Table of Contents

Diseases	Page
Alpha-1 Antitrypsin Deficiency	**2**
Amyotophic Lateral Sclerosis (Lou Gehrig Disease)	**3**
Androgen Insensitivity Syndrome	**6**
Antithrombin Pittsburgh	**8**
Apert Syndrome	**9**
Aspartylglucosaminuria	**10**
Canavan Disease	**11**
Ceroid Lipfuscinosis, Neuronal 1 (CLN1)	**12**
Cowden Disease 1	**13**
Creutzfekdt-Jakob Disease	**14**
Crouzon Syndrome	**15**
Cystic Fibrosis	**17**
Emery-Dreifuss Muscular Dystrophy	**19**
Fatal Familial Insomnia	**20**
Gaucher Disease	**21**
Gerstmann-Straussler Disease	**25**
Hemochromatosis	**26**
Hereditary Nonpolyposis Colorectal Cancer, type 2	**28**
Ladd Syndrome	**30**
Lynch Syndrome	**31**
Lesch-Nyhan Syndrome	**32**

Diseases (continued)	Page
Lipodystrophy, Familial Partial Type 2	**34**
Macrocephaly/Autism Syndrome	**35**
Mandibuloacral Dysplasia with Type A Lipodystrophy	**36**
Metachromatic Leukodystrophy	**37**
Mucopolysaccharidosis Type VII	**40**
Neurofibromatosis, Type 1; NF1	**42**
Neurofibromatosis, Type 2; NF2	**43**
Parkinson Disease 7, early-onset; PARK7	**44**
Pfeiffer Syndrome	**45**
Phenylketonuria (PKU)	**46**
Porphyria:Acute Intermittent	**48**
Porphyria: Congenital Erythropoitic	**50**
Porphyria: Cutanea Tarda and Hepatoerythropoietic	**52**
Retinoblastoma	**54**
Rett Syndrome	**55**
Sandhoff Disease	**56**
Stiff Skin Syndrome	**57**
Tay-Sachs Disease	**58**
Von Willebrand Disease, Type 2	**60**
References	**62**

Alpha-1 Antitrypsin Deficiency

Gene - SERPIN PEPTIDASE INHIBITOR; SERPINA1

OMIM - 107400 gene
OMIM - 613490 disease

Allele

107400.0011 (see table below)

Structures

WT structure in PDB (3NE4) amino acids (aa) 24-393
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0011	PI Z	SERPINA1, GLU342LYS ON M1A	[rs28929474]	[rs28929474]	[RCV000255454...]	

Amyotophic Lateral Sclerosis (Lou Gehrig's Disease)

Gene - SUPEROXIDE DISMUTASE 1; SOD1

OMIM - 147450 gene
OMIM - 105400 disease

Alleles

Many mutant alleles (see table below).

Stuctures

WT structure in PDB (1PU0) aa 1-153
9 crystalized mutant structures highlighted green below
Create other mutant structures using PyMOL software

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0001	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, GLY37ARG	[rs121912431]	-	[RCV000015874]	
.0002	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, LEU38VAL	[rs121912432]	-	[RCV000015875]	**2WZ5 1.5A aa 1-153**
.0003	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, GLY41SER	[rs121912433]	-	[RCV000015876]	
.0004	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, GLY41ASP	[rs121912434]	-	[RCV000015877]	
.0005	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, HIS43ARG	[rs121912435]	-	[RCV000015878]	**1PTZ 1.8A aa 1-153**

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0006	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, GLY85ARG	[rs121912436]	-	[RCV000015880]	**2VR6 1.3A aa 1-153**
.0007	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, GLY93CYS	[rs121912437]	-	[RCV000015881]	
.0008	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, GLY93ALA	[rs121912438]	-	[RCV000015882]	**2ZKY 2.40 aa 2-153**
.0009	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, GLU100GLY	[rs121912439]	-	[RCV000015883]	
.0010	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, LEU106VAL	[rs121912440]	-	[RCV000015879]	
.0011	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, ILE113THR	[rs74315452]	-	[RCV000015884]	**1UXL 1.6 aa 1-153**
.0012	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, ALA4VAL	[rs121912442]	-	[RCV000015885]	**1UXM 1.9 aa 1-153**
.0013	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, HIS46ARG	[rs121912443]	-	[RCV000281824...]	**1OEZ 2.15 aa 1-153**
.0014	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, ALA4THR	[rs121912444]	-	[RCV000015887]	
.0017	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, LEU144SER	[rs121912446]	-	[RCV000015891]	
.0018	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, ALA145THR	[rs121912447]	-	[RCV000015892]	
.0020	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, CYS6PHE	[rs121912448]	-	[RCV000015894]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0021	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, ILE151THR	[rs121912449]	-	[RCV000015895]	
.0022	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, GLU21LYS	[rs121912450]	-	[RCV000015896]	
.0023	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, SER134ASN	[rs121912451]	[rs121912451]	[RCV000015897]	**1OZU 1.3A aa 1-153**
.0024	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, LEU84VAL	[rs121912452]	[rs121912452]	[RCV000015898]	
.0025	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, GLY16SER	[rs121912453]	-	[RCV000015899]	
.0028	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, GLY72SER	[rs121912455]	[rs121912455]	[RCV000015902]	
.0029	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, GLY12ARG	[rs121912456]	-	[RCV000015903]	
.0030	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, PHE45CYS	[rs121912457]	-	[RCV000015904]	
.0031	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, HIS80ARG	[rs121912458]	-	[RCV000015905]	**3H2Q 1.85A aa 1-153**
.0033	AMYOTROPHIC LATERAL SCLEROSIS 1	SOD1, GLY93ARG	[rs121912437]	-	[RCV000015907]	

Androgen Insensitivity Syndrome

Gene – ANDROGEN RECEPTOR; AR

OMIM - 313700 gene
OMIM - 300068 disease

Alleles

Many (see table below) – numbering differs from structure in PDB (see red number for comparison in structure).

Structures

WT structure in PDB (2AM9) aa 670-918, x-ray crystal of Lipid Binding Domain in complex with testosterone.
The numbering differs from some alleles to structure. The number in red corresponds to the structure.
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0003	ANDROGEN INSENSITIVITY, COMPLETE	AR, ARG773CYS **774**	-	-	[RCV000010478]	
.0005	ANDROGEN INSENSITIVITY, COMPLETE	AR, VAL866MET	[rs137852564]	-	[RCV000010480]	
.0012	ANDROGEN INSENSITIVITY, COMPLETE	AR, MET786VAL **787**	[rs137852570]	-	[RCV000010488]	
.0015	ANDROGEN INSENSITIVITY, COMPLETE	AR, ARG773HIS **774**	[rs137852572]	-	[RCV000010493]	
.0017	ANDROGEN INSENSITIVITY, COMPLETE	AR, VAL865MET **866**	-	-	[RCV000010480]	
.0028	ANDROGEN INSENSITIVITY, COMPLETE	AR, LEU676PRO **677**	[rs137852579]	-	[RCV000010506]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0034	ANDROGEN INSENSITIVITY, COMPLETE	AR, LEU707ARG	[rs137852585]	-	[RCV000010511]	
.0039	ANDROGEN INSENSITIVITY, COMPLETE	AR, MET780ILE	[rs137852589]	-	[RCV000010516]	
.0050	ANDROGEN INSENSITIVITY SYNDROME	AR, LEU712PHE	[rs137852595]	-	[RCV000010526]	
.0052	ANDROGEN INSENSITIVITY SYNDROME	AR, SER865PRO	[rs137852597]	-	[RCV000010528]	
.0053	ANDROGEN INSENSITIVITY SYNDROME	AR, PHE856LEU	[rs137852598]	[rs137852598]	[RCV000010490]	
.0055	ANDROGEN INSENSITIVITY, COMPLETE	AR, HIS689PRO	[rs137852599]	-	[RCV000010530]	
.0057	ANDROGEN INSENSITIVITY, COMPLETE	AR, GLY743GLU	[rs137852600]	-	[RCV000010533]	

Antithrombin Pittsburgh

Gene - SERPIN PEPTIDASE INHIBITOR; SERPINA1
MET358ARG creates new function for protein

OMIM - 107400 gene
OMIM - 613490 disease

Allele
107400.0026 (see table below)

Structures
WT structure in PDB (3NE4) aa 24-393
2 structures of the mutant - green in table.
Mutant structure in PDB (1OO8) aa 24-393
Mutant structure bound to trypsin (1OPH) aa 26-418

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0026	PI PITTSBURGH 'ANTITHROMBIN' PITTSBURGH	SERPINA1, MET358ARG	[rs121912713]	[rs17580]	[RCV000148878...]	**1OO8 2.65A aa 24-393** **1OPH 2.3A aa 26-418**

Apert Syndrome

Gene – FIBROBLAST GROWTH FACTOR RECEPTOR 2; FGFR2

OMIM - 176943 gene
OMIM - 101200 disease

Alleles

Three mutant alleles in table below.

Structures

WT structure in PDB (LEV2) aa 150-363 ligand binding domain D2, D3 Crystallized structure of the 2 mutant proteins with mutations in the ligand binding domain highlighted in green.
Create other mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0010	APERT SYNDROME ENDOMETRIAL CANCER, SOMATIC, INCLUDED	FGFR2, SER252TRP	[rs79184941]	[rs79184941]	[RCV000014191...]	
.0011	APERT SYNDROME	FGFR2, PRO253ARG	[rs77543610]	-	[RCV000014193]	**1II4 2.7A aa 147-366**
.0017	APERT SYNDROME	FGFR2, SER252PHE	[rs121918498]	-	[RCV000014201]	**1IIL 2.3A aa 147-266**

Aspartylglucosaminuria

Gene – ASPARTYLGLUCOSAMINURIA; AGU

OMIM - 613228 gene
OMIM - 208400 disease

Alleles

Mutant alleles in table below.

Structures

WT structure in PDB (1APY) aa 3-323 – Signal peptide included in the sequence, but not in the structure.
Numbering is off by 23 aa. Correct aa number for the structure in red.
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0001	ASPARTYL-GLUCOSAMINURIA, FINNISH TYPE	AGA, CYS163SER **140**	[rs121964904]	[rs121964904]	[RCV000000243]	
.0002	ASPARTYL-GLUCOSAMINURIA	AGA, GLY302ARG **279**	[rs121964905]	-	[RCV000000244]	
.0003	ASPARTYL-GLUCOSAMINURIA	AGA, CYS306ARG **283**	[rs121964906]	-	[RCV000000245]	
.0004	ASPARTYL-GLUCOSAMINURIA	AGA, GLY60ASP **37**	[rs121964907]	-	[RCV000000246]	
.0005	ASPARTYL-GLUCOSAMINURIA	AGA, ALA101VAL **78**	[rs121964908]	[rs121964908]	[RCV000000247]	
.0012	ASPARTYL-GLUCOSAMINURIA	AGA, SER72PRO **49**	[rs121964909]	-	[RCV000000253]	

Canavan Disease

Gene – ASPARTOACYLASE; ASPA

OMIM - 608034 gene
OMIM - 271900 disease

Alleles

Mutant alleles in table below.

Structures

WT structure in PDB (2O4H) aa 10-310
Crystallized structure of 2 mutant proteins highlighted in green.
Create other mutant structures using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0001	CANAVAN DISEASE	ASPA, GLU285ALA	[rs28940279]	[rs28940279]	[RCV000002723]	**4NFR 3.0A aa 10-310**
.0002	CANAVAN DISEASE	ASPA, CYS152ARG	[rs104894548]	-	[RCV000002724]	
.0003	CANAVAN DISEASE	ASPA, ALA305GLU	[rs28940574]	[rs28940574]	[RCV000002725]	
.0008	CANAVAN DISEASE	ASPA, TYR231CYS	[rs104894550]	[rs104894550]	[RCV000002730]	**4TNU 2.9A aa 10-310**
.0010	CANAVAN DISEASE	ASPA, GLU24GLY	[rs104894551]	-	[RCV000002732]	
.0011	CANAVAN DISEASE	ASPA, ASP249VAL	[rs104894552]	[rs104894552]	[RCV000002733]	

Ceroid Lipfuscinosis, Neuronal 1 (CLN1)

Gene – PALMITOYL-PROTEIN THIOESTERASE 1; PPT1

OMIM - 600722 gene
OMIM - 256730 disease

Alleles

Mutant alleles in table below.

Structures

WT structure in PDB (3GRO) aa 31-305
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0001	CEROID LIPOFUSCINOSIS, NEURONAL, 1	PPT1, ARG122TRP	[rs137852695]	[rs137852695]	[RCV000188724...]	
.0002	CEROID LIPOFUSCINOSIS, NEURONAL, 1	PPT1, THR75PRO	[rs137852696]	[rs137852696]	[RCV000188709...]	
.0003	CEROID LIPOFUSCINOSIS, NEURONAL, 1	PPT1, ASP79GLY	[rs137852697]	[rs137852697]	[RCV000009452]	
.0004	CEROID LIPOFUSCINOSIS, NEURONAL, 1	PPT1, LEU219GLN	[rs137852698]	-	[RCV000009453]	
.0009	CEROID LIPOFUSCINOSIS, NEURONAL, 1	PPT1, GLY108ARG	[rs137852701]	-	[RCV000009458]	
.0010	CEROID LIPOFUSCINOSIS, NEURONAL, 1	PPT1, CYS45TYR	[rs137852702]	-	[RCV000009459]	

Cowden Disease 1

Gene – PHOSPHATASE AND TENSIN HOMOLOG; PTEN

OMIM - 601728 gene
OMIM - 158350 disease

Alleles

Mutant alleles in table below

Structures

WT structure in PDB (1D5R) aa 14-281 and 313-351
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0001	COWDEN DISEASE 1	PTEN, GLY129GLU	[rs121909218]	-	[RCV000008255]	
.0017	COWDEN DISEASE 1	PTEN, ARG130GLN	[rs121909229]	-	[RCV000131067...]	
.0023	COWDEN DISEASE 1	PTEN, CYS124SER	-	-	[RCV000008283]	

Creutzfekdt-Jakob Disease

Gene – PRION PROTEIN; PRNP

OMIM - 176640 gene
OMIM - 123400 disease

Alleles

Mutant alleles in well-established C-terminal structure in table below.

Structures

WT structure in PDB (4KML)
aa 1-123 (less structured) and aa 117-225 (well structured).
The crystal is built as the N-terminal structure of one molecule interacting with the C-terminal of a second molecule.
Therefore, there is no overlap between the two segments.
Create mutant structures using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0014	CREUTZFELDT-JAKOB DISEASE	PRNP, VAL210ILE	[rs74315407]	[rs74315407]	[RCV000020255...]	
.0016	CREUTZFELDT-JAKOB DISEASE	PRNP, VAL180ILE	[rs74315408]	[rs74315408]	[RCV000014344...]	
.0023	CREUTZFELDT-JAKOB DISEASE	PRNP, ARG208HIS	[rs74315412]	[rs74315412]	[RCV000014352...]	

Crouzon Syndrome

Gene –FIBROBLAST GROWTH FACTOR RECEPTOR 2; FGFR2

OMIM - 176943 gene
OMIM - 123500 disease

Alleles

There are several mutations associated with Crouson syndrome found in the ligand binding domain (see table below). There is one mutant allele found in the kinase domain that has a crystal structure highlighted green.

Structures

WT structures in PDB (LEV2 and 2PSQ)
LEV2 - (aa 150-363 ligand binding domain D2, D3)
Create other mutant structure using PyMOL software.
2PSQ – (aa 468-765 kinase domain) gap aa 582-594
(2PZP) mutant structure crystallized kinase domain.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0004	CROUZON SYNDROME	FGFR2, TYR340HIS	[rs121918489]	-	[RCV000014175]	
.0005	CROUZON SYNDROME	FGFR2, SER354CYS	[rs121918490]	-	[RCV000014176]	
.0008	CROUZON SYNDROME	FGFR2, TYR328CYS	[rs121918493]	-	[RCV000014189]	
.0009	CROUZON SYNDROME	FGFR2, SER347CYS	[rs121918494]	-	[RCV000014190]	
.0013	CROUZON SYNDROME	FGFR2, CYS342TRP	[rs121918496]	-	[RCV000014195]	
.0020	CROUZON SYNDROME	FGFR2, LYS292GLU	[rs121918500]	-	[RCV000014204]	
.0021	CROUZON SYNDROME	FGFR2, TRP290ARG	[rs121918501]	-	[RCV000014205]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0022	CROUZON SYNDROME	FGFR2, TRP290GLY	[rs121918501]	-	[RCV000014206]	
.0034	CROUZON SYNDROME SCAPHOCEPHALY, MAXILLARY RETRUSION, AND MENTAL RETARDATION, INCLUDED	FGFR2, LYS526GLU	[rs121918507]	-	[RCV000014220...]	**2PZP 2.4A aa 468-766 gap 581-595**

Cystic Fibrosis

Gene – CYSTIC FIBROSIS; CF

OMIM - 602421 gene
OMIM - 219700 disease

Alleles

Mutant alleles are listed in the table below.
A deletion of aa 508 is the most common mutation associated with CF.
It is included because there is a crystal structure available, which is highlighted green below.

Structures

WT structure in PDB (2PZE).
This is a structure of the nucleotide-binding domain of CFTR aa 387-646.
It contains a deletion of aa 405-436 to solubilize the peptide for crystallization.
2PZF - Crystal structure of most common mutant allele of CF.
This is a signal aa mutation instead of a missense mutation.
Create other mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0001	CYSTIC FIBROSIS BRONCHIECTASIS WITH OR WITHOUT ELEVATED SWEAT CHLORIDE 1, MODIFIER OF, INCLUDED	CFTR, PHE508DEL (rs113993960)	[rs113993960]	-	[RCV000058929...]	**2PZF 2.0A aa 386-646** Similar to WT structure above with aa 508 deletion.
.0007	CYSTIC FIBROSIS	CFTR, ALA455GLU	[rs74551128]	[rs74551128]	[RCV000007531]	
.0010	CYSTIC FIBROSIS	CFTR, SER549ASN	[rs121908755]	[rs121908755]	[RCV000007536...]	
.0011	CYSTIC FIBROSIS	CFTR, SER549ILE	[rs121908755]	[rs121908755]	[RCV000007537]	
.0012	CYSTIC FIBROSIS	CFTR, SER549ARG	[rs121908757, rs121909005]	[rs121908757]	[RCV000211129...]	
.0013	CYSTIC FIBROSIS	CFTR, GLY551ASP	[rs75527207]	[rs75527207]	[RCV000211289...]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0015	CYSTIC FIBROSIS	CFTR, ALA559THR	[rs75549581]	[rs75549581]	[RCV000319242...]	
.0016	CYSTIC FIBROSIS	CFTR, ARG560THR	[rs80055610]	[rs80055610]	[RCV000007533...]	
.0017	CYSTIC FIBROSIS	CFTR, TYR563ASN	[rs121909006]	-	[RCV000007534]	
.0018	CYSTIC FIBROSIS	CFTR, PRO574HIS	[rs121908758]	-	[RCV000007539]	
.0028	CYSTIC FIBROSIS	CFTR, GLY458VAL	[rs121909009]	-	[RCV000007552]	
.0037	CYSTIC FIBROSIS	CFTR, GLY551SER	[rs121909013]	-	[RCV000211256...]	
.0046	CYSTIC FIBROSIS	CFTR, VAL520PHE	[rs77646904]	-	[RCV000007570]	
.0051	CYSTIC FIBROSIS	CFTR, SER492PHE	[rs121909017]	-	[RCV000007575]	
.0052	CYSTIC FIBROSIS	CFTR, ARG560LYS	[rs80055610]	[rs80055610]	[RCV000007576]	
.0069	CYSTIC FIBROSIS	CFTR, ALA534GLU	[rs121909022, rs387906368]	[rs387906368]	[RCV000007593]	
.0083	CYSTIC FIBROSIS	CFTR, GLY480CYS	[rs79282516]	-	[RCV000078977...]	
.0090	CYSTIC FIBROSIS	CFTR, ILE556VAL	[rs75789129]	[rs75789129]	[RCV000007617...]	
.0094	CYSTIC FIBROSIS	CFTR, GLN524HIS	-	-	[RCV000007612]	
.0121	CYSTIC FIBROSIS	CFTR, ARG553GLN	[rs121909044]	-	[RCV000007646]	
.0130	CYSTIC FIBROSIS	CFTR, ALA445GLU	-	-	[RCV000007656]	
.0136	CYSTIC FIBROSIS	CFTR, ALA561GLU	[rs121909047]	-	[RCV000007662]	

Emery-Dreifuss Muscular Dystrophy, autosomal dominant (EDMD2)

Gene – **LAMIN A/C; LMNA**

OMIM - 150330 gene
OMIM - 81350 disease

Alleles

Mutant alleles in the globular domain of Lamin A/C associated with EDMD2 shown in the table below.

Structures

WT structure in PDB (1IFR) aa 432-544 globular domain
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0002	EMERY-DREIFUSS MUSCULAR DYSTROPHY, AUTOSOMAL DOMINANT	LMNA, ARG453TRP	[rs58932704]	-	[RCV000057273...]	
.0004	EMERY-DREIFUSS MUSCULAR DYSTROPHY, AUTOSOMAL DOMINANT	LMNA, LEU530PRO	[rs60934003]	-	[RCV000057333...]	

Fatal Familial Insomnia

Gene – PRION PROTEIN; PRNP

OMIM - 176640 gene
OMIM - 600072 disease

Alleles

Mutant allele (D178N) in well established C-terminal structure in table below.
MET129 is a polymorphism in PRNP.
The VAL129 allele with the D178N mutation leads to a different phenotype (Creutzfeldt-Jacobs Disease).

Structures

WT structure in PDB (4KML)
aa 1-123 (less structured) and aa 117-225 (well structured).
The crystal is built as the N-terminal structure of one molecule interacting with the C-terminal of a second molecule.
Therefore, there is no overlap between the two segments.
Mutant structures available:
3HEQ (D178N and MET129)
3HJX (D178N and VAL129) – if interested in the comparison between the polymorphism.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0010	FATAL FAMILIAL INSOMNIA CREUTZFELDT-JAKOB DISEASE, INCLUDED	PRNP, ASP178ASN AND MET129	[rs74315403]	-	[RCV000014337...]	**3HEQ aa 126-228**

Gaucher Disease (Perinatal lethal, type I, type II and type III)

Gene – GLUCOSIDASE, BETA, ACID; GBA

OMIM - 606463 gene
OMIM - (608013) (230800) (230900) (231000) each disease type in parenthesis

Alleles

Many mutant alleles in table below - sorted by disease type.

Structures

WT structure in PDB (2NT0) aa 1-497
Create mutant structure using PyMOL software

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0037	GAUCHER DISEASE, PERINATAL LETHAL	GBA, HIS311ARG	[rs78198234]	-	[RCV000004569]	
.0039	GAUCHER DISEASE, PERINATAL LETHAL	GBA, VAL398PHE	[rs121908310]	-	[RCV000004544]	
.0041	GAUCHER DISEASE, PERINATAL LETHAL	GBA, ARG257GLU	[rs78973108]	[rs78973108]	[RCV000079357...]	
.0042	GAUCHER DISEASE, PERINATAL LETHAL	GBA, ARG131LEU	[rs80356763]	[rs80356763]	[RCV000004574...]	
.0044	GAUCHER DISEASE, PERINATAL LETHAL	GBA, PHE251LEU	[rs121908313]	-	[RCV000004577]	
.0010	GAUCHER DISEASE, TYPE I	GBA, PHE216TYR	[rs74500255]	-	[RCV000004537]	
.0012	GAUCHER DISEASE, TYPE I	GBA, LYS157GLN	[rs121908297]	-	[RCV000004539]	
.0016	GAUCHER DISEASE, TYPE I	GBA, PRO289LEU	[rs121908298]	-	[RCV000004547]	
.0019	GAUCHER DISEASE, TYPE I	GBA, PRO122SER	[rs121908299]	-	[RCV000004550]	
.0020	GAUCHER DISEASE, TYPE I	GBA, TYR212HIS	[rs121908300]	-	[RCV000004551]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0021	GAUCHER DISEASE, TYPE I	GBA, GLY478SER	[rs121908301]	-	[RCV000004552]	
.0022	GAUCHER DISEASE, TYPE I	GBA, ARG496HIS	[rs75822236]	-	[RCV000020153...]	
.0024	GAUCHER DISEASE, TYPE I	GBA, VAL15LEU	[rs121908302]	-	[RCV000004556]	
.0025	GAUCHER DISEASE, TYPE I	GBA, GLY46GLU	[rs77829017]	[rs77829017]	[RCV000004532]	
.0027	GAUCHER DISEASE, TYPE I	GBA, PHE216VAL	[rs121908303]	-	[RCV000004559]	
.0028	GAUCHER DISEASE, TYPE I	GBA, ALA309VAL	[rs78396650]	-	[RCV000004560]	
.0029	GAUCHER DISEASE, TYPE I	GBA, TRP312CYS	[rs121908304]	-	[RCV000004561]	
.0032	GAUCHER DISEASE, TYPE I	GBA, SER364THR	[rs121908307]	[rs121908307]	[RCV000004564]	
.0036	GAUCHER DISEASE, TYPE I	GBA, PRO401LEU	[rs74598136]	-	[RCV000004568]	
.0045	GAUCHER DISEASE, TYPE I	GBA, LEU371VAL	[rs121908314]	-	[RCV000004578]	
.0004	GAUCHER DISEASE, TYPE I GAUCHER DISEASE, PERINATAL LETHAL, INCLUDED	GBA, ARG119GLN	[rs79653797]	[rs79653797]	[RCV000004519...]	
.0017	GAUCHER DISEASE, TYPE I GAUCHER DISEASE, TYPE II, INCLUDED	GBA, THR323ILE	[rs76539814]	-	[RCV000041967...]	
.0008	GAUCHER DISEASE, TYPE I GAUCHER DISEASE, TYPE II, INCLUDED GAUCHER DISEASE, TYPE III, INCLUDED	GBA, ARG463CYS	[rs80356771]	-	[RCV000004528...]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0026	GAUCHER DISEASE, TYPE I GAUCHER DISEASE, TYPE III, INCLUDED	GBA, ASN188SER	[rs364897]	[rs364897]	[RCV000004557...]	
.0040	GAUCHER DISEASE, TYPE I GAUCHER DISEASE, TYPE III, INCLUDED	GBA, GLY377SER	[rs121908311]	[rs121908311]	[RCV000004572...]	
.0043	GAUCHER DISEASE, TYPE I GAUCHER DISEASE, TYPE III, INCLUDED	GBA, LYS79ASN	[rs121908312]	-	[RCV000079344...]	
.0003	GAUCHER DISEASE, TYPE I	GBA, ASN370SER	[rs76763715]	-	[RCV000004515...]	
.0002	GAUCHER DISEASE, TYPE II	GBA, PRO415ARG	[rs121908295]	-	[RCV000004514]	
.0030	GAUCHER DISEASE, TYPE II	GBA, GLY325ARG	[rs121908305]	-	[RCV000079331...]	
.0031	GAUCHER DISEASE, TYPE II	GBA, CYS342GLY	[rs121908306]	-	[RCV000004563]	
.0001	GAUCHER DISEASE, TYPE II GAUCHER DISEASE, TYPE III, INCLUDED GAUCHER DISEASE, TYPE I, INCLUDED	GBA, LEU444PRO	[rs421016]	-	[RCV000004511...]	
.0007	GAUCHER DISEASE, TYPE III	GBA, ASP409VAL	[rs77369218]	-	[RCV000020149...]	
.0035	GAUCHER DISEASE, TYPE III	GBA, ARG353GLY	[rs121908308]	-	[RCV000004567]	
.0005	GAUCHER DISEASE, TYPE III GAUCHER DISEASE, TYPE I, INCLUDED	GBA, VAL394LEU	[rs80356769]	[rs80356769]	[RCV000004520...]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0013	GAUCHER DISEASE, TYPE III GAUCHER DISEASE, TYPE II, INCLUDED GAUCHER DISEASE, TYPE I, INCLUDED	GBA, PHE213ILE	[rs381737]	-	[RCV000004542...]	
.0006	GAUCHER DISEASE, TYPE IIIC GAUCHER DISEASE, TYPE I, INCLUDED GAUCHER DISEASE, TYPE II, INCLUDED GAUCHER DISEASE, TYPE III, INCLUDED GAUCHER DISEASE, PERINATAL LETHAL, INCLUDED	GBA, ASP409HIS	[rs77369218, rs1064651]	-	[RCV000004522...]	

Gerstmann-Straussler Disease

Gene – PRION PROTEIN; PRNP

OMIM - 176640 gene
OMIM - 137440 disease

Alleles

Mutant alleles in well-established C-terminal structure in table below.

Structures

WT structure in PDB (4KML)
aa 1-123 (less structured) and aa 117-225 (well structured).
The crystal is built as the N-terminal structure of one molecule interacting with the C-terminal of a second molecule.
Therefore, there is no overlap between the two segments.
2 mutant structures highlighted in green.
Create other mutant structures using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0011	GERSTMANN-STRAUSSLER DISEASE	PRNP, PHE198SER	[rs74315405]	-	[RCV000020252...]	**3HER 1.85A aa 125-226** **3HES 2.0A aa 125-226**
.0012	GERSTMANN-STRAUSSLER DISEASE	PRNP, GLN217ARG	[rs74315406]	-	[RCV000014341...]	
.0021	GERSTMANN-STRAUSSLER DISEASE	PRNP, GLY131VAL	[rs74315410]	[rs74315410]	[RCV000014351]	
.0024	GERSTMANN-STRAUSSLER DISEASE SPONGIFORM ENCEPHALOPATHY WITH NEUROPSYCHIATRIC FEATURES, INCLUDED	PRNP, HIS187ARG	[rs74315413]	-	[RCV000014353...]	
.0026	GERSTMANN-STRAUSSLER	PRNP, ALA133VAL	[rs74315415]	-	[RCV000014356]	

Hemochromatosis

Gene – HFE GENE; HFE

OMIM - 613609 gene
OMIM - 235200 disease

Alleles

Mutant alleles in table below.

Structures

WT structure in PDB (1A6Z) aa 4-275 bound to beta-2 microglobulin chains. Four protein chains are in this structure chains A and C correspond to HFE and either chain can be utilized for analysis. Numbering is different. Structure corresponds to mature protein, -22aa from the initial methionine. The numbers corresponding to the structure are in red. Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0001	HEMOCHROMATOSIS, TYPE 1 PORPHYRIA CUTANEA TARDA, SUSCEPTIBILITY TO, INCLUDED	HFE, CYS282TYR **260**	[rs1800562]	[rs1800562]	[RCV000210820...]	
.0002	HEMOCHROMATOSIS, TYPE 1 MICROVASCULAR COMPLICATIONS OF DIABETES, INCLUDED	HFE, HIS63ASP **41**	[rs1799945]	[rs1799945]	[RCV000000027...]	
.0003	HEMOCHROMATOSIS, TYPE 1	HFE, SER65CYS **43**	[rs1800730]	[rs1800730]	[RCV000290779...]	
.0007	HEMOCHROMATOSIS, TYPE 1	HFE, GLN127HIS **105**	[rs28934595]	-	[RCV000000034]	
.0009	HEMOCHROMATOSIS, TYPE 1	HFE, ILE105THR **83**	[rs28934596]	-	[RCV000000029]	
.0010	HEMOCHROMATOSIS, TYPE 1	HFE, GLY93ARG **71**	[rs28934597]	[rs28934597]	[RCV000000030]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0011	HEMOCHROMATOSIS, TYPE 1	HFE, GLN283PRO **261**	[rs111033563]	-	[RCV000000036]	

Hereditary Nonpolyposis Colorectal Cancer Type 2

Gene –MutL, E. COLI, HOMOLOG OF, 1; MLH1

OMIM - 120436 gene
OMIM - 609310 disease

Alleles

Mutant alleles in table below.

Structures

WT structures in PDB (4P7A) aa 3-336
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0002	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 2	MLH1, SER44PHE	[rs63751109]	-	[RCV000018608...]	
.0008	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 2	MLH1, HIS329PRO	[rs63750710]	-	[RCV000075954...]	
.0011	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 2 MISMATCH REPAIR CANCER SYNDROME, INCLUDED	MLH1, GLY67TRP	[rs63750206]	-	[RCV000018619...]	
.0017	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 2	MLH1, THR117MET	[rs63750781]	-	[RCV000018626...]	
.0029	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 2	MLH1, GLY67GLU	[rs63749939]	-	[RCV000132445...]	
.0030	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 2	MLH1, ARG265CYS	[rs63751194]	-	[RCV000022502...]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0034	MISMATCH REPAIR CANCER SYNDROME	MLH1, LEU73ARG	[rs397514684]	-	[RCV000213759...]	
.0002	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 2	MLH1, SER44PHE	[rs63751109]	-	[RCV000018608...]	

Ladd Syndrome

Gene –FIBROBLAST GROWTH FACTOR RECEPTOR 2; FGFR2

OMIM - 176943 gene
OMIM - 149730 disease

Alleles

Two mutant alleles in table below. They are both contained in the kinase domain.

Structures

WT structures in PDB (2PSQ)
2PSQ – (aa 468-765 kinase domain)
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0035	LADD SYNDROME	FGFR2, ALA648THR	[rs121918508]	-	[RCV000014222]	
.0037	LADD SYNDROME	FGFR2, ALA628THR	[rs121918509]	-	[RCV000014224]	

Lynch Syndrome; Hereditary Nonpolyposis Colorectal Cancer, Type 1

Gene – MutS, E. COLI, HOMOLOG OF, 2; MSH2

OMIM - 609309 gene
OMIM - 120435 disease

Alleles

Mutant alleles in table below.

Structures

WT structures in PDB (2O8B) aa 1-854
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0001	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 1	MSH2, PRO622LEU	[rs28929483]	-	[RCV000001823...]	
.0004	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 1	MSH2, HIS639TYR	[rs28929484]	-	[RCV000001826...]	
.0007	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 1	MSH2, ARG524PRO	[rs63751207]	-	[RCV000256140...]	
.0012	COLORECTAL CANCER, HEREDITARY, NONPOLYPOSIS, TYPE 1	MSH2, ALA636PRO	[rs63750875]	[rs63750875]	[RCV000130428...]	

Lesch-Nyhan Syndrome

Gene – HYPOXANTHINE GUANINE PHOSPHORIBOSYLTRANSFERASE 1; HPRT1

OMIM - 308000 gene
OMIM - 300322 disease

Alleles

Mutant alleles in table below.

Structures

WT structures in PDB (5HIA) use chain A, aa 2-101 and 120-217 number in corresponding structure in red.
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0006	LESCH-NYHAN SYNDROME HPRT DETROIT	HPRT, LEU41PRO **40**	[rs137852480]	-	[RCV000010723...]	
.0008	LESCH-NYHAN SYNDROME HPRT FLINT	HPRT, PHE74LEU 73	[rs137852481]	-	[RCV000010727...]	
.0012	LESCH-NYHAN SYNDROME HPRT MIDLAND	HPRT, VAL130ASP **129**	[rs137852483]	-	[RCV000010734...]	
.0015	LESCH-NYHAN SYNDROME HPRT NEW BRITON	HPRT, PHE199VAL **198**	[rs137852486]	-	[RCV000010741...]	
.0016	LESCH-NYHAN SYNDROME HPRT NEW HAVEN	HPRT, GLY70GLU **69**	[rs137852487]	-	[RCV000010743...]	
.0017	LESCH-NYHAN SYNDROME HPRT YALE	HPRT, GLY71ARG **70**	[rs137852488]	-	[RCV000010745...]	
.0019	LESCH-NYHAN SYNDROME	HPRT, HIS203ASP	[rs137852490]	-	[RCV000010747]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0020	LESCH-NYHAN SYNDROME	HPRT, ARG44LYS	[rs137852491]	-	[RCV000010748]	
.0036	LESCH-NYHAN SYNDROME, HPRT MONTREAL	HPRT, MET56THR	[rs137852495]	-	[RCV000010764...]	
.0037	LESCH-NYHAN SYNDROME	HPRT, MET143LYS **142**	[rs137852496]	-	[RCV000010765]	
.0055	LESCH-NYHAN SYNDROME HPRT TOKYO	HPRT, GLY140ASP **139**	[rs137852503]	-	[RCV000010789...]	
.0060	LESCH-NYHAN SYNDROME, GOUT, HPRT-RELATED,	HPRT, ARG48HIS **47**	[rs387906725]	-	[RCV000022877...]	

Lipodystrophy, Familial Partial Type 2

Gene – LAMIN A/C; LMNA

OMIM - 150330 gene
OMIM - 151660 disease

Alleles

Mutant alleles in the globular domain of Lamin A/C associated with lipodystrophy (FPT2) shown in the table below.

Structures

WT structure in PDB (1IFR) aa 432-544 globular domain
A structure for one mutant allele is available, shown highlighted green below.
Create other structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0011	LIPODYSTROPHY, FAMILIAL PARTIAL, TYPE 2	LMNA, ARG482TRP	[rs57920071]	-	[RCV000015579...]	**3GEF 1.5A aa 435-552**
.0012	LIPODYSTROPHY, FAMILIAL PARTIAL, TYPE 2	LMNA, ARG482LEU	[rs11575937]	[rs11575937]	[RCV000015580...]	
.0015	LIPODYSTROPHY, FAMILIAL PARTIAL, TYPE 2	LMNA, GLY465ASP	[rs61282106]	-	[RCV000015584...]	

Macrocephaly/Autism Syndrome

Gene – PHOSPHATASE AND TENSIN HOMOLOG; PTEN

OMIM - 601728 gene
OMIM - 605309 disease

Alleles

Mutant alleles in table below.

Structures

WT structure in PDB (1D5R) aa 14-281 and 313-351
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0037	MACROCEPHALY/ AUTISM SYNDROME	PTEN, HIS93ARG	[rs121909238]	-	[RCV000008298...]	
.0038	MACROCEPHALY/ AUTISM SYNDROME	PTEN, ASP252GLY	[rs121909239]	-	[RCV000008299]	
.0042	MACROCEPHALY/ AUTISM SYNDROME	PTEN, THR167ASN	[rs397514559]	-	[RCV000032872]	
.0043	MACROCEPHALY/ AUTISM SYNDROME	PTEN, THR131ILE	[rs397514560]	-	[RCV000032873]	

Mandibuloacral Dysplasia with Type A Lipodystrophy

Gene – LAMIN A/C; LMNA

OMIM - 150330 gene
OMIM - 248370 disease

Alleles

Mutant alleles in the globular domain of Lamin A/C associated with mandibuloacral dysplasia with type A lipodystrrophy shown in the table below.

Structures

WT structure in PDB (1IFR) aa 432-544 globular domain
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0026	MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY	LMNA, ARG527CYS	[rs57318642]	-	[RCV000192240...]	
.0037	MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY	LMNA, ALA529VAL	[rs60580541]	-	[RCV000015608...]	
.0046	MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY	LMNA, ALA529THR	[rs121912494]	-	[RCV000057331...]	

Metachromatic Leukodystrophy

Gene – CEREBROSIDE 3-SULFATASE; ARSA (ARYLSULFATASE A)

OMIM - 607574 gene
OMIM - 250100 disease

Alleles

Mutant alleles in table below.

Structures

WT structure in PDB (1AUK) aa 19-503
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0005	METACHROMATIC LEUKODYSTROPHY, ADULT	ARSA, GLY99ASP	[rs74315455]	-	[RCV000003198...]	
.0010	METACHROMATIC LEUKODYSTROPHY, LATE-ONSET	ARSA, ARG84GLN	[rs74315458]	[rs74315458]	[RCV000003205...]	
.0011	METACHROMATIC LEUKODYSTROPHY, LATE INFANTILE	ARSA, GLY309SER	[rs74315459]	[rs74315459]	[RCV000003206]	
.0013	METACHROMATIC LEUKODYSTROPHY, SEVERE	ARSA, GLY86ASP	[rs74315460]	-	[RCV000003207]	
.0014	METACHROMATIC LEUKODYSTROPHY, SEVERE	ARSA, SER96LEU	[rs199476371]	-	[RCV000003208...]	
.0015	METACHROMATIC LEUKODYSTROPHY	ARSA, GLY122SER	[rs74315461]	[rs74315461]	[RCV000078945...]	
.0016	METACHROMATIC LEUKODYSTROPHY, SEVERE	ARSA, PRO136LEU	[rs74315462]	[rs74315462]	[RCV000003210]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0018	METACHROMATIC LEUKODYSTROPHY	ARSA, GLY154ASP	[rs74315463]	-	[RCV000003212]	
.0020	METACHROMATIC LEUKODYSTROPHY	ARSA, PRO167ARG	[rs74315465]	-	[RCV000003214]	
.0022	METACHROMATIC LEUKODYSTROPHY	ARSA, ALA212VAL	[rs74315467]	[rs74315467]	[RCV000003216]	
.0023	METACHROMATIC LEUKODYSTROPHY	ARSA, ALA224VAL	[rs74315468]	-	[RCV000343115...]	
.0024	METACHROMATIC LEUKODYSTROPHY	ARSA, PRO231THR	[rs74315469]	-	[RCV000003218]	
.0025	METACHROMATIC LEUKODYSTROPHY	ARSA, ARG244CYS	[rs74315470]	[rs74315470]	[RCV000003219]	
.0026	METACHROMATIC LEUKODYSTROPHY, SEVERE	ARSA, GLY245ARG	[rs74315471]	[rs74315471]	[RCV000020321...]	
.0027	METACHROMATIC LEUKODYSTROPHY, SEVERE	ARSA, THR274MET	[rs74315472]	[rs74315472]	[RCV000169246...]	
.0029	METACHROMATIC LEUKODYSTROPHY	ARSA, ARG288CYS	[rs74315473]	[rs74315473]	[RCV000003223]	
.0030	METACHROMATIC LEUKODYSTROPHY, SEVERE	ARSA, SER295TYR	[rs74315474]	-	[RCV000003224]	
.0032	METACHROMATIC LEUKODYSTROPHY, SEVERE	ARSA, ASP335VAL	[rs74315475]	[rs74315475]	[RCV000003226...]	
.0033	METACHROMATIC LEUKODYSTROPHY, SEVERE	ARSA, ARG370TRP	[rs74315476]	[rs74315476]	[RCV000003227...]	
.0037	METACHROMATIC LEUKODYSTROPHY	ARSA, ARG390TRP	[rs74315480]	[rs74315480]	[RCV000003231]	
.0042	METACHROMATIC LEUKODYSTROPHY, ADULT	ARSA, PRO135LEU	[rs121434215]	-	[RCV000003235]	
.0043	METACHROMATIC LEUKODYSTROPHY ADULT	ARSA, THR286PRO	[rs28940894]	[rs28940894]	[RCV000003236]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0044	METACHROMATIC LEUKODYSTROPHY, LATE INFANTILE	ARSA, GLU253LYS	[rs74315483]	-	[RCV000364541...]	
.0046	METACHROMATIC LEUKODYSTROPHY, LATE INFANTILE	ARSA, CYS300PHE	[rs74315484]	-	[RCV000003239...]	
.0047	METACHROMATIC LEUKODYSTROPHY, JUVENILE	ARSA, PRO425THR	[rs74315485]	-	[RCV000003240]	

Mucopolysaccharidosis Type VII

Gene – BETA-GLUCURONIDASE; GUSB

OMIM - 611499 gene
OMIM - 253220 disease

Alleles

Mutant alleles in table below.

Structures

WT structure in PDB (3HN3) aa 22-632
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0001	MUCOPOLYSACCHARIDOSIS, TYPE VII	GUSB, ALA619VAL	[rs121918172]	[rs121918172]	[RCV000000941]	
.0002	MUCOPOLYSACCHARIDOSIS, TYPE VII	GUSB, ARG382CYS	[rs121918173]	[rs121918173]	[RCV000000942...]	
.0003	MUCOPOLYSACCHARIDOSIS, TYPE VII	GUSB, ARG216TRP	[rs121918174]	[rs121918174]	[RCV000000943]	
.0004	MUCOPOLYSACCHARIDOSIS, TYPE VII	GUSB, ALA354VAL	[rs121918175]	[rs121918175]	[RCV000000944]	
.0005	MUCOPOLYSACCHARIDOSIS, TYPE VII	GUSB, ARG611TRP	[rs121918176]	[rs121918176]	[RCV000000945]	
.0006	MUCOPOLYSACCHARIDOSIS, TYPE VII	GUSB, PRO148SER	[rs121918177]	-	[RCV000000946]	
.0007	MUCOPOLYSACCHARIDOSIS, TYPE VII	GUSB, TYR495CYS	[rs121918178]	-	[RCV000000947]	
.0012	MUCOPOLYSACCHARIDOSIS, TYPE VII	GUSB, LEU176PHE	[rs121918181]	[rs121918181]	[RCV000000953...]	
.0013	MUCOPOLYSACCHARIDOSIS, TYPE VII	GUSB, LYS350ASN	[rs121918182]	[rs121918182]	[RCV000000954]	
.0014	MUCOPOLYSACCHARIDOSIS, TYPE VII	GUSB, ARG577LEU	[rs121918183]	-	[RCV000000950]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0015	MUCOPOLYSACCHARIDOSIS, TYPE VII	GUSB, TRP627CYS	[rs121918184]	-	[RCV000000955]	

Neurofibromatosis, Type I; NF1

Gene – NEUROFIBROMIN 1; NF1

OMIM - 613113 gene
OMIM - 162200 disease

Alleles

Mutant alleles contained in the GAP domain in table below.

Structures

WT structure in PDB (1NF1) aa 1206-1529, GAP related domain
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0005	NEUROFIBROMATOSIS, TYPE I	NF1, LYS1423GLU	[rs137854550]	[rs137854550]	[RCV000000364]	
.0016	NEUROFIBROMATOSIS, TYPE I	NF1, ARG1391SER	[rs137854554]	-	[RCV000000376]	
.0022	NEUROFIBROMATOSIS, TYPE I	NF1, ARG1276PRO	[rs137854556]	-	[RCV000000381]	
.0041	NEUROFIBROMATOSIS, TYPE I	NF1, LEU1243PRO	[rs137854564]	-	[RCV000000402]	

Neurofibromatosis Type 2 (NF2)

Gene – NEUROFIBROMATOSIS, TYPE II; NF2
MERLIN, SCHWANNOMIN; SCH

OMIM - 607379 gene
OMIM - 101000 disease

Alleles

There is one mutant allele in the FERM domain, see the table below.

Structures

WT structure in PDB (1H4R) aa 20-313 (FERM domain)
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0016	NEUROFIBROMATOSIS, TYPE II	NF2, PHE62SER	[rs121434261]	-	[RCV000003458]	

Parkinson Disease 7, Early-Onset; PARK7

Gene – ONCOGENE DJ1; DJ1

OMIM - 602533 gene
OMIM - 606324 disease

Alleles

Mutant alleles in table below.

Structures

WT structure in PDB (2OR3) aa 2-188
One mutant crystal structure in green.
Create other mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0002	PARKINSON DISEASE 7, AUTOSOMAL RECESSIVE EARLY-ONSET	PARK7, LEU166PRO	[rs28938172]	-	[RCV000007480]	
.0003	PARKINSON DISEASE 7, AUTOSOMAL RECESSIVE EARLY-ONSET	PARK7, MET26ILE	[rs74315351]	-	[RCV000007481]	**2RK4 1.15A aa 2-187**
.0004	PARKINSON DISEASE 7, AUTOSOMAL RECESSIVE EARLY-ONSET	PARK7, ASP149ALA	[rs74315352]	[rs74315352]	[RCV000007482]	
.0005	PARKINSON DISEASE 7, AUTOSOMAL RECESSIVE EARLY-ONSET	PARK7, GLU64ASP	[rs74315353]	-	[RCV000007483]	

Pfeiffer syndrome

Gene –FIBROBLAST GROWTH FACTOR RECEPTOR 2; FGFR2

OMIM - 176943 gene
OMIM - 101600 disease

Alleles

There are four mutations associated with Pfeiffer syndrome found in the ligand binding domain (see the table below).
There is one mutant allele found in the kinase domain that has a crystal structure.

Structures

WT structures in PDB (LEV2 and 2PSQ)
LEV2 - (aa 150-363 ligand binding domain D2, D3)
Create mutant structure using PyMOL software.
2PSQ – (aa 468-765 kinase domain)
(2Q0B) mutant structure in kinase domain

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0012	PFEIFFER SYNDROME	FGFR2, THR341PRO	[rs121918495]	-	[RCV000014194]	
.0019	PFEIFFER SYNDROME	FGFR2, TRP290CYS	[rs121918499]	-	[RCV000014203]	
.0029	PFEIFFER SYNDROME GASTRIC CANCER, SOMATIC, INCLUDED	FGFR2, SER267PRO	[rs121918505]	-	[RCV000014213...]	
.0033	PFEIFFER SYNDROME	FGFR2, GLU565ALA	[rs121918506]	-	[RCV000014219]	**2Q0B 2.9A aa 458-768**
.0039	PFEIFFER SYNDROME	FGFR2, ASP321ALA	[rs121918510]	-	[RCV000014227]	

Phenylketonuria (PKU)

Gene – PHENYLALANINE HYDROXYLASE; PAH

OMIM - 612349 gene
OMIM - 261600 disease

Alleles

Mutant alleles (aa 117-42) in table below.

Structures

WT structure in PDB (4ANP) aa 117-425
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0002	PHENYLKETONURIA	PAH, ARG408TRP	[rs5030858]	[rs5030858]	[RCV000078507...]	
.0003	PHENYLKETONURIA	PAH, LEU311PRO	[rs62642936]	[rs62642936]	[RCV000000608...]	
.0004	PHENYLKETONURIA	PAH, GLU280LYS	[rs62508698]	-	[RCV000078532...]	
.0006	PHENYLKETONURIA	PAH, ARG261GLN	[rs5030849]	-	[RCV000078530...]	
.0007	PHENYLKETONURIA	PAH, ARG252TRP	[rs5030847]	-	[RCV000089079...]	
.0010	PHENYLKETONURIA	PAH, ARG158GLN	[rs5030843]	-	[RCV000078522...]	
.0012	PHENYLKETONURIA	PAH, PRO281LEU	[rs5030851]	[rs5030851]	[RCV000078534...]	
.0013	PHENYLKETONURIA	PAH, TYR204CYS	[rs62514927]	[rs62514927]	[RCV000000621...]	
.0014	PHENYLKETONURIA	PAH, ARG243GLN	[rs62508588]	-	[RCV000000622...]	
.0016	PHENYLKETONURIA	PAH, ARG413PRO	[rs79931499]	[rs79931499]	[RCV000000623...]	
.0023	PHENYLKETONURIA	PAH, SER273PHE	[rs62514953]	[rs62514953]	[RCV000000629...]	
.0026	PHENYLKETONURIA	PAH, LEU255SER	[rs62642930]	-	[RCV000000631...]	
.0028	PHENYLKETONURIA	PAH, ALA259VAL	[rs118203921]	[rs118203921]	[RCV000000633...]	
.0029	PHENYLKETONURIA	PAH, TYR277ASP	[rs78655458]	[rs78655458]	[RCV000000634...]	
.0032	PHENYLKETONURIA	PAH, SER349ARG	-	-	[RCV000000637]	
.0035	PHENYLKETONURIA	PAH, GLU221GLY	[rs62514934]	[rs62514934]	[RCV000000640...]	
.0038	PHENYLKETONURIA	PAH, ARG408GLN	[rs5030859]	[rs5030859]	[RCV000088806...]	
.0039	PHENYLKETONURIA	PAH, PHE299CYS	[rs62642933]	[rs62642933]	[RCV000089148...]	
.0041	PHENYLKETONURIA	PAH, SER349PRO	[rs62508646]	-	[RCV000000646...]	
.0045	PHENYLKETONURIA	PAH, VAL388MET	[rs62516101]	-	[RCV000088774...]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0047	PHENYLKETONURIA	PAH, PRO244LEU	[rs118203923]	[rs118203923]	[RCV000000652...]	
.0062	PHENYLKETONURIA	PAH, PRO407LEU	[rs62644473]	-	[RCV000000667...]	

Porphyria: Acute Intermittent

Gene – HYDROXYMETHYLBILANE SYNTHASE; HMBS PORPHOBILINOGEN DEAMINASE; PBGD

OMIM - 609806 gene
OMIM - 176000 disease

Alleles

Mutant alleles in table below.

Structures

WT structure in PDB (3ECR) aa 21-359 (gaps 59-79 and 261-264). Numbering in structure off by 3 aa. See corrections for structure in red below.
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0004	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **119** ARG116TRP	[rs118204094]	-	[RCV000001510]	
.0005	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **170** ARG167GLN	[rs118204095]	[rs118204095]	[RCV000001511]	
.0006	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **176** ARG173GLN	[rs118204096]	-	[RCV000001512]	
.0008	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **152** ARG149GLN	[rs118204098]	-	[RCV000001514]	
.0009	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **258** LEU245ARG	[rs118204099]	-	[RCV000001515]	
.0013	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **170** ARG167TRP	[rs118204101]	[rs118204101]	[RCV000001521]	
.0014	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **170** ARG167LEU	[rs118204095]	[rs118204095]	[RCV000001522]	
.0015	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **29** ARG26HIS	[rs118204103]	-	[RCV000001508]	
.0016	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **34** ALA31THR	[rs118204104]	-	[RCV000001519]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0017	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **37** GLN34LYS	[rs118204105]	-	[RCV000001520]	
.0023	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **114** GLY111ARG	[rs118204107]	-	[RCV000001529]	
.0025	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **108** LEU177ARG	[rs118204108]	-	[RCV000001531]	
.0026	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **204** ARG201TRP	[rs118204109]	-	[RCV000001527]	
.0027	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **226** GLU223LYS	[rs118204110]	-	[RCV000001532]	
.0029	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **250** CYS247ARG	[rs118204111]	-	[RCV000001534]	
.0031	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **253** GLU250LYS	[rs118204112]	-	[RCV000001536]	
.0032	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **255** ALA252THR	[rs118204113]	-	[RCV000001537]	
.0033	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **255** ALA252VAL	[rs118204114]	[rs118204114]	[RCV000001538]	
.0034	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **259** HIS256ASN	[rs118204115]	-	[RCV000001539]	
.0038	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **219** GLY216ASP	[rs118204116]	-	[RCV000001543]	
.0045	PORPHYRIA, ACUTE INTERMITTENT	HMBS, **84** LEU81PRO	[rs118204119]	-	[RCV000001550]	

Porphyria: Congenital Erythropoitic

Gene – UROPORPHYRINOGEN III SYNTHASE; UROS

OMIM - 606938 gene
OMIM - 263700 disease

Alleles

Mutant alleles in table below.

Structures

WT structure in PDB (1JR2) aa 1-260
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0001	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	UROS, CYS73ARG	[rs121908012]	[rs121908012]	[RCV000003948]	
.0002	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	UROS, PRO53LEU	[rs121908013]	-	[RCV000003949]	
.0003	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	UROS, ALA66VAL	[rs28941774]	-	[RCV000003950]	
.0004	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	UROS, THR62ALA	[rs28941775]	[rs28941775]	[RCV000003951]	
.0005	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	UROS, THR228MET	[rs121908014]	[rs121908014]	[RCV000003952]	
.0006	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	UROS, LEU4PHE	[rs121908015]	[rs121908015]	[RCV000003953]	
.0009	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	UROS, VAL82PHE	[rs121908016]	[rs121908016]	[RCV000003956]	
.0010	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	UROS, GLY188ARG	[rs121908017]	-	[RCV000003957]	
.0011	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	UROS, GLU81ASP	[rs121908018]	-	[RCV000003958]	
.0012	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	UROS, GLY188TRP	[rs121908017]	-	[RCV000003959]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0017	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	UROS, GLY225SER	[rs121908020]	[rs121908020]	[RCV000003964]	
.0020	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	UROS, PRO248GLN	[rs121908021]	-	[RCV000003967]	

Porphyria: Cutanea Tarda and Hepatoerythropoietic

Gene – UROPORPHYRINOGEN DECARBOXYLASE; UROD

OMIM - 613521 gene
OMIM - (176100**)** disease

Alleles – Mutant alleles in table below.

Structures
WT structure in PDB (1URO) aa 10-366
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0001	PORPHYRIA CUTANEA TARDA	UROD, GLY281VAL	[rs121918057]	[rs121918057]	[RCV000000082]	
.0002	PORPHYRIA, HEPATO-ERYTHROPOIETIC PORPHYRIA CUTANEA TARDA	UROD, GLY281GLU	[rs121918057]	[rs121918057]	[RCV000240661...]	
.0004	PORPHYRIA, HEPATO-ERYTHROPOIETIC	UROD, GLU167LYS	[rs121918058]	-	[RCV000000086]	
.0005	PORPHYRIA, HEPATO-ERYTHROPOIETIC	UROD, ARG292GLY	[rs121918059]	[rs121918059]	[RCV000000087]	
.0006	PORPHYRIA, HEPATO-ERYTHROPOIETIC	UROD, PRO62LEU	[rs121918060]	-	[RCV000000088]	
.0007	PORPHYRIA, HEPATO-ERYTHROPOIETIC	UROD, TYR311CYS	[rs121918061]	[rs121918061]	[RCV000000089]	
.0008	PORPHYRIA CUTANEA TARDA	UROD, GLU314GLU	[rs121918062]	-	[RCV000000090]	
.0009	PORPHYRIA CUTANEA TARDA	UROD, MET165ARG	[rs121918063]	-	[RCV000000091]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0010	PORPHYRIA CUTANEA TARDA	UROD, LEU195PHE	[rs121918064]	-	[RCV000000092]	
.0011	PORPHYRIA CUTANEA TARDA	UROD, ASN304LYS	[rs121918065]	[rs121918065]	[RCV000000093]	
.0012	PORPHYRIA CUTANEA TARDA	UROD, ARG332HIS	[rs121918066]	[rs121918066]	[RCV000000094]	
.0001	PORPHYRIA CUTANEA TARDA	UROD, GLY281VAL	[rs121918057]	[rs121918057]	[RCV000000082]	

Retinoblastoma

Gene – RB1 GENE; RB1

OMIM - 614041 gene
OMIM - 180200 disease

Alleles

Mutant alleles in table below.

Structure

WT structure in PDB (2R7G) aa 380-578 (small gap 502-509) and aa 643-785
Create mutant structure using PyMOL software

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0004	RETINOBLASTOMA	RB1, SER567LEU	[rs137853292]	-	[RCV000013947]	
.0019	RETINOBLASTOMA	RB1, ARG661TRP	[rs137853294]	[rs137853294]	[RCV000013962...]	
.0024	RETINOBLASTOMA	RB1, CYS712ARG	[rs137853296]	-	[RCV000013968]	

Rett Syndrome

Gene – METHYL-CpG-BINDING PROTEIN 2; MECP2

OMIM - 300005 gene
OMIM - 312750 disease

Alleles
Mutant alleles in the methyl-CpG binding domain listed in the table below.

Structure

WT structure in PDB (3C2I) aa 77-167 (Methyl-CpG binding domain)
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0001	RETT SYNDROME, ZAPPELLA VARIANT RETT SYNDROME, INCLUDED	MECP2, ARG133CYS	[rs28934904]	-	[RCV000012578...]	
.0002	RETT SYNDROME	MECP2, PHE155SER	[rs28934905]	-	[RCV000012579]	
.0007	RETT SYNDROME ENCEPHALOPATHY, NEONATAL SEVERE, DUE TO MECP2 MUTATION, INCLUDED	MECP2, THR158MET	[rs28934906]	-	[RCV000133129...]	
.0008	RETT SYNDROME	MECP2, ARG106TRP	[rs28934907]	-	[RCV000012585...]	
.0027	RETT SYNDROME	MECP2, LEU100VAL	[rs28935168]	-	[RCV000168679...]	
.0036	RETT SYNDROME, ZAPPELLA VARIANT	MECP2, PRO152ALA	[rs179363900]	-	[RCV000012618...]	

Sandhoff Disease

Gene – HEXOSAMINIDASE B; HEXB

OMIM - 606873 gene
OMIM - 268800 disease

Alleles

Sandhoff alleles, associated with the B chain, are in the table below.

Structures

WT structure in PDB (2GJX) A chain aa 23-528, B chain 54-552
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0006	SANDHOFF DISEASE, JUVENILE TYPE	HEXB, TYR456SER	[rs121907982]	[rs121907982]	[RCV000004081]	
.0007	SANDHOFF DISEASE, JUVENILE TYPE	HEXB, PRO417LEU	[rs28942073]	[rs28942073]	[RCV000004082...]	
.0009	SANDHOFF DISEASE, ADULT TYPE	HEXB, ARG505GLN	[rs121907983]	[rs121907983]	[RCV000004083]	
.0012	SANDHOFF DISEASE, INFANTILE TYPE	HEXB, SER62LEU	[rs820878]	[rs820878]	[RCV000004086]	
.0014	SANDHOFF DISEASE, CHRONIC	HEXB, PRO504SER	[rs121907985]	[rs121907985]	[RCV000004088]	

Stiff Skin Syndrome

Gene – FIBRILLIN 1; FBN1

OMIM - 134797 gene
OMIM - 184900 disease

Alleles

Mutant alleles are in the table below.

Structures

WT structure in PDB (1UZK) aa 1486-1647
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0052	STIFF SKIN SYNDROME	FBN1, CYS1564SER	[rs267606800]	-	[RCV000017935]	
.0053	STIFF SKIN SYNDROME	FBN1, CYS1577GLY	[rs267606801]	-	[RCV000017936]	
.0054	STIFF SKIN SYNDROME	FBN1, GLY1594ASP	[rs267606798]	-	[RCV000017937...]	

Tay-Sachs Disease

Gene – HEXOSAMINIDASE A; HEXA

OMIM - 606869 gene
OMIM - 272800 disease

Alleles

Tay-Sachs alleles, associated with the A chain, are in the table below.

Structures

WT structure in PDB (2GJX) a chain aa 23-528, b chain aa 54-552
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0004	TAY-SACHS DISEASE	HEXA, GLU482LYS	[rs121907952]	-	[RCV000004096]	
.0006	TAY-SACHS DISEASE, B1 VARIANT HEXA, DN ALLELE	HEXA, ARG178HIS	[rs28941770]	-	[RCV000004100...]	
.0007	TAY-SACHS DISEASE, B1 VARIANT HEXA, CZECHOSLOVAKIAN ALLELE	HEXA, ARG178CYS	[rs121907953]	[rs121907953]	[RCV000004102...]	
.0008	GM2-GANGLIOSIDOSIS, ADULT	HEXA, GLY269SER	[rs121907954]	[rs121907954]	[RCV000168285...]	
.0009	GM2-GANGLIOSIDOSIS, JUVENILE	HEXA, ARG504HIS	[rs121907955]	[rs121907955]	[RCV000004099]	
.0010	GM2-GANGLIOSIDOSIS, JUVENILE	HEXA, ARG499HIS	[rs121907956]	[rs121907956]	[RCV000210735...]	
.0011	TAY-SACHS DISEASE	HEXA, ARG170GLN	[rs121907957]	-	[RCV000004106...]	
.0012	TAY-SACHS DISEASE	HEXA, TRP420CYS	[rs121907958]	[rs121907958]	[RCV000004107]	
.0013	TAY-SACHS DISEASE, JUVENILE	HEXA, GLY250ASP	[rs121907959]	[rs121907959]	[RCV000004108]	

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0015	GM2-GANGLIOSIDOSIS, CHRONIC	HEXA, ARG504CYS	[rs28942071]	[rs28942071]	[RCV000169084...]	
.0017	TAY-SACHS DISEASE	HEXA, SER210PHE	[rs121907961]	-	[RCV000004114]	
.0024	TAY-SACHS DISEASE	HEXA, ARG178LEU	[rs28941770]	-	[RCV000004118]	
.0028	GM2-GANGLIOSIDOSIS, ADULT-ONSET	HEXA, ARG499CYS	[rs121907966]	-	[RCV000169417...]	
.0030	TAY-SACHS DISEASE	HEXA, TRP485ARG	[rs121907968]	-	[RCV000004123]	
.0038	TAY-SACHS DISEASE, B1 VARIANT	HEXA, ASP258HIS	[rs121907971]	-	[RCV000004130]	
.0039	TAY-SACHS DISEASE	HEXA, ARG170TRP	[rs121907972]	[rs121907972]	[RCV000004131]	
.0041	GM2-GANGLIOSIDOSIS, LATE ONSET	HEXA, LYS197THR	[rs121907973]	[rs121907973]	[RCV000004133]	
.0043	TAY-SACHS DISEASE	HEXA, PHE211SER	[rs121907974]	-	[RCV000004135]	
.0044	TAY-SACHS DISEASE	HEXA, LEU127ARG	[rs121907975]	-	[RCV000004136]	
.0045	TAY-SACHS DISEASE	HEXA, HIS204ARG	[rs121907976]	-	[RCV000004137]	
.0047	TAY-SACHS DISEASE	HEXA, MET301ARG	[rs121907977]	[rs121907977]	[RCV000004139]	
.0048	TAY-SACHS DISEASE	HEXA, GLY454SER	[rs121907978]	-	[RCV000004140]	
.0049	TAY-SACHS DISEASE	HEXA, LEU39ARG	[rs121907979]	-	[RCV000004141]	
.0053	GM2-GANGLIOSIDOSIS, LATE ONSET	HEXA, TYR180HIS	[rs28941771]	-	[RCV000004145]	
.0055	GM2-GANGLIOSIDOSIS, SUBACUTE	HEXA, TRP474CYS	[rs121907981]	[rs121907981]	[RCV000004147]	
.0056	TAY-SACHS DISEASE	HEXA, LEU451VAL	[rs28940871]	-	[RCV000004148]	
.0057	GM2-GANGLIOSIDOSIS, SUBACUTE	HEXA, VAL324VAL	[rs28942072]	-	[RCV000004149]	
.0058	TAY SACHS DISEASE, MILD	HEXA, CYS58TYR	[rs387906949]	[rs387906949]	[RCV000023580]	

Von Willebrand Disease, Type 2

Gene – VON WILLEBRAND FACTOR; VWF

OMIM 613160 gene
OMIM 613554 disease

Alleles

Mutant alleles in table below correspond to the 3 A domains that have been crystallized.

Structures

WT structures in PDB. 3 structures available for the 3 A domains.
5BV8 (VWR A1 domain) aa 1261-1469
3ZQK (VWF A2 domain) aa 1494-1674
4DMU (VWF A3 domain) aa 1686-1873
WT structure for each allele is listed in the last column.
Create mutant structure using PyMOL software.

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0001	VON WILLEBRAND DISEASE, TYPE 2A	VWF, ILE1628THR	[rs61750584]	-	[RCV000086808...]	3ZQK
.0002	VON WILLEBRAND DISEASE, TYPE 2A	VWF, ARG1597TRP	[rs61750117]	-	[RCV000000309...]	3ZQK
.0003	VON WILLEBRAND DISEASE, TYPE 2A	VWF, VAL1607ASP	[rs61750579]	-	[RCV000086803...]	3ZQK
.0004	VON WILLEBRAND DISEASE, TYPE 2B	VWF, TRP1313CYS	[rs61749392]	-	[RCV000000311...]	5BV8
.0005	VON WILLEBRAND DISEASE, TYPE 2B	VWF, ARG1306TRP	[rs61749384]	-	[RCV000000312...]	5BV8
.0006	VON WILLEBRAND DISEASE, TYPE 2B	VWF, ARG1308CYS	[rs61749387]	-	[RCV000000313...]	5BV8
.0007	VON WILLEBRAND DISEASE, TYPE 2B	VWF, VAL1316MET	[rs61749397]	[rs61749397]	[RCV000000314...]	5BV8
.0008	VON WILLEBRAND DISEASE, TYPE 2B	VWF, ARG1341GLN	[rs61749403]	-	[RCV000086721...]	5BV8

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar	PDB structures
.0009	VON WILLEBRAND DISEASE, TYPE 2A	VWF, SER1613PRO	[rs61750581]	-	[RCV000000316...]	3ZQK
.0019	VON WILLEBRAND DISEASE, TYPE 2A	VWF, CYS1272ARG	[rs61749372]	-	[RCV000000328...]	5BV8
.0020	VON WILLEBRAND DISEASE, TYPE 2B	VWF, VAL1314LEU	[rs61749393]	-	[RCV000000329...]	5BV8
.0022	VON WILLEBRAND DISEASE, TYPE 2A	VWF, PHE1514CYS	[rs61750101]	-	[RCV000086775...]	3ZQK
.0030	VON WILLEBRAND DISEASE, TYPE 2M	VWF, SER1285PHE	[rs61749380]	-	[RCV000000339...]	5BV8
.0033	VON WILLEBRAND DISEASE, TYPE 2B	WVF, PRO1266LEU	[rs61749370]	-	[RCV000086676...]	5BV8
.0040	VON WILLEBRAND DISEASE, TYPE 2CB	VWF, TRP1745CYS	-	-	[RCV000024002]	4DMU
.0042	VON WILLEBRAND DISEASE, TYPE 2CB	VWF, SER1783ALA	[rs267607353]	[rs267607353]	[RCV000024004...]	4DMU

References for Database and Links

OMIM

Amberger, J. S., Bocchini, C. A., Schiettecatte, F., Scott, A. F., & Hamosh, A. (2015). OMIM.org: Online Mendelian Inheritance in Man (OMIM(®)), an online catalog of human genes and genetic disorders. *Nucleic Acids Research, 43*(Database issue), D789-D798. doi:10.1093/nar/gku1205

Hamosh, A., Scott, A. F., Amberger, J. S., Bocchini, C. A., & McKusick, V. A. (2005). Online Mendelian Inheritance in Man (OMIM), a knowledgebase of human genes and genetic disorders. *Nucleic Acids Research, 33*(Database Issue), D514-D517. doi:10.1093/nar/gki033

https://www.omim.org/

RSCB-PDB

Berman, H. M., Westbrook, J., Feng, Z., Gilliland, G., Bhat, T. N., Weissig, H., . . . Bourne, P. E. (2000). The Protein Data Bank. *Nucleic Acids Research, 28*(1), 235-242. doi:10.1093/nar/28.1.235

http://www.rcsb.org/pdb/home/home.do

PyMOL

Schrodinger, LLC. (2015). *The PyMOL Molecular Graphics System, Version 1.8* **https://www.pymol.org/**

Ensembl (dbSNP)

YZerbino,D.R., Achuthan,P., Akanni,W., Amode,M.R., Barrell,D., Bhai,J., Billis,K., Cummins,C., Gall,A., Giron,C.G. ....Paul Flicek (2017) Ensembl 2018. Nucleic Acids Res., doi:10.1093/nar/gkx1098.

http://www.ensembl.org.

The Exome Aggregation Consorium (ExAC)

Karczewski, K. J., Weisburd, B., Thomas, B., Solomonson, M., Ruderfer, D. M., Kavanagh, D., . . . MacArthur, D. G. (2017). The ExAC browser: displaying reference data information from over 60 000 exomes. *Nucleic Acids Research, 45*(Database issue), D840-D845. doi:10.1093/nar/gkw971

Lek, M., Karczewski, K. J., Minikel, E. V., Samocha, K. E., Banks, E., Fennell, T., . . . Exome Aggregation, C. (2016). Analysis of protein-coding genetic variation in 60,706 humans. *Nature, 536*(7616), 285-291. doi:10.1038/nature19057

http://exac.broadinstitute.org/

NCBI Clinical Variant database (ClinVar)

Landrum, M. J., Lee, J. M., Benson, M., Brown, G., Chao, C., Chitipiralla, S., . . . Maglott, D. R. (2016). ClinVar: public archive of interpretations of clinically relevant variants. *Nucleic Acids Research, 44*(Database issue), D862-D868. doi:10.1093/nar/gkv1222

Landrum, M. J., Lee, J. M., Riley, G. R., Jang, W., Rubinstein, W. S., Church, D. M., & Maglott, D. R. (2014). ClinVar: public archive of relationships among sequence variation and human phenotype. *Nucleic Acids Research, 42*(Database issue), D980-D985. doi:10.1093/nar/gkt1113

https://www.ncbi.nlm.nih.gov/clinvar/