Болезни и фенотипы

Василий Евгеньевич Раменский

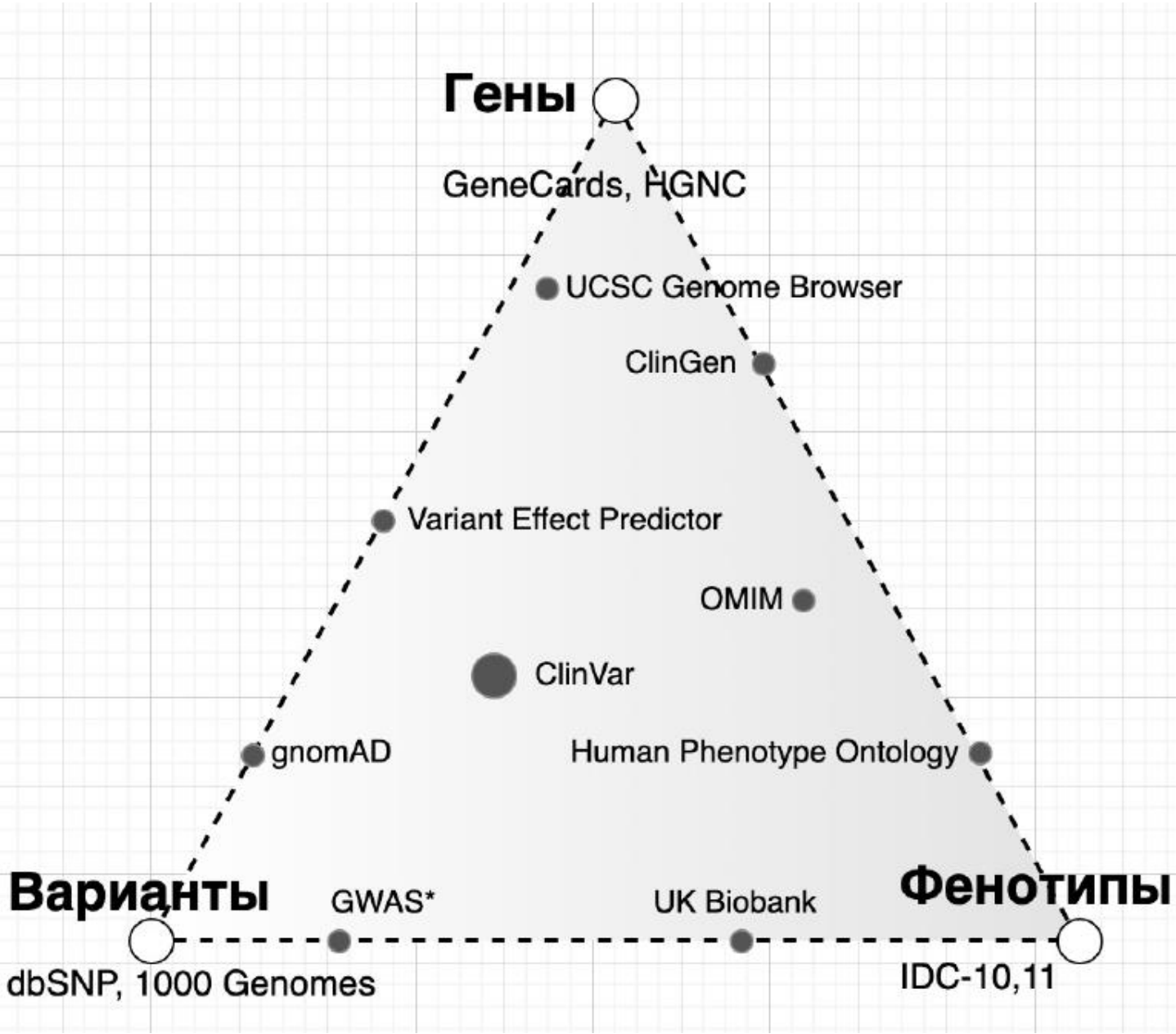
Анастасия Александровна Жарикова и Мария Ильинична Зайченока

НМИЦ Терапии и профилактической медицины

Факультет биоинженерии и биоинформатики МГУ

Институт искусственного интеллекта МГУ

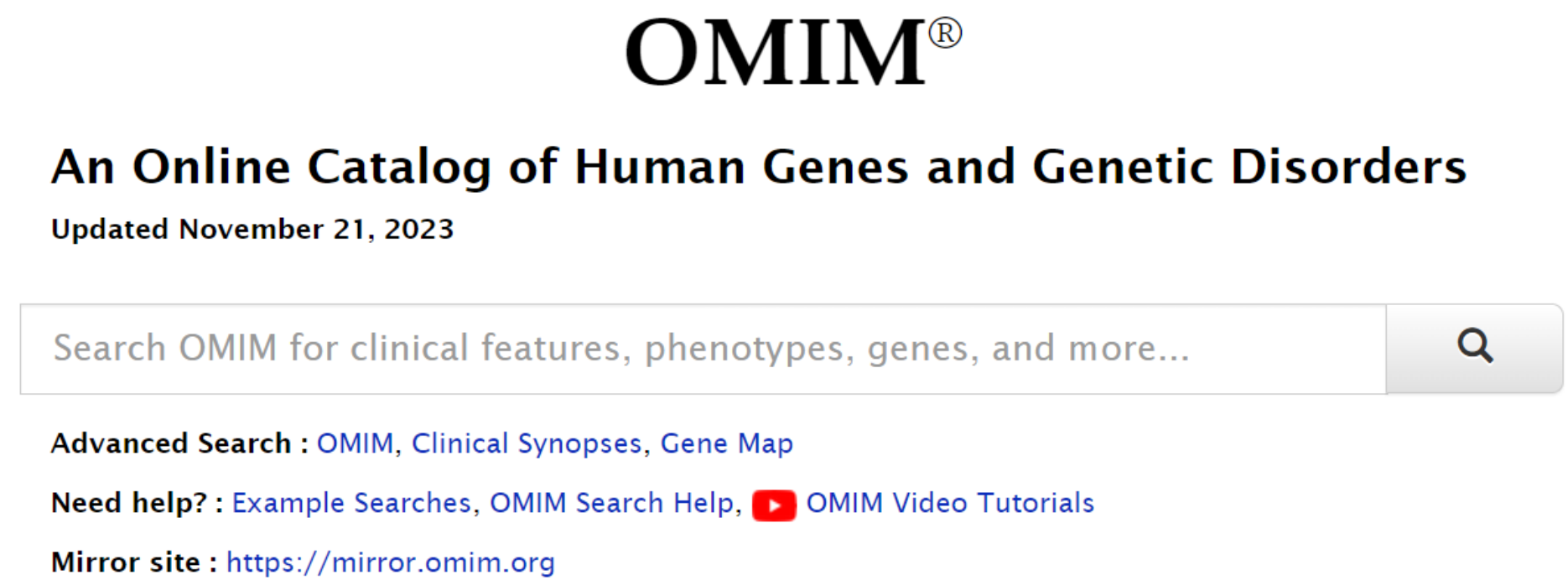
2024


Гены
GeneCards, HGNC
UCSC Genome Browser
ClinGen
Variant Effect Predictor
OMIM
ClinVar
gnomAD
Human Phenotype Ontology
Варианты
GWAS*
UK Biobank
Фенотипы
dbSNP, 1000 Genomes
IDC-10,11

OMIM

Online Mendelian Inheritance in Man (**OMIM**)

https://www.omim.org/



Коды MIM

Диапазон кода заболевания зависит от типа наследования:

- 100000—299999 — аутосомные заболевания (создано до 15 мая 1994 года);
- 300000—399999 — X-сцепленные заболевания;
- 400000—499999 — Y-сцепленные заболевания;
- 500000—599999 — Митохондриальные заболевания;
- 600000 и выше — аутосомные заболевания (создано после 15 мая 1994 года).

wiki

OMIM

OMIM Entry Statistics

Number of Entries in OMIM (Updated November 21st, 2023) :

MIM Number Prefix	Autosomal	X Linked	Y Linked	Mitochondrial	Totals
Gene description *	16,285	769	51	37	17,142
Gene and phenotype, combined +	21	0	0	0	21
Phenotype description, molecular basis known #	6,330	381	5	34	6,750
Phenotype description or locus, molecular basis unknown %	1,391	112	4	0	1,507
Other, mainly phenotypes with suspected mendelian basis	1,640	100	3	0	1,743
Totals	25,667	1,362	63	71	27,163

OMIM

OMIM Morbid Map Scorecard (Updated November 21st, 2023) :

Total number of phenotypes* for which the molecular basis is known	7,450
Total number of genes with phenotype-causing mutation	4,859

* Phenotypes include (1) single-gene mendelian disorders and traits; (2) susceptibilities to cancer and complex disease (e.g., BRCA1 and familial breast-ovarian cancer susceptibility, 113705.0001, and CFH and macular degeneration, 134370.0008); (3) variations that lead to abnormal but benign laboratory test values ("nondiseases") and blood groups (e.g., lactate dehydrogenase B deficiency, 150100.0001 and ABO blood group system, 110300.0001); and (4) select somatic cell genetic disease (e.g., GNAS and McCune-Albright syndrome, 139320.0008 and IDH1 and glioblastoma multiforme, 147700.0001.)

OMIM

Distribution of Phenotypes across Genes (Updated November 21st, 2023) :

Number of genes with 1 phenotype	3,410
Number of genes with 2 phenotypes	880
Number of genes with 3 phenotypes	316
Number of genes with 4+ phenotypes	253

OMIM

Dissected OMIM Morbid Map Scorecard (Updated November 21st, 2023) :

Class of phenotype	Phenotype	Gene *
Single gene disorders and traits	6,392	4,495
Susceptibility to complex disease or infection	677	500
"Nondiseases"	151	118
Somatic cell genetic disease	237	131

*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.)

OMIM

OMIM Update List

Updates since the database was placed on the web in December 1995

2023	Jan	Feb	Mar	Apr	May	Jun	Jul	Aug	Sep	Oct	Nov
New	54	38	41	53	48	39	20	42	32	37	37
Updated	422	257	531	415	310	380	415	449	316	411	240

2022	Jan	Feb	Mar	Apr	May	Jun	Jul	Aug	Sep	Oct	Nov	Dec
New	50	42	49	51	44	41	45	44	42	44	40	35
Updated	434	484	634	641	461	447	383	359	479	383	450	339

2021	Jan	Feb	Mar	Apr	May	Jun	Jul	Aug	Sep	Oct	Nov	Dec
New	30	44	44	50	53	52	52	53	39	50	41	46
Updated	443	469	642	508	372	433	403	593	371	359	355	411

OMIM

OMIM Update List for November 2023

November 21st, 2023

New Entries:

#620610 OOCYTE/ZYGOTE/EMBRYO MATURATION ARREST 21; OZEMA21
#620629 OPTIC ATROPHY 16; OPA16
*620630 TRANSMEMBRANE PROTEIN 170A; TMEM170A

New Clinical Synopses:

#620603 IMMUNODEFICIENCY 114, FOLATE-RESPONSIVE; IMD114
#620609 LONG-OLSEN SYNDROME; LNGOS

Updated Entries:

#145001 HYPERPARATHYROIDISM 2 WITH JAW TUMORS; HRPT2
#146255 HYPOPARATHYROIDISM, SENSORINEURAL DEAFNESS, AND RENAL DYSPLASIA SYNDROME; HDRS
#165500 OPTIC ATROPHY 1; OPA1
#165510 OPTIC ATROPHY 13 WITH RETINAL AND FOVEAL ABNORMALITIES; OPA13
*600424 SOLUTE CARRIER FAMILY 19 (FOLATE TRANSPORTER), MEMBER 1; SLC19A1
*603078 CHECKPOINT KINASE 1; CHEK1
#606159 NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 3; NBIA3
#606593 LIG4 SYNDROME
#606612 MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH OR WITHOUT IMPAIRED INTELLECTUAL DEVELOPMENT), TYPE B, 5; MDDGB5

OMIM

- Поиск можно начинать с
  - Гена
  - Заболевания
  - Варианта (rs121918383) (могут быть не все варианты)

Упражнение

- Найдите в базе данных OMIM заболевание hypercholesterolemia

OMIM

hypercholesterolemia | Options | View Results as: Gene Map Table | Clinical Synopsis | Display: Highlights

Search: 'hypercholesterolemia '
Results: 137 entries. Show 100 | Download As | « First | ‹ Previous | Next › | Last »

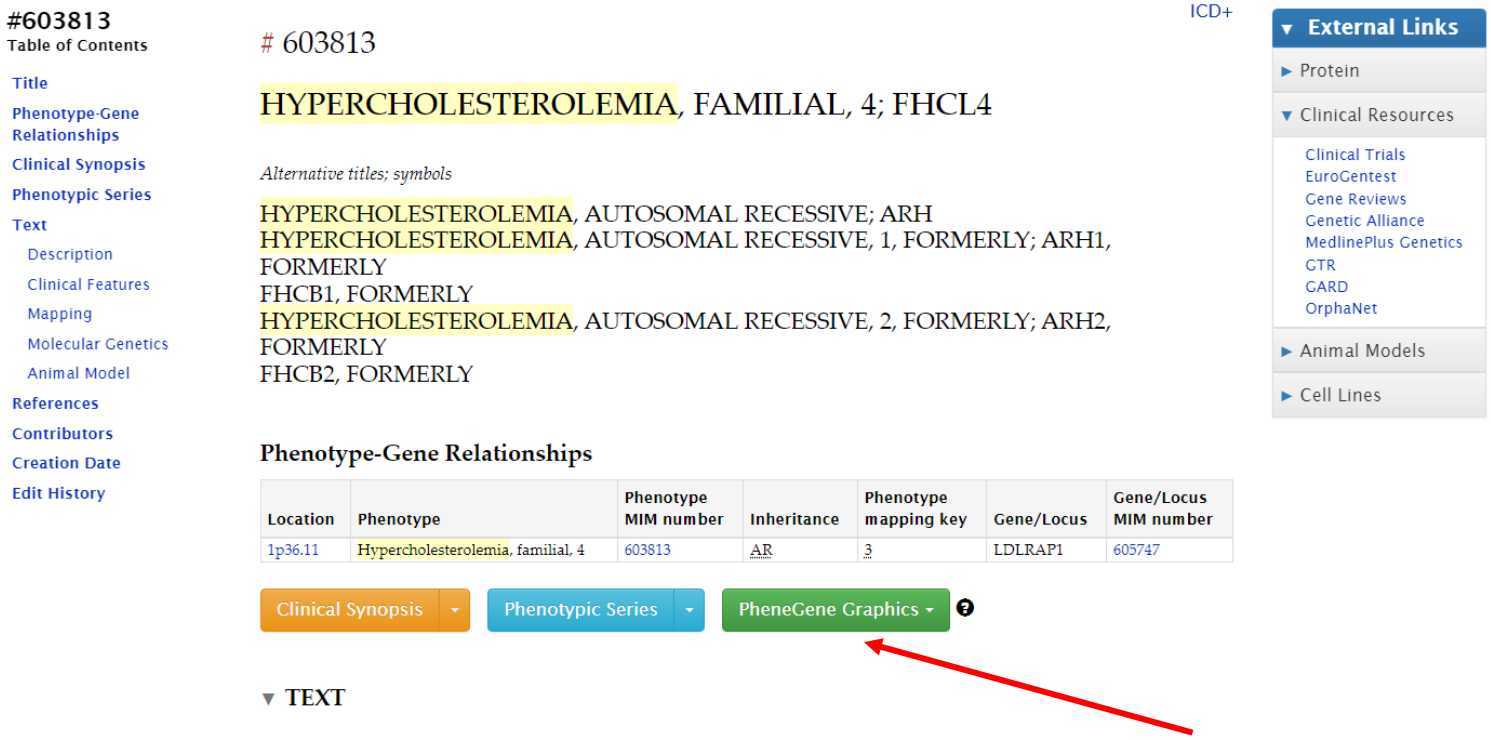
1: # 603813. HYPERCHOLESTEROLEMIA, FAMILIAL, 4; FHCL4
Cytogenetic location: 1p36.11
Matching terms: (hypercholesterolaemia | hypercholesterolemia)
► Phenotype-Gene Relationships ► Phenotypic Series ► ICD+ ► Links

2: # 144010. HYPERCHOLESTEROLEMIA, FAMILIAL, 2; FHCL2
Cytogenetic location: 2p24.1
Matching terms: (hypercholesterolaemia | hypercholesterolemia)
► Phenotype-Gene Relationships ► Phenotypic Series ► ICD+ ► Links

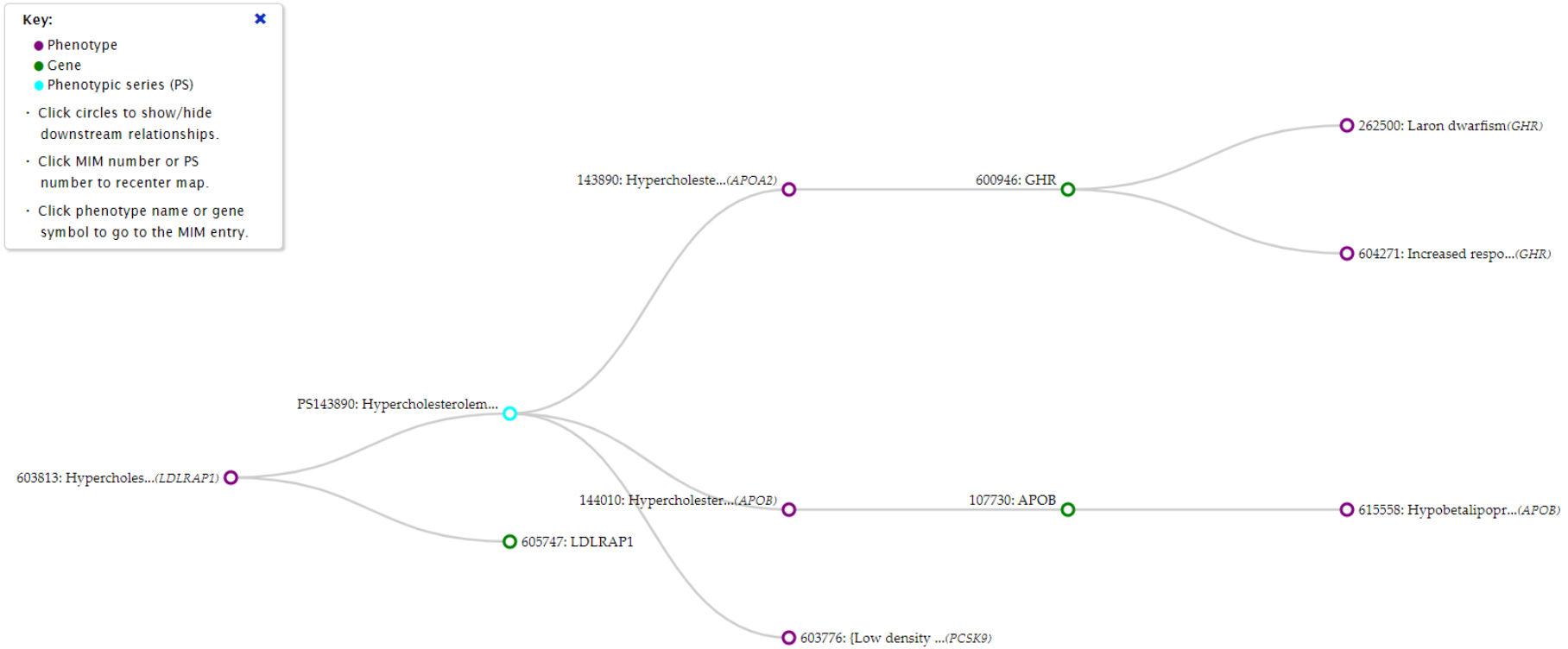
3: # 603776. HYPERCHOLESTEROLEMIA, FAMILIAL, 3; FHCL3
LOW DENSITY LIPOPROTEIN CHOLESTEROL LEVEL QUANTITATIVE TRAIT LOCUS 1, INCLUDED; LDLCQ1, INCLUDED
Cytogenetic locations: 1p32.3,
Matching terms: (hypercholesterolaemia | hypercholesterolemia)
► Phenotype-Gene Relationships ► Phenotypic Series ► Links

OMIM

- Для фенотипа текстовое (!) описание

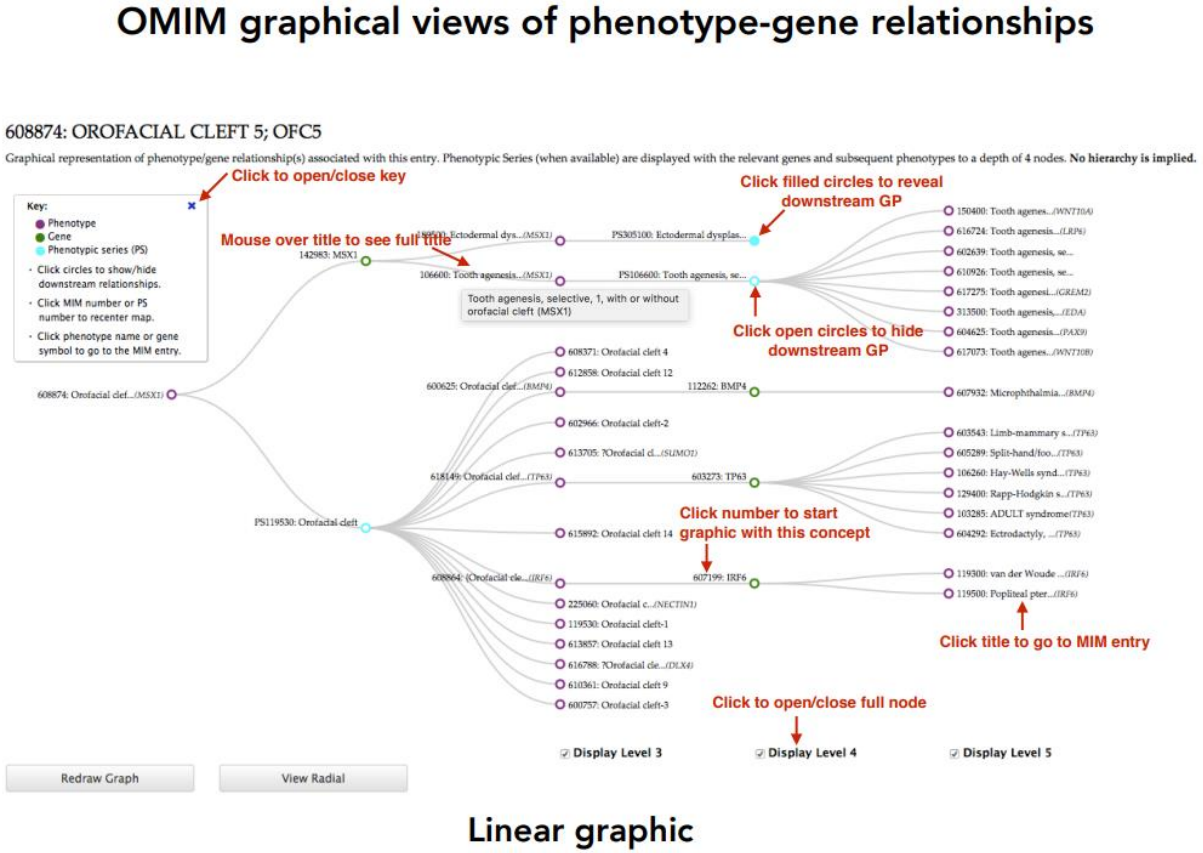


OMIM


Key:
Phenotype
Gene
Phenotypic series (PS)
· Click circles to show/hide downstream relationships.
· Click MIM number or PS number to recenter map.
· Click phenotype name or gene symbol to go to the MIM entry.
603813: Hypercholes...(LDLRAP1)
PS143890: Hypercholesterolem...
143890: Hypercholeste...(APOA2)
600946: GHR
262500: Laron dwarfism(GHR)
604271: Increased respo...(GHR)
144010: Hypercholester...(APOB)
107730: APOB
615558: Hypobetalipopr...(APOB)
605747: LDLRAP1
603776: {Low density ...(PCSK9)

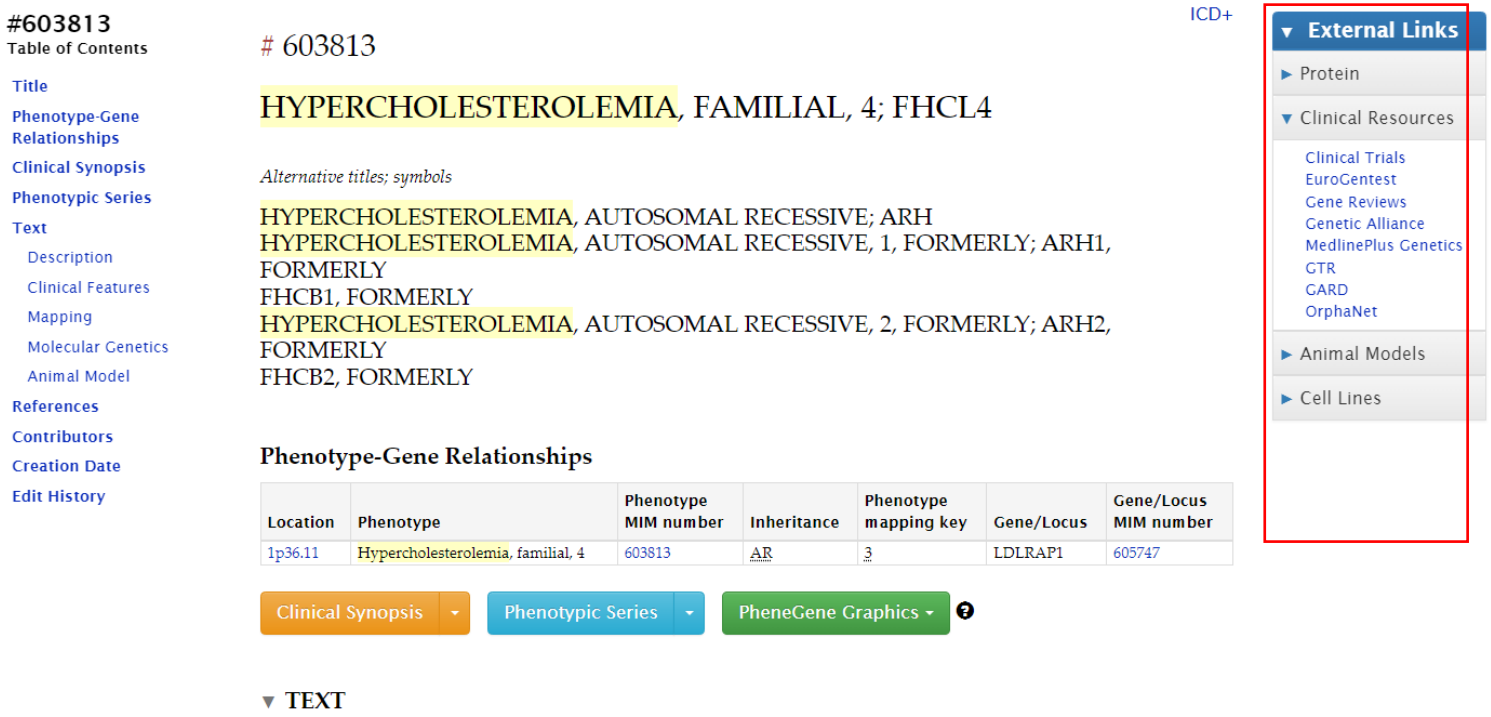
OMIM

- https://www.omim.org/static/omim/pdf/OMIM_graphics.pdf



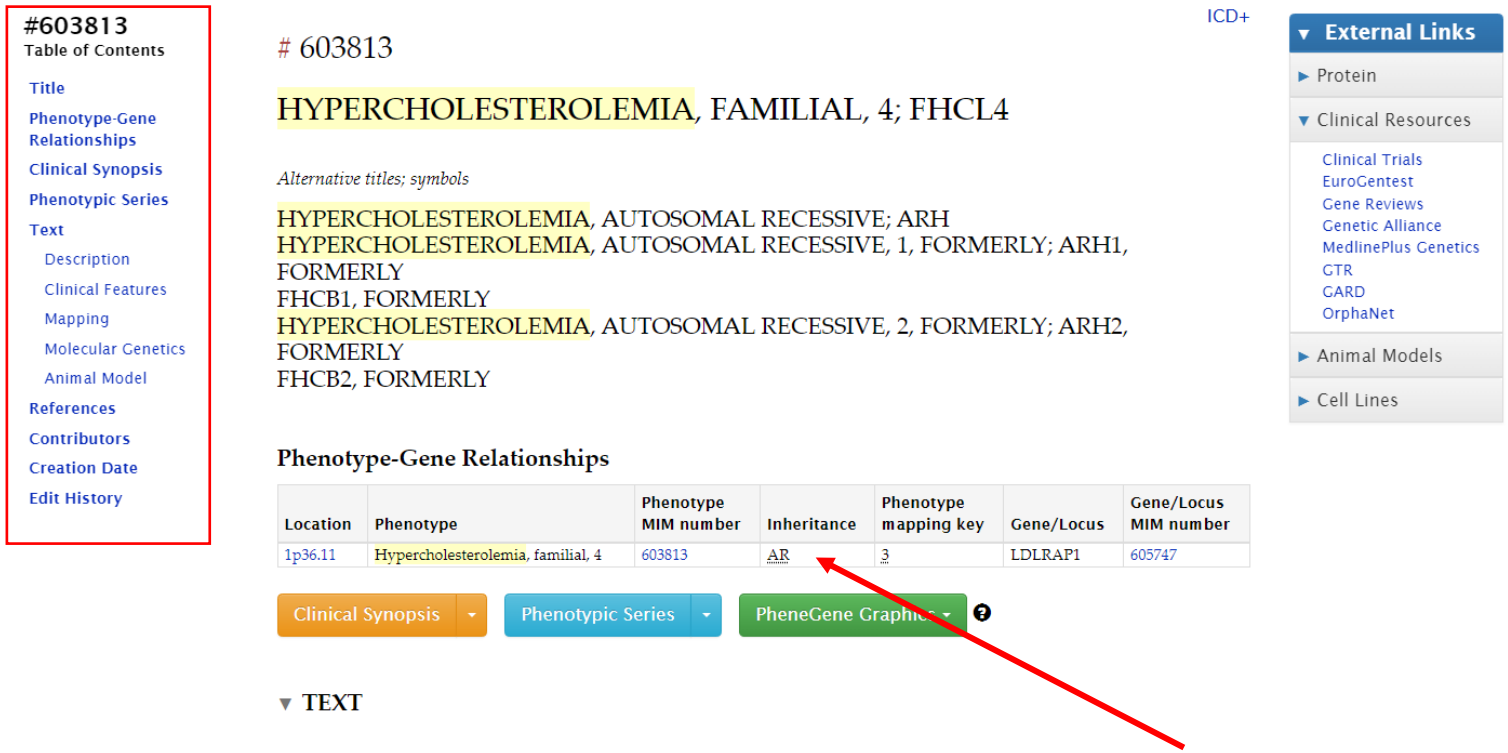
OMIM

- External Links
- Очень много перекрестных ссылок на ресурсы



OMIM

- Для фенотипа текстовое (!) описание



OMIM

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

- Описание фенотипа

▼ **TEXT**

A number sign (#) is used with this entry because autosomal recessive familial hypercholesterolemia-4 (FHCL4) is caused by homozygous or compound heterozygous mutation in the ARH gene (LDLRAP1; 605747) on chromosome 1p36.

▼ **Description**

Autosomal recessive familial hypercholesterolemia-4 (FCHL4) is a rare monogenic disease characterized by very high levels of low-density lipoprotein (LDL) cholesterol (usually above 400 mg/dl) and increased risk of premature atherosclerotic cardiovascular disease (summary by Sanchez-Hernandez et al., 2018).

OMIM

- Посмотрите, как устроен раздел Clinical Features
- Описания клинических случаев с ссылками на источники

Clinical Features

Zuliani et al. (1995) described a consanguineous Sardinian family in which a brother and sister had a severe form of hypercholesterolemia with the clinical features of familial hypercholesterolemia (FH; 143890) homozygotes, including severely elevated plasma low density lipoprotein (LDL) cholesterol, tuberous and tendon xanthomata, and premature atherosclerosis. However, LDL receptor (LDLR; 606945) activity measured in skin fibroblasts was normal, as was LDL binding ability. Haplotype segregation analysis excluded involvement of the LDLR and apolipoprotein B (APOB; 107730) genes in the pathogenesis of the disorder. Consanguinity, absence of vertical transmission, and bimodal distribution of plasma cholesterol levels in the kindred were consistent with autosomal recessive inheritance. Sitosterolemia (see 210250) and pseudohomozygous hyperlipidemia (see 144250) were ruled out.

OMIM

- Выявление геномного локуса, ассоциированного с фенотипом

Mapping

Eden et al. (2001) performed a genomewide scan with polymorphic genetic markers in the 2 families reported by Norman et al. (1999). In both pedigrees, a single region of approximately 12 cM on 1p36-p35, designated FHCB2, fulfilled the criteria for homozygous inheritance of alleles in the affected offspring but not their unaffected sibs. The combined lod score was 5.3 in these unrelated families.

Using 4 ARH families, including 2 previously studied by Zuliani et al. (1995, 1999), Garcia et al. (2001) mapped the ARH locus to a 1-cM interval on chromosome 1p35 extending from D1S1152 to D1S2885. Garcia et al. (2001) identified 6 mutations in a gene encoding a putative adaptor protein (LDLRAP1; 605747) mapping to this region. They found no linkage to 15q25-q26, the locus that Ciccarese et al. (2000) had found to be associated with ARH using one of the same families.

OMIM

- Исследования молекулярно-генетических аспектов фенотипа

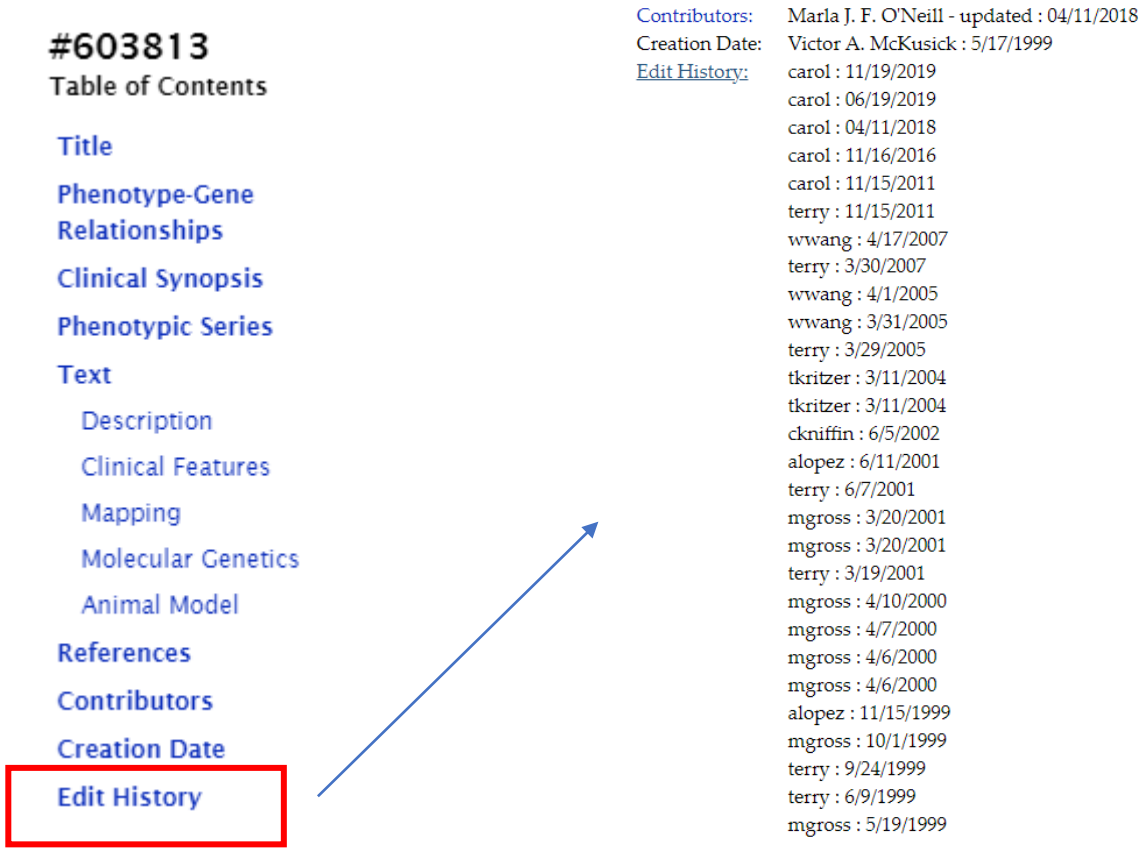
▼ Molecular Genetics

In affected individuals from 6 families with autosomal recessive hypercholesterolemia, including the 2 Sardinian families originally reported by Zuliani et al. (1995) and Zuliani et al. (1999) and a Lebanese family previously described by Khachadurian and Uthman (1973), as well as another Lebanese family, an Iranian family, and an American family, Garcia et al. (2001) identified homozygous mutations in the ARH gene (LDLRAP1; see 605747.0001-605747.0006). The nonsense mutation (W22X; 605747.0001) and 1-bp insertion (605747.0002) that were detected in the 2 original Sardinian families were also identified in homozygosity or compound heterozygosity in 10 additional unrelated Sardinian ARH probands, and neither mutation was found in 50 normolipidemic Sardinians. The authors suggested that the finding of 2 mutations accounting for ARH in 12 Sardinian families represented genetic drift on the island of Sardinia.

Arca et al. (2002) screened the entire coding sequence of LDLRAP1 in 40 unrelated individuals from around the world who had hypercholesterolemia and at least 1 normocholesterolemic parent. They identified 4 Italian probands who were homozygous for the same 1-bp insertion (605747.0002) that had previously been identified in Sardinian patients. No mutations were identified in the other 36 probands.

OMIM

- Информация пополняется новыми исследованиями, наблюдениями и т.п.



#603813
Table of Contents

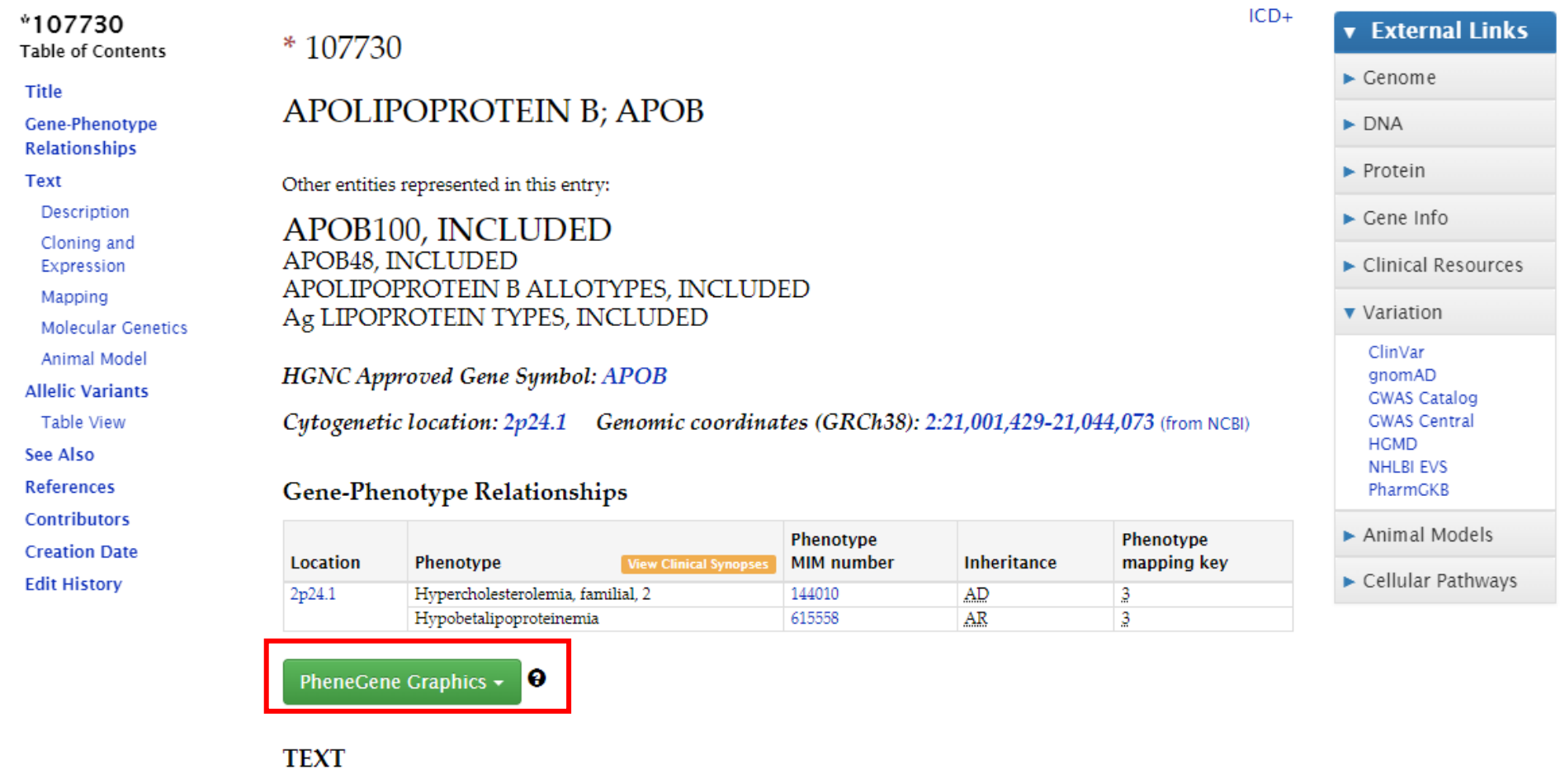
- Title
- Phenotype-Gene Relationships
- Clinical Synopsis
- Phenotypic Series
- Text
  - Description
  - Clinical Features
  - Mapping
  - Molecular Genetics
  - Animal Model
- References
- Contributors
- Creation Date
- Edit History

Contributors: Marla J. F. O'Neill - updated : 04/11/2018
Creation Date: Victor A. McKusick : 5/17/1999
Edit History:
carol : 11/19/2019
carol : 06/19/2019
carol : 04/11/2018
carol : 11/16/2016
carol : 11/15/2011
terry : 11/15/2011
wwang : 4/17/2007
terry : 3/30/2007
wwang : 4/1/2005
wwang : 3/31/2005
terry : 3/29/2005
tkritzer : 3/11/2004
tkritzer : 3/11/2004
ckniffin : 6/5/2002
alopez : 6/11/2001
terry : 6/7/2001
mgross : 3/20/2001
mgross : 3/20/2001
terry : 3/19/2001
mgross : 4/10/2000
mgross : 4/7/2000
mgross : 4/6/2000
mgross : 4/6/2000
alopez : 11/15/1999
mgross : 10/1/1999
terry : 9/24/1999
terry : 6/9/1999
mgross : 5/19/1999

Упражнение

- Найдите в базе данных OMIM ген APOB
- Обсудите структуру результата поиска и записи, аналогичные поиску по заболеваниям

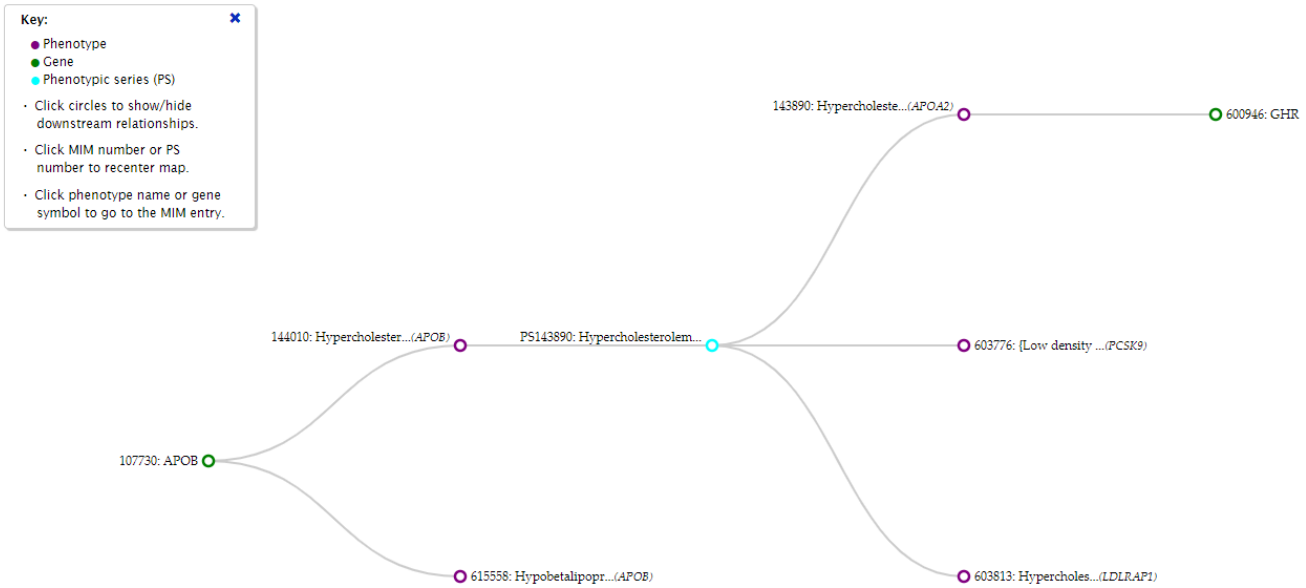
OMIM


*107730
Table of Contents
Title
Gene-Phenotype Relationships
Text
Description
Cloning and Expression
Mapping
Molecular Genetics
Animal Model
Allelic Variants
Table View
See Also
References
Contributors
Creation Date
Edit History
* 107730
APOLIPOPROTEIN B; APOB
Other entities represented in this entry:
APOB100, INCLUDED
APOB48, INCLUDED
APOLIPOPROTEIN B ALLOTYPES, INCLUDED
Ag LIPOPROTEIN TYPES, INCLUDED
HGNC Approved Gene Symbol: APOB
Cytogenetic location: 2p24.1 Genomic coordinates (GRCh38): 2:21,001,429-21,044,073 (from NCBI)
Gene-Phenotype Relationships
Location
Phenotype
View Clinical Synopses
Phenotype MIM number
Inheritance
Phenotype mapping key
2p24.1
Hypercholesterolemia, familial, 2
144010
AD
3
Hypobetalipoproteinemia
615558
AR
3
PheneGene Graphics
TEXT
ICD+
External Links
Genome
DNA
Protein
Gene Info
Clinical Resources
Variation
ClinVar
gnomAD
GWAS Catalog
GWAS Central
HGMD
NHLBI EVS
PharmGKB
Animal Models
Cellular Pathways

OMIM

107730:

Graphical representation of phenotype/gene relationship(s) associated with this entry. Phenotypic Series (when available) are displayed with the relevant genes and subsequent phenotypes to a depth of 4 nodes. A quick reference overview and guide (PDF). **No hierarchy is implied.** Feedback



OMIM

*107730
Table of Contents

ICD+

* 107730

APOLIPOPROTEIN B; APOB

Other entities represented in this entry:

APOB100, INCLUDED
APOB48, INCLUDED
APOLIPOPROTEIN B ALLOTYPES, INCLUDED
Ag LIPOPROTEIN TYPES, INCLUDED

HGNC Approved Gene Symbol: APOB

Cytogenetic location: 2p24.1 Genomic coordinates (GRCh38): 2:21,001,429-21,044,073 (from NCBI)

Gene-Phenotype Relationships

Location	Phenotype View Clinical Synopses	Phenotype MIM number	Inheritance	Phenotype mapping key
2p24.1	Hypercholesterolemia, familial, 2	144010	AD	3
	Hypobetalipoproteinemia	615558	AR	3

PheneGene Graphics

TEXT

External Links
- Genome
- DNA
- Protein
- Gene Info
- Clinical Resources
- Variation
  - ClinVar
  - gnomAD
  - GWAS Catalog
  - GWAS Central
  - HGMD
  - NHLBI EVS
  - PharmGKB
- Animal Models
- Cellular Pathways

OMIM

Human Genetics Knowledge for the World

Каждая запись – текстовое описание варианта с ссылкой на публикацию и перекрестными ссылками на записи в OMIM (например, для фенотипа)

APOLIPOPROTEIN B; APOB

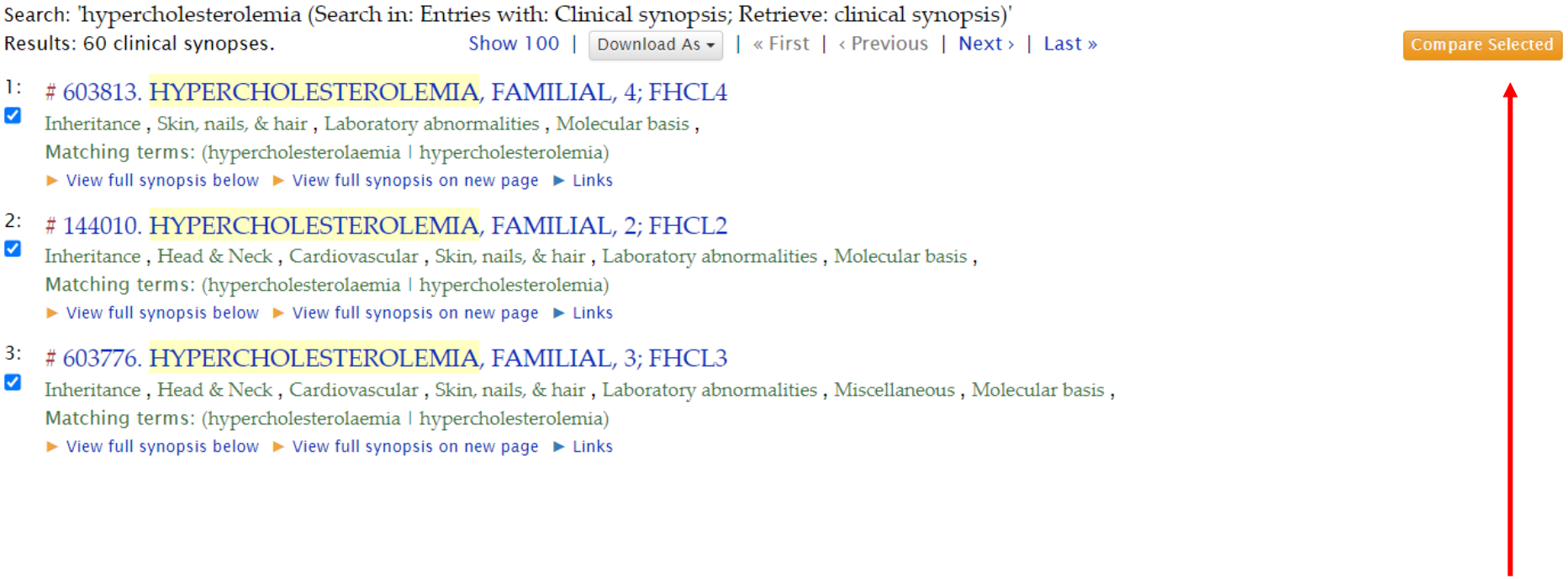
Allelic Variants (22 Selected Examples) :

All ClinVar Variants

Number	Phenotype	Mutation	SNP	gnomAD	ClinVar
.0001	HYPOBETALIPOPROTEINEMIA, FAMILIAL	APOB, 4-BP DEL, NT5391	rs281865425	-	RCV000019470...
.0002	HYPOBETALIPOPROTEINEMIA, FAMILIAL, ASSOCIATED WITH APOB39	APOB, 1-BP DEL, FS1799TER	rs397514255	rs397514255	RCV000019471
.0003	HYPOBETALIPOPROTEINEMIA, FAMILIAL	APOB, ARG1306TER	rs121918383	rs121918383	RCV000019472...
.0004	HYPOBETALIPOPROTEINEMIA, FAMILIAL, ASSOCIATED WITH APOB40	APOB, VAL1829CYS	rs121918384	rs121918384	RCV000019473...
.0005	HYPOBETALIPOPROTEINEMIA, FAMILIAL, ASSOCIATED WITH APOB90 OR APOB89	APOB, GLU4034ARG	rs121918385	-	RCV000019474
.0006	HYPOBETALIPOPROTEINEMIA, FAMILIAL, ASSOCIATED WITH APOB46	APOB, ARG2058TER	rs121918386	rs121918386	RCV000019476...
.0007	HYPOBETALIPOPROTEINEMIA, FAMILIAL, ASSOCIATED WITH APOB87	APOB, 1-BP DEL, 12032G	rs387906569	rs387906569	RCV000019477
.0008	HYPOBETALIPOPROTEINEMIA, FAMILIAL, ASSOCIATED WITH APOB31	APOB, 1-BP DEL, 1425G	rs397514256	rs397514256	RCV000019478
.0009	HYPERCHOLESTEROLEMIA, FAMILIAL, 2	APOB, ARG3500GLN	rs5742904	rs5742904	RCV000019479...
.0010	HYPOBETALIPOPROTEINEMIA, FAMILIAL	APOB, EX21DEL	-	-	RCV000019475
.0011	APOB POLYMORPHISM IN SIGNAL PEPTIDE	APOB, INS AND DEL	-	-	RCV000251913...
.0012	HYPOBETALIPOPROTEINEMIA, FAMILIAL	APOB, LEU3041TER	rs121918387	-	RCV000019481...
.0013	HYPOBETALIPOPROTEINEMIA, NORMOTRIGLYCERIDEMIC	APOB, GLN2252TER	rs121918388	rs121918388	RCV001837438
.0014	HYPOBETALIPOPROTEINEMIA, FAMILIAL, ASSOCIATED WITH APOB32	APOB, GLN1450TER	rs121918389	rs121918389	RCV000019483
.0015	HYPOBETALIPOPROTEINEMIA, FAMILIAL	APOB, ARG2495TER	rs121918390	rs121918390	RCV000019484...
.0016	HYPOBETALIPOPROTEINEMIA, FAMILIAL	APOB, 1-BP DEL, NT11840	rs587776852	rs587776852	RCV000019485...
.0017	HYPERCHOLESTEROLEMIA, FAMILIAL, 2	APOB, ARG3531CYS	rs12713559	rs12713559	RCV000019486...
.0018	HYPOBETALIPOPROTEINEMIA, FAMILIAL	APOB, IVS7AS, A-G, -2	rs1572800245	-	RCV000019487...
.0019	HYPOBETALIPOPROTEINEMIA, FAMILIAL	APOB, 1-BP DEL, 4432T	-	-	RCV000019488
.0020	HYPOBETALIPOPROTEINEMIA, NORMOTRIGLYCERIDEMIC	APOB, 4-BP DEL, NT36491	-	-	RCV001837441
.0021	HYPOBETALIPOPROTEINEMIA, NORMOTRIGLYCERIDEMIC	APOB, TYR1173TER	rs121918391	-	RCV001837442...
.0022	HYPOBETALIPOPROTEINEMIA, FAMILIAL	APOB, 2-BP INS, 825GG	rs606231236	-	RCV000032601

OMIM

- Сравнить несколько записей



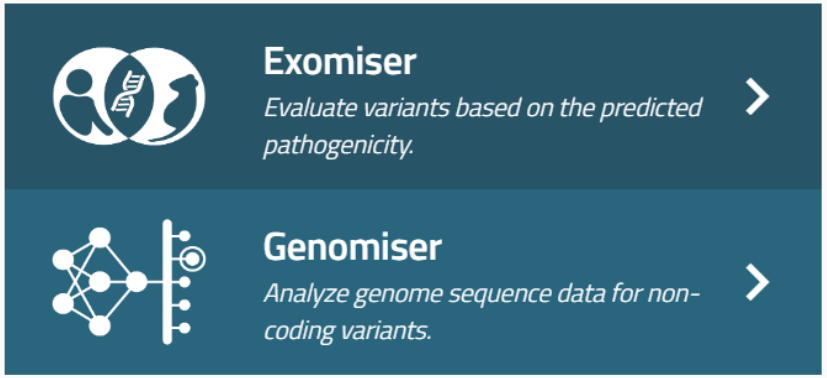
OMIM

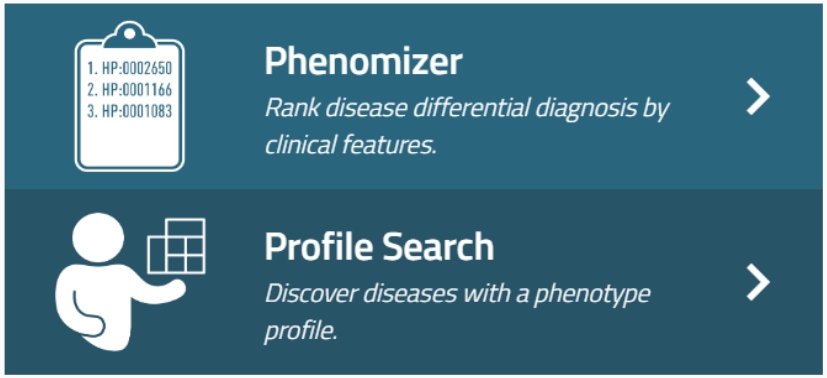
- Результат сравнения

NUMBER	# 603813 ▼	# 144010 ▼	# 603776 ▼
TITLE	HYPERCHOLESTEROLEMIA, FAMILIAL, 4; FHCL4	HYPERCHOLESTEROLEMIA, FAMILIAL, 2; FHCL2	HYPERCHOLESTEROLEMIA, FAMILIAL, 3; FHCL3
GENE	*LDLRAP1* - 605747	*APOB* - 107730	*PCSK9* - 607786
INHERITANCE (in 3/3)	- Autosomal recessive	- Autosomal dominant	- Autosomal dominant
HEAD & NECK (in 2/3) ▼		*Eyes* - Corneal arcus - Xanthelasma	*Eyes* - Arcus corneae
CARDIOVASCULAR (in 2/3) ▼		*Heart* - Coronary artery disease	*Heart* - Coronary artery disease
SKIN, NAILS, & HAIR (in 3/3) ▼	*Skin* - Xanthomas	*Skin* - Tendinous xanthomas - Planar xanthomas (in homozygotes)	*Skin* - Xanthelasmas - Tendinous xanthomata
LABORATORY ABNORMALITIES (in 3/3) ▼	- Hypertriglyceridemia - Very high low-density lipoprotein (LDL) cholesterol (>400 mg/dL) - High total cholesterol (>600 mg/dL)	- Hypercholesterolemia - Abnormal LDL	- High total cholesterol High LD cholesterol
MISCELLANEOUS (in 1/3) ▼			- Elevated cholesterol levels evident before age 20
MOLECULAR BASIS (in 3/3) ▼	- Caused by mutation in the low density lipoprotein receptor adaptor protein 1 gene (LDLRAP1, 605747.0001)	- Caused by mutation in the apolipoprotein B gene (APOB, 107730.0001)	- Caused by mutation in the proprotein convertase, subtilisin/kexin-type, 9 gene (PCSK9, 607786.0001)

HPO

- https://hpo.jax.org/app/





HPO

- Поиск от
  - Фенотипа
  - Заболевания
  - Гена

Упражнение

- Найдите в базе HPO гиперхолестеринемию по OMIM ID: 603813

HPO

Hypercholesterolemia, autosomal recessive OMIM:603813 MONDO:0011374

An autosomal recessive condition caused by mutation(s) in the LDLRAP1 gene, encoding low density lipoprotein receptor adaptor protein 1. The phenotype is similar to that of familial hypercholesterolemia, but generally considered to be a milder form of hypercholesterolemia.

Export Associations | Report Entry Issue

HPO Associations | Gene Associations

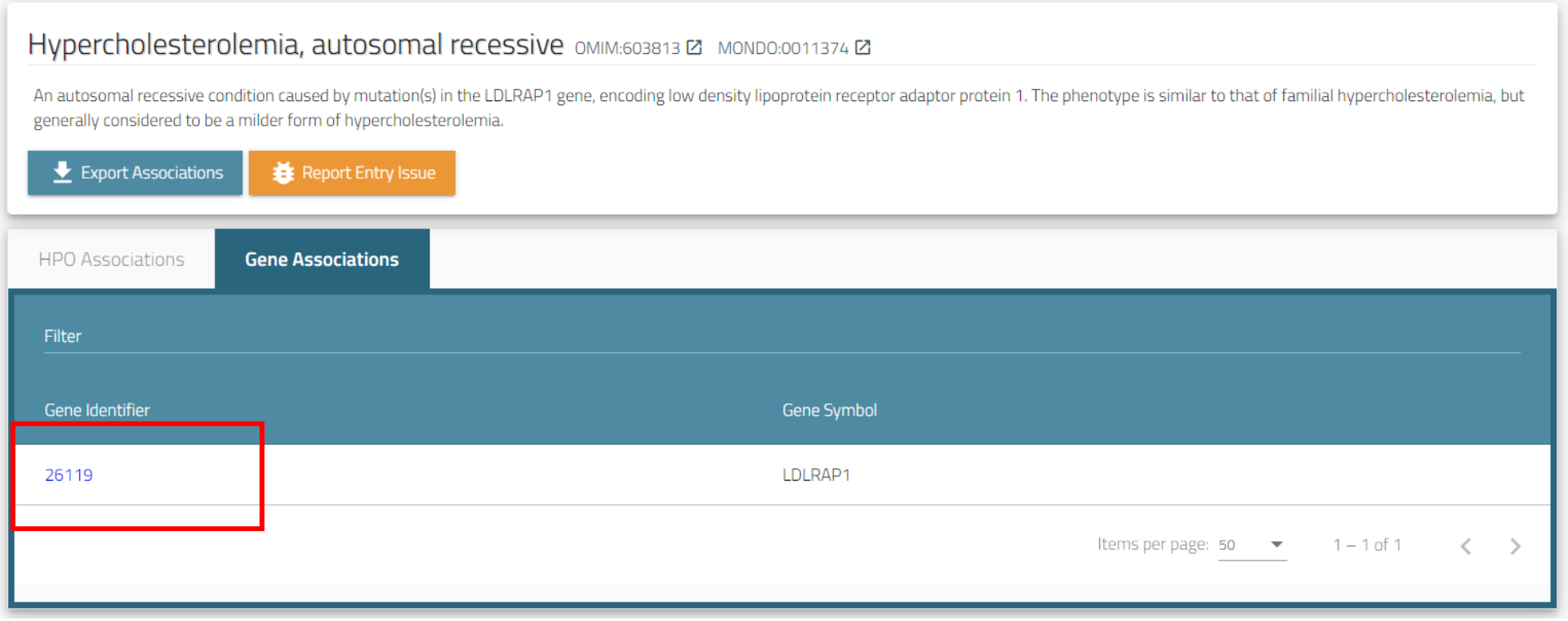
Inheritance [1 annotation]

Term Identifier	Term Name	Onset	Frequency	Source(s)
HP:0000007	Autosomal recessive inheritance			PubMed

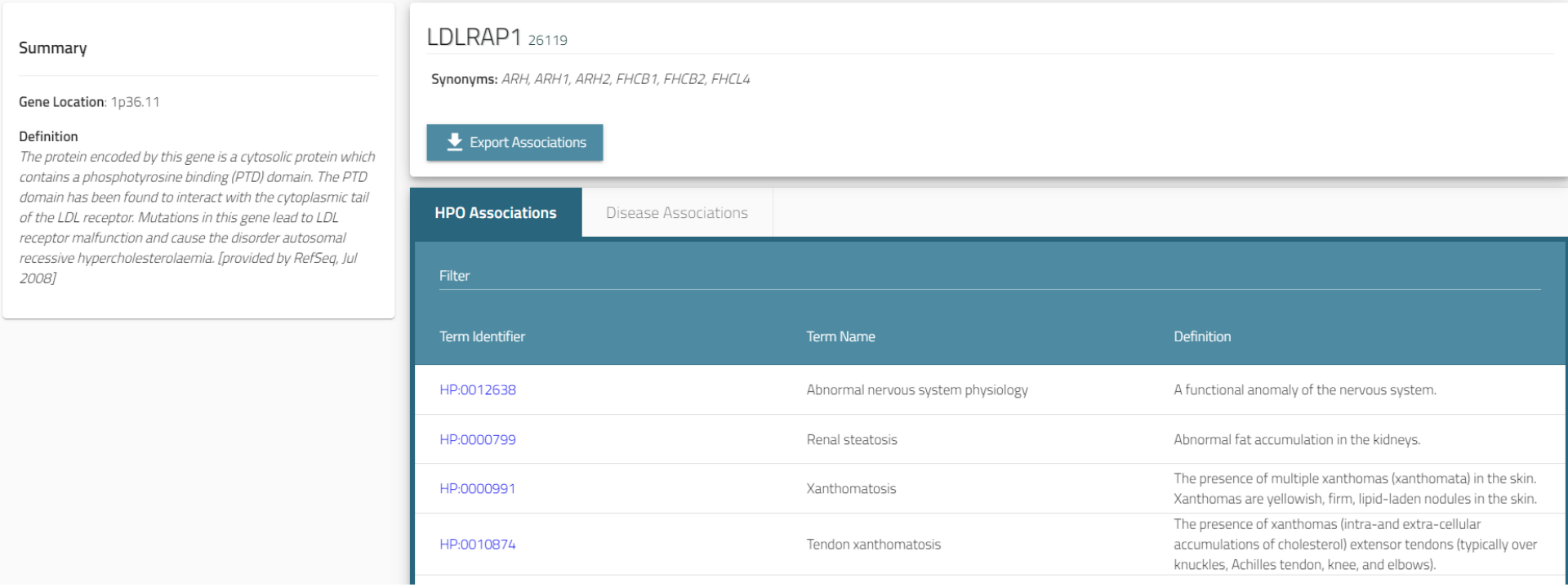
Cardiovascular [1 annotation]

Term Identifier	Term Name	Onset	Frequency	Source(s)
HP:0002621	Atherosclerosis			OMIM

HPO


Hypercholesterolemia, autosomal recessive OMIM:603813 MONDO:0011374
An autosomal recessive condition caused by mutation(s) in the LDLRAP1 gene, encoding low density lipoprotein receptor adaptor protein 1. The phenotype is similar to that of familial hypercholesterolemia, but generally considered to be a milder form of hypercholesterolemia.
Export Associations
Report Entry Issue
HPO Associations
Gene Associations
Filter
Gene Identifier
Gene Symbol
26119
LDLRAP1
Items per page: 50
1 – 1 of 1

HPO



Summary

Gene Location: 1p36.11

Definition

The protein encoded by this gene is a cytosolic protein which contains a phosphotyrosine binding (PTD) domain. The PTD domain has been found to interact with the cytoplasmic tail of the LDL receptor. Mutations in this gene lead to LDL receptor malfunction and cause the disorder autosomal recessive hypercholesterolaemia. [provided by RefSeq, Jul 2008]

LDLRAP1 26119

Synonyms: *ARH, ARH1, ARH2, FHCB1, FHCB2, FHCL4*

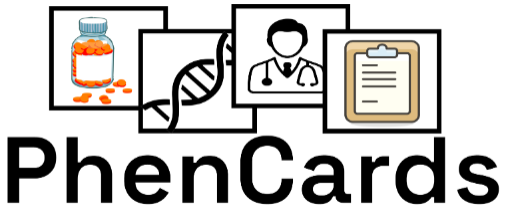
Export Associations

HPO Associations | Disease Associations

Filter

Term Identifier	Term Name	Definition
HP:0012638	Abnormal nervous system physiology	A functional anomaly of the nervous system.
HP:0000799	Renal steatosis	Abnormal fat accumulation in the kidneys.
HP:0000991	Xanthomatosis	The presence of multiple xanthomas (xanthomata) in the skin. Xanthomas are yellowish, firm, lipid-laden nodules in the skin.
HP:0010874	Tendon xanthomatosis	The presence of xanthomas (intra-and extra-cellular accumulations of cholesterol) extensor tendons (typically over knuckles, Achilles tendon, knee, and elbows).

PhenCards



- https://phencards.org/
- PhenCards is a web server for linking human phenotype information to biomedical knowledge

Упражнение

- Найдите в PhenCards ген, заболевание или фенотип
- Обратите внимание на выдачу
- Обсудите, ссылки на какие ресурсы выведены в результате поиска

Mondo

- https://www.ebi.ac.uk/ols4/ontologies/mondo
- слайды

Mondo Disease Ontology

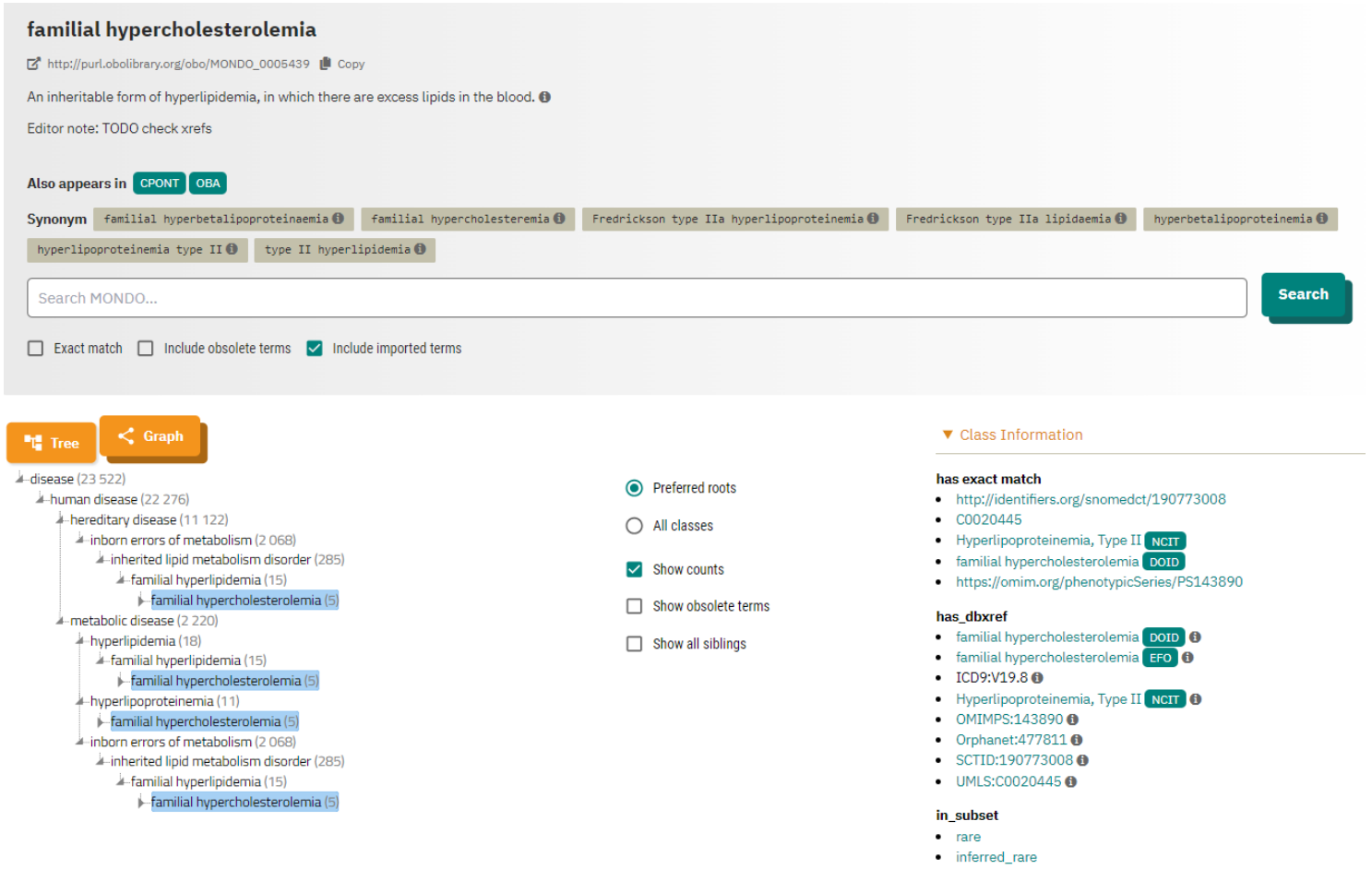
Version 2024-02-06

A semi-automatically constructed ontology that merges in multiple disease resources to yield a coherent merged ontology.

Disease term feature	Count
Total number of terms	22,157
Database cross references	104,479
Term definitions	15,443
Exact synonyms	66,247
Related synonyms	30,661
Narrow (more specific) synonyms	2,214
Broad (more general) synonyms	847

Disease type	Count (Concepts)
Rare diseases	10,443
Infectious diseases	1,240
Cancers (including neoplasms)	4,298
Mendelian diseases	11,380

Mondo


familial hypercholesterolemia
http://purl.obolibrary.org/obo/MONDO_0005439 Copy
An inheritable form of hyperlipidemia, in which there are excess lipids in the blood.
Editor note: TODO check xrefs
Also appears in CPONT OBA
Synonym familial hyperbetalipoproteinaemia familial hypercholesteremia Fredrickson type IIa hyperlipoproteinemia Fredrickson type IIa lipidaemia hyperbetalipoproteinemia hyperlipoproteinemia type II type II hyperlipidemia
Search MONDO...
Search
Exact match
Include obsolete terms
Include imported terms
Tree
Graph
disease (23 522)
human disease (22 276)
hereditary disease (11 122)
inborn errors of metabolism (2 068)
inherited lipid metabolism disorder (285)
familial hyperlipidemia (15)
familial hypercholesterolemia (5)
metabolic disease (2 220)
hyperlipidemia (18)
familial hyperlipidemia (15)
familial hypercholesterolemia (5)
hyperlipoproteinemia (11)
familial hypercholesterolemia (5)
inborn errors of metabolism (2 068)
inherited lipid metabolism disorder (285)
familial hyperlipidemia (15)
familial hypercholesterolemia (5)
Preferred roots
All classes
Show counts
Show obsolete terms
Show all siblings
Class Information
has exact match
http://identifiers.org/snomedct/190773008
C0020445
Hyperlipoproteinemia, Type II NCIT
familial hypercholesterolemia DOID
https://omim.org/phenotypicSeries/PS143890
has_dbxref
familial hypercholesterolemia DOID
familial hypercholesterolemia EFO
ICD9:V19.8
Hyperlipoproteinemia, Type II NCIT
OMIMPS:143890
Orphanet:477811
SCTID:190773008
UMLS:C0020445
in_subset
rare
inferred_rare

Mondo

Mondo uses the following ontologies as sources or for cross-references (xrefs)/alignments.

Source	ID Space/URI Prefix	Role	Website
OMIM Phenotypes	OMIM	Source	www.omim.org
OMIM Phenotypic Series	OMIMPS	Source	www.omim.org
Orphanet	Orpha	Source	https://www.orpha.net/consor/cgi-bin/index.php
SNOMED (disorder subset)	SCTID	xref/Alignments	www.snomed.org
National Cancer Institute Thesaurus (disease/disorder subset)	NCIT	Source	https://ncit.nci.nih.gov/ncitbrowser/
Genetic and Rare Diseases Information Center	GARD	Source	https://rarediseases.info.nih.gov/
Medical Subject Headings	MESH	xref/Alignments	https://id.nlm.nih.gov/mesh/
Unified Medical Language System	UMLS	xref/Alignments	https://www.nlm.nih.gov/research/umls/index.html
ICD - ICD-9 - International Classification of Diseases	ICD9	xref/Alignments	https://www.cdc.gov/nchs/icd/icd9.htm
ICD - ICD-10 - International Classification of Diseases	ICD10	xref/Alignments	https://www.cdc.gov/nchs/icd/icd10cm.htm
Experimental Factor Ontology	EFO	xref/Alignments*	https://www.ebi.ac.uk/efo/
Disease Ontology	DO	Source	http://www.obofoundry.org/ontology/doid.html
Mental Functioning Ontology	MF	Source	http://www.obofoundry.org/ontology/mf.html
MedGen	MEDGEN	xref/Alignments	https://www.ncbi.nlm.nih.gov/medgen/
Ontology for General Medical Science	OGMS	xref/Alignments	https://github.com/OGMS/ogms
Medical Dictionary for Regulatory Activities	MeDRA	xref/Alignments	https://www.meddra.org/
OncoTree	ONCOTREE	xref/Alignments	http://oncotree.mskcc.org/#/home

ЗАДАНИЕ

- Выберите себе заболевание для работы с ним в семестре
- Зафиксируйте свой выбор в таблице до 11 марта 2024 включительно

- В конце семестра нужно будет написать отчет