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Searching Online Mendelian Inheritance in Man (OMIM): A Knowledgebase of Human Genes and Genetic Phenotypes

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Abstract

Online Mendelian Inheritance in Man (OMIM) at OMIM.org is the primary repository of comprehensive, curated information on genes and genetic phenotypes and the relationships between them. This unit provides an overview of the types of information in OMIM and optimal strategies for searching and retrieving the information. OMIM.org has links to many related and complementary databases providing easy access to exploring more information on a topic. The relationship between genes and genetic disorders is highlighted in this unit. The basic protocol explains searching OMIM both from a gene then clinical features perspective. Two alternate protocols provide strategies for viewing gene-phenotype relationships as a gene map table and clinical features as a Quick View or Side-by-Side format. OMIM.org is updated nightly and the MIMmatch service, described in the Support Protocol, provides a convenient way to follow updates to entries, gene-phenotype relationships, and collaborate with other researchers.

Keywords

OMIM; human genetic disorders; molecular genetics; disease gene discovery

INTRODUCTION

Online Mendelian Inheritance in Man (OMIM®) is a continuation of Dr. Victor McKusick's *Mendelian Inheritance in Man* (MIM) (McKusick, 1998), a seminal medical genetics resource used primarily by physicians and other professionals interested in genetic disorders, by genetics researchers, and by advanced students in science and medicine. It provides concise textual information based on the peer-reviewed biomedical literature on over 15,500 genes, 26,200 allelic variants, and 7,800 genetic phenotypes. In addition, OMIM.org provides tabular views of phenotype-gene relationships (gene map), overviews of clinical features of phenotypes (clinical synopses), phenotypic series, and a convenient way to follow updates and collaborate (MIMmatch). All entries in OMIM contain extensive targeted

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Internet Resources

<http://omim.org>

The OMIM Web site.

<http://omim.org/help/faq>

Frequently asked questions (FAQ) about OMIM

links to external data resources. The overall structure of OMIM is shown in Figure 1.2.1 and illustrates the intersections of gene and phenotype information.

The Basic Protocol describes a basic text search and overview of the layout of search results. Alternate Protocol 1 explains searching the Gene Map to reveal unique displays of the intersection between gene and phenotype. Alternate Protocol 2 explains searching clinical information to display the Quick View and Side-by-Side views of the Clinical Synopses. The Support Protocol describes how to register for MIMmatch alerts.

BASIC PROTOCOL: SEARCHING OMIM

[OMIM.org](https://omim.org) is the official website for OMIM information. It has been optimized to retrieve and display information from OMIM in a straightforward and clear manner. [OMIM.org](https://omim.org) supports free-text and field-directed searches. Detailed help, example searches, and tips are available from every screen. The top of the **OMIM home page** (<https://omim.org>) shows the black menu bar that is common across all [OMIM.org](https://omim.org) web pages. From this menu a user can always access Statistics, Download/API registration, Contact Us, MIMmatch access, Help/FAQ, and Tips specific for the page (?). Statistics include the Update List of new and added entries, number of entries in OMIM, and number of phenotype-gene relationships cataloged by OMIM. Below the search box is access to advanced search pages, example searches, search help, and a tutorial.

Necessary Resources

Hardware—Any Internet-connected computer, tablet, or mobile device

Software—Up-to-date Web browsers such as Chrome, FireFox, Safari, Microsoft Explorer or Edge

Performing an OMIM search for genes

1. If, for example, you are interested in potassium channel genes, you can enter your search in a number of ways. For example, *+potassium +channel* (352 entries retrieved), *KCN** (333 entries retrieved), or *"potassium channel"* (with quotes *potassium* and *channel* must appear as a phrase, 276 entries retrieved). Of note, entering *potassium channel* without qualifiers applies both Boolean AND and OR to the search. Entries satisfying the AND operator are given higher relevance (1,334 entries retrieved). OMIM searches are vocabulary enhanced to accommodate synonyms, British spelling, and user-selected thesaurus terms. Figure 1.2.2 shows an **entry retrieval page** for a search on *+potassium +channel* and is described below.
 - A. Blue buttons “Gene Map Table” and “Clinical Synopsis” to the right of the search box navigate the search results to the Gene Map display and Clinical Synopsis (see Alternate Protocols 1 and 2). If thesaurus terms are available for the search, they will be displayed below the search box.

- B. The text below the green line shows the total number of entries retrieved with that search; an option to expand the number of titles displayed; an option to download the search results, and retrieval navigation buttons.
 - C. Each entry in OMIM has a unique and stable MIM number. The MIM *number* has a *prefix* denoting whether the entry represents a gene or phenotype. A mouse-over reminds the user what they mean. The number is followed by the preferred *MIM title* and *symbol*. Clicking on the title brings up the full display of that entry. Below the title is the *cytogenetic location* and *genomic coordinates*; clicking on either of them takes the user to that gene/locus in OMIM's gene map or in UCSCs Genome Browser.
 - D. Colored triangles precede links to reveal on-page information and access to *Gene-phenotype/Phenotype-gene Relationships*, *ICD+* codes, *Phenotypic series*, and *Links* (opened for entry 600937). ICD+ and Phenotypic series links will not appear in gene entries. Access to this information is also present on the entry full view.
2. Clicking on the title for 176260 opens the **entry full view** for this gene (Figure 3). Note that the search terms are highlighted, and there is a green bar next to the Description paragraph denoting information that has been changed or added in the last 3 month. Highlights and change bars can be toggled off by selecting the option to the right of the search box.
- A. Following the number and preferred title are alternative names that may have been given to the entry. The HGNC-approved gene symbol display and link are above the mapping information links and Gene-Phenotype relationship table.
 - B. Left of the title is the entry navigation *Table of Contents*. Selecting the heading takes the user directly to that section in the entry. Note that *Allelic Variants* are listed in a gene entry and can be viewed as descriptive text or in a **Table View** (Figure 1.2.4). Both versions have links, when available, to dbSNP, ExAC, and ClinVar.
 - C. To the right of the title are *External Links*. The topically organized pull-down menus in this stack contain links to over 55 resources including genome browsers, DNA and protein sequence databases, model organism databases, pathway database, etc. Each link takes the user directly to the relevant information in that resource.
 - D. The *plus sign* at the end of text paragraphs takes the user to articles in PubMed that are related to the citation(s) in the MIM entry paragraph. This is an easy way to find additional articles that may be available on a specific topic.

Performing an OMIM search for phenotypes (including disorders) or clinical features

1. Information in a phenotype entry is different from that in a gene entry. Organizationally, each phenotype in a unique phenotype-gene relationship has its own OMIM entry and number. Clinical features in an entry are specific to that phenotype-gene relationship. To show how a class of related phenotypes lies across the genome, OMIM has *Phenotypic Series*. Users can search by disorder name or clinical features. For example, starting at the OMIM.org home page, search on *hypogonadism and microcornea*. Optional thesaurus terms related to hypogonadism (e.g., hypogenitalism, micropenis, small testis, etc.) are displayed under the search box that could be selected to further broaden a search. For the purposes of this protocol, select 600118 by clicking on the title, Warburg Micro syndrome 1 (Figure 1.2.5).
 - A. The ICD+ link shows the available disease identifiers. The External Links have robust *Clinical Resources* (shown), *Animal Model*, and *Cell Lines* sections. These links appear only if there is information in the external resource that is relevant to the MIM entry.
 - B. The *Clinical Synopsis* (a feature-based list) and *Phenotypic Series* links can display contents in-page (down triangle, shown here) or in a new page (main button).
 - C. If the user is a *MIMmatch* member, MIMmatch options will be displayed below the Table of Contents to the left of the entry. The user has options to follow updates to the entry or share interest. This entry has 15 individuals sharing their interest. The link provides details and a way to share contact information.
 - D. The *Phenotypic Series* shows the genetic heterogeneity of Warburg Micro syndrome (Figure 1.2.6).

GUIDELINES FOR UNDERSTANDING ENTRY STRUCTURE AND CONTENT

[Copy Editor: Rather than having one Guidelines section following the last protocol, the authors have included protocol-specific guidelines after each protocol. CP style can accommodate this.]

Number

Each OMIM entry is given a unique and stable six-digit number based on the chromosomal location. Autosomal entries start with a 1, 2 or 6, X-linked entries start with a 3, Y-linked with a 4, and mitochondrial with a 5. Numbers that start with a 1 or 2 do not refer to a mode of inheritance.

MIM numbers are preceded by a prefix:

An **asterisk (*)** before an entry number indicates a gene.

A **number symbol** (#) before an entry number indicates that it is a descriptive entry, usually of a phenotype, and does not represent a unique locus. The reason for the use of the number symbol is given in the first paragraph of the entry. Discussion of any gene(s) related to the phenotype resides in another entry(ies) as described in the first paragraph.

A **plus sign** (+) before an entry number indicates that the entry contains the description of a gene of known sequence and a phenotype.

A **percent sign** (%) before an entry number indicates that the entry describes a confirmed mendelian phenotype or phenotypic locus for which the underlying molecular basis is not known.

No symbol before an entry number generally indicates a description of a phenotype for which the mendelian basis, although suspected, has not been clearly established or that the separateness of this phenotype from that in another entry is unclear.

A **caret** (^) before an entry number means the entry was removed from the database or moved to another entry as indicated.

Title

Each entry has a "Preferred title" with symbol. Synonymous names and symbols that have been used in the literature are included as "Alternative title(s)". "Included titles" mark placement of information on a topic that does not warrant a separate entry (e.g., fusion genes).

Text

Information in the main text of an OMIM entry is put under standard headings (e.g., Description, Clinical Features, Cloning and Expression, etc.) that can be targeted in fielded searches (see online search help 1.5; https://omim.org/help/search#1_5).

Allelic Variants

This section reports mutations and variations in the gene that are found to underlie a phenotype. OMIM includes only select variants. Criteria for inclusion include the first mutation to be discovered, high population frequency, distinctive phenotype, historic significance, unusual mechanism of mutation, unusual pathogenetic mechanism, and distinctive inheritance (e.g., dominant with some mutations, recessive with other mutations in the same gene). Most of the allelic variants represent disease-causing mutations. A few polymorphisms are included, many of which show a positive correlation with particular common disorders. Effective January 2013, based on a rigorous assessment of published reports, OMIM creates phenotype-gene relationships that are "established," "provisional," or of "unknown significance." An established relationship may be based on (1) the existence of multiple, unrelated individuals with pathogenic variants in the same gene; (2) variants that segregate with the phenotype in multiplex families; and/or (3) variants that occur de novo in a statistically significant number of individuals. Functional data and/or animal models support the causality but are not required. A provisional relationship may be based on (1) only one multiplex family reported to have variants in a single gene and the variants

segregate with the phenotype in the family, and (2) there is supportive functional data such as in vitro enzyme activity, a comparable phenotype in a model organism experiment, or an animal model. In this case the gene-phenotype relationship is qualified by noting that the variant has been identified in only 1 family. In rare instances, a similar gene-phenotype relationship may be established on the basis of a single patient if there is robust supporting phenotype and functional data. A variant of unknown significance may be created if a report identifies only 1 patient or family and provides no supporting functional data.

References

All information in an OMIM entry is cited. The references are listed in alphabetical order in this section. There are links to PubMed and to the full text of the article, when available.

ALTERNATE PROTOCOL 1: SEARCHING OMIM'S GENE MAP

OMIM's Gene Map is a table of the genes and loci in OMIM organized by chromosome. OMIM currently has entries for over 15,500 genes and focuses on gene-phenotype relationships. The tabular format provides easy access to multiple gene-phenotype relationships in a single view. The table includes the cytogenetic location, genomic coordinates, gene/locus symbol and title, gene/locus MIM#, comments, and homologous mouse gene. If there is a phenotype(s) associated with a gene/locus, there is a column for phenotype, the phenotype MIM#, the inheritance mode, and the phenotype mapping key. Mousing over the number will display the definition of the key. A summary of the definitions are the following: (1) the disorder was positioned by mapping of the wildtype gene; (2) the disease phenotype itself was mapped; (3) the molecular basis of the disorder is known; (4) the disorder is a chromosome deletion or duplication syndrome. The following describes accessing the gene map table view of a standard search OMIM then restricting the view to genes with phenotypes and then a direct search of the gene map by genomic coordinates to show the phenotypes within a region.

Necessary Resources

Hardware—Any Internet-connected computer, tablet, or mobile device

Software—Up-to-date Web browsers such as Chrome, FireFox, Safari, Microsoft Explorer or Edge

View phenotypes associated with a gene-based search

1. Go to <https://omim.org> and type a gene symbol or words like “potassium channel” (with quotes) in the search box. Then select search.
2. From the retrieval entry page, select the Gene Map Table button to the right of the search box (Figure 1.2.2). This will display the search retrieval in table form in the Gene Map viewer.
3. Select the Phenotype Only Entries toggle to display only those entries from the search that have phenotypes (Figure 1.2.7).

Perform a genomic coordinate search

1. Go to <https://omim.org> and select Gene Map from the advanced search options below the search box.
2. Type in a chromosome number followed by a genomic region (e.g., 18:23,234,12-43,221,000) or cytogenetic location (e.g., 18p11.3). The display will include all genes and loci in OMIM that are in or overlap the search region (Figure 1.2.8). The explanation of the phenotype mapping key and download options are displayed.

ALTERNATE PROTOCOL 2: SEARCHING OMIM CLINICAL SYNOPSES

Each OMIM phenotype entry has a clinical synopsis that contains a list of the salient clinical features described for the phenotype. These features are curated from the biomedical literature and are specific to unique gene-phenotype combinations. This protocol describes accessing the Clinical Synopsis Quick View format and the Side-by-Side viewer.

Necessary Resources

Hardware—Any Internet-connected computer, tablet, or mobile device

Software—Up-to-date Web browsers such as Chrome, FireFox, Safari, Microsoft Explorer or Edge

Accessing the OMIM Clinical Synopsis quick view and side-by-side view

1. Go to <https://omim.org> and type microcornea AND hypogonadism in the search box. Then select search.
2. From the entry retrieval page, select the Clinical Synopsis button to the right of the search box (see Figure 1.2.2).
3. Entries from the search with Clinical Synopses are displayed in the Quick View format. Mouse over the anatomical headings to view the features (Figure 1.2.9).
4. Check the box under the entry retrieval number to select the entries to compare side-by-side.
5. Click the Compare Selected button to reveal the selected synopses side-by-side (Figure 1.2.10). The display of an Elements of Morphology feature definition photo for brachycephaly is displayed.

SUPPORT PROTOCOL 1: Registering for and using MIMmatch

MIMmatch is a service available through [OMIM.org](https://omim.org) that provides easy ways to stay up-to-date and collaborate. MIMmatch users can (1) designate entries they wish to follow and to receive email alerts when the entries are updated; (2) find other researchers who may share their interest in certain entries; (3) receive a daily update on any new gene-phenotype relationships established in OMIM; and/or (4) save search queries. Your name will appear only to other MIMmatch users and only on those entries you designate. All MIMmatch

alerts for a user are collected in a single email, and user's information is not shared without the user's consent.

Necessary Resources

Hardware—Any Internet-connected computer, tablet, or mobile device

Software—Up-to-date Web browsers such as Chrome, FireFox, Safari, Microsoft Explorer or Edge

1. Go to <https://omim.org> and select *MIMmatch* from the menu items on the black bar at the top of the page. Enter the required information on the entry form and click "create account". An email will be sent to you requiring you to activate the newly created account.

Note: MIMmatch options described below will not appear on the website unless you are logged into MIMmatch.

2. When you sign into MIMmatch from the MIMmatch option on the black bar at the top of every [OMIM.org](https://omim.org) page, you will be taken to your MIMmatch dashboard page. From this page you can check the box to be notified of new gene-phenotype relationships and will see 3 tabs: (1) "MIM Entries Flagged" for following or sharing interest; (2) "Phenotypic Series Flagged" for following updates to phenotypic series; and (3) "Saved Searches".
3. Once you have established a MIMmatch account and are logged in, you can populate your "MIM Entries Flagged" when you search OMIM. For example, from a search of *+hypogonadism +microcornea*, you can select "Notify me on update" or "Share my interest" by clicking on the MIMmatch link below the desired entry. You can view others who have chosen to share their interest on this entry from the "Others interested in this entry" link below the MIM titles on a search retrieval page (Figure 1.2.11). The MIMmatch options also appear below the Table of Contents in the full entry (Figure 1.2.3).
4. The "Save search" option supports an easy way to perform the same query of OMIM on a regular basis. A query can be saved by selecting "Save search" from the *Options* link to the right of the search box.

SUPPORT PROTOCOL: Download and API

From the Downloads link in the black bar, users can access *mim2gene.txt*, a tab-delimited file linking MIM numbers with NCBI Gene IDs, Ensembl Gene IDs, and HGNC Approved Gene Symbols. Access to additional OMIM data and the API is available after registration. The API is robust and includes the functionality of the website. Online help that is specific for API use is available under the Help menu option in the black menu bar at the top of the page.

COMMENTARY

Background Information

OMIM entries are based on the peer-reviewed biomedical literature and are expertly curated into structured entries. These executive summaries are a great place to learn about genes and phenotypes, and users are encouraged to explore the copious links both within OMIM entries and to the many linked external resources. Should a user have additional questions about specific information in an OMIM entry, there are links directly to the source reference for further investigation and clarification. The knowledgebase is updated nightly, and the website is designed to display on desktop, tablet, and mobile devices.

Acknowledgments

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Literature Cited

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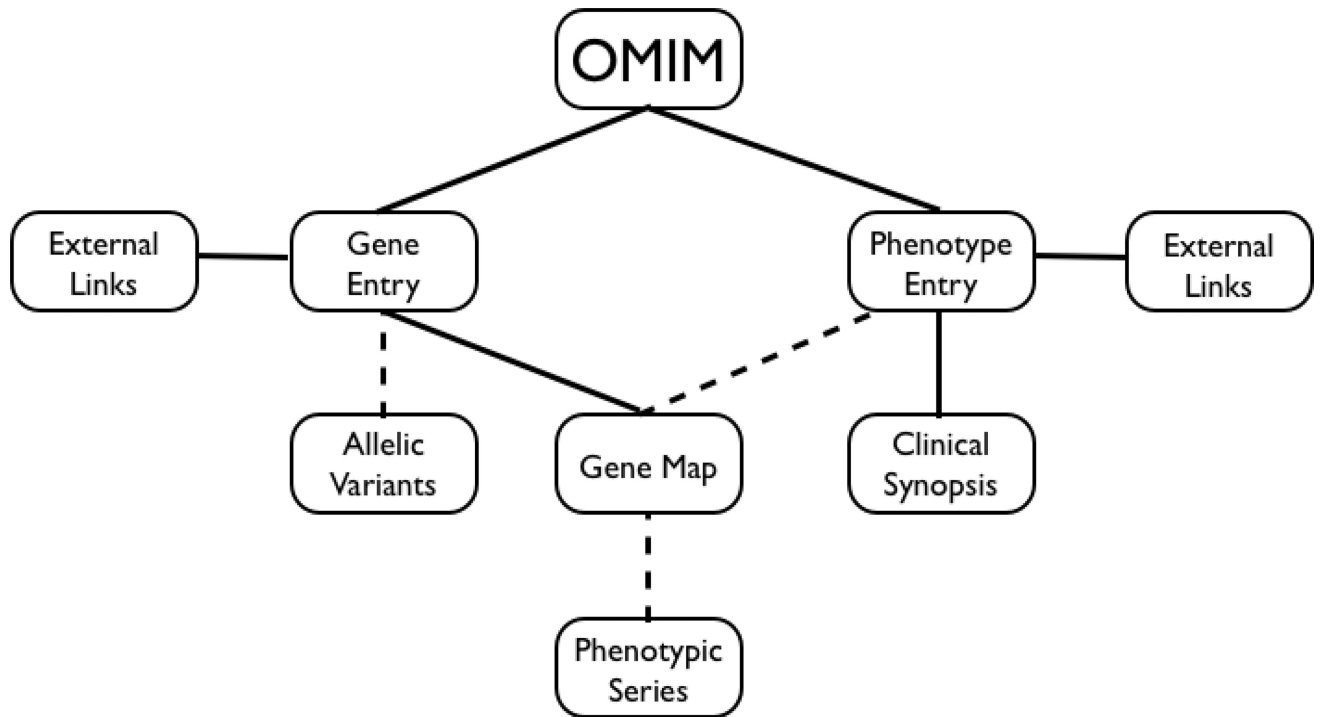


Figure 1.2.1.

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.

Search: '+potassium +channel'

Results: 352 entries. [Show 100](#) | [Download As](#) | [« First](#) | [« Previous](#) | [Next](#) | [Last »](#)

1: * 176260. F [Gene description](#) [NNE](#)L, VOLTAGE-GATED, SHAKER-RELATED SUBFAMILY, MEMBER 1; KCNA1
 Cytogenetic location: 12p13.32, Genomic coordinates (GRCh38): 12:4,909,906-4,918,255
 Matching terms: potassium, channel
[▶ Gene-Phenotype Relationships](#) [▶ Links](#)

2: * 600937. POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 11; KCNJ11
 Cytogenetic location: 11p15.1, Genomic coordinates (GRCh38): 11:17,385,245-17,389,330
 Matching terms: potassium, channel
[▶ Gene-Phenotype Relationships](#) [▼ Links](#)

Links								
Testing	Genome	DNA	Protein	Gene Info	Clinical Resources	Variation	Animal Models	Cellular Pathways
Gene	Ensembl	Ensembl	HPRD	BioGPS	Gene Tests	1000	IMPC	
Tests	NCBI Map Viewer	NCBI RefSeq	Human Protein	Ensembl	Genetics Home	Genome	KOMP	KEGG
GTR	UCSC Genome	UCSC Genome	Atlas	GeneCards	Reference	ClinVar	MGI Mouse Gene	Reactome
NextGxDx	Browser	Browser	UniProt	Gene	GTR	ExAC Beta	NCBI	
				Ontology	NextGxDx	GWAS	HomoloGene	
				KEGG		Catalog	ZFin	
				NCBI Gene		GWAS		
				UCSC		Central		
						HGMD		
						HGVS		
						NHLBI EVS		
						PharmGKB		

3: * 607542. POTASSIUM CHANNEL, VOLTAGE-GATED, KQT-LIKE SUBFAMILY, MEMBER 1; KCNQ1
 Cytogenetic location: 11p15.5-p15.4, Genomic coordinates (GRCh38): 11:2,444,990-2,849,109
 Matching terms: potassium, channel

Figure 1.2.2.

Search result page from a search of +potassium +channel at OMIM.org. Targeted links for entry 600937 are displayed by clicking the “Links” button below the entry title. The mouse-over explanation of the MIM number prefix (in this case “Gene description”) is also displayed.

Search: +potassium +channel

Display: Highlights Change Bars

***176260**

POTASSIUM CHANNEL, VOLTAGE-GATED, SHAKER-RELATED SUBFAMILY, MEMBER 1; KCNA1

Alternative titles; symbols
MK1, MOUSE, HOMOLOG OF KV1.1

HGNC Approved Gene Symbol: KCNA1

Cytogenetic location: 12p13.32 Genomic coordinates (GRCh38): 12:4,909,906-4,918,255 (from NCBI)

Gene-Phenotype Relationships

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key
12p13.32	Episodic ataxia/myokymia syndrome	160120	AD	3

TEXT

Description

Potassium channels represent the most complex class of voltage-gated ion channels from both functional and structural standpoints. Present in all eukaryotic cells, their diverse functions include maintaining membrane potential, regulating cell volume, and modulating electrical excitability in neurons. The delayed rectifier function of potassium channels allows nerve cells to efficiently repolarize following an action potential. In *Drosophila*, 4 sequence-related K⁺ channel genes—Shaker, Shaw, Shal, and Shal—have been identified (Chandy et al., 1990; McPherson et al., 1991). [Click this 'reference-plus' icon to see articles related to this paragraph in PubMed.](#)

Cloning and Expression

By PCR of genomic DNA with primers based on regions conserved between *Drosophila* Shaker and

External Links

- Genome
- DNA
- Protein
- Gene Info
- Clinical Resources
- Variation
 - 1000 Genome
 - ClinVar
 - ExAC Beta
 - GWAS Central
 - HGMD
 - HGVS
 - NHLBI EVS
 - PharmGKB
- Animal Models
- Cellular Pathways

Figure 1.2.3. Entry full view for 176260 displaying highlighted search terms, green entry change bar (left of description paragraph), and Reference Plus link.

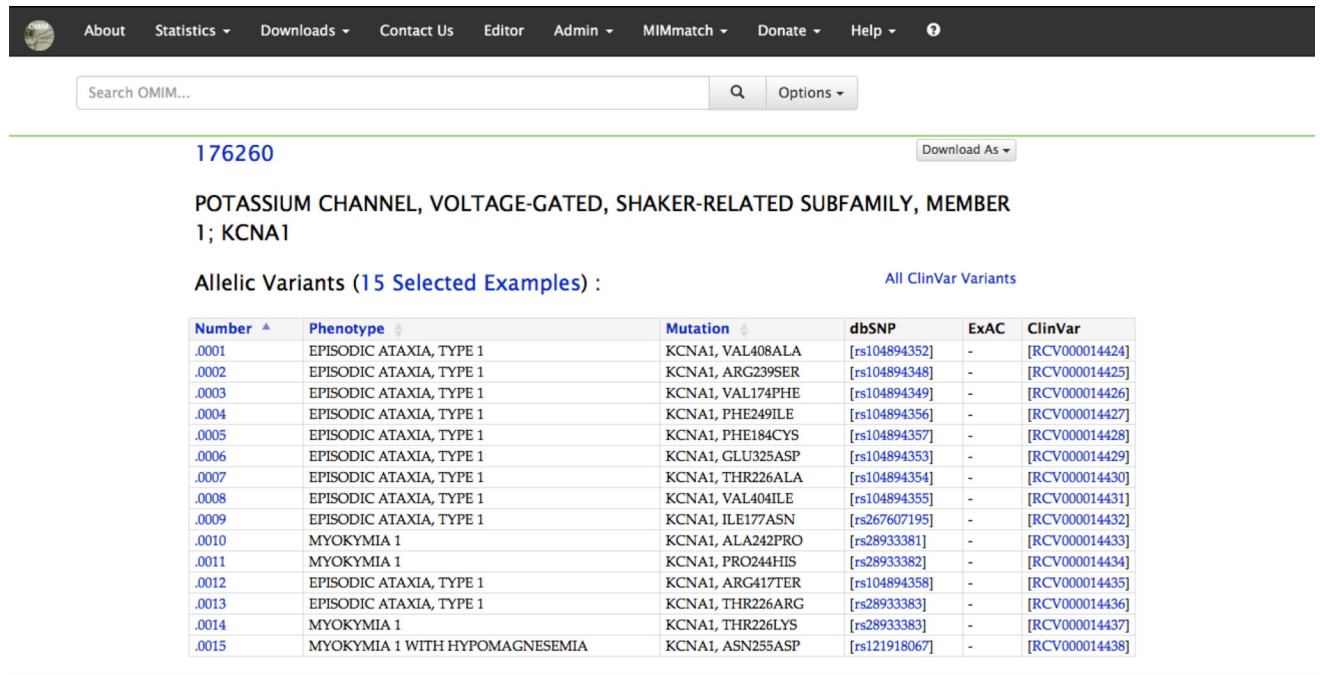
Author Manuscript

Author Manuscript

Author Manuscript

Author Manuscript

Author Manuscript



Search OMIM...

176260

POTASSIUM CHANNEL, VOLTAGE-GATED, SHAKER-RELATED SUBFAMILY, MEMBER 1; KCNA1

Allelic Variants (15 Selected Examples) : [All ClinVar Variants](#)

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar
.0001	EPISODIC ATAXIA, TYPE 1	KCNA1, VAL408ALA	[rs104894352]	-	[RCV000014424]
.0002	EPISODIC ATAXIA, TYPE 1	KCNA1, ARG239SER	[rs104894348]	-	[RCV000014425]
.0003	EPISODIC ATAXIA, TYPE 1	KCNA1, VAL174PHE	[rs104894349]	-	[RCV000014426]
.0004	EPISODIC ATAXIA, TYPE 1	KCNA1, PHE249ILE	[rs104894356]	-	[RCV000014427]
.0005	EPISODIC ATAXIA, TYPE 1	KCNA1, PHE184CYS	[rs104894357]	-	[RCV000014428]
.0006	EPISODIC ATAXIA, TYPE 1	KCNA1, GLU325ASP	[rs104894353]	-	[RCV000014429]
.0007	EPISODIC ATAXIA, TYPE 1	KCNA1, THR226ALA	[rs104894354]	-	[RCV000014430]
.0008	EPISODIC ATAXIA, TYPE 1	KCNA1, VAL404ILE	[rs104894355]	-	[RCV000014431]
.0009	EPISODIC ATAXIA, TYPE 1	KCNA1, ILE177ASN	[rs267607195]	-	[RCV000014432]
.0010	MYOKYMIA 1	KCNA1, ALA242PRO	[rs28933381]	-	[RCV000014433]
.0011	MYOKYMIA 1	KCNA1, PRO244HIS	[rs28933382]	-	[RCV000014434]
.0012	EPISODIC ATAXIA, TYPE 1	KCNA1, ARG417TER	[rs104894358]	-	[RCV000014435]
.0013	EPISODIC ATAXIA, TYPE 1	KCNA1, THR226ARG	[rs28933383]	-	[RCV000014436]
.0014	MYOKYMIA 1	KCNA1, THR226LYS	[rs28933383]	-	[RCV000014437]
.0015	MYOKYMIA 1 WITH HYPOMAGNESEMIA	KCNA1, ASN255ASP	[rs121918067]	-	[RCV000014438]

Figure 1.2.4.

Allelic variant table view showing dbSNP, ExAC, and ClinVar links

The screenshot displays the OMIM entry for #600118, Warburg Micro Syndrome 1. The page includes a search bar with the query 'hypogonadism and microcornea', a navigation menu, and a table of contents. The main content area shows the title 'WARBURG MICRO SYNDROME 1; WARBM1' and alternative titles. A table of Phenotype-Genes is provided below.

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key	Gene/Locus	Gene/Locus MIM number
2q21.3	Warburg micro syndrome 1	600118	AR	3	RAB3GAP1	602536

Below the table, there are buttons for 'Clinical Synopsis' and 'Phenotypic Series'. The 'Phenotypic Series' section is expanded, showing inheritance (Autosomal recessive), growth (Short stature), and head & neck features (Microcephaly, Micrognathia, Large ears, Microphthalmia, Microcornea, Congenital cataract, Optic atrophy, Ptosis, Deep-set eyes). The MIMmatch section is also visible, with options to 'Notify me on update' and 'Share my interest'.

Red arrows in the image point to the 'ICD+' dropdown menu (showing ORPHA: 2510 and DO: 0060237), the 'Phenotypic Series' button, and the 'MIMmatch' section.

Figure 1.2.5. Full view of entry 600118 showing the ICD+ coding numbers, in-page view of the *clinical synopsis*, and MIMmatch information.

Download As ▾

Phenotypic Series

Warburg micro syndrome - PS600118 - 4 Entries [View corresponding clinical synopses as a table](#)

Location ▲	Phenotype ▾	Inheritance	Phenotype mapping key ▾	Phenotype MIM number ▾	Gene/Locus ▾	Gene/Locus MIM number ▾
1q41	Warburg micro syndrome 2	AR	3	614225	RAB3GAP2	609275
2q21.3	Warburg micro syndrome 1	AR	3	600118	RAB3GAP1	602536
10p12.1	Warburg micro syndrome 3	AR	3	614222	RAB18	602207
20p13	Warburg micro syndrome 4	AR	3	615663	TBC1D20	611663

Figure 1.2.6.

Phenotypic Series for Warburg micro syndrome. The sort can be changed by clicking on the triangle links in the table headers. Explanation of the Inheritance and Phenotype mapping keys are available through a mouse-over pop-up.

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Author Manuscript

Author Manuscript

Search: '+potassium +channel (Entries with: gene map locus; Phenotype only entries; Retrieve: gene map)'
Results: 166 entries. [Show 100](#) | [Download As](#) | [« First](#) | [« Previous](#) | [Next >](#) | [Last >](#)

View Results as: [Entries](#)
Display: Highlights

Phenotype Only Entries All Entries

Genomic context table	Location (from NCBI, GRCh38)	Gene/Locus	Gene/Locus name	Gene/Locus MIM number	Phenotype	Phenotype MIM number	Inheritance	Pheno map key	Comments	Mouse symbol (from MGI)
1:	1:2,019,328 1p36.33	GABRD, GEFSP5, EIG10, EJM7	Gamma-aminobutyric acid (GABA) A receptor, delta	137163	[Epilepsy, juvenile myoclonic, susceptibility to]	613060	AD	3		Gabrd
					[Epilepsy, idiopathic generalized, 10]	613060	AD	3		
					[Epilepsy, generalized, with febrile seizures plus, type 5, susceptibility to]	613060	AD	3		
2:	1:11,106,530 1p36.22	MTOR, FRAP1, SKS	Mechanistic target of rapamycin	601231	Smith-Kingsmore syndrome	616638	AD	3		Mtor
3:	1:16,021,990 1p36.13	CLCNKA	Chloride channel, kidney, A	602024	Bartter syndrome, type 4b, digenic	613090	DR	3	11kb from CLCNKB; simultaneous mutation in CLCNKA and CLCNKB	Clcnkb
4:	1:16,043,735 1p36.13	CLCNKB	Chloride channel, kidney, B	602023	Bartter syndrome, type 4b, digenic	613090	DR	3	unequal crossingover with CLCNKA	Clcnka
					Bartter syndrome, type 3	607364	AR	3		

Figure 1.2.7.

OMIM Gene map with phenotype only view selected.

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Search: '18:23,234,12-43,221,000'
Results: 121 entries.

Show 100 | Download As | « First | < Previous | Next > | Last » | Phenotype Only Entries | All Entries

Genomic context table	Location (from NCBI, GRCh38)	Gene/Locus	Gene/Locus name	number	Phenotype	Phenotype MIM number	Inheritance	Pheno map key	Comments	Mouse symbol (from MGI)
1:	18:0 18p11.3-p11.2	AA1	Alopecia areata 1	104000	Alopecia areata 1	104000	Mu	2	max lod at D18S967	
2:	18:0 18p	DEL18p, C18DELp	Chromosome 18p deletion syndrome	146390	Chromosome 18p deletion syndrome	146390	AD	4		
3:	18:0 18p11.32-p11.31	DFNB46	Deafness, neurosensory, autosomal recessive 46	609647	Deafness, autosomal recessive 46	609647	AR	2	between D18S59 and D18S391	
4:	18:0 18p11	DYT15	Dystonia-15, myoclonic	607488	Dystonia-15, myoclonic	607488	AD	2		
5:	18:0 18p	DYT7	Dystonia-7 (torsion dystonia, adult-onset, focal)	602124	Dystonia-7, torsion	602124	AD	2		
6:	18:0 18pter-p11.21	ERV1	Oncogene ERV1; endogenous retrovirus-1	131150						
7:	18:0 18p11	IBD21	Inflammatory bowel disease 21	612354	[Inflamm disease 2			2	associated with rs2542151	
8:	18:0 18p	MAFD1, BPAD, MD1	Major affective disorder 1	125480	[Major affective disorder 1]	125480	AD	2	?also 18q	

2 - The disorder was placed on the map by statistical methods

Figure 1.2.8.
OMIM Gene map results from a genomic coordinate search, 18:23,234,12-43,221,000.

Search OMIM... View Results as: Display: Highlights Feature IDs

Would you also like: hypogonadism micropenis Add All
 "small testis" "hypoplastic scrotum"

Search: 'microcornea and hypogonadism (Entries with: clinical synopsis; Retrieve: clinical synopsis)'
 Results: 15 clinical synopses. [Show 100](#) | [Download As](#) | [« First](#) | [« Previous](#) | [Next »](#) | [Last »](#)

- # 600118. **WARBURG MICRO SYNDROME 1; WARBM1**
 Inheritance, Growth, Head & Neck, Genitourinary, Skeletal, Skin, nails, & hair, Neurologic, Molecular basis,
 Matching terms: hypogonadism, microcornea
[View full synopsis below](#) [View full synopsis on new page](#) [Links](#)
- # 248800. **MARINESCO-SJOGREN SYNDROME; MSS**
 Inheritance, Growth, Head & Neck, **Skeletal**, Muscle, soft tissues, Neurologic, Endocrine features, Laboratory abnormalities, Miscellaneous, Molecular basis,
 Matching terms:
[View full synopsis](#) [View full synopsis on new page](#) [Links](#)
 Skeletal deformities due to severe myopathy and hypotonia
 Kyphosis
 Scoliosis
- # 614222. **WARBURG MICRO SYNDROME 3; WARBM3**
 Inheritance, Growth, Head & Neck, Genitourinary, Skeletal, Skin, nails, & hair, Muscle, soft tissues, Neurologic, Molecular basis,
 Matching terms: Cubitus valgus
[View full synopsis](#) [View full synopsis on new page](#) [Links](#)
 Coxa valga
 Short metacarpals
- # 212720. **MARINESCO-SJOGREN SYNDROME; MSS**
 Inheritance, Growth, Head & Neck, Genitourinary, Skeletal, Skin, nails, & hair, Neurologic, Endocrine features,
 Matching terms: hypogonadism, microcornea
[View full synopsis below](#) [View full synopsis on new page](#) [Links](#)
 Short metatarsals
- # 615663. **WARBURG MICRO SYNDROME 4; WARBM4**
 Inheritance, Growth, Head & Neck, Genitourinary, Skeletal, Skin, nails, & hair, Muscle, soft tissues, Neurologic, Miscellaneous, Molecular basis,
 Matching terms: hypogonadism, microcornea

Figure 1.2.9. Clinical Synopsis Quick View with feature mouse over the anatomical headings to pop-up the features.

NUMBER	# 234050	# 604168	# 212720	# 600118
TITLE	TRICHOThIODYSTROPHY 4, NONPHOTOSENSITIVE; TTD4	CONGENITAL CATARACTS, FACIAL DYSMORPHISM, AND NEUROPATHY; CCFDN	MARTSOLF SYNDROME	WARBURG MICRO SYNDROME 1; WARBMI
INHERITANCE	- Autosomal recessive	- Autosomal recessive	- Autosomal recessive	- Autosomal recessive
GROWTH		Height - Short stature	Height - Short stature - Height less than 5th percentile Weight - Weight less than 5th percentile	Height - Short stature
HEAD & NECK	Other - Growth retardation Head - Microcephaly Face - Short smooth philtrum (rare) - Retrognathia, mild (rare) Ears - Large ears Eyes - Brittle and sparse eyebrows - Brittle and sparse eyelashes - Nystagmus (rare) - Optic atrophy (rare) - Epicanthal folds (rare) - Microcornea (in some patients) - Microphthalmia (rare) - Nystagmus (in some patients) - Infantile cataract (in some patients) - Decreased best corrected visual acuity (in some patients)	Face - Facial dysmorphism - Prominent midface - Thickening of perioral tissues - Mandibular retrognathism Eyes - Congenital cataracts - Microcorneas - Nystagmus	Head - Brachycephaly - Microcephaly Face - Mild maxillary hypoplasia - Micrognathia - Short philtrum Ears - Prominent antitragus - Posteriorly rotated ears Eyes - Cataracts - Epicanthal fold - Downward-slanting palpebral fissures	Ears - Large ears Eyes - Microphthalmia - Microcornea - Congenital cataract - Optic atrophy - Ptosis - Deep-set eyes

Figure 1.2.10. Clinical Synopsis Side-by-Side view with Elements of Morphology *brachycephaly* feature photo pop-up open.

The screenshot displays the MIMmatch website interface. At the top, there is a navigation bar with links for About, Statistics, Downloads, Contact Us, Editor, Admin, MIMmatch, Donate, and Help. Below this is a search bar containing the query '+hypogonadism +microcornea'. To the right of the search bar are buttons for 'Options' and 'View Results as: Gene Map Table Clinical Synopsis'. Below the search bar, there are suggestions for related terms: 'hypogonitalism', 'micropenis', 'small testis', and 'hypoplastic scrotum'. A dropdown menu is open, showing options: 'Advanced Search', 'OMIM', 'Clinical Synopses', 'Gene Map', 'Search History', and 'Save Search'. The 'Save Search' option is circled in red. The search results section shows 'Search: '+hypogonadism +microcornea'' and 'Results: 15 entries.' The first result is '# 600118. WARBURG MICRO SYNDROME 1; WARBM1' with a cytogentic location of 2q21.3. The second result is '# 614222. WARBURG MICRO SYNDROME 3; WARBM3' with a cytogentic location of 10p12.1. Both results include matching terms and navigation links for Phenotype-Gene Relationships, Phenotypic Series, ICD+, Links, and MIMmatch. The MIMmatch link in the first result is also circled in red. There are also options to 'Notify me on update' and 'Share my interest'.

Figure 1.2.11.
MIMmatch options.