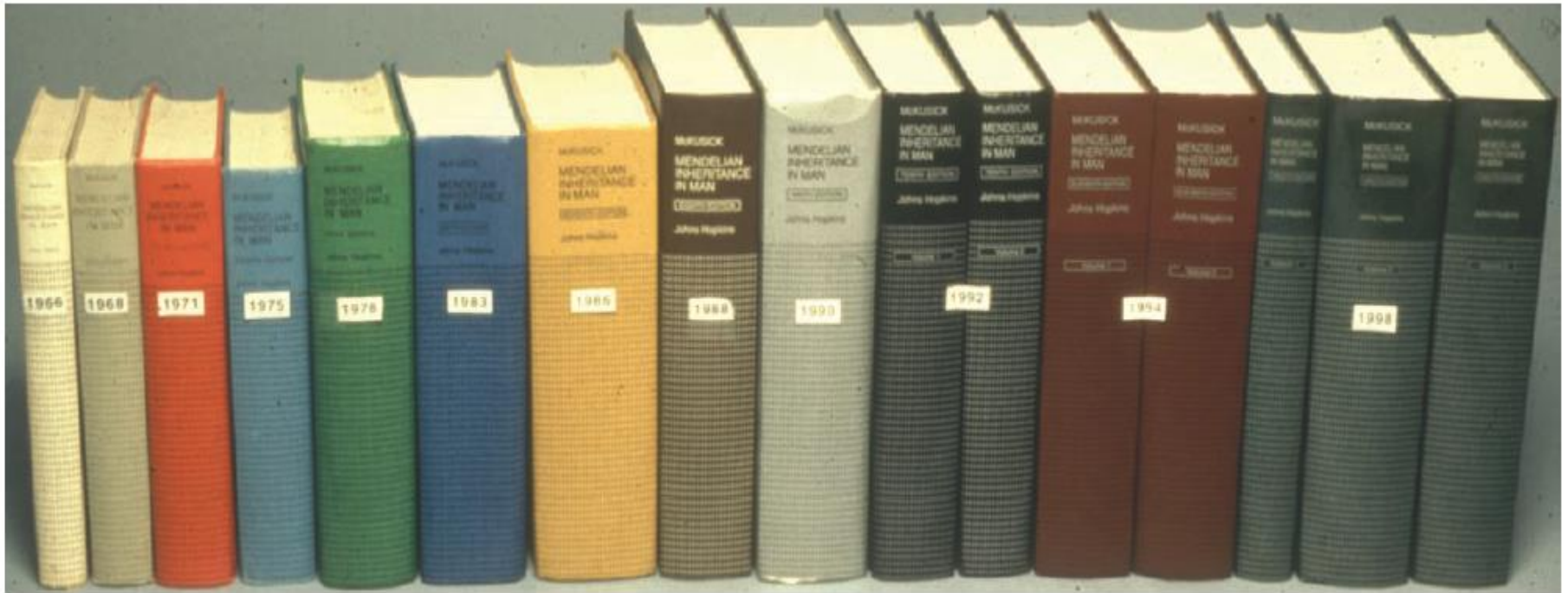
Figure 1. Twelve print editions of MIM, the first published in 1966 and the most recent, in three volumes, published in 1998

OMIM.org: Online Mendelian Inheritance in Man (OMIM®), an online catalog of human genes and genetic disorders

Joanna S. Amberger[1,*], Carol A. Bocchini[1], François Schiettecatte[2], Alan F. Scott[1] and Ada Hamosh[1]

[1]McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine, Baltimore, MD 21287, USA and [2]FS Consulting, LLC, Salem, MA 01970, USA

50 YEARS OMIM

Human Genetics Knowledge for the World

https://omim.org

https://mirror.omim.org

OMIM®

Online Mendelian Inheritance in Man®

An Online Catalog of Human Genes and Genetic Disorders

Updated May 19, 2017

ABO	🔍

Advanced Search : OMIM, Clinical Synopses, Gene Map

Need help? : Example Searches, OMIM Search Help, OMIM Tutorial

Mirror site : mirror.omim.org

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ABO   🔍   Options ▾

View Results as: **Gene Map Table**
Clinical Synopsis ❓
Display: ☑ Highlights

Search: 'ABO'
Results: 89 entries.   Show 100 | Download As ▾ | « First | ‹ Previous | Next › | Last »

1: * 110300. **ABO** GLYCOSYLTRANSFERASE; **ABO**
TRANSFERASE A, ALPHA 1-3-N-ACETYLGALACTOSAMINYLTRANSFERASE, INCLUDED
Cytogenetic location: 9q34.2, Genomic coordinates (GRCh38): 9:133,255,175-133,275,213
Matching terms: abo
▶ Gene-Phenotype Relationships ▶ Links

2: # 616093. BLOOD GROUP, **ABO** SYSTEM
Cytogenetic location: 9q34.2
Matching terms: abo
▶ Phenotype-Gene Relationships ▶ ICD+ ▶ Links

3: 110250. BLOOD GROUP--**ABO** SUPPRESSOR
Matching terms: abo
▶ Links

4: # 193400. VON WILLEBRAND DISEASE, TYPE 1; VWD1
Cytogenetic location: 12p13.31
Matching terms: abo
▶ Phenotype-Gene Relationships ▶ ICD+ ▶ Links

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*110300
Table of Contents

* 110300

ABO GLYCOSYLTRANSFERASE; ABO

Alternative titles; symbols

ABO HISTO-BLOOD GROUP GLYCOSYLTRANSFERASES

Other entities represented in this entry:

TRANSFERASE A, ALPHA 1-3-N-ACETYLGALACTOSAMINYLTRANSFERASE, INCLUDED
TRANSFERASE B, ALPHA 1-3-GALACTOSYLTRANSFERASE, INCLUDED

HGNC Approved Gene Symbol: ABO

Cytogenetic location: 9q34.2   *Genomic coordinates (GRCh38): 9:133,255,175-133,275,213* (from NCBI)

Gene-Phenotype Relationships

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key
9q34.2	[Blood group, ABO system]	616093		3

▼ External Links

► Genome

► DNA

► Protein

► Gene Info

► Clinical Resources

▼ Variation

  1000 Genome
  ClinVar
  ExAC
  gnomAD
  GWAS Catalog
  GWAS Central
  HGMD
  HGVS
  Locus Specific DBs
  NHLBI EVS
  PharmGKB

► Animal Models

► Cellular Pathways

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ICD+

ICD+   ✕

SNOMEDCT: 63915006

▶ Protein

▼ Clinical Resources

Clinical Trials
GTR

▶ Animal Models

#616093
Table of Contents

Title

Phenotype-Gene Relationships

Text

  Description

  Inheritance

  Molecular Genetics

  History

See Also

References

Creation Date

Edit History

616093

BLOOD GROUP, ABO SYSTEM

Alternative titles; symbols

ABO BLOOD GROUP SYSTEM

Phenotype-Gene Relationships

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key	Gene/Locus	Gene/Locus MIM number
9q34.2	[Blood group, ABO system]	616093		3	ABO	110300

▼ TEXT

A number sign (#) is used with this entry because the ABO blood group system is based on variation in the ABO gene (110300) on chromosome 9q34.2.

1.2 What numbering system is used in the OMIM database?

Each OMIM entry is given a unique six-digit number as summarized below:

1----- (100000-) 2----- (200000-) Autosomal 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)

Allelic variants (mutations; see 1.4) are designated by the MIM number of the entry, followed by a decimal point and a unique 4-digit variant number. For example, allelic variants in the factor IX gene (300746) are numbered 300746.0001 through 300746.0101.

1.3 What do the symbols preceding a MIM number represent?

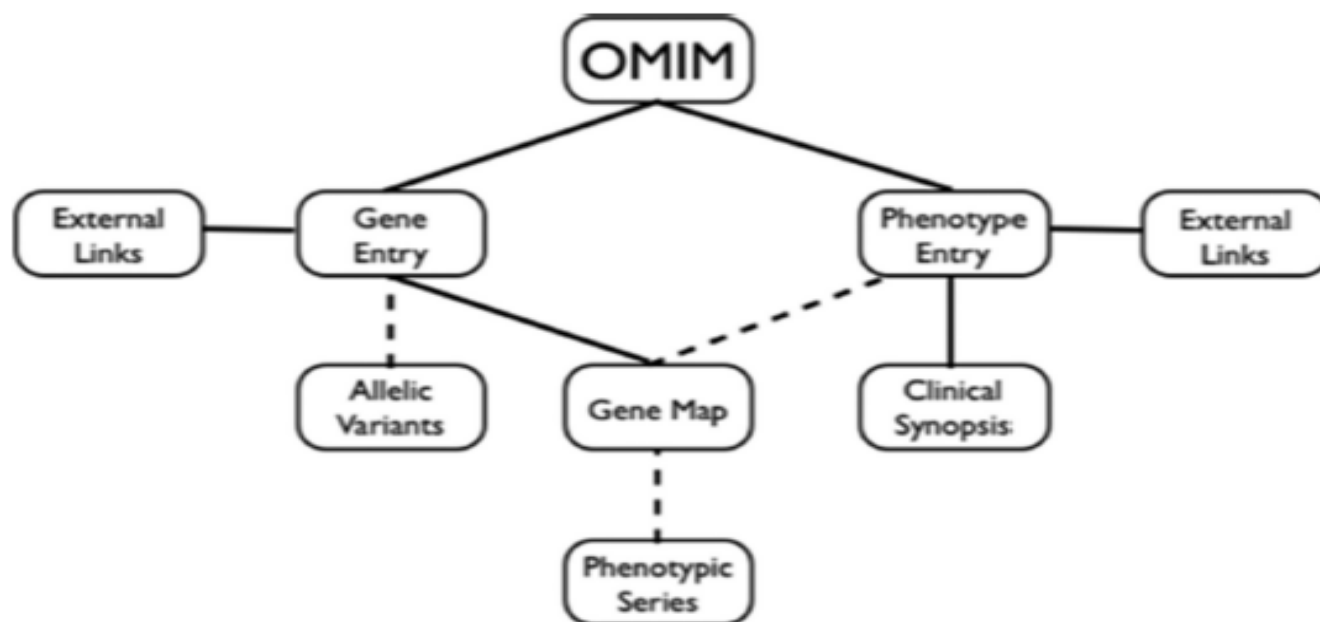| Prefix |
| --- |
| * Gene description |
| + Gene and phenotype, combined |
| # Phenotype description, molecular basis known |
| % Phenotype description or locus, molecular basis unknown |
| Other, mainly phenotypes with suspected mendelian basis |

Figure 1. Diagram of OMIM content. Dashed lines indicate that not all genes have allelic variants; not all phenotypes are mapped; and mapped phenotypes are not necessarily part of a Phenotypic Series.

About   Statistics ▾   Downloads ▾   Contact Us   MIMmatch   Donate ▾   Help ▾   ❓

achondrogenesis   🔍   Options ▾   Display: ☐ Highlights

#200600   ICD+
Table of Contents

Title   # 200600

Phenotype-Gene
Relationships   ACHONDROGENESIS, TYPE IA; ACG1A

Clinical Synopsis
                                    Alternative titles; symbols
Phenotypic Series
                                    ACHONDROGENESIS, HOUSTON-HARRIS TYPE
Text

  Description   **Phenotype-Gene Relationships**

  Clinical Features

  Molecular Genetics

  History

See Also

References

Contributors

Creation Date

Edit History

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key	Gene/Locus	Gene/Locus MIM number
14q32.12	Achondrogenesis, type IA	200600	AR	3	TRIP11	604505

Clinical Synopsis ▾   **Phenotypic Series** ▾

▾ **TEXT**

Search OMIM...   🔍   Options ▾   Display: ☐ Highlights   ☐ Feature IDs

ICD+

#200600
Table of Contents

MIM Entry

Phenotypic Series

200600

ACHONDROGENESIS, TYPE IA; ACG1A

INHERITANCE
- Autosomal recessive

GROWTH
Height
- Dwarfism, marked micromelic
Other
- Short trunk

HEAD & NECK
Nose
- Flat nasal bridge 👤
- Short nose 👤
- Anteverted nares 👤
Neck
- Short neck 👤

#200600
Table of Contents

MIM Entry

Phenotypic Series

ICD+

200600

ACHONDROGENESIS, TYPE IA; ACG1A

INHERITANCE
- Autosomal recessive [SNOMEDCT: 258211005] [UMLS: C0441748] [HPO: HP:0000007]

GROWTH
Height
- Dwarfism, marked micromelic [UMLS: C3150094]
Other
- Short trunk [SNOMEDCT: 95429007] [UMLS: C0521527]

HEAD & NECK
Nose
- Flat nasal bridge [UMLS: C1836542] [HPO: HP:0005280] 👤
- Short nose [UMLS: C1854114] [HPO: HP:0003196] 👤
- Anteverted nares [SNOMEDCT: 708670007] [UMLS: C1840077] [HPO: HP:0000463] 👤
Neck
- Short neck [SNOMEDCT: 95427009] [UMLS: C0521525] [HPO: HP:0000470] 👤

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Download As ▾

Phenotypic Series

Achondrogenesis - PS200600 - 3 Entries

View corresponding clinical synopses as a table

Location ▲	Phenotype	Inheritance	Phenotype mapping key	Phenotype MIM number	Gene/Locus	Gene/Locus MIM number
5q32	Achondrogenesis Ib	AR	3	600972	SLC26A2	606718
12q13.11	Achondrogenesis, type II or hypochondrogenesis	AD	3	200610	COL2A1	120140
14q32.12	Achondrogenesis, type IA	AR	3	200600	TRIP11	604505

View corresponding clinical synopses as a table

Phenotype Mapping Key
1 - The disorder is placed on the map due to its association with a gene, but the underlying defect is not known.
2 - The disorder was placed on the map by statistical methods.
3 - The molecular basis of the disorder is known.
4 - A contiguous gene duplication or deletion syndrome in which multiple genes are involved.

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Download As ▾

Excel File
Tab-delimited File

Phenotypic Series Titles

Phenotypic Series Title	Phenotypic Series Number
Abdominal obesity-metabolic syndrome	PS605552
Achondrogenesis	PS200600
Acne inversa	PS142690
Acrodysostosis	PS101800
Adams-Oliver syndrome	PS100300
Advanced sleep phase syndrome	PS604348
Agammaglobulinemia	PS601495
Aicardi-Goutieres syndrome	PS225750
Alagille syndrome	PS118450
Alopecia, isolated	PS203655
Alopecia-mental retardation syndrome	PS203650
Alternating hemiplegia of childhood	PS104290
Amelogenesis imperfecta	PS104500

Download As ▾

Phenotypic Series

Alopecia, isolated - PS203655 - 8 Entries

View corresponding clinical synopses as a table

Location ▲	Phenotype	Inheritance	Phenotype mapping key	Phenotype MIM number	Gene/Locus	Gene/Locus MIM number
3q26	Alopecia, androgenetic, 1	AD	2	109200	AFA1	109200
8p21.3	Alopecia universalis	AR	3	203655	HR	602302
16q11-q22	Alopecia areata 2	AD, AR	2	610753	AA2	610753
18p11.3-p11.2	Alopecia areata 1	Mu	2	104000	AA1	104000
20p11.22	Alopecia, androgenetic, 3		2	612421	AGA3	612421
Xq11-q12	Alopecia, androgenetic, 2		2	300710	AGA2	300710
Not Mapped	Alopecia, focal			104110	ALPF	104110
Not Mapped	Alopecia, congenital			300042	ALPC	300042

View corresponding clinical synopses as a table

Phenotype Mapping Key
1 - The disorder is placed on the map due to its association with a gene, but the underlying defect is not known.
2 - The disorder was placed on the map by statistical methods.
3 - The molecular basis of the disorder is known.
4 - A contiguous gene duplication or deletion syndrome in which multiple genes are involved.

OMIM Entry Statistics

Number of Entries in OMIM (Updated May 19th, 2017) :

MIM Number Prefix	Autosomal	X Linked	Y Linked	Mitochondrial	Totals
Gene description *	14,789	717	49	35	15,590
Gene and phenotype, combined +	77	0	0	2	79
Phenotype description, molecular basis known #	4,643	318	4	31	4,996
Phenotype description or locus, molecular basis unknown %	1,476	124	5	0	1,605
Other, mainly phenotypes with suspected mendelian basis	1,672	111	2	0	1,785
Totals	22,657	1,270	60	68	24,055

OMIM Gene Map Statistics

OMIM Morbid Map Scorecard (Updated May 19th, 2017) :

Total number of phenotypes* for which the molecular basis is known	5,982
Total number of genes with phenotype-causing mutation	3,734

Distribution of Phenotypes across Genes (Updated May 19th, 2017) :

Number of genes with 1 phenotype	2,535
Number of genes with 2 phenotypes	707
Number of genes with 3 phenotypes	262
Number of genes with 4+ phenotypes	230

Dissected OMIM Morbid Map Scorecard (Updated May 19th, 2017) :

Class of phenotype	Phenotype	Gene *
Single gene disorders and traits	4,942	3,352
Susceptibility to complex disease or infection	702	501
"Nondiseases"	143	113
Somatic cell genetic disease	210	119

*Some genes may be counted more than once because mutations in a gene may cause more than one phenotype and the phenotypes may be of different classes (e.g., activating somatic BRAF mutation underlying cancer, 164757.0001. and germline BRAF mutation in Noonan syndrome, 164757.0022.)

Search OMIM...   🔍   Options ▾

OMIM Data Downloads

Name ▲	Änderungsdatum	Typ	Größe
genemap.txt	21.08.2014 15:13	Text Document	1,806 KB
genemap_key.txt	21.08.2014 15:13	Text Document	8 KB
genemap2.txt	21.08.2014 15:14	Text Document	1,721 KB
mim2gene.txt	21.08.2014 15:14	Text Document	568 KB
morbidmap.txt	21.08.2014 15:14	Text Document	520 KB
omim.txt	22.05.2017 00:59	Text Document	180,521 KB
omim.txt.Z	21.08.2014 15:22	Z-Datei	77,404 KB

OMIM data are updated nightly and are available for download followi
keep a record of downloads for funding purposes* and to notify users
without registration to help interconnectivity of MIM numbers among

File Name	File Description
mim2gene.txt	A tab-delimited file linking MIM numbers with NCBI

, and governmental agencies can register

. This includes the morbid map information

s genemap.txt above) sorted alphabetically by

including additional information such as

```
           0         10        20        30        40        50        60        70
 1  *RECORD*
 2  *FIELD* ·NO
 3  100050
 4  *FIELD* ·TI
 5  100050 ·AARSKOG ·SYNDROME, ·AUTOSOMAL ·DOMINANT
 6  *FIELD* ·TX
 7
 8  DESCRIPTION
 9
10  Aarskog ·syndrome ·is ·characterized ·by ·short ·stature ·and ·facial, ·limb, ·and
11  genital ·anomalies. ·One ·form ·of ·the ·disorder ·is ·X-linked ·(see ·305400),
12  but ·there ·is ·also ·evidence ·for ·autosomal ·dominant ·and ·autosomal
13  recessive ·(227330) ·inheritance ·(summary ·by ·Grier ·et ·al., ·1983).
14
15  CLINICAL ·FEATURES
16
17  Grier ·et ·al. ·(1983) ·reported ·father ·and ·2 ·sons ·with ·typical ·Aarskog
18  syndrome, ·including ·short ·stature, ·hypertelorism, ·and ·shawl ·scrotum.
19  Stretchable ·skin ·was ·present ·in ·these ·patients.
20
```

wget -O OMIM_CS_%1.html http://www.omim.org/clinicalSynopsis/%1

```
################################# CRAWLER WARNING #####################################
#
# - The terms of service and the robots.txt file disallows crawling the site
#   except for the crawlers listed in the robots.txt file, see
#   https://omim.org/help/agreement and https://omim.org/robots.txt for
#   more information.
#
# - A number of data file are available for download at https://omim.org/downloads.
#
# - We have an API you can learn about at https://omim.org/api and https://omim.org/help/api,
#   this provides access to the data in XML, JSON, Python and Ruby formats.
#
# - You should feel free to contact us at https://omim.org/contact to figure out the best
#   approach to getting the data you need.
#
# - YOUR IP ADDRESS WILL BE PERMANENTLY BLOCKED SHOULD YOU CHOOSE TO IGNORE THIS
#   AND CRAWL THE SITE ANYWAY.
#
################################# CRAWLER WARNING #####################################



# Crawl delay, every two seconds
Crawl-delay: 2
```

https://omim.org/robots.txt

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Application Program Interface (API) Access to OMIM

In addition to searching OMIM through the website, OMIM offers a programmatic interface in the form of a REST-based API against which requests can be made over HTTP. The OMIM website is built on this API so any data and functionality available in the website is also available from the API. The data are updated nightly, and the response can be formatted in XML, JSON, JSONP, Python, or Ruby.

For-profit companies or **anyone** planning to **redisplay or incorporate OMIM data** into software MUST secure a license to these files. Registering below will initiate this process, and you can expect to hear from the JHU licensing office.

Please ensure that you describe accurately how you plan to use OMIM so that we can process your registration. If registration is approved you will receive an email containing an API key which will allow you access to the website functions will be automatically rejected.

Johns Hopkins University holds the copyright to OMIM including the collective data contained in OMIM for any commercial purposes, will not develop a derivative data to a third party without first obtaining a license from Johns Hopkins University to do so. I will not use or share the data to a third party without first obtaining a license from Johns Hopkins University to do so. I will use OMIM in my registration. If OMIM data are used in research, I will notify OMIM.

I accept : ☐

First Name : [Required...]

Last Name : [...]

[Required...]

[Required...]

[Required...]

Please enter your institutional email address (By entering *.com address, you give a permission for a JHU Tech licensing representative to contact you). Email addresses from generic email providers such as Gmail, Yahoo, Hotmail, Live, MSN, icloud, 126.com, 163.com or qq.com will be automatically rejected.

An API key is required to access the OMIM API. This unique KEY will be generated upon registration, must be renewed yearly, and must be included with every request. Johns Hopkins University reserves the right to revoke the key at its discretion.

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OMIM API

Overview:

The OMIM API URLs are organized in a very simple fashion:

`/api/[handler]?[parameters]`

`/api/[handler]/[component]?[parameters]`

`/api/[handler]/[action]?[parameters]`

The handler refers to the data object, such as an entry or a clinical synopsis.

The component is optional and refers to a specific data component within a data object for example references within an entry.

The action is optional and refers to an action to be performed on a data object, such as a search for entries.

For basic 'GET's, the component or action are usually optional.

The parameters would include things such as MIM numbers, data retrieval options and data formatting options.

Since this is a read-only database, 'GET' is the only HTTP method for public access, all other HTTP methods will return an error (except for the apiKey handle which supports 'POST' and 'DELETE').

Handlers:

2.1	entry
2.2	clinicalSynopsis
2.3	geneMap
2.4	search
2.5	html
2.6	apiKey
2.7	dump

NCBI   Resources ☑ How To ☑   Sign in to NCBI

OMIM | OMIM ▼ | | Search |
                        Limits   Advanced   Help

OMIM

OMIM is a comprehensive, authoritative compendium of human genes and genetic phenotypes that is freely available and updated daily. OMIM is authored and edited at the McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine, under the direction of Dr. Ada Hamosh. Its official home is omim.org.

Using OMIM

Getting Started

FAQ

OMIM tools

OMIM API

Related Resources

ClinVar

Gene

GTR

MedGen

Last updated on: 20 May 2017

OMIM Frequently Asked Questions (FAQs)

1.12 Where can I access OMIM's website?

OMIM's primary website is available at https://omim.org/. A mirror site is available at https://mirror.omim.org/.

1.13 What reference assembly does OMIM use for the genomic coordinates?

OMIM uses genomic reference build GRCh38 (Patch 4) available at ftp://ftp.ncbi.nih.gov/gene/DATA /gene2refseq.gz (218 MB file).

1.14 What is a Phenotypic Series?

A Phenotypic Series is a tabular view of genetic heterogeneity of similar phenotypes across the genome. The link is available under the Phenotype-Gene mini-table in many phenotype entries. A list of disorders with a phenotypic series is available here.

HGNC
HUGO Gene Nomenclature Committee

Search everything ▼ | Search symbols, keywords or IDs | 🔍

Use * to search with a root symbol (eg ZNF*) ⓘ

Home | **Downloads** | **Gene Families** | **Tools** | **Useful Links** | **About** | **Newsletters** | **Contact Us** | **Help** | **VGNC** | **Request Symbol**

Symbol Report: VPS13A ⓘ

APPROVED SYMBOL ⓘ	VPS13A
APPROVED NAME ⓘ	vacuolar protein sorting 13 homolog A
HGNC ID ⓘ	HGNC:1908
PREVIOUS SYMBOLS & NAMES ⓘ	CHAC, "chorea acanthocytosis", "vacuolar protein sorting 13 homolog A (S. cerevisiae)", "vacuolar protein sorting 13A (yeast)"
SYNONYMS ⓘ	"chorein", KIAA0986
LOCUS TYPE ⓘ	gene with protein product
CHROMOSOMAL LOCATION ⓘ	9q21.2
HCOP ⓘ	Orthology Predictions for VPS13A

omim.org

@OmimOrg

This is the official feed for omim.org, OMIM is a comprehensive, authoritative compendium of human genes and genetic phenotypes.

📍 Baltimore, MD

🔗 omim.org

📅 Joined May 2011

📷 Photos and videos

Tweets   Tweets & replies   Media

🔁 omim.org Retweeted

Welch Library @WelchLibrary · Apr 27

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Disease to Gene: Making the Connections

A Symposium Celebrating OMIM's 50th Anniversary

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Human Genetics Knowledge for the World

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An Online Catalog of Human Genes & Genetic Disorders

Materials prepared by:

Jennifer Williams, Ph.D.

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MEDICINE

National Human
Genome Research
Institute

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Version 1

Learn to use Online Mendelian Inheritance in Man, or OMIM, a catalog of human genes and genetic conditions. OMIM is a foundational resource in genomics and is valuable for clinician and biomedical researchers. OMIM links and data are found at sites all around the bioinformatics sphere, but understanding the full scope of OMIM's data and resources enable the most comprehensive understanding of human phenotypes and disea OMIM contains full-text summaries of information from the scien literature, and provides extensive links to the literature resource and other genomic resource tools as well. Use OMIM as a comprehensive first stop to find important information and gene links for human Mendelian disorders.

You will learn:

- ways to perform both simple and advanced searches
- how to navigate and customize output displays to best serve your needs
- methods to view OMIM data organized by either genes or disorders

| LAUNCH Online Tutorial | PowerPoint Slides | Slide Handouts | Hands-On Exercises | LINK Visit the Resource |

Einheitlicher Bewertungsmaßstab (EBM)

11 Humangenetische Gebührenordnungspositionen

11233 **Ausführliche humangenetische Beurteilung** wegen evidentem genetischen und/oder teratogenen Risiko von bis zu 20 Minuten Dauer

Die Berechnung der Gebührenordnungsposition 11233 setzt die Angabe des phänotypischen OMIM-Kodes oder, falls kein Eintrag in OMIM vorliegt, ersatzweise die Angabe der Art der Erkrankung voraus.

11304 **Schriftliches wissenschaftlich begründetes ärztliches Gutachten zum Antrag des Versicherten auf Durchführung einer Mutationssuche nach den Gebührenordnungspositionen 11449 oder 11514**

Obligater Leistungsinhalt

- Beschreibung des konkreten Untersuchungsumfangs mit tabellarischer Auflistung von
    - Genname(n) einschl. Angabe der kodierenden Sequenzlänge,
    - Gennummer(n) nach OMIM,

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