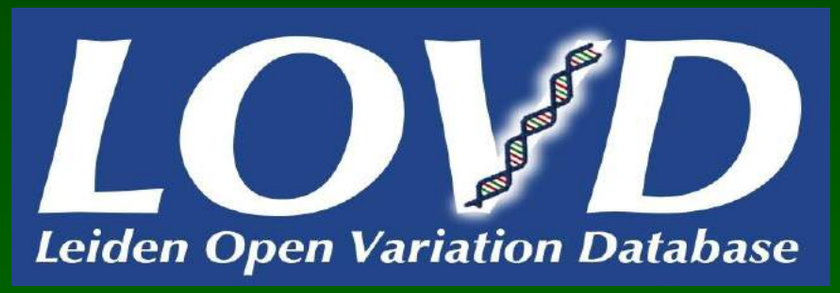
Gene variant databases

sharing information

(LSDBs)

Johan den Dunnen



My affiliations

THE HUMAN VARIOME PROJECT

get all variants/consequences found easily accessible

HGVS HUMAN GENOME VARIATION SOCIETY

standards for variant description and databases

LOVD Leiden Open Variation Database

software for web-based gene databases

L U M C

DNA diagnostics

..is based on

SHARING *what we know on the* ***relation***
between ***variants in genes*** *&* ***phenotypes***

without sharing, no DNA diagnostics

..do we share ?
..are databases supported ?

DNA diagnostics

- **the work**

analyse sample (sequence)
identify variants

check what others found
draw initial conclusion
..perform additional experiments
RNA, protein, cells, computational, animal model, ..
plan to publish results

share with colleagues > submit to database

DNA diagnostics

- **the work**

analyse sample (sequence)
identify variants

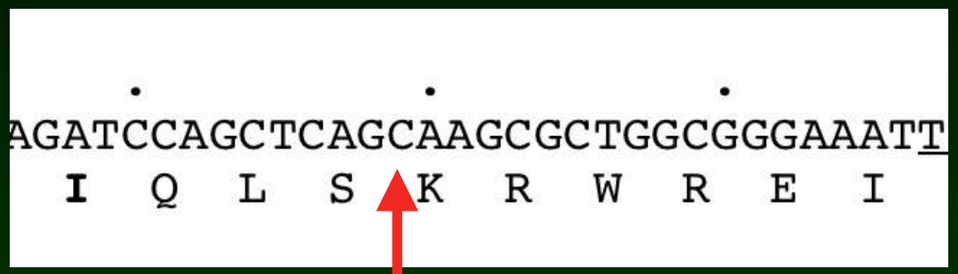
share with colleagues > submit to database

check what others found
draw initial conclusion
..perform additional experiments
RNA, protein, cells, computational, animal model, ..
plan to publish results

It seems so simple...

Example

DMD gene



c.5859C>T r.(?) p(Ser1953=)

found in diagnosis

prenatal
at risk family for DMD
no definite diagnosis

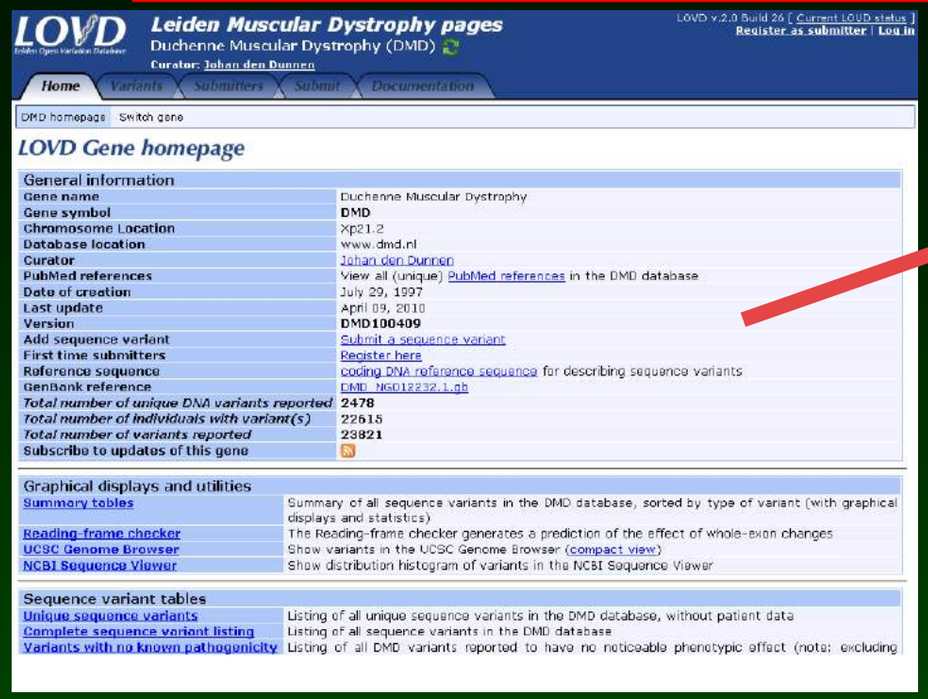
found in diagnosis

WES, trio analysis
male parent
45y, healthy

you may have life saving information,
do you realize this??

©Johan vd Leij

...privacy



LMNA : c.?

...we have identified a variant in this gene in a patient with a certain phenotype.

For privacy reasons we are not able to share more information.

For details please contact us.

VUS

Variant of Unsufficient Sharing

Organisations

- **HVP & HGVS**
  - ***Human Variome Project***
  - ***Human Genome Variation Society***
    - umbrella organisations promoting collecting genome variation & establishing standards

- **GA4GH**
  - ***Global Alliance for Genomics and Health***
    - recent initiative, similar goals
    - active on many aspects

- **EBI & NCBI**
  - ***reference sequences***
  - ***genome browsers***
  - ***variant databases***

Databases require?

- **standard to describe phenotypes**
  HPO (Human Phenotype Ontology)

- **standard to describe variants in DNA**
  HGVS nomenclature

- **databases**
  sharing information

DNA variant DBs

- **central databases**

inch deep, mile wide

HGMD

lists first published report > no frequencies
pathogenic only (most)

OMIM

list firsts report(s) & some interesting cases

dbSNP, EVA

all variants (originally mainly non pathogenic)

many others

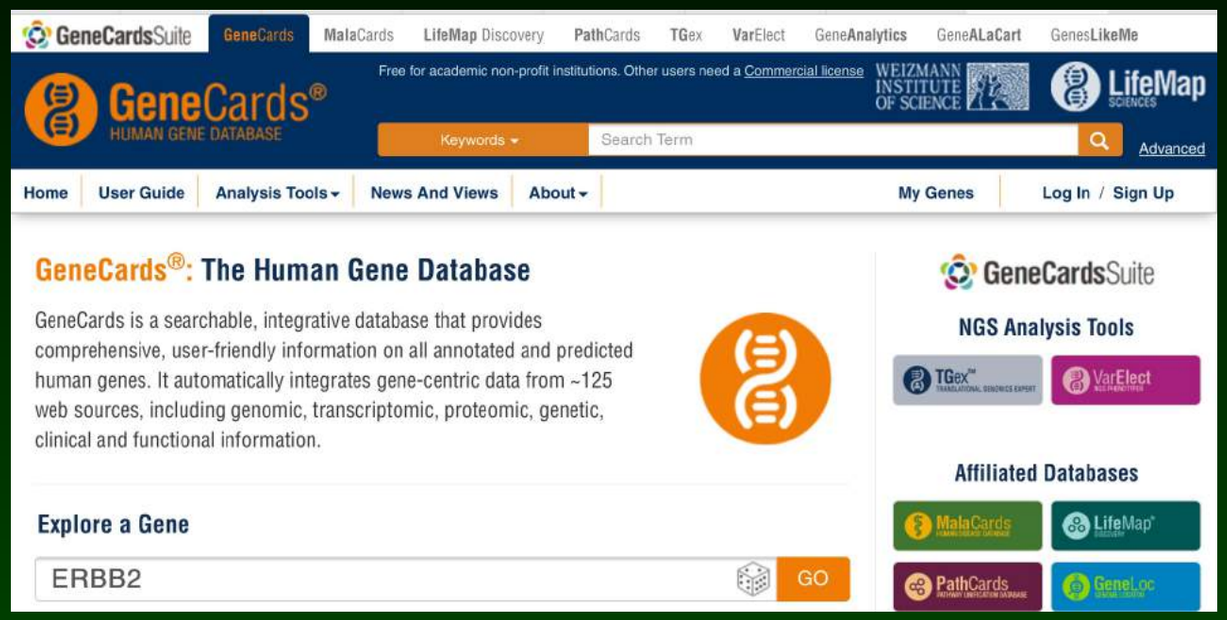
COSMIC, DBVar, Decipher, ...

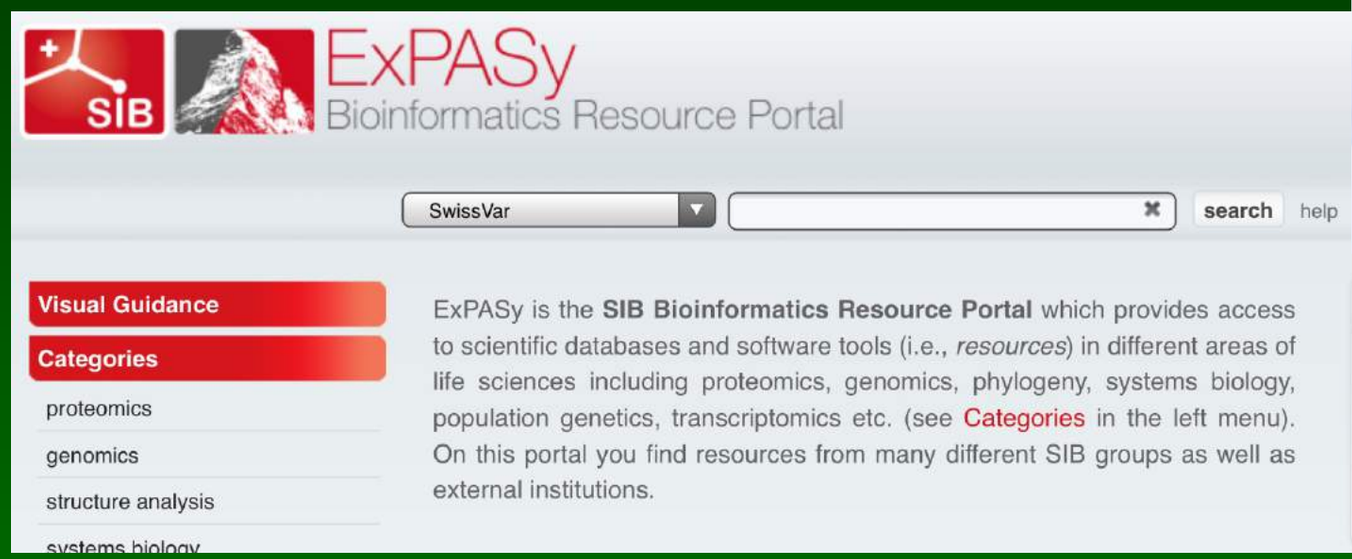
- **gene variant databases**

all details per gene
all variants
unpublished data from direct DB submission

inch wide, mile deep

Other databases

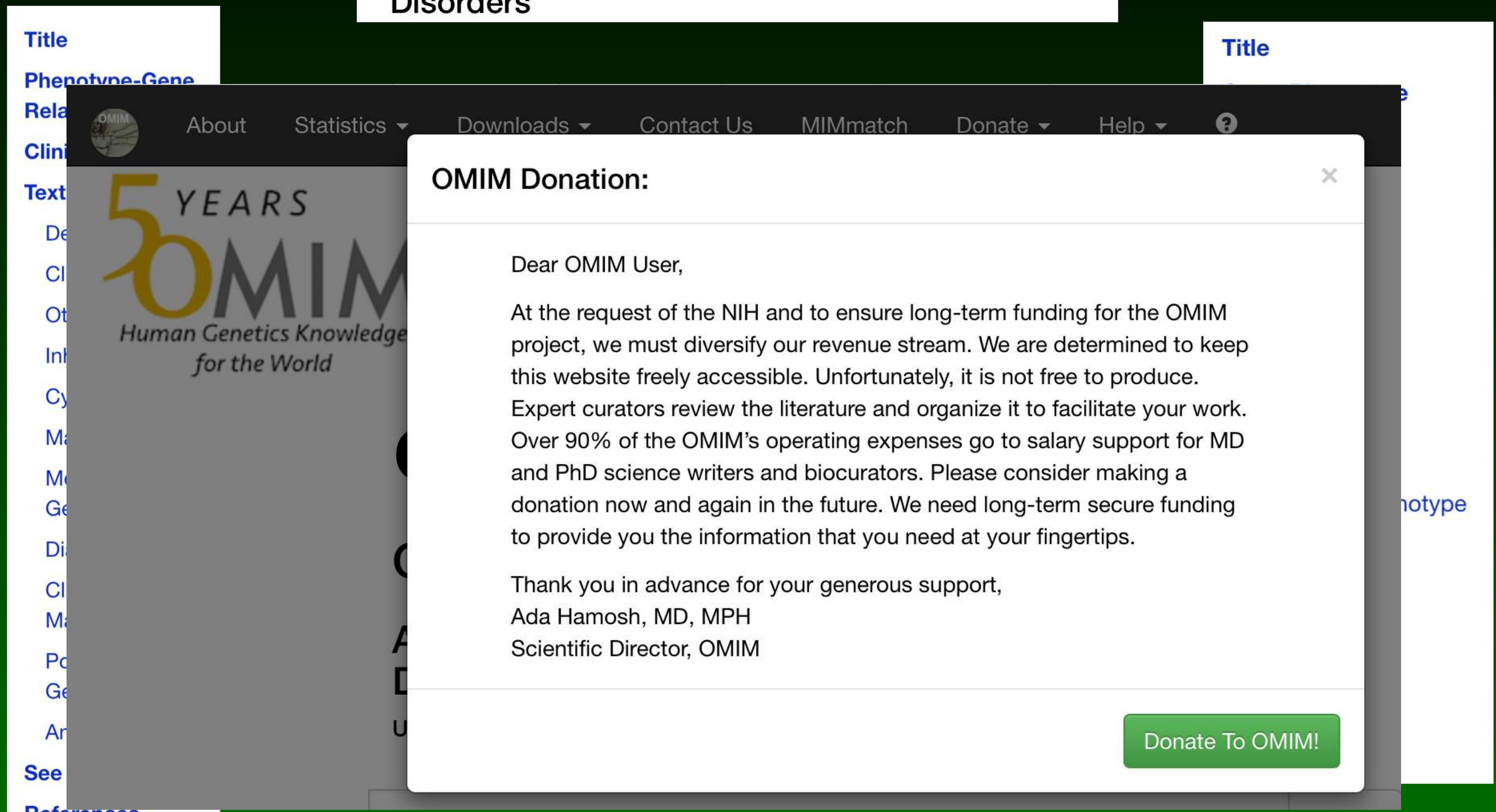




OMIM®

Online Mendelian Inheritance in Man®

An Online Catalog of Human Genes and Genetic Disorders



Definition

Locus Specific DataBase (LSDB)

A listing of sequence variants in a specific gene causing a Mendelian disorder or a change in phenotype, curated by an expert in that gene

(gene variant database)

LOVD
Leiden Open Variation Database

MutaDATABASE

UMD
The Universal Mutation Database

MUTbase

CCAAGAGATATATCT
TCACTTAGACCTCAC
AAGTCAGGGCAGAGC
CTCCTGAGGAGAAGT
GGTTACAAGACAGGT
TGCCTATTGGTCTAT

ClinVar

ClinVar aggregates inf

LSDBs

- **gene variant databases** ***(LSDBs)***

use HGVS description variants
link to other relevant sources
store ALL variants
from publications AND direct submissions
detailed phenotype data (...often limited)
active curator searching, asking, adding, ...

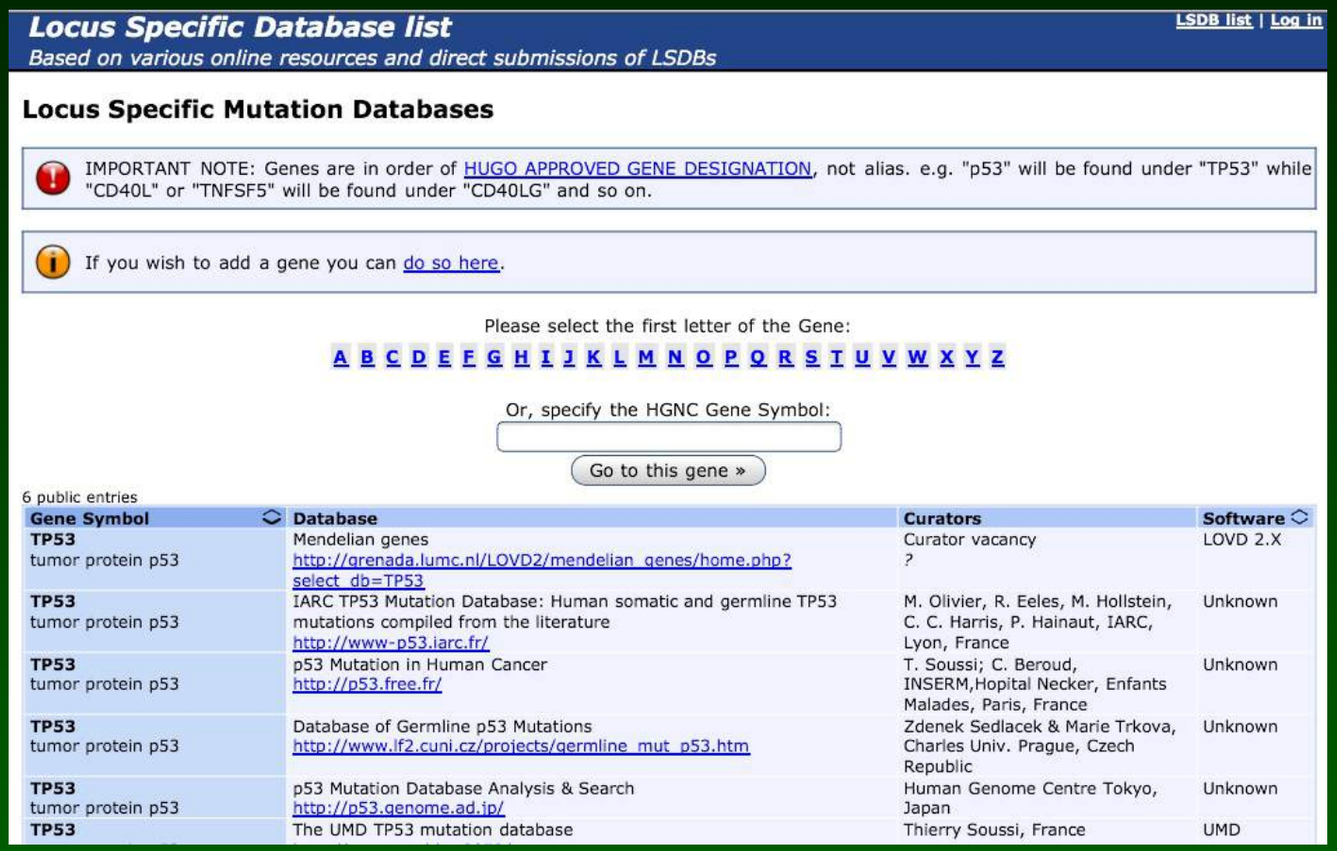
variable formats / quality / updating

A hit means data on consequences for function are available

...but not a DB for all genes

Is there a DB?

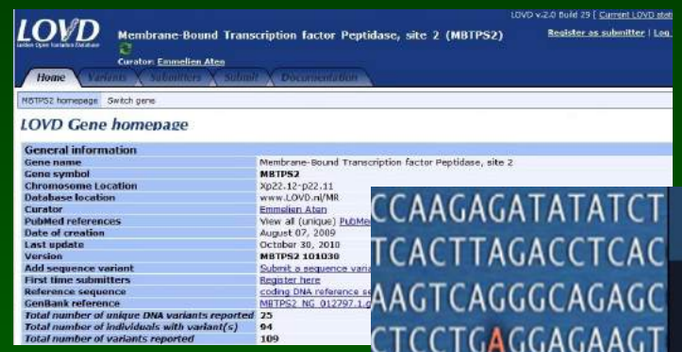
try
GeneSymbol .variome.org

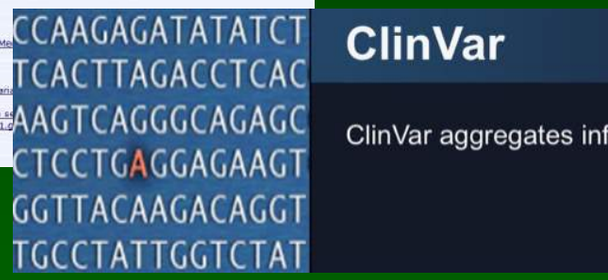


TP53.variome.org

2 second acknowledgement

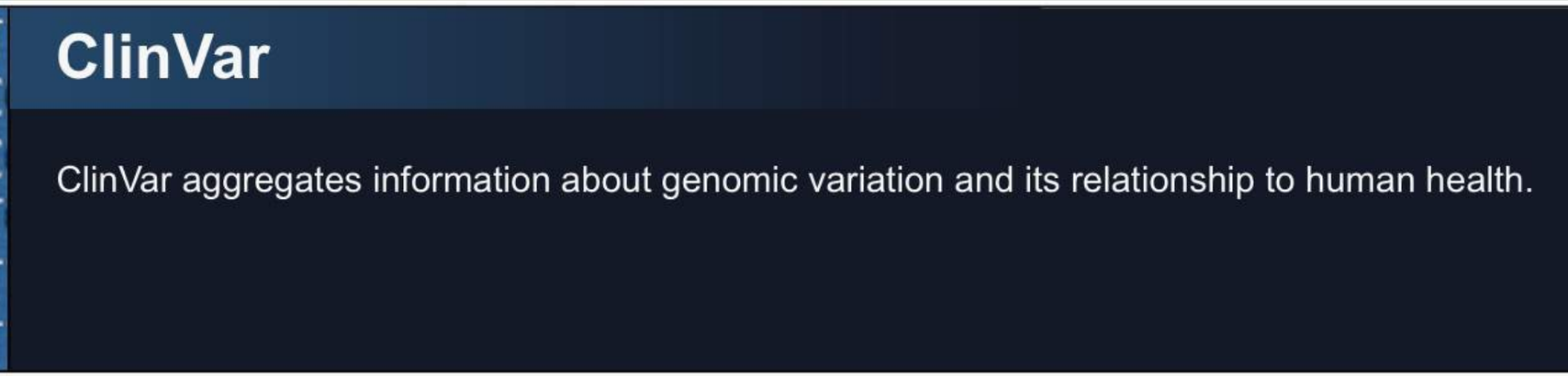
always a hit in
LOVD and ClinVar



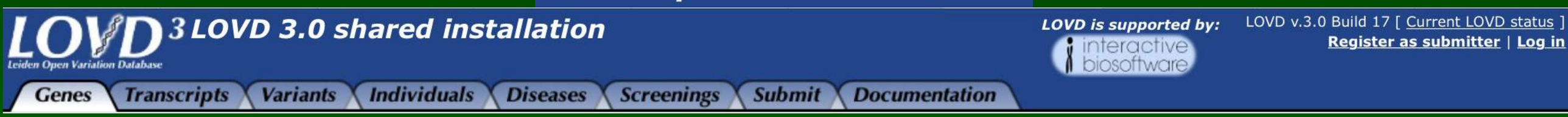


when not ...?

Collaboration



collaboration initiated, exchange data



approaching other databases to merge their data

Your country ?

xx.LOVD.org

xx = country code

Malaysia DNA Variant Database

Based on: LOVD Leiden Open Variation Database

MY

Home | Variants in individuals from Malaysia | Variants by submitters from Malaysia

This resource automatically retrieves information from our LOVD3 shared installation. The information retrieved is based on the geographic information per country (here for **Malaysia**).

Variants shown are either linked to the country of origin of an *individual* (patient), or to the country of origin of the *submitter* of ta. Note the difference: data from an *individual* in Germany can be submitted by a *submitter* from Belgium.

Queries can be submitted by using the search boxes in the column's header.

Home | Variants in individuals from Malaysia | Variants

244 entries on 10 pages. Showing entries 1 - 25.

25 per page | **Legend** | « First | ‹ Prev | 1 | 2 | 3 | 4

Gene	Transcript	Effect	DNA change (cD
ABCC11	NM_032583.3	+/+	c.538G>A
ABCC11	NM_032583.3	+/+	c.538G>A
ABCC11	NM_032583.3	+/+	c.538G>A
ABCC11	NM_032583.3	+/+	c.538G>A
APC	NM_000038.5	+/+	c.847C>T

244 variants

Home | Variants in individuals from Malaysia | Variants

46 entries on 2 pages. Showing entries 1 - 25.

25 per page | **Legend** | « First | ‹ Prev | 1 | 2 | Next ›

Gene	Transcript	Effect	DNA change (cD
BRCA1	NM_007294.3	?/+	c.181T>C
BRCA1	NM_007294.3	+/+	c.115T>C
BRCA1	NM_007294.3	+/+	c.115T>C
BRCA1	NM_007294.3	-/-	c.2521C>T
BRCA1	NM_007294.3	?/+	c.190T>C
BRCA1	NM_007294.3	?/?	c.4211T>G
BRCA1	NM_007294.3	?/?	c.5057A>G
BRCA1	NM_007294.3	?/?	c.5057A>G
BRCA1	NM_007294.3	?/?	c.5072C>A

46 submissions

Your country ?

xx.LOVD.org

xx = country code

Singapore DNA Variant Database

Based on: LOVD Leiden Open Variation Database

SG

Home | Variants in individuals from Singapore | Variants by submitters from Singapore

This resource automatically retrieves information from our LOVD3 shared installation. The information retrieved is based on the geographic information per country (here for **Singapore**).

Variants shown are either linked to the country of origin of an *individual* (patient), or to the country of origin of the *submitter* of the data. Note the difference: data from an *individual* in Germany can be submitted by a *submitter* from Belgium.

Queries can be submitted by using the search boxes in the column's header.

Home | Variants in individuals from Singapore | Variants by submitte

78 entries on 4 pages. Showing entries 1 - 25.

25 per page | **Legend** | « First | ‹ Prev | 1 | 2 | 3 | 4 | Next › | Last »

Gene	Transcript	Effect	DNA change (cDNA)
APC	NM_000038.5	+/.	c.1226_1229delinsAAA
BRCA1	NM_007294.3	+/.	c.(-20+1_-19-1)_(80+1_81-1)del
BRCA1	NM_007294.3	+/.	c.(4185+1_4186-1)_(4357+1_4358-1)dup
BRCA1	NM_007294.3	+/.	c.1392del
BRCA1	NM_007294.3	+/.	c.1405del

78 variants

Home | Variants in individuals from Singapore | Variants by su

18 entries on 1 page. Showing entries 1 - 18.

25 per page | **Legend**

Gene	Transcript	Effect	DNA change (cDNA)
APC	NM_000038.5	+/.	c.1226_1229delinsAAA
LDLR	NM_000527.4	?/.	c.1060G>A
NPR2	NM_003995.3	+?/.	c.1352-1G>A
NPR2	NM_003995.3	+?/.	c.142G>T
NPR2	NM_003995.3	+?/.	c.1167G>T

18 submissions

Your country ?

Home | Variants in individuals from Malaysia | Variants

46 entries on 2 pages. Showing entries 1 - 25.

25 per page | **Legend** | « First | ‹ Prev | 1 | 2 | Next ›

Gene	Transcript	Effect	DNA change (cD
BRCA1	NM_007294.3	?/+	c.181T>C
BRCA1	NM_007294.3	+/+	c.115T>C
BRCA1	NM_007294.3	+/+	c.115T>C
BRCA1	NM_007294.3	-/-	c.2521C>T
BRCA1	NM_007294.3	?/+	c.190T>C
BRCA1	NM_007294.3	?/?	c.4211T>G
BRCA1	NM_007294.3	?/?	c.5057A>G
BRCA1	NM_007294.3	?/?	c.5057A>G
BRCA1	NM_007294.3	?/?	c.5072C>A

LSDB-in-a-Box

- **software**
  - *Open Source, platform independent*
  - *freely available* > *http:// www.LOVD.nl*
  - *follows existing HGVS guidelines*
    - *LSDB content, description of variants*

- **fully WWW-based**
  - *display of data*
  - *data management*
    - *access levels incl. manager, curator, submitter*

- **runs on any platform**
  - *laptop / PC, intranet, internet*

- **make any database**
  - *pre-programmed for DNA / RNA / protein*

LOVD
Leiden Open Variation Database

©Ivo Fokkema

free hosting on Leiden servers

LOVD connections

.nl

(can be country nodes)

contact through central site

LSDB list, web service (exome queries)

data flow

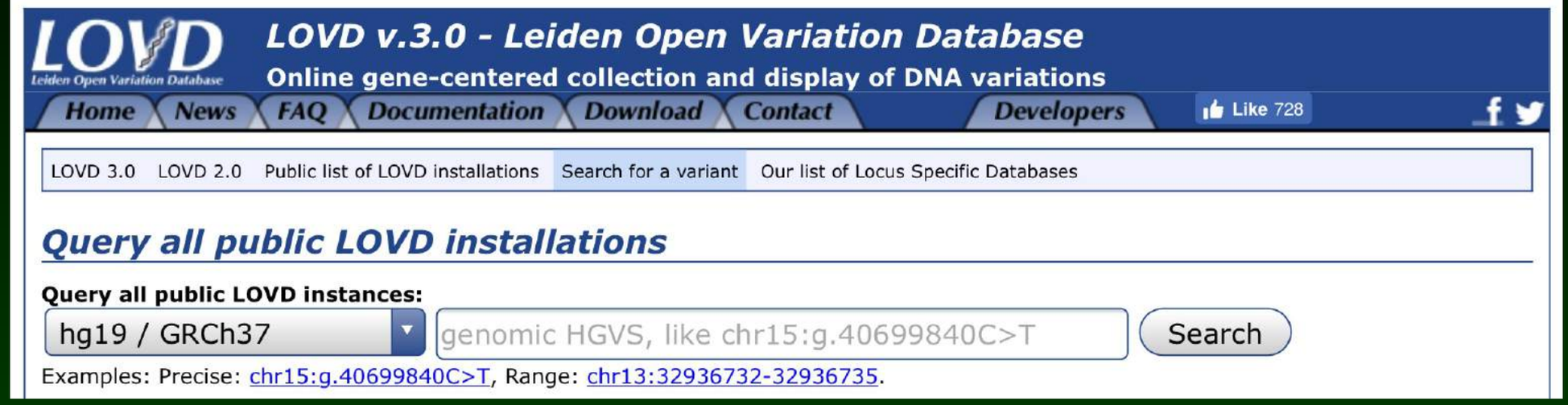
lab > gene variant database > central repository

LOVDs world-wide

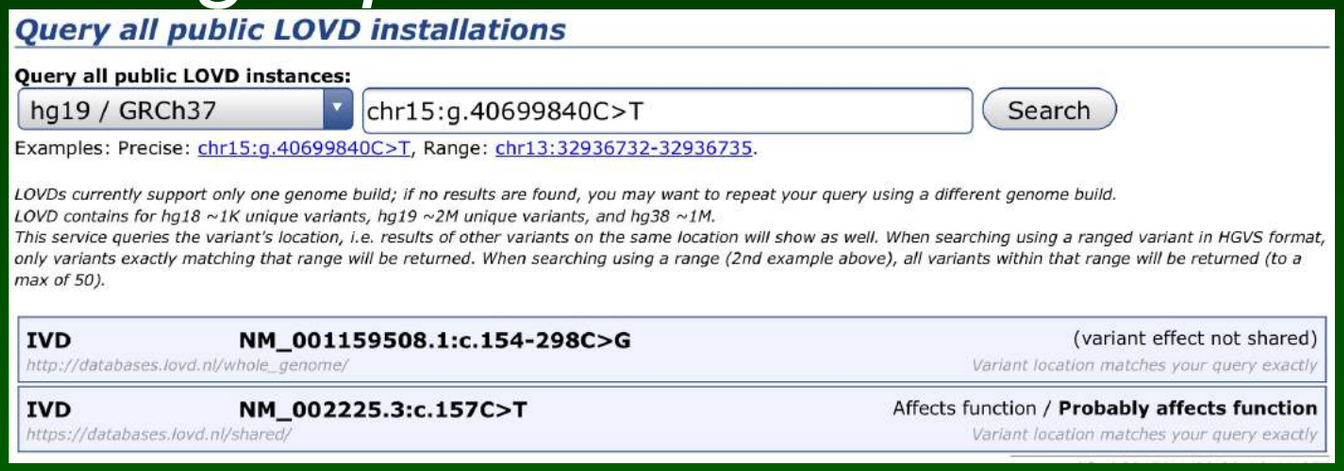
Aug. 2018

In total: 292,384,028 variants (3,953,852 unique) in 763,965 individuals in 85 LOVD installations.			
http://bipmed.iqm.unicamp.br/snparray/	LOVD 3.0-20	17391 genes	
BIPMed SNP Array	A1BG-AS1,A1CF,A2M,A2M-AS1,A2ML1,A2MP1,A3GALT2,A4GALT,A4GN...		902273 unique
http://bipmed.iqm.unicamp.br/	LOVD 3.0-21	20930 genes	66158522 variants
BIPMed WES	A1BG,A1BG-AS1,A1CF,A2M,A2M-AS1,A2ML1,A2MP1,A3GALT2,A4GALT...		622610
http://databases.lovd.nl/whole_genome/	LOVD 3.0-20a	22002 genes	1998175
Whole genome datasets	A1BG,A1BG-AS1,A1CF,A2M,A2M-AS1,A2ML1,A2MP1,A4GALT,A4GNT,A...		1998135
https://databases.lovd.nl/shared/	LOVD 3.0-21	22980 genes	757275
Global Variome shared LOVD	A1BG,A1BG-AS1,A1CF,A2LD1,A2M,A2M-AS1,A2ML1,A2MP1,A3GALT2,...		173628
http://proteomics.bio21.unimelb.edu.au/lovd/	LOVD 3.0-07	14772 genes	239690 variants
LOVD - Leiden Open Variation Database	A1BG,A1BG-AS1,A1CF,A2M,A2ML1,A4GALT,A4GNT,AAAS,AACS,AACSP...		152241
http://bipmed.iqm.unicamp.br/cfa/	LOVD 3.0-21	41 genes	134333
Craniofacial anomalies	APOC2,APOC4-APOC2,AXIN2,BCL3,BMP4,CLPTM1,DVL2,ERBB2,FGF22...		257 unique
https://ab-openlab.csir.res.in/mitolsdb/	LOVD 2.0-35	37 genes	112662 variants
MitoLSDB	MTATP6,MTATP8,MTCO1,MTCO2,MTCO3,MTCYB,MTND1,MTND2,MTND3,M...		4660 unique
http://www.insight-database.org/	LOVD 3.0-21	17 genes	36826 variants
International Society for Gastrointestinal Hereditary Tumours Database	APC,BMPR1A,EPCAM,FBXO11,GALNT12,MLH1,MLH3,MSH2,MSH3,MSH6,...		5836 unique
http://HCI-LOVD.hci.utah.edu	LOVD 2.0-33	8 genes	26167 variants
LOVD - human mismatch repair genes	MLH1,MLH1_priors,MSH2,MSH2_priors,MSH6,MSH6_priors,PMS2,P...		26167 unique

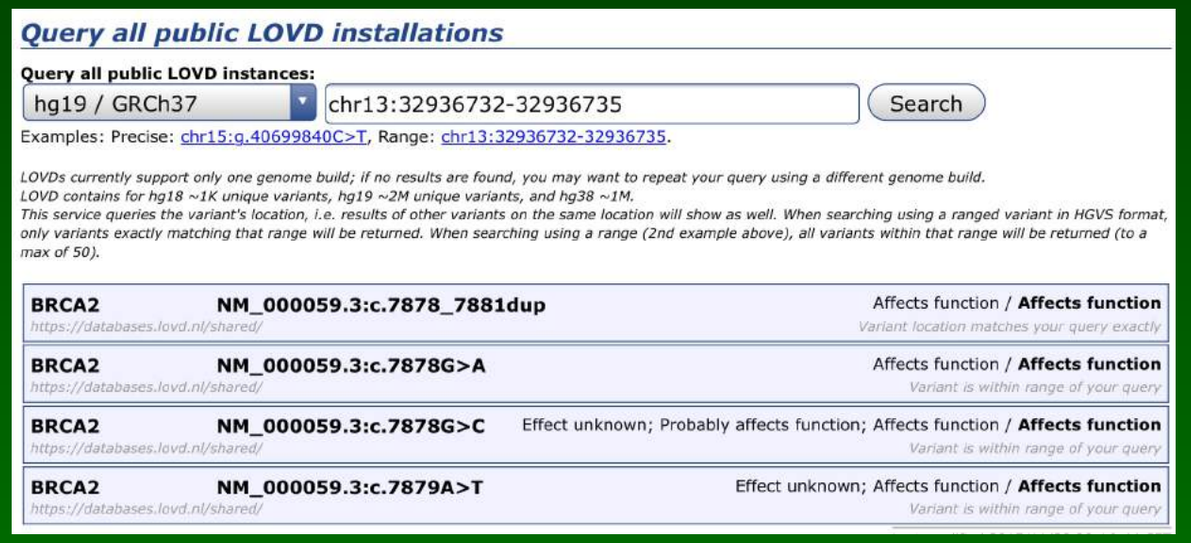
Query all LOVDs



using a position



LOVDs have an API

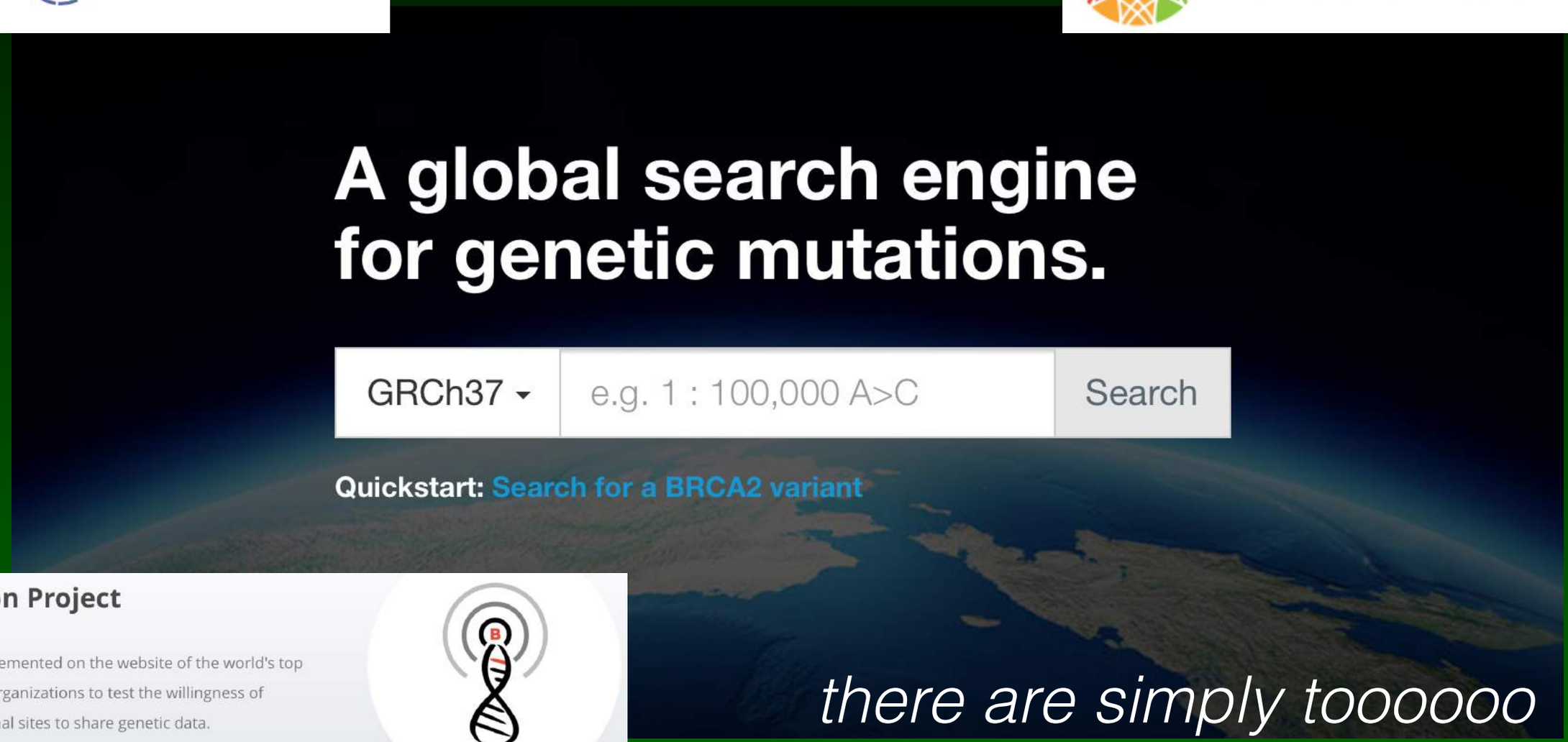


using a range

Beacons

Beacon Network

Global Alliance for Genomics & Health



Beacon Project

Being implemented on the website of the world's top genomic organizations to test the willingness of international sites to share genetic data.

there are simply toooooo many databases

Global Alliance for Genomics & Health

THE HUMAN VARIOME PROJECT
an NGO official partner of UNESCO
sharing data • reducing disease

brcaexchange.org

BRCA Challenge

The BRCA Challenge aims to advance understanding of the genetic basis of breast and other cancers using data from around the world.

BRCA CHALLENGE

BRCA Exchange

Expert Reviewed

HOME VARIANTS COMMUNITY

search for "c.1105G>A", "brca1" or "IVS7+1037T>C"

BRCA Exchange
a public BRCA variant portal
OPEN
17+

What's New Version History
Followed variant update notifications are now produced through a more robust local background-fetch mechanism, removing the reliance on push notifications. Various small bugfixes and user-interface more
4mo ago
Version 1.3.12

Preview

This is the BRCA Exchange Clinical Space, and contains only expert-reviewed variant classifications shown here represent the initial set submitted by the ENIGMA consortium, ~1000 (primarily missens BRCA2. The Clinical space will become more fully populated pending additional expert review.

Show All Public Data

Show Filters

2 matching variants of which 1 matched on synonyms

c.1105G>A

Gene	HGVS Nucleotide	HGVS Protein	Protein Abbrev	BIC Designation	Clinical Significance
BRCA1	c.1105G>A	p.(Asp369Asn)	D369N	1224G>A	Benign / Little Clinical Significance
BRCA1	c.993G>C	p.(Arg331Ser)	-	-	Not Yet Classified

Search

search for "c.1105G>A" or "brc...

20644 variants legend

Gene	HGVS Nucleotide
BRCA1	c.4358-2692G>A
BRCA2	c.775delA
BRCA1	c.117T>A
BRCA2	c.7341T>C
BRCA1	c.134+1508G>A
BRCA2	c.7544C>T
BRCA1	c.825_828delCAC...
BRCA2	c.9118-1G>A
BRCA1	c.5333-18T>G

LOVD 3 Leiden Open Variation Database | THE HUMAN VARIOME PROJECT

Shared database
DMD (dystrophin)

Curator: Johan den Dunnen

Genes | Transcripts | Variants | Individuals | Diseases | Screenings | Submit | Documentation

DMD gene homepage

This database is one of the gene variant databases from the Leiden Muscular Dystrophy pages.

General information

Gene symbol	DMD
Gene name	dystrophin
Chromosome	X
Chromosomal band	p21.2
Imprinted	Unknown
Genomic reference	NG_012232.1
Transcript reference	NM_004006.2
Exon/intron information	NM_004006.2 exon/intron table
Associated with diseases	all-in-one, BMD, BMD/DMD, CMD-3B, DMD
Citation reference(s)	-
Refseq URL	Genomic reference sequence
Curators (1)	**Johan den Dunnen**
Total number of public variants reported	15850
Unique public DNA variants reported	4925
Individuals with public variants	36693
Hidden variants	1468
Notes	This database is one of the gene variant databases from the: Leiden Muscular Dystrophy pages©. When refering to this database please cite Aartsma-Rus et al. (2006). Muscle Nerve. 34:135-144 and/or White SJ, den Dunnen JT (2006). Cytogenet.Genome Res. 115: 240-246.
Date created	July 29, 1997
Date last updated	March 09, 2018
Version	DMD:180309

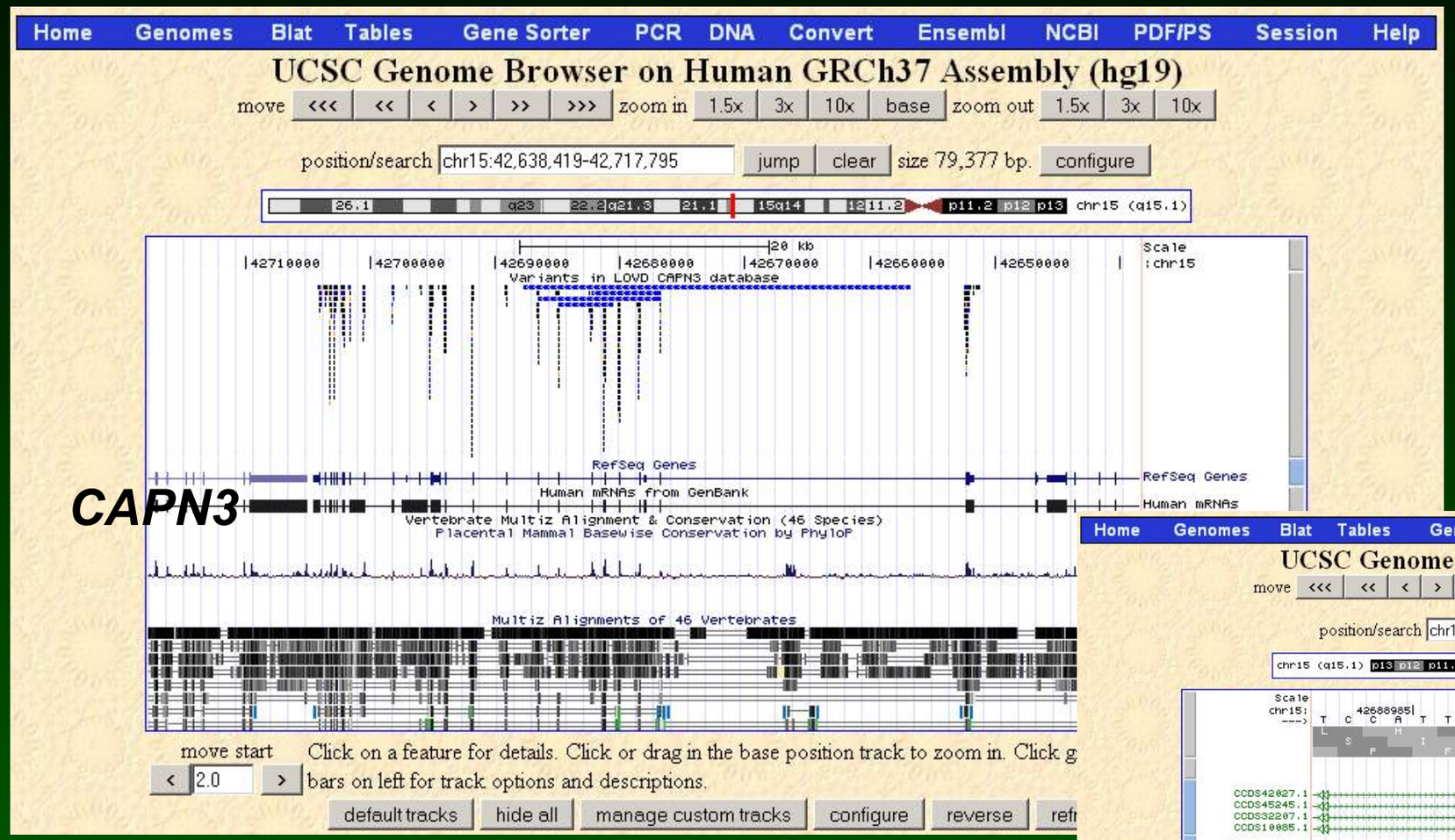
Graphical displays and utilities

Graphs	Graphs displaying summary information of all variants in the database »
Reading frame checker	The Reading-frame checker generates a prediction of the effect of whole-exon changes. Active for: NM_004006.2.
UCSC Genome Browser	Show variants in the UCSC Genome Browser (full view, compact view)
Ensembl Genome Browser	Show variants in the Ensembl Genome Browser (full view, compact view)
NCBI Sequence Viewer	Show distribution histogram of variants in the NCBI Sequence Viewer

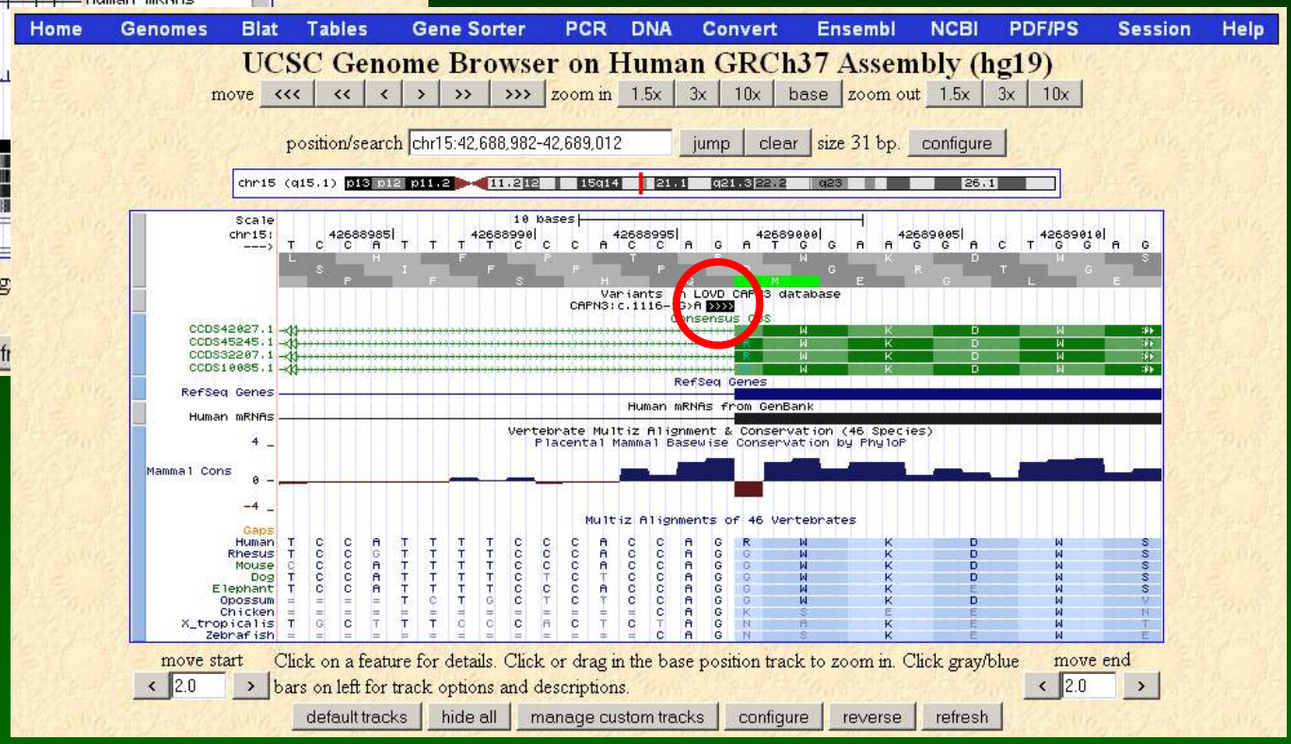
Links to other resources

Homepage URL	http://www.LOVD.nl/DMD
External URL	the Leiden Muscular Dystrophy pages Orphanet
HGNC	2928
Entrez Gene	1756
PubMed articles	DMD
OMIM - Gene	300377
OMIM - Diseases	BMD (dystrophy, muscular, Becker type (BMD)) CMD-3B (cardiomyopathy, dilated, type 3B (CMD-3B)) DMD (dystrophy, muscular, Duchenne type (DMD))
HGMD	DMD
GeneCards	DMD
GeneTests	DMD

LOVD to genome



CAPN3



Genome browsers

genome browsers start to accept HGVS descriptions

immediately jumping to position/change in genome

Request:	Genome Browser Response:
chr7	Displays all of chromosome 7
chrUn_gl000212	Displays all of the unplaced contig gl000212
20p13	Displays region for band p13 on chr 20
chr3:1-1000000	Displays first million bases of chr 3, counting from p-arm telomere
chr3:1000000+2000	Displays a region of chr3 that spans 2000 bases, starting with position 1000000
RH18061;RH80175 15q11;15q13 rs1042522;rs1800370	Displays region between genome landmarks, such as the STS markers RH18061 and RH80175, or chromosome bands 15q11 to 15q13, or SNPs rs1042522 and rs1800370. This syntax may also be used for other range queries, such as between uniquely determined ESTs, mRNAs, refSeqs, etc.
D16S3046	Displays region around STS marker D16S3046 from the Genethon/Marshfield maps. Includes 100,000 bases on each side as well.
AA205474	Displays region of EST with GenBank accession AA205474 in BRCA1 cancer gene on chr 17
AC008101	Displays region of clone with GenBank accession AC008101
AF083811	Displays region of mRNA with GenBank accession number AF083811
PRNP	Displays region of genome with HUGO Gene Nomenclature Committee identifier PRNP
NM_017414	Displays the region of genome with RefSeq identifier NM_017414
NP_059110	Displays the region of genome with protein accession number NP_059110
pseudogene mRNA	Lists transcribed pseudogenes, but not cDNAs
homeobox caudal	Lists mRNAs for caudal homeobox genes
zinc finger	Lists many zinc finger mRNAs
kruppel zinc finger	Lists only kruppel-like zinc fingers
huntington	Lists candidate genes associated with Huntington's disease
zahler	Lists mRNAs deposited by scientist named Zahler
Evans,J.E.	Lists mRNAs deposited by co-author J.E. Evans

LOVD - Leiden Open Variation Database

Growth Hormone Receptor (GHR)

LOVD v.2.0 Build 11 [Current LOVD status]
Register as submitter | Log in

Home | Variants | Submitters | Submit | Documentation

visitor

GHR homepage | Switch gene

LOVD - Leiden Open Variation Database

Growth Hormone Receptor (GHR)

LOVD v.2.0 Build 11 [Current LOVD status]
Welcome, Cecilia Camacho-Hübner Camacho-Hübner
Your account | Your submissions | Log out

Home | Variants | Submitters | Submit | Documentation

submitter *colleague*

GHR homepage | Switch gene

LOVD - Leiden Open Variation Database

Insulin-like Growth Factor 1 (IGF1)

LOVD v.2.0 Build 11 [Current LOVD status]
Welcome, Jan Maarten Wit
Your account | Log out

Home | Variants | Submitters | Submit | Configuration | Documentation

curator *collaborator*

Configuration | Switch gene | Curate | Find & Replace | Add column | Edit columns | Edit gene db | Empty gene db | Download | Import

---

LOVD - Leiden Open Variation Database

Signal Transducer and Activator of Transcription 5B (STAT5B)

LOVD v.2.0 Build 11 [Current LOVD
Welcome, Johan den D
Your account | L

Home | Variants | Submitters | Submit | Configuration | Setup | Documentation

DB manager

Setup | Settings | Add col | Edit cols | New col | All cols | New link | Edit links | Scan | Modules | New gene | Edit genes | New user | Edit users | New sub | Edit subs | Lo

LOVD Setup

Leiden Open Variation Database		Statistics				Variants				
Installed:2008-06-12	Updated:2008-09-01	Users:12	Submitters:8	Genes:5	Log entries:395	Total:92	Submitted:24	Non public:2	Marked:8	Public

Show your data

Leiden, Netherlands
Carli Tops
Leiden University Medical Center
Clinical Genetics

84 entries in MLH1

Path.	Exon	DNA change	DNA_reported	RNA change	Protein
+/?	01	c.18_34del17	-	r.(?)	p.Val7_ArgfsX18
+/?	01	c.18_34del17	-	r.(?)	p.Val7_ArgfsX18
+/?	01	c.18_34del17	-	r.(?)	p.Val7_ArgfsX18
?/?	01	c.102_103delGA	-	r.(?)	p.Glu34AspfsX3

You have not sent any submissions to the MLH3 gene database yet!

60 entries in MSH2

Path.	Exon	DNA change	DNA_reported	RNA change	Protein
?/?	00	c.(?_-68)_*+?del	del 1_16	-	p.0?
?/?	00	c.(?_-68)_*+?del + MSH6 (1)	del MSH2 ex1_16, delTACSTD1-15,-27, del MSH6	-	p.0?
?/?	00	c.(?_-68)_*+?del	del 1_16	-	p.0?

62 entries in MSH6

Path.	Exon	DNA change	DNA_reported	RNA change	Protein	Codon_nr
?/?	00	c.1-?_*+?del + MSH2 (1)	del MSH2 ex1_16, delTACSTD1-15,-27, del MSH6	-	p.0?	-
?/?	01	c.1-18G>T	-	-	p.?	-
?/?	01	c.1-?_457+?del	deletion of 21.6 kb around exon 1+2	-	p.Met1_Gly153>ValfsX21	-

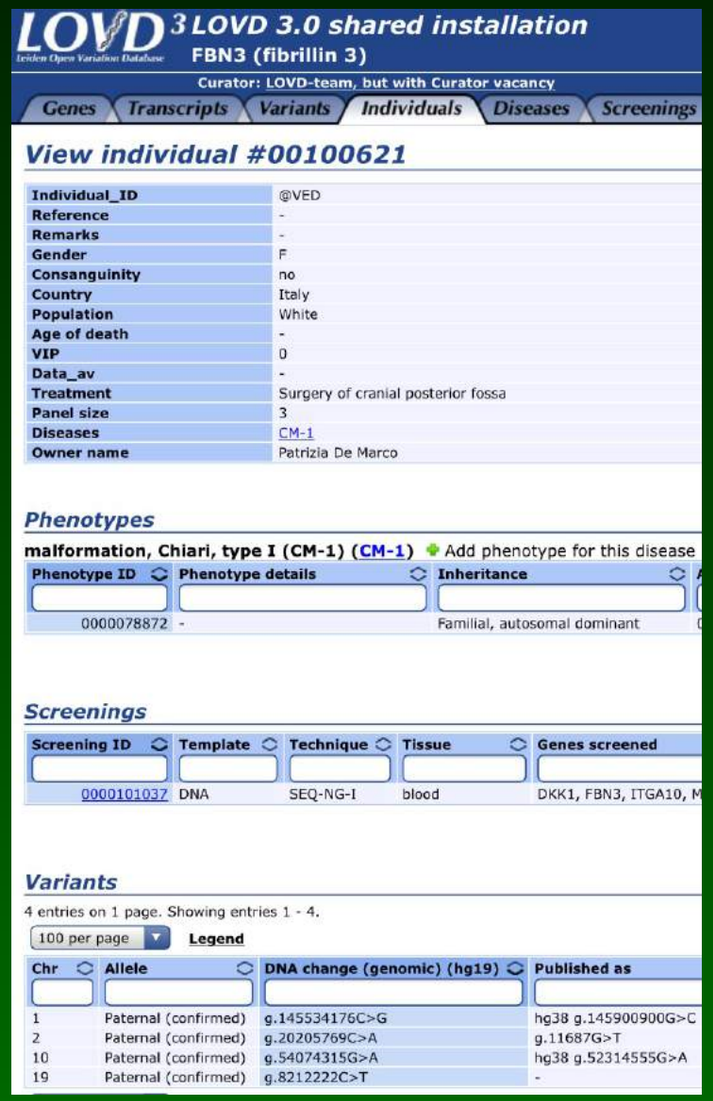
You have not sent any submissions to the PMS1 gene database yet!

9 entries in PMS2

Path.	Exon	DNA change	DNA_reported	RNA change	Protein
+/?	03	c.219_220dup + MLH1 (2)	-	r.(?)	p.Gly74ValfsX3
?/?	07i	c.?	g.46227+?_47919-?ins	r.(?)	-
?/?	08	c.861_864delACAG + c.1688G>T, MLH1 (1)	c.856delG	r.(?)	p.Arg287SerfsX19

4 databases in 1

- **Individuals**
  - *details on the Individual*
  - *fields fixed per database installation*
- **Phenotypes**
  - *details on Phenotype(s) per Individual*
    - *one Individual can have several Phenotypes*
  - *fields variable per Phenotype*
- **Screenings**
  - *details on Screening(s) performed*
  - *fields fixed per database installation*
- **Variants**
  - *details on Variant(s) identified*
  - *fields variable per Gene*



LOVD Leiden Open Variation Database

LUMC

4 in 1: Individual

Genes | Transcripts | Variants | Individuals | Diseases | Screenings | Submit

Individual #00054908

Variants | Individuals | Diseases | Screenings

Individual_ID	-
Reference	Journal: O'Rawe 2015
Remarks	2-generation family, 3 affected brothers, unaffected heterozygous carrier mother
Gender	M
Consanguinity	-
Country	Colombia
Population	-
Age/Death	>9y (later than 9 years)
VIP	0
Data_av	-
Treatment	-
Panel size	3
Diseases	ID
Owner name	Johan den Dunnen

LOVD Leiden Open Variation Database

LUMC

4 in 1: Phenotype

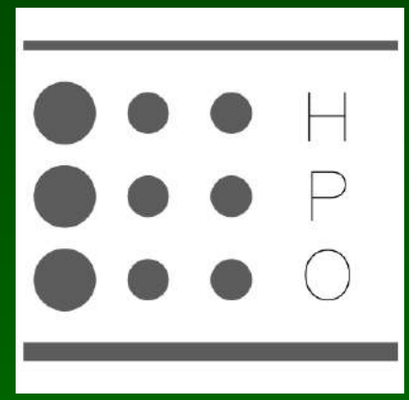
Phenotype #0000041575

Individual ID	00054908
Associated disease	ID
Diagnosis/Initial	intellectual disability
Diagnosis/Definite	MRXS-33
Inheritance	Familial, X-linked recessive
Age/Examination	09y (9 years)
Birth_Details	36w, caesarian section (HP:0011410), weight 4480 (1.85), height 52 (0.48), OFC 36.5 (1.02)
Age/Diagnosis	-
Age/Onset	-
Phenotype/Onset	-
Phenotype details	seizures (4 hours after birth), lactic acidosis.; postnatal growth retardation (HP:0008897); delayed gross motor development (HP:0002194); delayed speech and language development (HP:0000750); oral-pharyngeal dysphagia (HP:0200136); prominent supraorbital ridges (HP:0000336); downslanted palpebral fissures (HP:0000494); prominent forehead (HP:0011220); sagging cheeks; long philtrum (HP:0000343); low-set ears (HP:0000369); protruding ear (HP:0000411); thickened helices (large earlobe HP:0009748); long face (HP:0000276); high arched palate (high palate HP:0000218); thin upper lip (thin upper lip vermilion HP:0000219); pointed chin (HP:0000307); broad upturned nose (anteverted nares HP:0000463); hypertelorism (HP:0000316); sacral dimple (HP:0000960); hirsutism (HP:0001007); frequent dermatitis & eczema (eczema HP:0000964); toenail dysplasia (HP:0100797); hearing impairment (HP:0000365); chromic otitis media (HP:0000389); strabismus (HP:0000486); constipation (HP:0002019); gastroesophageal reflux (HP:0002020); microcephaly (HP:0000252); cerebellar atrophy (low cerebral white matter volume); hypoplasia of the corpus callosum (HP:0002079); seizures (HP:0001250); generalized hypotonia (HP:0001290); non-ambulatory; sleep-wake cycle disturbance (HP:0006979); osteopenia (HP:0000938); unusual gluteal crease with sacral caudal remnant/sacral dimple (abnormal sacral segmentatino, HP:0008468), prominent protruding coccyx (HP:0008472); distal joint hypermobility (HP:0001382); kyphosis (HP:0002808); scoliosis (HP:0002650); short neck (HP:0000470); hydronephrosis; autistic behaviors (HP:0000729); intellectual disability (HP:0001249); not present -HP:0000490, -HP:0000496, -HP:0000579, -HP:0000739, -HP:0001057, -HP:0001251, -HP:0001257, -HP:0001264, -HP:0001511, -HP:0002119, -HP:0002141, -HP:0007018, -HP:0007375
Intellectual_dis	-
Speech	-
Development	-
Owner name	Johan den Dunnen

Variants | Individuals | Diseases | Screenings

HPO

HP:
-HP:
?HP:



LOVD
Leiden Open Variation Database

LUMC

links to files

- *internet*
- *local*

LOVD - Leiden Open Variation Database

Insulin-like Growth Factor 1 Receptor (IGF1R)

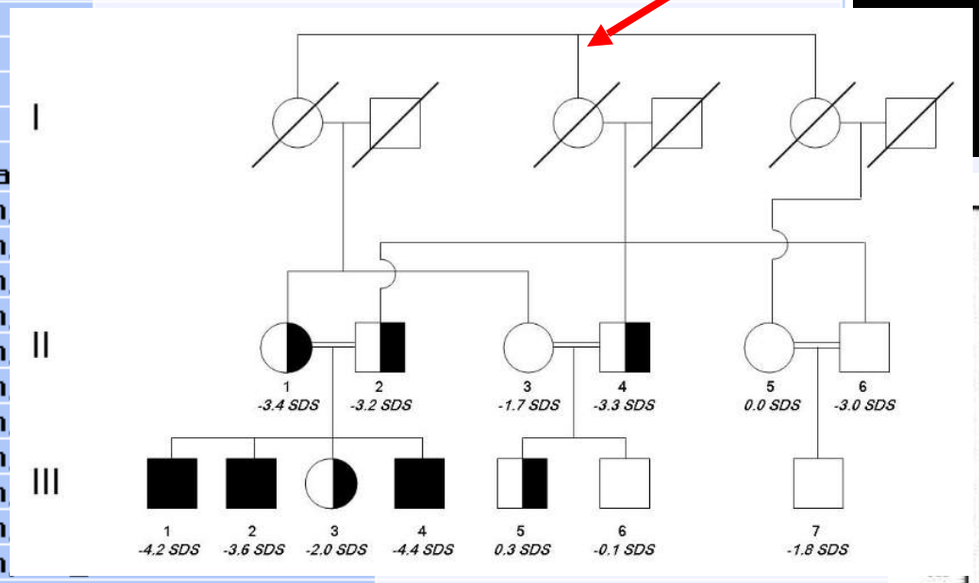
Home | Variants | Submitters | Submit | Configuration | Setup

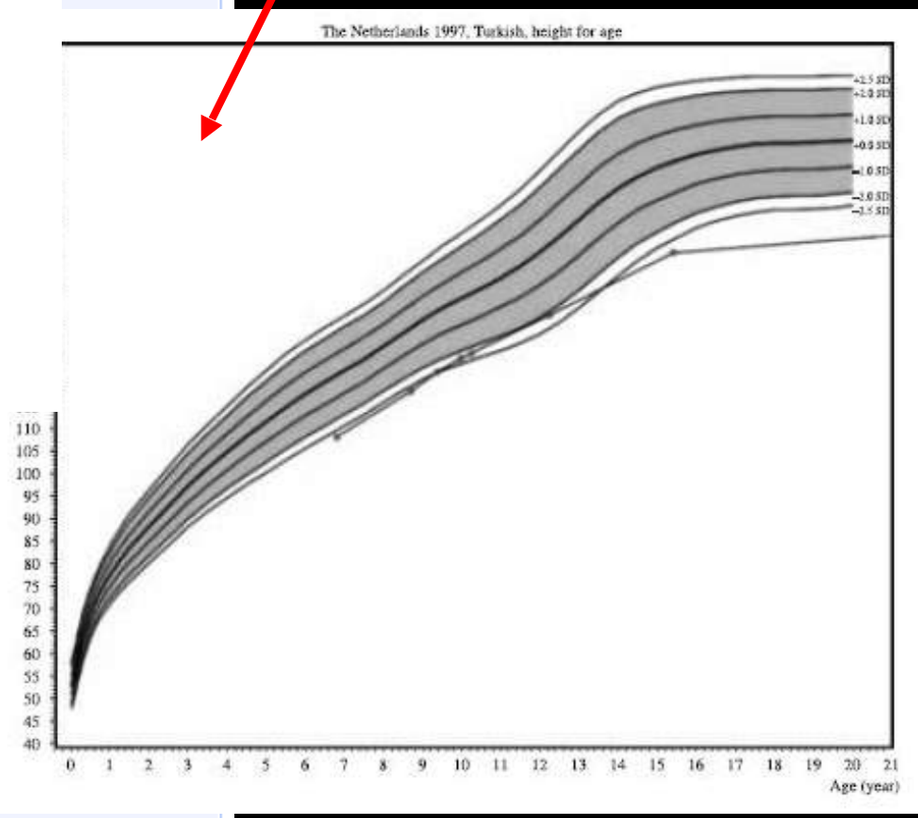
LOVD - Variant listings

Patient data (#0000070)	
Patient ID	p.A
Reference	Netherlands:Leiden
Date_Of_Birth	-
Gender	F
Origin/Geograph	
Origin/Ethnic	
Residence/Count	
Referring_doctor	
Visit_1st/Date_a	
Visit_1st/Compla	
Neonatal/Gestat	
Neonatal/Length	
Neonatal/Weight	
Neonatal/Head_	
Neonatal/Feedin	
Growth_Referenc	
Puberty	
Puberty_menarc	
PM/milestones	
PM/hearing	
PM/hearBEP	
PM/hearAudio	
PM/blindness	
Clinical	
Medication	

Father/Birth	-
Father/Adult	?,184.7 (
Father/Head	-
Father/Puberty	unknown
Father/Clinical	no
Mother/Birth	-
Mother/Adult	?,176.6 1.34
Mother/Head	
Mother/Puberty	
Mother/Clinical	
Father/Father	
Father/Mother	
Mother/Father	
Mother/Mother	
Length	
Length/Curve	
Length/SitStand	
Length/SitStandF	
Weight_BMI	
Weight_BMIRef	
Head	
HeadRef	
Dysmorphism/Fa	
Dysmorphism/Ot	
Neuro	
Syndrome	
Radiology	
Skeletal	

Patient ID	18463107.III2
Disease	-
Remarks	3 siblings 3 generation family (III-2), growth curve
Reference	Nederland:Leiden
# Reported	1
Geographic origin	-
Ethnic origin	Kurdish
Date_Of_Birth	1985-12-28





Protein/GH_stim2	-
Protein/IGF_gen1	-
Protein/IGF_gen2	-
Protein/IGF_gen3	-
Treatment_GH	?,?,12,1mg/m2/day, 197,?,1.02
Treatment_IGF1	-
Carbohydrate_metab	-
Carbohydrate_OGTT	-
Bone/BMDyoung	-
Bone/BMDadult	-
Remarks	W.A. Ester et al, table 1 shows the and after GH therapy. Patient A also had delayed dentition and hypocanthal folds.

4 in 1: Screening

Genes | Transcripts | Variants | Individuals | Diseases | Screenings | Submit | Documentation

Screening #0000054861

Individual ID	00054908
Template	DNA
Technique	SEQ;SEQ-NG
Tissue	-
Remarks	WES
Variants found?	1
Owner name	Johan den Dunnen

Variants | Individuals | Diseases | Screenings

Genes screened

Symbol	Gene	Chr	Band	Transcript
TAF1	TAF1 RNA polymerase II, TATA box binding protein (TBP)-associated factor, 250kDa	X	q13.1	

Variants found

1 entry on 1 page. Showing entry 1.

100 per page **Legend**

Chr	Allele	DNA change (genomic) (hg19)	DNA change (hg38)	Published as
X	Maternal (confirmed)	g.70602671C>T	-	-

100 per page **Legend**

LOVD Leiden Open Variation Database

LUMC

4 in 1: Variants

curator LOVD team, but with curator vacancy

Genes | Transcripts | Variants | Individuals | Diseases | Screenings | Submit | Documentation

Genomic variant #0000084884

Individual ID	00054908
Chromosome	X
Allele	Maternal (confirmed)
Affects function (as reported)	Affects function
Affects function (by curator)	Not classified
DNA change (genomic) (Relative to hg19 / GRCh37)	g.70602671C>T
DNA change (hg38)	-
Published as	-
ISCN	-
DB-ID	TAF1_000024 See all 4 reported entries
Variant remarks	-
Reference	Journal: O'Rawe 2015
ClinVar ID	-
dbSNP ID	-
Germline/De novo/Somatic	Germline
Segregation	yes
Frequency	-
Re-site	-
VIP	0
Methylation	-
Average frequency (large NGS studies)	Variant not found in online data sets
Owner	Johan den Dunnen

Gene	TAF1
Transcript ID	NM_004606.3
Affects function (as reported)	Affects function
Affects function (by curator)	Not classified
Exon	12
DNA change (cDNA)	c.1786C>T
Class.	-
RNA change	r.(?)
Protein	p.(Pro596Ser)

Variants | Individuals | Diseases | Screenings

LOVD Leiden Open Variation Database

LUMC

1 individual / many variants

LOVD 3 Leiden Open Variation Database — THE HUMAN VARIOME PROJECT

Shared database

CYP2D6 (cytochrome P450, family 2, subfamily

Curators: Andrea Gaedigk and Lisa Kalman

Genes | Transcripts | Variants | Individuals | Diseases | Screenings

Individual #00074429

Individual_ID	-
Reference	PubMed: Buermans 2017, Journal: Buermans 2017
Remarks	-
Gender	-
Consanguinity	no
Country	Netherlands
Population	-
Age/Death	-
VIP	0
Data_av	-
Treatment	-
Panel size	1
Diseases	DMB-i
Owner name	Henk Buermans

23 entries on 1 page. Showing entries 1 - 23.

100 per page | Legend

Chr	Allele	DNA change (genomic) (hg19)	DNA change (hg38)
22	Parent #2	g.42522071C=	g.42126069A>C
22	Parent #2	g.42522071C=	g.42126069A>C
22	Parent #2	g.42522613C>G	g.42126611C>G
22	Parent #2	g.42523003G>A	g.42127001G>A
22	Parent #2	g.42523209C>T	g.42127207C>T
22	Parent #2	g.42523409G=	g.42127407T>G
22	Parent #2	g.42523943A=	g.42127941G>A
22	Parent #2	g.42525132G=	g.42129130C>G
22	Parent #1	g.42525625C>T	g.42129623C>T
22	Parent #2	g.42525772G>A	g.42129770G>A
22	Both (homozygous)	g.42525952C=	g.42129950A>C
22	Parent #2	g.42526484A=	g.42130482C>A
22	Parent #2	g.42526549C=	g.42130547T>C
22	Parent #2	g.42526561_42526562=	g.[42130559T>G;42130560C>G]
22	Parent #2	g.42526567G=	g.42130565A>G
22	Parent #2	g.42526571C=	g.42130569G>C
22	Parent #2	g.42526573T=	g.42130571G>T
22	Parent #2	g.42526580G=	g.42130578C>G
22	Parent #2	g.42527533G=	g.42131531G>A
22	Parent #1	g.42527887delT	g.42131885delT
22	Parent #2	g.42528028C=	g.42132026T>C
22	Parent #1	g.42528030T[21]	g.42132049delT
22	Parent #2	g.42528030T[25]	g.42132047_42132049dup

LOVD Leiden Open Variation Database

LUMC

Databases and RNA

RNA ?

NCBI Resources How To

ClinVar ClinVar Advanced

Home | About | Data use and maintenance | Using the website

NM_033337.2(CAV3):c.158G>A (p.Ser53Asn)

NM_033337.2(CAV3):c.158G>A (p.Ser53Asn)

Variant type:	single nucleotide variant
Cytogenetic location:	3p25
Genomic location:	Chr3:8745569 (on Assembly GRCh38) Chr3:8787255 (on Assembly GRCh37)
Protein change:	S53N
HGVS:	NG_008797.2:g.16760G>A NM_033337.2:c.158G>A NC_000003.12:g.8745569G>A NC_000003.11:g.8787255G>A ...more
Links:	Leiden Muscular Dystrophy (CAV3): CAV3_00054 dbSNP: 199476326
NCBI 1000 Genomes Browser:	rs199476326
Molecular consequence:	NM_033337.2:c.158G>A: missense variant [Sequence Ontology SO:0001583]
Functional consequence:	unknown functional consequence

HGV Database

managed by Human Genome Variation powered by figshare

The HGV database is a fully searchable online database of genome variations published in peer-reviewed Data Reports in Human Genome Variation (ISSN 2054-345X). You can search and filter by different variables, including specific disease, gene, population or region.

Gene name	*KRT5*
Gene/Locus MIM number	148040
Mutation type	missense
Disease / Phenotype	Epidermolysis Bullosa Simplex Koebner type
Phenotype MIM number	131900
GenBank accession number	NM_000424.3
Mutation (HGVS format)	c.T974A
Protein alteration	p.L325H
Codon / Base change	CTC-CAC
Chromosome	12q13.13

(June 2014)

ClinVar DMD

825.	NM_004006.2(**DMD**):c.3637A>G (p.Lys1213Glu) *GRCh37:* ChrX:32466722 *GRCh38:* ChrX:32448605	**DMD**	not specified	Uncertain significance (Feb 1, 2016)	criteria provided, single submitter
826.	NM_004006.2(**DMD**):c.3631G>T (p.Glu1211Ter) *GRCh37:* ChrX:32466728 *GRCh38:* ChrX:32448611	**DMD**	Becker muscular dystrophy	Pathogenic (Nov 1, 1997)	no assertion criteria provided
827.	NM_004006.2(**DMD**):c.3604-12T>A *GRCh37:* ChrX:32466767 *GRCh38:* ChrX:32448650	**DMD**	not specified	Likely benign (Aug 12, 2016)	
828.	NM_004006.2(**DMD**):c.3604-14T>C *GRCh37:* ChrX:32466769 *GRCh38:* ChrX:32448652	**DMD**	not specified	Likely benign (Nov 16, 2016)	
829.	NM_004006.2(**DMD**):c.3603+15dupA *GRCh37:* ChrX:32472764 *GRCh38:* ChrX:32454647	**DMD**	not specified	Likely benign (Apr 28, 2015)	

NCBI Resources How To

ClinVar | ClinVar | Search ClinVar for gene symbols, HGVS expressior

Advanced

Home | About | Access | Help | Submit | Statistics | FTP

NM_004006.2(DMD):c.3631G>T (p.Glu1211Ter)

Variation ID: 11280

Review status: ☆ ☆ ☆ ☆ (0/4) no assertion criteria provided

Interpretation

Go to:

Clinical significance: Pathogenic

Last evaluated: Nov 1, 1997

Number of submission(s): 1

Condition(s): Becker muscular dystrophy [MedGen - Orphanet - OMIM]

See supporting ClinVar records

Allele(s)

Go to:

NM_004006.2(DMD):c.3631G>T (p.Glu1211Ter)

LOVD DMD

27	c.3628_3665del	-	r.3628_3665del	p.Lys1210*
27	c.3630delA	-	r.(?)	p.(Glu1211Lysfs*4)
27	c.3631G>T	3839G>T	r.[3631g>u, 3604_3786del, 3604_4071del]	p.[Glu1211*; Arg1202_1262del; Arg1202_1357del]
27	c.3679C>T	-	r.(?)	p.(Gln1227*)
27	c.3697delC			p.(Gln1233Lysfs*4)
27	c.3700G>T			p.(Glu1234*)
27	c.3705C>T (Reported 2 times)			p.(=)

Patient data (#0006974)	
Phenotype	muscular dystrophy, Becker (BMD)
Phenotype additional	-
Reference	Japan:Kobe
Remarks	-
Geographic origin	Japan
Ethnic origin	-
Gender	M
Inheritance	unknown
Consanguinity	-
Fam_Pat	-
# reported	1
CK level	-
Protein data	-
Submitter	Masafumi Matsuo

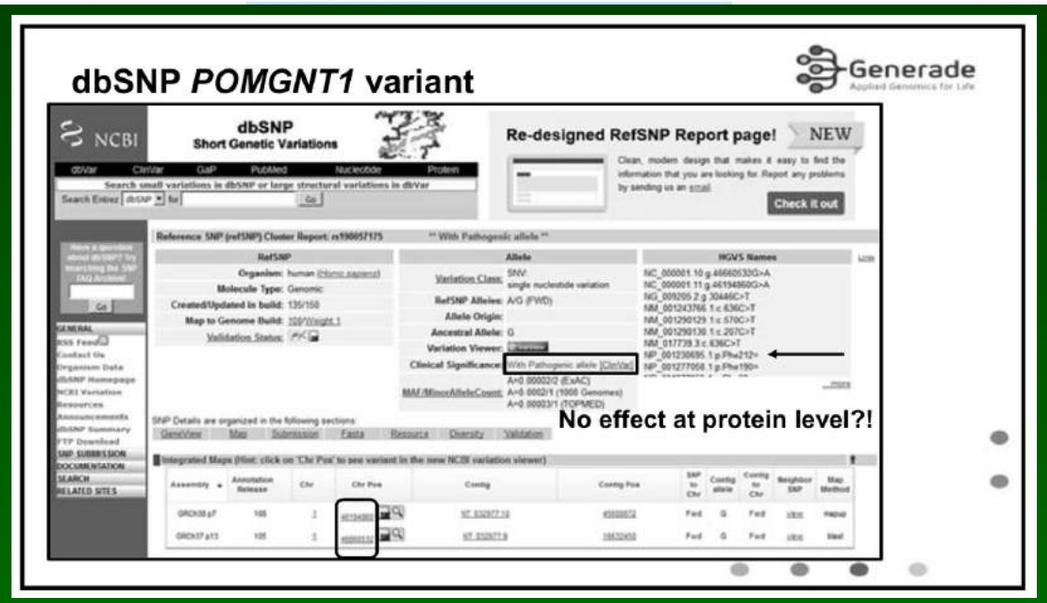
Variant data	
Allele	Parent #1
Reported pathogenicity	Pathogenic
Concluded pathogenicity	Unknown
Exon	27
DNA change	c.3631G>T (View in UCSC Genome Browser, Ensembl)
Var_pub_as	3839G>T
RNA change	r.[3631g>u, 3604_3786del, 3604_4071del]
Protein change	p.[Glu1211*; Arg1202_1262del; Arg1202_1357del]
DB-ID	DMD_00074
Variant remarks	10% diff.splice
Genet_ori	germline (inherited)
Segregation	-
Reference	Shiga, Takeshima 2010, (OMIM 0074)
Template	DNA, RNA
Technique	RT-PCR, SEQ, SSCA
Frequency	-
RE-site	-

825.	NM_004006.2(DMD):c.3637A>G (p.Lys1213Glu) GRCh37: ChrX:32466722 GRCh38: ChrX:32448605	DMD	not specified	Uncertain significance (Feb 1, 2016)	criteria provided, single submitter
826.	NM_004006.2(DMD):c.3631G>T (p.Glu1211Ter) GRCh37: ChrX:32466728 GRCh38: ChrX:32448611	DMD	Becker muscular dystrophy	Pathogenic (Nov 1, 1997)	no assertion criteria provided
827.	NM_004006.2(DMD):c.3604-12T>A GRCh37: ChrX:32466767 GRCh38: ChrX:32448650	DMD	not specified	Likely benign (Aug 12, 2016)	criteria provided, single submitter
828.	NM_004006.2(DMD):c.3604-14T>C GRCh37: ChrX:32466769 GRCh38: ChrX:32448652	DMD	not specified	Likely benign (Nov 16, 2016)	criteria provided, single submitter
829.	NM_004006.2(DMD):c.3603+15dupA GRCh37: ChrX:32472764 GRCh38: ChrX:32454647	DMD	not specified	Likely benign (Apr 28, 2015)	criteria provided, single submitter

LOVD Leiden Open Variation Database and RNA

POMGNT1 database

+/.	7	c.594C>G	-	r.(?)	p.(Ser198Arg)	g.46660574G>C
+/.	7	c.630G>T	-	r.(?)	p.(Trp210Cys)	g.46660538C>A
+/+?	7	c.636C>T	-	r.spl	p.?	g.46660532G>A
+/.	7	c.636C>T	-	r.535_652del	p.Asp179Valfs*23	g.46660532G>A
+/.	7	c.636C>T	-	r.(?)	p.(=)	g.46660532G>A
+/.	7	c.636C>T	-	r.535_652del	p.Asp179Valfs*23	g.46660532G>A
+/.	7	c.636C>T	-	r.(=)	p.(=)	g.46660532G>A
+/.	7	c.643C>T	-	r.535_652del	p.Asp179Valfs*23	g.46660525G>A
+/+?	7	c.643C>T	-	r.535_652del	p.Asp179Valfs*23	g.46660525G>A
+?/+?	7i	c.652+1G>A	-	r.spl	p.?	g.46660515C>T
+/.	7i	c.652+1G>A	-	r.spl?	p.?	g.46660515C>T



In vitro record

LOVD 3 Leiden Open Variation Database — THE HUMAN VARIOME PROJECT

Shared database

BRCA1 (breast cancer 1, early onset)

LOVD is supported by: interactive biosoftware

LOVD v.3.0 Build 21 [Curr

Register as su

Effect	Exon	DNA change (cDNA)	RNA change	Protein	Germline/De novo/Somatic	FunctionalAnalysis/Technique
					vitro	
+/.	1i_3i	c.(-20+1_-19-1)_(134+1_135-1)del	r.spl	p.Met1_Cys47del	In vitro (cloned)	mRNA analysis (RT-PCR)
-?/.	1i	c.-19-22_-19-21dup	r.=	?	In vitro (cloned)	Splicing reporter minigene
-?/.	1i	c.-19-10T>C	r.=	?	In vitro (cloned)	Splicing reporter minigene
-/.	1i	c.-19-10T>C	r.=	?	In vitro (cloned)	mRNA analysis (RT-PCR)
?/.	2	c.-3G>C	?	?	In vitro (cloned)	?
-/.	2	c.19C>T	-	p.Arg7Cys	In vitro (cloned)	E3 Ub-ligase activity assay BRCA1/RING-BARD1
-/.	2	c.32T>C	-	p.Val11Ala	In vitro (cloned)	E3 Ub-ligase activity assay BRCA1/RING-BARD1
?/.	2	c.32T>C	?	p.Val11Ala	In vitro (cloned)	?
?/.	2	c.32T>C	-	p.Val11Ala	In vitro (cloned)	peptide binding ability BRCA1/RING-BARD1
-/.	2	c.43A>C	-	p.Ile15Leu	In vitro (cloned)	evolutionary conservation analysis
-/.	2	c.43A>C	-	p.Ile15Leu	In vitro (cloned)	E3 Ub-ligase activity assay BRCA1/RING-BARD1
+/.	2	c.44T>C	-	p.Ile15Thr	In vitro (cloned)	evolutionary conservation analysis
?/.	2	c.44T>C	-	p.Ile15Thr	In vitro (cloned)	E3 Ub-ligase activity assay BRCA1/RING-BARD1
-/.	2	c.44T>C	-	p.Ile15Thr	In vitro (cloned)	peptide binding ability BRCA1/RING-BARD1

BRCA1 variants tested in in vitro functional assays

SUMMARY record

LOVD 3 LOVD 3.0 shared installation
BRCA1 (breast cancer 1, early onset)
LOVD is supported by: interactive biosoftware
LOVD v.3.0 Build 19a [Current LOVD
Register as submitter |

+/+	11	c.981_982del	r.(?)	p.(Cys328*)	-
+/+	11	c.984_985insC	r.(?)	p.(Asn329Glnfs*2)	-
+/+	11	c.984_988del	r.(?)	p.(Cys328*)	-
-/-	11	c.1001C>T	r.(?)	p.(Pro334Leu)	-
+/+	11	c.1008dup	r.(?)	p.(Glu337Argfs*9)	-
+/+	11	c.1012A>T	r.(?)	p.(Lys338*)	-
+/+	11	c.1016del	r.(?)	p.(Lys339Argfs*2)	-
+/+	11	c.1016dup	r.(?)	p.(Val340Glyfs*6)	-
+/+	11	c.1016_1017insC	r.(?)	p.(Lys339Asnfs*7)	-
+/+	11	c.1017_1018insA	r.(?)	p.(Val340Serfs*6)	-

nstallation
early onset)
LOVD is supported by: interactive biosoftware
LOVD v.3.0 Build 19a [Current LOVD
Register as submitter |

o encode protein	ENIGMA classification criteria	-	SUMMARY record
o encode protein	ENIGMA classification criteria	-	SUMMARY record
o encode protein	ENIGMA classification criteria	-	SUMMARY record
terior :torial :holds for class as per Plon 2008 (PMID:18951446). Class 1 based on posterior probability = 0.000000271	ENIGMA classification criteria, {ENIGMA:BRCA[1] [c.1001C%3ET]}, PubMed: Lindor 2012	-	SUMMARY record
Variant allele predicted to encode truncated non-functional protein	ENIGMA classification criteria	-	SUMMARY record
Variant allele predicted to encode truncated non-functional protein	ENIGMA classification criteria	-	SUMMARY record
Variant allele predicted to encode truncated non-functional protein	ENIGMA classification criteria	-	SUMMARY record
Variant allele predicted to encode truncated non-functional protein	ENIGMA classification criteria	-	SUMMARY record
Variant allele predicted to encode truncated non-functional protein	ENIGMA classification criteria	-	SUMMARY record

ENIGMA variant interpretation committee

(BRCA associated or not, 5 classes)

CLASSIFICATION record

View genomic variant #0000146002

Chromosome	1
Allele	Parent #1
Affects function (reported)	Effect unknown
Affects function (concluded)	Not classified
DNA change (genomic) (Relative to hg19 / GRCh37)	g.898757G>C
Published as	-
ISCN	-
DB-ID	KLHL17_000001
Variant remarks	-
Reference	-
dbSNP ID	
Germline/Somatic/De novo	CLASSIFICATION record
Segregation	
Frequency	-
Re-site	-
VIP	0
Methylation	-
Average frequency (large NGS studies)	Variant not found in online data sets
Owner	VKGL-NL_Leiden

Variant on transcripts

Gene	Transcript	Affects function	Exon	DNA change (cDNA)	Class.	RNA change	Protein
KLHL17	NM_198317.2	?/.	8	c.1228G>C	-	r.(?)	p.(Asp410His)

VKGL initiative

no data on Individual (disease)

Germline/Somatic/De novo

CLASSIFICATION

Owner

VKGL

VKGL-NL_AMC

VKGL-NL_Groningen

VKGL-NL_Rotterdam

VKGL-NL_Utrecht

VKGL-NL_AMC

VKGL-NL_Utrecht

VKGL-NL_Groningen

VKGL-NL_Leiden

Effect	Exon	DNA change (cDNA)	Owner
			vkgl
-/.	14	c.1635A>G	VKGL-NL_Utrecht
-/.	14	c.1635A>G	VKGL-NL_AMC
-/.	14	c.1635A>G	VKGL-NL_Groningen

Dutch clinical labs sharing all variants and their classification

LOVD Leiden Open Variation Database

LUMC

VKGL initiative

LOVD 3 — Leiden Open Variation Database — THE HUMAN VARIOME PROJECT

Shared database
DMD (dystrophin)

LOVD is supported by: interactive biosoftware

LOVD v.3.0 Build 21 [Current LOVD stat
Register as submitter | Lo

48 entries on 1 page. Showing entries 1 - 48.

100 per page — Legend

Effect	Reported	Exon	DNA change (cDNA)	Class.	RNA change	Protein	DNA change (genomic) (hg19)	DNA change (hg38)
?/.	2	6	c.434G>A	VUS	r.(?)	p.(Arg145Gln)	g.32834681C>T	g.32816564C>T
-/., -?/.	3	8i	c.832-18C>G	benign, likely benign	r.(?)	p.(=)	g.32716133G>C	g.32698016G>C
-/., -?/.	3	8i	c.832-17C>A	benign, likely benign	r.(?)	p.(=)	g.32716132G>T	g.32698015G>T
-?/., ?/.	2	9	c.842G>C	likely benign, VUS	r.(?)	p.(Ser281Thr)	g.32716105C>G	g.32697988C>G
-?/.	2	10	c.1095A>C	likely benign	r.(?)	p.(Gln365His)	g.32663135T>G	g.32645018T>G
-/.	3	14	c.1635A>G	benign	r.(?)	p.(=)	g.32591931T>C	g.32573814T>C
?/.	2	16	c.1945C>T	VUS	r.(?)	p.(Arg649Trp)	g.32583866G>A	g.32565749G>A
-/.	2	17	c.2143A>T	benign	r.(?)	p.(Thr715Ser)	g.32563301T>A	g.32545184T>A
-/.	2	17i	c.2168+13T>C	benign	r.(?)	p.(=)	g.32563263A>G	g.32545146A>G
-?/., -/.	6	20	c.2391T>G	likely benign, benign	r.(?)	p.(Asn797Lys)	g.32509625A>C	g.32491508A>C
-?/.	2	20	c.2490C>T	likely benign	r.(?)	p.(=)	g.32509526G>A	
-/., -?/.	3	20i	c.2623-11C>G	benign, likely benign	r.(?), r.(=)	p.(=)	g.32503227G>C	

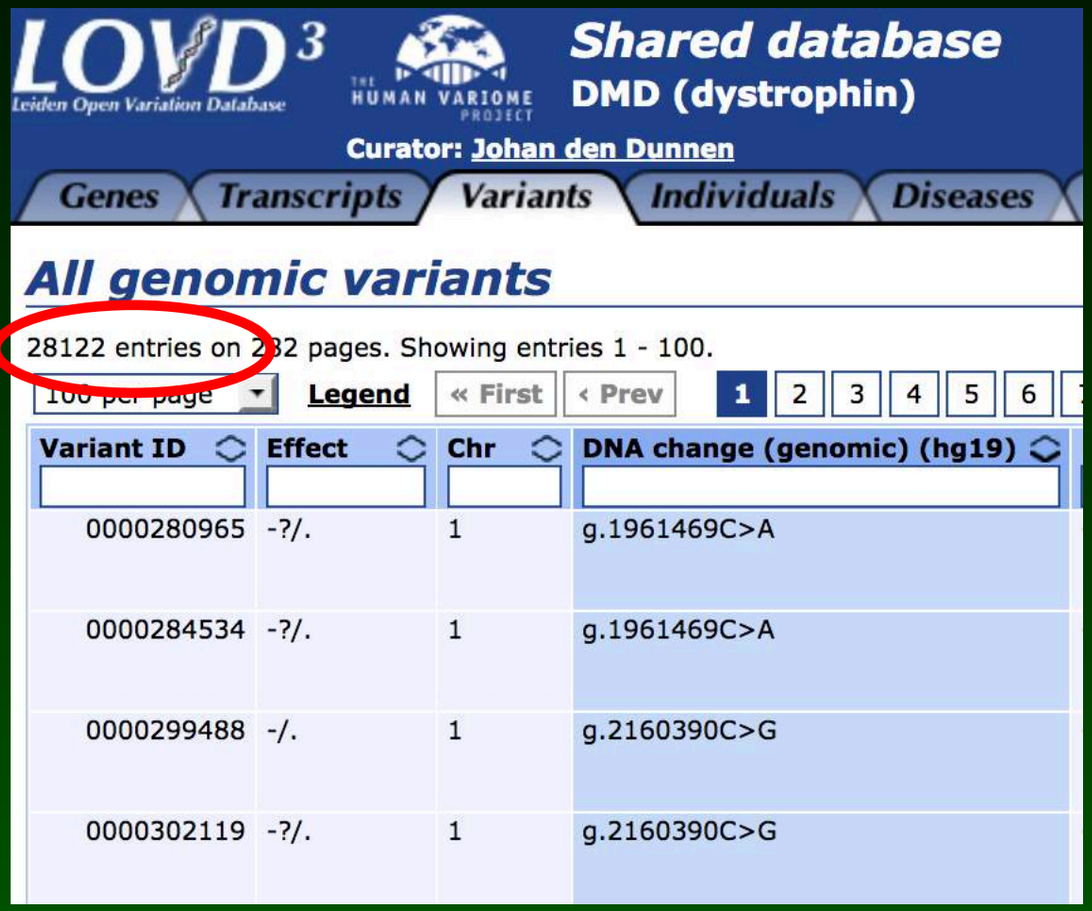
Effect	Exon	DNA change (cDNA)	Class.	RNA change	Protein
-?/.	26i	c.3603+15dup	likely benign	r.(?)	p.(=)
-/.	26i	c.3603+15dup	benign	r.(?)	p.(=)
-/.	26i	c.3603+15dup	benign	r.(?)	p.(=)
-/.	26i	c.3603+15dup	benign	r.(?)	p.(=)
-/.	26i	c.3603+15dup	benign	r.(?)	p.(=)

DNA change (hg38)	Published as
g.32454647dup	3603+15_3603+16insA
g.32454647dup	3234+15_3234+16insA
g.32454647dup	3234+14_3234+15insA
g.32454647dup	3234+13_3234+14insA
g.32454647dup	3603+2_3603+3insA

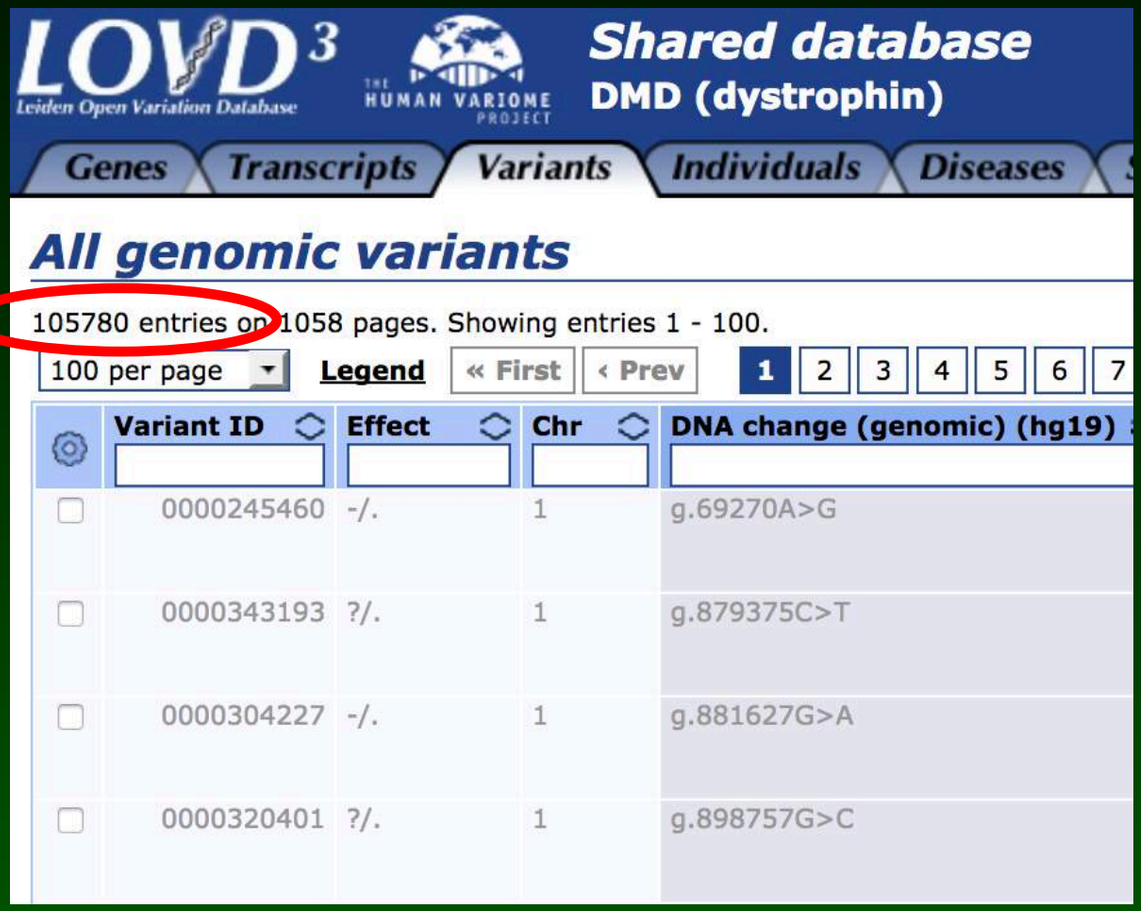
LOVD Leiden Open Variation Database

LUMC

VKGL initiative



public: 28,122



total: 105,780

Dutch clinical labs sharing all variants
*and their **consensus** classification*

1139 variants no consensus
29 variants opposite classification

LOVD Leiden Open Variation Database

LUMC

Minimal reporting

LOVD Leiden Open Variation Database

Leiden Muscular Dystrophy pages

Protein O-Mannosyltransferase 1 (POMT1)

Home | Variants | Submitters | Submit | Documentation

View unique variants | Search unique variants | View all contents | Full database search | Variant listing based on patient origin | Database statistics | Switch gene

LOVD - Variant listings

Unhide all columns

291 public entries

100 entries per page

Exon	DNA change	Disease	Reference	Geographic origin
00	c.? + c.1790_1791del	LIS-II	JdD	(FR)
02	c.-6T>G + c.752G>A, c.942C>T, c.2110dupG, c.*41C>T	WWS	JdD	IT
02	c.78G>A + c.1148+16G>A, c.2234G>A	MDC	United States:Marshfield, WI	-
02	c.85A>C + c.1864C>T	MDC	United States:Iowa City	US
02	c.122-5dupT	-	JdD	-
02	c.122+5G>A + c.2115G>A	LIS-II	JdD	(FR)
02	c.122+417GT(15_23)	-	JdD	-
03	c.123-5dupT + 18 others	FCMD, MEB	Portugal:Porto	PT
03	c.129C>T	-	JdD	-
03	c.145_146ins290145_160dup	WWS	JdD	FR

variant + contact details

How improve sharing?

- **...make variant submission obligatory**

  for publication

  for approved grants

  for clinical lab
  as part of QC
  to get accreditation

...to get a sample sequenced

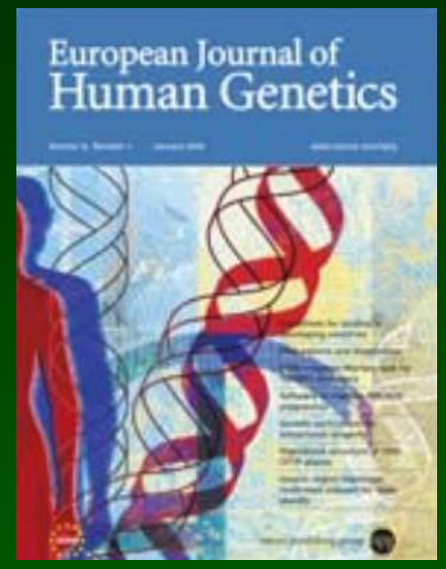
+ QC standards for LSDBs

Journals

Human Mutation
Variation, Informatics, and Disease

OFFICIAL JOURNAL HGV HUMAN GENOME

- **Human Mutation**
  first to demand using HGVS nomenclature
  first to demand database submission
  before accepting a paper for publication



- **European Journal of Human Genetics**
  demanding HGVS nomenclature & DB submission
  ...and checking every paper !!

collaboration with LOVD Leiden Open Variation Database

Adopt a gene !

CV

Curator for the ... gene variant database.

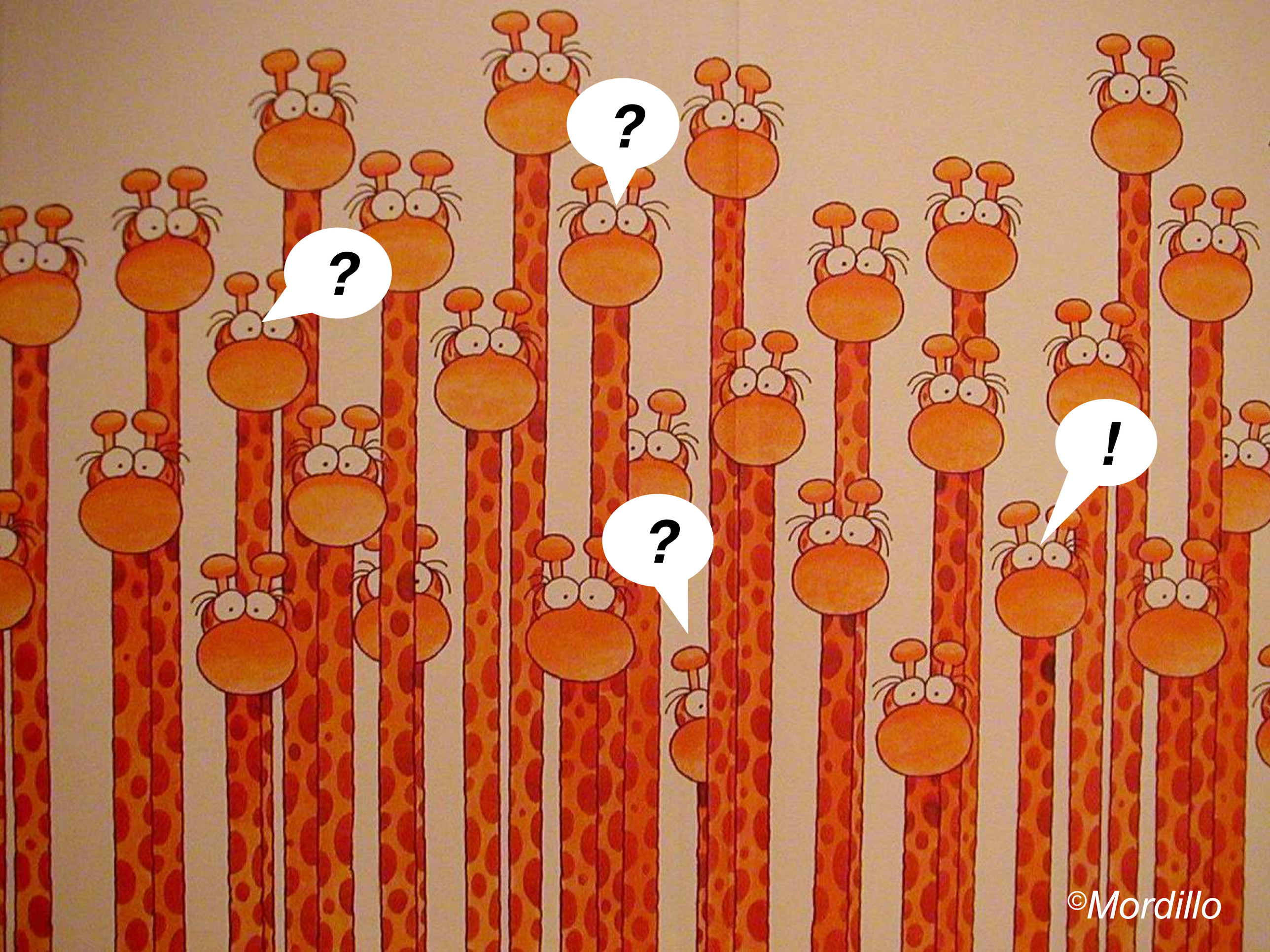
many orphan genes

THE HUMAN VARIOME PROJECT

HGV HUMAN GENOME VARIATION SOCIETY

LOVD Leiden Open Variation Database

become a
foster parent
database curator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