Appendix 1.

Genetic disorders in Arab populations indexed in the CTGA Database (June, 2008).

OMIM #	Name	Country*
100100	Abdominal Muscles, Absence of, with Urinary Tract Abnormality and Cryptorchidism	Le O Sa T U
100300	Adams-Oliver Syndrome	U
100800	Achondroplasia	Le U
101000	Neurofibromatosis, Type II	O
101200	Apert Syndrome	Le Mo Sa
101400	Saethre-Chotzen Syndrome	U
101600	Pfeiffer Syndrome	O
102730	Adenosine Deaminase, Elevated, Hemolytic Anemia due to	Li
103050	Adenylosuccinase Deficiency	Mo
104290	Alternating Hemiplegia of Childhood	O
104300	Alzheimer Disease	P
104530	Amelogenesis Imperfecta, Hypoplastic Type	J
105250	Amyloidosis, Primary Cutaneous	Le O Sa
105400	Amyotrophic Lateral Sclerosis 1	T
105830	Angelman Syndrome	U
106100	Angioedema, Hereditary	U
106300	Spondyloarthropathy, Susceptibility to, 1	B Eg Jo Le O P Sa Sy U
106700	Total Anomalous Pulmonary Venous Return 1	OU
107300	Antithrombin III Deficiency	A
107320	Antiphospholipid Syndrome	B O Sa
107600	Aplasia Cutis Congenita	O Q Sa T U
108110	Arthrogryposis Multiplex Congenita	O Sa U
108120	Arthrogryposis, Distal, Type 1	O U
108300	Stickler Syndrome, Type I	U
108800	Atrial Septal Defect 1	Le Li O Q Su U
109150	Machado-Joseph Disease	Y
109400	Basal Cell Nevus Syndrome	Eg
109650	Behcet Syndrome	A B Er K O Sa
109800	Bladder Cancer	Eg
110100	Blepharophimosis, Ptosis, and Epicanthus Inversus	Sa
113000	Brachydactyly, Type B1	J
113100	Brachydactyly, Type C	I
113600	Branchial Cleft Anomalies	J O
113610	Branchial Myoclonus with Spastic Paraparesis and Cerebellar Ataxia	K
113620	Branchial Clefts with Characteristic Facies, Growth Retardation, Imperforate Nasolacrimal Duct, and Premature Aging	Le
113900	Progressive Familial Heart Block, Type I	Le
114000	Caffey Disease	Sa
114290	Campomelic Dysplasia	O U
114480	Breast Cancer	B J O S U
114500	Colorectal Cancer	K Sy U
114900	Carcinoid Tumors, Intestinal	O
115150	Cardiofaciocutaneous Syndrome	Eg
115200	Cardiomyopathy, Dilated, 1A	O U
115210	Cardiomyopathy, Familial Restrictive, 1	U
115430	Carpal Tunnel Syndrome	J O
117000	Central Core Disease of Muscle	A
117550	Sotos Syndrome	P Sa U
117650	CerebrocostOdibular Syndrome	U
118450	Alagille Syndrome	K
118650	Chondrodysplasia Punctata, Autosomal Dominant	U
118800	Paroxysmal Nonkinesigenic Dyskinesia 1	O
119100	Split-Hand/Foot Malformation with Long Bone Deficiency 1	A P Sa Sy U
119300	Van der Woude Syndrome	K T
119530	Orofacial Cleft 1	J O Sy U
120000	Coarctation of Aorta	O U
120330	Papillorenal Syndrome	O
120435	Lynch Syndrome I	Le Sy
121050	Contractural Arachnodactyly, Congenital	O
122100	Corneal Dystrophy, Juvenile Epithelial, of Meesmann	Sa
122470	Cornelia de Lange Syndrome	K O
123400	Creutzfeldt-Jakob Disease	Eg Li O T
125630	Dermodistortive Urticaria	Le
125700	Diabetes Insipidus, Neurohypophyseal Type	P
125851	Maturity-Onset Diabetes of the Young, Type II	O
125853	Diabetes Mellitus, Noninsulin-Dependent	B Er J Le O U
127300	Leri-Weill Dyschondrosteosis	D K
130020	Ehlers-Danlos Syndrome, Type III	Sa
130070	Ehlers-Danlos Syndrome, Progeroid Form	Q
130650	Beckwith-Wiedemann Syndrome	Ma O
130710	Emphysema, Congenital Lobar	K O

OMIM #	Name	Country*
131445	Ependymoma, Familial	P U
131760	Epidermolysis Bullosa Herpetiformis, Dowling-Meara Type	P
132700	Cylindromatosis, Familial	B
133200	Erythrokeratodermia Variabilis	U
133450	Ewing Sarcoma Breakpoint Region 1	K O Sa U
135100	Fibrodysplasia Ossificans Progressiva	Eg Sy T
136760	Frontonasal Dysplasia	Eg Sa U
137215	Gastric Cancer	J O
137280	Gastritis, Familial Giant Hypertrophic	J
137580	Gilles De La Tourette Syndrome	B K Sa
137750	Glaucoma 1, Open Angle, A	O Sa
137800	Glioma of Brain, Familial	P O
139090	Gray Platelet Syndrome	P Sa
139393	Guillain-Barre Syndrome, Familial	O U
140300	Hashimoto Thyroiditis	P So Su Sy T U
141200	Hematuria, Benign Familial	B U
141800	Hemoglobin - Alpha Locus 1	A B C K Ma Mo O Sa Su T U Y
141900	Hemoglobin - Beta Locus	A B C Eg I K Le Mo O P Q Sa Su Sy T U Y
142250	Hemoglobin, Gamma G	A Sa U
142309	Hemoglobin--Variants for which the Chain Carrying the Mutation is Unknown or Uncertain	Sa
142340	Diaphragmatic Hernia, Congenital	B Eg O Sa U
142470	Heterocellular Hereditary Persistence of Fetal Hemoglobin	Sa
142623	Hirschsprung Disease, Susceptibility to, 1	B I Mo O P Sa T U
142900	Holt-Oram Syndrome	B J O Sa U
143100	Huntington Disease	B Mo O Su
143400	Multicystic Renal Dysplasia, Bilateral	B O U
143465	Attention Deficit-Hyperactivity Disorder	O
143890	Hypercholesterolemia, Autosomal Dominant	B Le P Sy U
144200	Palmoplantar Keratoderma, Epidermolytic	K
145420	Hypertelorism, Teebi Type	K
145500	Hypertension, Essential	Eg Er Mo O Su U
145600	Malignant Hyperthermia, Susceptibility To, 1	B Sa
145900	Hypertrophic Neuropathy of Dejerine-Sottas	Le
146000	Hypochondroplasia	U
146110	Hypogonadotropic Hypogonadism	K O
146390	Chromosome 18p Deletion Syndrome	U
146450	Hypospadias, Autosomal	B O
147050	IgE Responsiveness, Atopic	B
147710	Intussusception	B O Q
147920	Kabuki Syndrome	P
148000	Kaposi Sarcoma	C O Su
148820	Waardenburg Syndrome, Type III	Eg Y
148900	Segmentation Syndrome 1	U
150250	Larsen Syndrome, Autosomal Dominant	T
150590	Leg Ulcers, Familial, of Juvenile Onset	I
150600	Legg-Calve-Perthes Disease	B Le T
150699	Leiomyoma, Uterine	Eg
150800	Leiomyoma, Hereditary Multiple, of Skin	Eg Er I J Le Mo P So Su U Y
150900	Lentigines	Le
151600	Leukonychia Totalis	P O
152200	Apolipoprotein(a)	Su
152700	Systemic Lupus Erythematosus	B Eg Le O Q U
153600	Macroglobulinemia, Waldenstrom, Susceptibility To, 1	B Eg
153670	Bernard-Soulier Syndrome, Benign Autosomal Dominant	A
154500	Treacher Collins-Franceschetti Syndrome	O
154570	Mannose 6-Phosphate Receptor Recognition Defect, Lebanese Type	Le
154700	Marfan Syndrome	O
155255	Medulloblastoma	Mo
155600	Melanoma, Cutaneous Malignant	J O
156240	Mesothelioma, Malignant	Eg O
156810	Microgastria-Limb Reduction Defects Association	Su U
158000	Monilethrix	?
158330	Mullerian Aplasia	O
160700	Myopia 2	O
160900	Dystrophia Myotonica 1	Eg P U Y
161200	Nail-Patella Syndrome	P
161550	Nasopharyngeal Carcinoma	B O
161900	Renal Failure, Progressive, with Hypertension	I
162091	Schwannomatosis	B O
162200	Neurofibromatosis, Type I	O T U
162700	Neutropenia, Chronic Familial	Y
163950	Noonan Syndrome 1	B Eg Le O U
164210	Hemifacial Microsomia	B P T
164400	Spinocerebellar Ataxia 1	O
164500	Spinocerebellar Ataxia 7	Mo
164750	Omphalocele	U

OMIM #	Name	Country*
166000	Enchondromatosis, Multiple	J Mo O
166200	Osteogenesis Imperfecta, Type I	U
166210	Osteogenesis Imperfecta, Type IIA	A Le O Su U
166800	Otosclerosis	T
167000	Suppressor of Tumorigenicity 8	B Sa U
167100	Pachydermoperiostosis	T U
167750	Pancreas, Annular	O U
168900	Patella, Chondromalacia Of	B
169500	Leukodystrophy, Adult-Onset, Autosomal Dominant	Su
169610	Pemphigus Vulgaris, Familial	J K Mo
170650	Periodontitis, Aggressive, 1	J
173000	Pilonidal Sinus	B J Sa
173650	Kindler Syndrome	A I Sa T
173800	Poland Syndrome	Sa U
173900	Polycystic Kidneys	B J
174400	Polydactyly, Preaxial I	J O Sa
174800	McCune-Albright Syndrome	T U
175100	Adenomatous Polyposis of the Colon	B Eg Mo P Sy Y
175200	Peutz-Jeghers Syndrome	B I
176000	Porphyria, Acute Intermittent	B K Mo O
176100	Porphyria Cutanea Tarda	T
176261	Potassium Channel, Voltage-Gated, ISK-Related Subfamily, Member 1	Le
176270	Prader-Willi Syndrome	Eg K Sa U
176670	Hutchinson-Gilford Progeria Syndrome	Eg Li U
176807	Prostate Cancer	B Eg J K O Sy U Y
176860	Protein C Deficiency, Congenital Thrombotic Disease due to	J O P
176880	Protein S, Alpha	Eg I J K O P Sa Su Sy U
177850	Pseudoxanthoma Elasticum, Forme Fruste	Mo
178550	Pulmonary Hemosiderosis	O T
178600	Pulmonary Hypertension, Primary	K O U
179010	Pyloric Stenosis, Infantile Hypertrophic 1	B J O Sa
179280	Radial-Renal Syndrome	Mo
179800	Renal Tubular Acidosis, Distal, Autosomal Dominant	T U
179850	Dowling-Degos Disease	U
180200	Retinoblastoma	Eg O Su Sy
180300	Rheumatoid Arthritis	O U
180849	Rubinstein-Taybi Syndrome	Le Mo Q U
180860	Silver-Russell Syndrome	Le
180960	S-Adenosylhomocysteine Hydrolase	T
181000	Sarcoidosis	Sa Sy
181450	Ulnar-Mammary Syndrome	Y
182212	Shprintzen-Goldberg Craniosynostosis Syndrome	B Le Mo
182260	Slipped Femoral Capital Epiphyses	B
182601	Spastic Paraplegia 4, Autosomal Dominant	T
182860	Spectrin, Alpha, Erythrocytic 1	A Mo T
182870	Spectrin, Beta, Erythrocytic	A
182940	Neural Tube Defects	Eg O U
183090	Spinocerebellar Ataxia 2	T
184095	Spondyloepiphyseal Dysplasia, Maroteaux Type	Le
184253	Spondylometaphyseal Dysplasia, Algerian Type	A
184700	Polycystic Ovary Syndrome 1	B J U
185100	Strabismus, Susceptibility to	Eg O
185300	Sturge-Weber Syndrome	O U
185900	Syndactyly, Type I	B
187100	Teeth, Supernumerary	Le
187300	Telangiectasia, Hereditary Hemorrhagic, of Rendu, Osler, and Weber	A B
187400	Testicular Torsion	B Sa
187500	Tetralogy of Fallot	B Le Li Ma O U
187600	Thanatophoric Dysplasia, Type I	Mo O U
188030	Thrombocytopenic Purpura, Autoimmune	Eg O Q Sa Su
188050	Thrombophilia	Eg I J K P Sa Su Sy
188400	DiGeorge Syndrome	Le Sa U
188455	Thyroglossal Duct Cyst, Familial	P
188470	Thyroid Carcinoma, Follicular	B J Mo T U
188550	Thyroid Carcinoma, Papillary	O
188580	Thyrotoxic Periodic Paralysis	B O Sa
189800	Preeclampsia/Eclampsia 1	K O
189960	Tracheoesophageal Fistula with or without Esophageal Atresia	O Sa U
190160	Thyroid Hormone Receptor, Beta	A
190685	Down Syndrome	B Eg Le O P Sa U
191100	Tuberous Sclerosis	O
191390	Ulcerative Colitis, Susceptibility to	B O
192132	ATPase, H+ Transporting, Lysosomal, 56/58-Kd, V1 Subunit B, Isoform 1	Mo Sa
192350	VATER Association	J O
192500	Long QT Syndrome 1	O T
192600	Cardiomyopathy, Familial Hypertrophic	Le

OMIM #	Name	Country*
193000	Vesicoureteral Reflux 1	K O Sa T U
193200	Vitiligo	J K O U
193230	Vitreoretinal Degeneration, Snowflake Type	A
193300	Von Hippel-Lindau Syndrome	Le
193700	Arthrogryposis, Distal, Type 2A	Mo
194070	Wilms Tumor 1	Eg U
194200	Wolff-Parkinson-White Syndrome	O
200400	Achalasia, Familial Esophageal	O Su
200700	Chondrodysplasia, Grebe Type	J O U
201000	Carpenter Syndrome	B Eg Le P
201100	Acrodermatitis Enteropathica, Zinc-Deficiency Type	Eg J
201300	Neuropathy, Hereditary Sensory and Autonomic, Type II	?
201450	Acyl-CoA Dehydrogenase, Medium-Chain, Deficiency Of	B
201460	Acyl-CoA Dehydrogenase, Long-Chain, Deficiency of	O
201710	Lipoid Congenital Adrenal Hyperplasia	P
201910	Adrenal Hyperplasia, Congenital, due to 21-Hydroxylase Deficiency	Eg J K O T U
202010	Adrenal Hyperplasia, Congenital, due to 11-Beta-Hydroxylase Deficiency	Mo Sa T
202370	Adrenoleukodystrophy, Autosomal Neonatal Form	Eg
202400	Afibrinogenemia, Congenital	I Mo
203100	Oculocutaneous Albinism, Type IA	B J Le O Q
203500	Alkaptonuria	Eg
203655	Alopecia Universalis Congenita	O
203700	Alpers Diffuse Degeneration of Cerebral Gray Matter with Hepatic Cirrhosis	P
203740	Alpha-Ketoglutarate Dehydrogenase Deficiency	A T
203750	Alpha-Methylacetoacetic Aciduria	T
203800	Alstrom Syndrome	A
204200	Ceroid Lipofuscinosis, Neuronal, 3	Mo
204500	Ceroid Lipofuscinosis, Neuronal, 2	Le
204870	Corneal Dystrophy, Gelatinous Drop-Like	T
205100	Amyotrophic Lateral Sclerosis 2, Juvenile	K Sa T
206500	Anencephaly	B I K Mo O P Sa T U
206920	Anophthalmia with Limb Anomalies	Le
207600	Takayasu Arteritis	B I Le T
207900	Argininosuccinic Aciduria	B O T
208050	Arterial Tortuosity Syndrome	Mo
208100	Arthrogryposis Multiplex Congenita, Neurogenic Type	P
208150	Pena-Shokeir Syndrome, Type I	O
208230	Arthropathy, Progressive Pseudorheumatoid, of Childhood	J
208250	Camptodactyly-Arthropathy-Coxa Vara-Pericarditis Syndrome	Sa
208300	Ascites, Chylous	O Sa
208400	Aspartylglucosaminuria	P T
208500	Asphyxiating Thoracic Dystrophy 1	Le O U
208850	Ataxia-Deafness-Retardation Syndrome	K
208870	Ataxia-Microcephaly-Cataract Syndrome	P
208900	Ataxia-Telangiectasia	Mo
209500	Atrichia with Papular Lesions	O P
209880	Autonomic Control, Congenital Failure of	Sa U
209900	Bardet-Biedl Syndrome	B K Le O Y
209920	Bare Lymphocyte Syndrome, Type II	A Mo T
210000	Behr Syndrome	I
210200	3-@Methylcrotonyl-CoA Carboxylase 1 Deficiency	T
210600	Seckel Syndrome 1	O Y
210745	Blepharophimosis with Ptosis, Syndactyly, and Short Stature	Y
211450	Bronchomalacia	O
211530	Bulbar Palsy, Progressive, with Sensorineural Deafness	Le O
211770	CAHMR Syndrome	Eg
211890	Campomelia, Cumming Type	Eg
211900	Calcinosis, Tumoral, with Hyperphosphatemia	Le
211960	Camptodactyly with Muscular Hypoplasia, Skeletal Dysplasia, and Abnormal Palmar Creases	Li Mo
211980	Lung Cancer	B K O Q Sa
212065	Congenital Disorder of Glycosylation, Type Ia	O
212110	Cardiomyopathy, Dilated, Autosomal Recessive	Sa
212112	Cardiomyopathy, Congestive, with Hypergonadotropic Hypogonadism	Le
212135	Cardioskeletal Syndrome, Kuwaiti Type	K
212138	Solute Carrier Family 25 (Carnitine/Acylcarnitine Translocase), Member 20	Sa
212140	Carnitine Deficiency, Systemic Primary	B
212720	Martsolf Syndrome	Eg
212750	Celiac Disease	Li O
213200	Spinocerebellar Ataxia, Autosomal Recessive 2	Le
213300	Joubert Syndrome 1	O P U
213700	Cerebrotendinous Xanthomatosis	Mo
213980	Cerebrofaciothoracic Dysplasia	Mo
214100	Zellweger Syndrome	A O Sa
214150	Cerebrooculofacioskeletal Syndrome	Eg O
214300	Klippel-Feil Syndrome, Autosomal Recessive	B I J
214400	Charcot-Marie-Tooth Disease, Type 4A	A Le Mo T

OMIM #	Name	Country*
214500	Chediak-Higashi Syndrome	B J Sa Sy
214700	Chloride Diarrhea, Familial	K
214800	CHARGE Syndrome	O T
215100	Rhizomelic Chondrodysplasia Punctata, Type 1	U
215518	Ciliary Discoordination due to Random Ciliary Orientation	Le
215700	Citrullinemia, Classic	K Mo O
216550	Cohen Syndrome	Le O Sa U
216900	Achromatopsia 2	I Mo
217070	Complement Component 7	Mo T Y
217080	Cone-Rod Dystrophy and Amelogenesis Imperfecta	? P
217095	Conotruncal Heart Malformations	K Le U
217400	Corneal Dystrophy and Perceptive Deafness	Mo
217990	Corpus Callosum, Agenesis of	O
218030	Cortisol 11-Beta-Ketoreductase Deficiency	O
218350	Craniofacial Dyssynostosis with Short Stature	K
218700	Hypothyroidism, Congenital, Nongoitrous, 2	O Sa
218800	Crigler-Najjar Syndrome	Sa T
219050	Cryptorchidism, Unilateral or Bilateral	J O
219200	Cutis Laxa, Autosomal Recessive, Type II	I O Sa
219250	Cutis Marmorata Telangiectatica Congenita	O
219550	Cysteine Peptiduria	I
219600	Cystic Disease of Lung	Y
219700	Cystic Fibrosis	B I J Li Mo O Sa Sy U Y
219721	Cystic Fibrosis with Helicobacter Pylori Gastritis, Megaloblastic Anemia, and Subnormal Mentality	K
219730	Cystic Kidney Disease with Ventriculomegaly	O
219800	Cystinosis, Nephropathic	P U
220100	Cystinuria	Li
220150	Hypouricemia, Renal	I
220200	Dandy-Walker Syndrome	O P Sa U
220210	Dandy-Walker-Like Malformation with Atrioventricular Septal Defect	Le
220290	Deafness, Neurosensory, Autosomal Recessive 1	Eg P T
220400	Jervell and Lange-Nielsen Syndrome	B Le Sa
220500	Deafness, Congenital, and Onychodystrophy, Recessive Form	O
220900	Deafness, Congenital, with Total Albinism	Mo
221745	Mitochondrial Deafness Modifier Gene 1	P
221950	Dextrocardia with Unusual Facies and Microphthalmia	P
222100	Diabetes Mellitus, Insulin-Dependent	B Eg I J Le Li Mo O P Sa So Su Sy U
222300	Wolfram Syndrome	O Su
222400	Diaphragmatic Hernia 2	Eg K Sa
222448	Donnai-Barrow Syndrome	Q Sa U
222700	Lysinuric Protein Intolerance	Mo Sa
222748	Dihydropyrimidinase	Le
222765	Rhizomelic Chondrodysplasia Punctata, Type 2	U Y
223000	Lactase Deficiency, Congenital	O
223100	Lactase Persistence	Eg J P Su
223400	Duodenal Atresia	I Le U
223800	Dyggve-Melchior-Clausen Disease	Eg Le Mo P
223900	Neuropathy, Hereditary Sensory And Autonomic, Type III	A
224120	Anemia, Dyserythropoietic Congenital, Type I	K Sa
224230	Dyskeratosis Congenita, Autosomal Recessive	Sy
224400	Dyssegmental Dysplasia, Rolland-Desbuquois Type	J Le P
224410	Dyssegmental Dysplasia, Silverman-Handmaker Type	Le
224500	Dystonia Musculorum Deformans 2	P
224700	Ebstein Anomaly	B Su
224900	Ectodermal Dysplasia, Anhidrotic	Mo U
225280	EEM Syndrome	Y
225400	Ehlers-Danlos Syndrome, Type VI	K
225500	Ellis-van Creveld Syndrome	Eg J O Sa U
225750	Aicardi-Goutieres Syndrome 1	A O
226300	Enteropathy, Protein-Losing	P
226400	Epidermodysplasia Verruciformis	A
226650	Epidermolysis Bullosa, Generalized Atrophic Benign	O U
226700	Epidermolysis Bullosa Letalis	Sa Su U Y
226730	Epidermolysis Bullosa with Pyloric Atresia	Sa T U
226980	Epiphyseal Dysplasia, Multiple, with Early-Onset Diabetes Mellitus	K O Sa T U
227090	Erythroderma, Lethal Congenital	O
227260	Facial Ectodermal Dysplasia	O U
227300	Factor V and Factor VIII, Combined Deficiency of	A T
227310	Factor V and Factor VIII, Combined Deficiency of, with Normal Protein C and Protein C Inhibitor	Sy
227320	Faciothoracogenital Syndrome	?
227330	Faciodigitogenital Syndrome, Recessive	K
227500	Factor VII Deficiency	Mo
227600	Factor X Deficiency	Mo O Su U
227645	Fanconi Anemia, Complementation Group C	I
227650	Fanconi Anemia	O Sa U
228000	Farber Lipogranulomatosis	T

OMIM #	Name	Country*
228250	Femur, Unilateral Bifid, with Monodactylous Ectrodactyly	P
228400	Fever, Familial Lifelong Persistent	Le
228520	Fibrochondrogenesis	Le O U
228550	Fibromatosis, Congenital Generalized	B Mo Su
228600	Fibromatosis, Juvenile Hyaline	Eg Sa
228980	Fleck Retina, Familial Benign	P
229200	Ehlers-Danlos Syndrome, Type VIB	Sy T
229300	Friedreich Ataxia 1	T
229400	Frontofacionasal Dysostosis	O U
229800	Fructosuria	?
229850	Fryns Syndrome	B C
230350	Galactose Epimerase Deficiency	Eg
230400	Galactosemia	A Eg O U
230500	GM1-Gangliosidosis, Type I	O U
230740	GAPO Syndrome	A Eg
230750	Gastroschisis	J O
230800	Gaucher Disease, Type I	O U
230900	Gaucher Disease, Type II	P
231005	Gaucher-Like Disease	P
231070	Geroderma Osteodysplastica	K Mo O P Sy U
231090	Hydatidiform Mole	Le
231300	Glaucoma 3, Primary Infantile, A	P U
231550	Achalasia-Addisonianism-Alacrima Syndrome	Sa
231670	Glutaric Acidemia I	K O U
231680	Multiple Acyl-CoA Dehydrogenation Deficiency	Eg O Sa U Y
232200	Glycogen Storage Disease I	B J O P U
232300	Glycogen Storage Disease II	I O P
232400	Glycogen Storage Disease III	Eg O
232500	Glycogen Storage Disease IV	Eg O
232700	Glycogen Storage Disease VI	P
233710	Granulomatous Disease, Chronic, Autosomal Recessive, Cytochrome b-Positive, Type II	J P
234050	Trichothiodystrophy, Nonphotosensitive 1	Mo
234820	Hemangiopericytoma, Malignant	B
235200	Hemochromatosis	A Eg T
235400	Hemolytic Uremic Syndrome, Atypical	J K P Sa U
235510	Hennekam Lymphangiectasia-Lymphedema Syndrome	O P T U
235730	Mowat-Wilson Syndrome	Mo U
236000	Hodgkin Lymphoma	B U
236100	Holoprosencephaly	O Sa U
236200	Homocystinuria	K O Q Sa U
236250	Homocystinuria due to Deficiency of N(5,10)-Methylenetetrahydrofolate Reductase Activity	B Sa
236450	Hutterite Cerebroosteonephrodysplasia Syndrome	Y
236600	Hydrocephalus	Eg J K Le O P Q Su Sy U
236670	Walker-Warburg Syndrome	P
236680	Hydrolethalus Syndrome 1	O
236700	McKusick-Kaufman Syndrome	Le
236730	Urofacial Syndrome	K
236792	L-2-Hydroxyglutaric Aciduria	Mo T
236800	Hydroxykynureninuria	A
237300	Carbamoyl Phosphate Synthetase I Deficiency, Hyperammonemia due to	P
237500	Dubin-Johnson Syndrome	B
237900	Hyperbilirubinemia, Transient Familial Neonatal	Y
238320	Hypergonadotropic Hypogonadism	O
238600	Hyperlipoproteinemia, Type I	U
239000	Paget Disease, Juvenile	I
239500	Hyperprolinemia, Type I	A
239710	Acrofrontofacionasal Dysostosis, Severe	K
239840	Hypertrichosis, Congenital Anterior Cervical, with Peripheral Sensory and Motor Neuropathy	P
241080	Hypogonadism, Diabetes Mellitus, Alopecia, Mental Retardation, and Electrocardiographic Abnormalities	Sa
241090	Hypogonadism, Primary, and Partial Alopecia	J K Le
241410	Hypoparathyroidism-Retardation-Dysmorphism Syndrome	O Q Sa
241550	Hypoplastic Left Heart Syndrome	Le O
242100	Ichthyosiform Erythroderma, Congenital, Nonbullous, 1	A Eg
242300	Ichthyosis, Lamellar, 1	Eg O Sa U
242650	Primary Ciliary Dyskinesia	J P Q Sa U
242870	Immunodeficiency, Partial Combined, with Absence of HLA Determinants and Beta-2-Microglobulin from Lymphocytes	A
243060	Infertility Associated with Multi-Tailed Spermatozoa and Excessive DNA	Li
243110	Interleukin 1, Defective T-Cell Response to	Le
243150	Intestinal Atresia, Multiple	Le U
243200	Intracranial Hypertension, Idiopathic	J Li O U
243310	Iris Coloboma with Ptosis, Hypertelorism, and Mental Retardation	O
243320	Intrinsic Factor and R Binder, Combined Congenital Deficiency of	A
243500	Isovaleric Acidemia	B O Sa
243600	Jejunal Atresia	K Le O P Sa U

OMIM #	Name	Country*
243700	Hyperimmunoglobulin E-Recurrent Infection Syndrome, Autosomal Recessive	O Sa
243800	Johanson-Blizzard Syndrome	Sa
244400	Kartagener Syndrome	P
244460	Kenny-Caffey Syndrome, Type 1	K Sa
245000	Papillon-Lefevre Syndrome	Eg J Q Sa Su
245200	Krabbe Disease	P Sa U
245552	Lambotte Syndrome	Mo
245570	Landau-Kleffner Syndrome	Le Sa T
245590	Laron Syndrome, Type II	P
245600	Larsen Syndrome, Recessive	Eg U
245800	Laurence-Moon Syndrome	B K O U Y
246200	Donohue Syndrome	Le U Y
246450	3-@Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency	Mo O Sa
246570	Fibular Aplasia, Tibial Campomelia, and Oligosyndactyly Syndrome	Mo
247100	Lipoid Proteinosis of Urbach and Wiethe	Le
248110	Macrosomia with Microphthalmia, Lethal	K
248250	Hypomagnesemia 3, Renal	? O
248300	Mal de Meleda	A P T U
248500	Mannosidosis, Alpha B, Lysosomal	P
248600	Maple Syrup Urine Disease	B Eg J K O Sa T U
248800	Marinesco-Sjogren Syndrome	K
248950	McDonough Syndrome	U
249000	Meckel Syndrome, Type 1	Eg K O P Sa T U
249100	Familial Mediterranean Fever	B Eg I J K Le Li Mo P Sa Sy U
249240	Megalencephaly with Dysmyelination	I
249270	Thiamine-Responsive Megaloblastic Anemia Syndrome	O P
249420	Frank-Ter Haar Syndrome	Sy
249500	Mental Retardation, Autosomal Recessive	A
250100	Metachromatic Leukodystrophy	O P
250220	Spondylometaphyseal Dysplasia, Sedaghatian Type	U Y
250450	Metaphyseal Dysplasia, Anetoderma, and Optic Atrophy	Eg
250790	Methemoglobinemia due to Deficiency of Cytochrome b5	Y
250800	Methemoglobinemia due to Deficiency of Methemoglobin Reductase	A
250950	3-@Methylglutaconic Aciduria, Type I	B O Sa
250951	3-@Methylglutaconic Aciduria, Type IV	I
251000	Methylmalonic Aciduria due to Methylmalonyl-CoA Mutase Deficiency	B O Sa
251170	Mevalonate Kinase	U
251200	Microcephaly, Primary Autosomal Recessive, 1	J O
251260	Nijmegen Breakage Syndrome	K U
251270	Microcephaly with Chorioretinopathy	K
251280	Microcephaly with Spastic Quadriplegia	P
251450	Desbuquois Syndrome	Mo U T
251600	Microphthalmia, Isolated 1	?
251850	Microvillus Inclusion Disease	I
252010	Mitochondrial Complex I Deficiency	Mo
252350	Moyamoya Disease 1	Le Li Sa T U
252500	Mucolipidosis II Alpha/Beta	P Sa U
252600	Mucolipidosis III Alpha/Beta	U
252650	Mucolipidosis IV	Su
252900	Mucopolysaccharidosis Type IIIA	O
252920	Mucopolysaccharidosis Type IIIB	Eg P Sy T U Y
253000	Mucopolysaccharidosis Type IVA	Eg O T
253200	Mucopolysaccharidosis Type VI	Eg J O U
253220	Mucopolysaccharidosis Type VII	A O
253250	Mulibrey Nanism	Eg
253270	Holocarboxylase Synthetase Deficiency	J
253290	Multiple Pterygium Syndrome, Lethal Type	Mo Sa
253300	Spinal Muscular Atrophy, Type I	B K Li O Sa U
253400	Spinal Muscular Atrophy, Type III	B K O Sa
253550	Spinal Muscular Atrophy, Type II	B K O Sa
253600	Muscular Dystrophy, Limb-Girdle, Type 2A	P
253601	Muscular Dystrophy, Limb-Girdle, Type 2B	Y
253700	Muscular Dystrophy, Limb-Girdle, Type 2C	A Eg Mo T
254130	Miyoshi Myopathy	T
254200	Myasthenia Gravis	Li O U
254210	Myasthenic Syndrome, Congenital, Associated With Episodic Apnea	I
254500	Myeloma, Multiple	U
254780	Myoclonic Epilepsy of Lafora	P
254800	Myoclonic Epilepsy of Unverricht and Lundborg	P
255120	Carnitine Palmitoyltransferase I Deficiency	O Sa
255800	Schwartz-Jampel Syndrome, Type 1	Eg O U
256000	Leigh Syndrome	Ma
256020	Nail-Patella-Like Renal Disease	P
256100	Nephronophthisis 1	K Sa U
256300	Nephrosis 1, Congenital, Finnish Type	J O Sa Su U
256370	Nephrotic Syndrome, Early-Onset, with Diffuse Mesangial Sclerosis	?

OMIM #	Name	Country*
256450	Hyperinsulinemic Hypoglycemia, Familial, 1	O Sa
256500	Netherton Syndrome	Eg Sa U
256520	Neu-Laxova Syndrome	Eg O
256540	Neuraminidase Deficiency with Beta-Galactosidase Deficiency	O T
256550	Neuraminidase Deficiency	O Sa
256700	Neuroblastoma	U
256800	Insensitivity to Pain, Congenital, with Anhidrosis	K Sa U
256850	Giant Axonal Neuropathy	T
257220	Niemann-Pick Disease, Type C1	I J U
257300	Mosaic Variegated Aneuploidy Syndrome	K
257320	Lissencephaly Syndrome, Norman-Roberts Type	Sa
257350	Nuchal Bleb, Familial	O
257920	Oculopalatoskeletal Syndrome	Su U
257980	Odontoonychodermal Dysplasia	Le
258315	Omodysplasia, Generalized Form	Sy U
258501	3-@Methylglutaconic Aciduria, Type III	I
258860	Orofaciodigital Syndrome, Type IV	Le
258870	Ornithine Aminotransferase Deficiency	A I Le
259420	Osteogenesis Imperfecta, Type III	U
259700	Osteopetrosis, Autosomal Recessive 1	B K Le P Sa Sy U
259730	Osteopetrosis, Autosomal Recessive 3	Eg K Sa T
259775	Raine Syndrome	Eg P Sa U
259900	Hyperoxaluria, Primary, Type I	B K O Sa U
260350	Pancreatic Carcinoma	O
260500	Papilloma of Choroid Plexus	O
260600	Pelizaeus-Merzbacher-Like Disease, Autosomal Recessive, 2	Y
260650	Pellagra-Like Syndrome	Su
260800	Pentosuria	Le
260920	Hyper-IgD Syndrome	P Q
261100	Megaloblastic Anemia 1	B Sa
261500	Peroxidase and Phospholipid Deficiency in Eosinophils	Su Y
261550	Persistent Mullerian Duct Syndrome, Types I and II	K O U
261600	Phenylketonuria	A B Eg K O T U Y
261630	Phenylketonuria II	T
261750	Phosphorylase Kinase Deficiency of Liver and Muscle, Autosomal Recessive	P
261800	Pierre Robin Syndrome	K O U
262400	Pituitary Dwarfism I	I Y
262500	Pituitary Dwarfism II	?
263200	Polycystic Kidney Disease, Autosomal Recessive	K Le O P Sa T U
263510	Short Rib-Polydactyly Syndrome, Type III	U
263610	Polyhydramnios, Chronic Idiopathic	B
263630	Polysyndactyly with Cardiac Malformation	O
263650	Popliteal Pterygium Syndrome, Lethal Type	Q U
263700	Porphyria, Congenital Erythropoietic	P
263800	Gitelman Syndrome	P
264090	Progeroid Syndrome, Neonatal	Le P
264300	17-@Beta Hydroxysteroid Dehydrogenase III Deficiency	P
264350	Pseudohypoaldosteronism, Type I, Autosomal Recessive	O
264480	Pseudotrisomy 13 Syndrome	U
264600	Pseudovaginal Perineoscrotal Hypospadias	J O U
264900	PTA Deficiency	I
265000	Multiple Pterygium Syndrome, Escobar Variant	K O P Sa U
265100	Pulmonary Alveolar Microlithiasis	Le
265380	Pulmonary Hypertension, Familial Persistent, of the Newborn	T
265430	Pulmonary Hypoplasia, Primary	O P Sa U
265800	Pycnodysostosis	Eg O P
265950	Pyloric Atresia	I Le Sa T U
266130	Glutathione Synthetase Deficiency	Eg O Sa U
266140	Pyropoikilocytosis, Hereditary	Sa
266150	Pyruvate Carboxylase Deficiency	Eg
266200	Pyruvate Kinase Deficiency of Red Cells	Le
266265	Congenital Disorder of Glycosylation, Type IIc	P
266600	Inflammatory Bowel Disease 1	B P Q Su
267000	Renal Hamartomas, Nephroblastomatosis, and Fetal Gigantism	Y
267300	Renal Tubular Acidosis, Distal, with Progressive Nerve Deafness	K Mo Sa
267430	Renal Tubular Dysgenesis	P
267450	Respiratory Distress Syndrome in Premature Infants	K O
267500	Reticular Dysgenesia	U
267700	Hemophagocytic Lymphohistiocytosis, Familial, 1	I Sa
268020	Retinitis Pigmentosa, Deafness, Mental Retardation, and Hypogonadism	Mo
268050	Retinopathy, Pigmentary, and Mental Retardation	Le
268130	Revesz Syndrome	Su U
268200	Rhabdomyolysis, Acute Recurrent	K
268210	Rhabdomyosarcoma 1	B Eg J O
268240	Rheumatic Fever-Related Antigen	Eg
268250	Rhizomelic Syndrome	?

OMIM #	Name	Country*
268310	Robinow Syndrome, Autosomal Recessive	Eg K O Sa U
268800	Sandhoff Disease	Le O Q U
269000	SC Phocomelia Syndrome	Le U
269150	Schinzel-Giedion Midface-Retraction Syndrome	Eg
269160	Schizencephaly	O So U
269250	Schneckenbecken Dysplasia	P U
269700	Lipodystrophy, Congenital Generalized, Type 2	Le O P
269950	Sideroblastic Anemia, Autosomal	Li
270200	Sjogren-Larsson Syndrome	Eg
270300	Peeling Skin Syndrome	K
270400	Smith-Lemli-Opitz Syndrome	Eg O
270550	Spastic Ataxia, Charlevoix-Saguenay Type	T
270750	Spastic Paraplegia 23	J
270800	Spastic Paraplegia 5A, Autosomal Recessive	J O T
271322	Spinocerebellar Degeneration with Slow Eye Movements	K P
271550	Spondyloenchondrodysplasia	I
271640	Spondyloepimetaphyseal Dysplasia with Joint Laxity	K
271665	Spondylometaepiphyseal Dysplasia, Short Limb-Hand Type	Eg U
271900	Canavan Disease	J Sa
272300	Sulfocysteinuria	A
272440	Syndactyly, Type I, with Microcephaly and Mental Retardation	T
272450	Syndesmodysplasic Dwarfism	A
272460	Spondylocarpotarsal Synostosis Syndrome	Le
272750	Tay-Sachs Disease, AB Variant	Sa
272800	Tay-Sachs Disease	Eg Le Mo O Sy U Y
272950	Teebi-Shaltout Syndrome	T
273150	Testes, Rudimentary	Le
273250	Testicular Regression Syndrome	T
273300	Testicular Tumors	B J K
273395	Tetra-Amelia, Autosomal Recessive	Le P
273800	Thrombasthenia of Glanzmann and Naegeli	I J O P Sa Su
274150	Thrombotic Thrombocytopenic Purpura, Congenital	O Q
274230	Thymoma, Familial	Y
274600	Pendred Syndrome	Eg U
275000	Graves Disease	D O T
275200	Hypothyroidism, Congenital, Nongoitrous, 1	Eg O Sy U
275210	Tight Skin Contracture Syndrome, Lethal	A
275350	Transcobalamin II Deficiency	Mo
275355	Squamous Cell Carcinoma, Head and Neck	O Su
275595	Trigonobrachycephaly, Bulbous Bifid Nose, Micrognathia, and Abnormalities of the Hands and Feet	P
275630	Triglyceride Storage Disease with Impaired Long-Chain Fatty Acid Oxidation	Eg
275900	Spastic Paraplegia 20, Autosomal Recessive	K
276300	Mismatch Repair Cancer Syndrome	Sa U
276600	Tyrosine Transaminase Deficiency	Sa
276700	Tyrosinemia, Type I	O
276820	Ulna and Fibula, Absence of, with Severe Limb Deficiency	J
276821	Ulnar Hypoplasia with Mental Retardation	?
276901	Usher Syndrome, Type IIA	T
276902	Usher Syndrome, Type III	Y
276903	Myosin VIIA	T Y
276905	Usher Syndrome, Type IIB	T
277000	Rokitansky-Kuster-Hauser Syndrome	B Mo O
277170	Varadi-Papp Syndrome	Eg Sa U
277300	Spondylocostal Dysostosis, Autosomal Recessive, 1	Eg P T U
277320	Visceral Myopathy, Familial, with External Ophthalmoplegia	I
277350	Vitamin A Metabolic Defect	Le
277440	Vitamin D-Dependent Rickets, Type II	B O Sa
277450	Vitamin K-Dependent Clotting Factors, Combined Deficiency of, 1 Chondrodysplasia Punctata with Coagulation Factor Deficiency	P
277460	Vitamin E, Familial Isolated Deficiency of	T
277465	Vitiligo, Progressive, with Mental Retardation and Urethral Duplication	A
277580	Waardenburg-Shah Syndrome	O T U
277600	Weill-Marchesani Syndrome, Autosomal Recessive	Le Sa
277700	Werner Syndrome	P
277900	Wilson Disease	O P
278000	Wolman Disease	B J T
278250	Wrinkly Skin Syndrome	O P Sa Sy U
278300	Xanthinuria, Type I	K Le
278700	Xeroderma Pigmentosum, Complementation Group A	Eg P T U
278720	Xeroderma Pigmentosum, Complementation Group C	Eg
279000	Young Syndrome	So Y
300000	Opitz Syndrome	B
300068	Androgen Insensitivity Syndrome	Eg O
300100	Adrenoleukodystrophy	Le Sa
300331	Thrombocytosis, Familial X-Linked	Sa

OMIM #	Name	Country*
300388	Polymicrogyria, Bilateral Perisylvian	B
300419	Mental Retardation, X-Linked 54	T
300530	Kawasaki Disease	B O Su Y
300624	Fragile X Mental Retardation Syndrome	B K Le O Sa
301090	Tetra-Amelia, X-Linked	P
301220	Pigmentary Disorder, Reticulate, with Systemic Manifestations	Le U
301500	Fabry Disease	U
301800	Anus, Imperforate	I Sa U
301900	Borjeson-Forssman-Lehmann Syndrome	Sa
302960	Chondrodysplasia Punctata 2, X-Linked Dominant	U
303350	MASA Syndrome	U
304050	Aicardi Syndrome	O
304100	Corpus Callosum, Partial Agenesis of, X-Linked	O
304790	Immunodysregulation, Polyendocrinopathy, and Enteropathy, X-Linked	Mo
305000	Dyskeratosis Congenita, X-linked	Eg I U
305600	Focal Dermal Hypoplasia	J O Sa
305800	Membranoproliferative Glomerulonephritis, X-Linked	B K Le O Q Sa U Y
305900	Glucose-6-Phosphate Dehydrogenase	A B C D Eg I J Le Li O P Sa So Su Sy T U Y
306700	Hemophilia A	B Eg I So
307000	Hydrocephalus due to Congenital Stenosis of Aqueduct of Sylvius	K Sa U
307030	Hyperglycerolemia	O
308050	Congenital Hemidysplasia with Ichthyosiform Erythroderma and Limb Defects	Eg
308205	Ichthyosis Follicularis, Atrichia, and Photophobia Syndrome	Le
308240	Lymphoproliferative Syndrome, X-Linked	B
308350	Infantile Spasm Syndrome, X-Linked	O Sa U
309200	Major Affective Disorder 2	I Y
309350	Melnick-Needles Syndrome	Eg
309400	Menkes Disease	U
309520	Lujan-Fryns Syndrome	Le
309550	Fragile Site Mental Retardation 1 Gene	T U
309900	Mucopolysaccharidosis Type II	Mo
310200	Muscular Dystrophy, Duchenne Type	Eg J
310400	Myotubular Myopathy 1	O Y
311200	Orofaciodigital Syndrome I	Sa
311250	Ornithine Transcarbamylase Deficiency, Hyperammonemia due to	O
312000	Panhypopituitarism, X-Linked	Q
312060	Properdin Deficiency, X-Linked	T
312550	Retinal Dysplasia, Primary	I
312750	Rett Syndrome	K O Sa Su
313900	Thrombocytopenia 1	Sa
580000	Deafness, Aminoglycoside-Induced	P
600060	Deafness, Neurosensory, Autosomal Recessive 2	T
600116	Parkinson Disease 2, Autosomal Recessive Juvenile	A T
600118	Warburg Micro Syndrome	Le
600146	Spastic Paraplegia 5B, Autosomal Recessive	T
600252	Lowry-Maclean Syndrome	K
600309	Atrioventricular Septal Defect	O
600360	Aplasia Cutis Congenita of Limbs, Recessive	Y
600631	Enuresis, Nocturnal 1	U
600669	Epilepsy, Idiopathic Generalized	Eg O
600737	Inclusion Body Myopathy 2, Autosomal Recessive	P
600794	Spinal Muscular Atrophy, Distal, Type V	A
600807	Asthma, Susceptibility To	O U
600850	Schizophrenia 4	A
600995	Nephrotic Syndrome, Steroid-Resistant, Autosomal Recessive	O Sa U
601067	Usher Syndrome, Type ID	Mo
601071	Deafness, Autosomal Recessive 9	Le
601161	Trisomy 18-Like Syndrome	U
601170	Muscular Dystrophy, Congenital, with Severe Central Nervous System Atrophy and Absence of Large Myelinated Fibers	U
601214	Naxos Disease	P
601277	Ichthyosis, Lamellar, 2	Mo
601367	Stroke, Ischemic	O Q
601386	Deafness, Autosomal Recessive 12	Sy
601446	Right Pelvic Kidney	U
601451	Nevo Syndrome	U
601537	Microcephaly, Retinitis Pigmentosa, and Sutural Cataract	Mo
601549	Alacrima	J
601552	Ectopia Lentis, Spontaneous Filtering Blebs, and Craniofacial Dysmorphism	Le
601553	Hypotrichosis, Congenital, with Juvenile Macular Dystrophy	Eg
601559	Stuve-Wiedemann Syndrome	O Su U Y
601596	Charcot-Marie-Tooth Disease, Type 4C	A
601606	Trichoepithelioma, Multiple Familial	Sa
601626	Leukemia, Acute Myeloid	O
601634	Neural Tube Defects, Folate-Sensitive	B O P Su U
601706	Yemenite Deaf-Blind Hypopigmentation Syndrome	Y

OMIM #	Name	Country*
602078	Fibrosis of Extraocular Muscles, Congenital, 2	Sa
602088	Nephronophthisis 2	P
602089	Hemangioma, Capillary Infantile	U
602097	Usher Syndrome, Type IE	Mo
602099	Amyotrophic Lateral Sclerosis 5	T
602247	Xanthomatosis, Susceptibility to	Sy
602400	Ichthyosis, Follicular Atrophoderma, Hypotrichosis, and Hypohidrosis	U
602401	Ectodermal Dysplasia, Hidrotic, Autosomal Recessive	Le
602459	Deafness, Autosomal Dominant Nonsyndromic Sensorineural 15	Eg Li T
602501	Megalencephaly-Cutis Marmorata Telangiectatica Congenita	Le
602522	Bartter Syndrome, Infantile, with Sensorineural Deafness	Le P
602557	Spondyloepimetaphyseal Dysplasia, Shohat Type	I
602722	Renal Tubular Acidosis, Distal, Autosomal Recessive	Le O Sa
603003	Bile Duct Cysts	B Eg Sa
603034	Endplate Acetylcholinesterase Deficiency	I P
603098	Deafness, Autosomal Recessive 13	Le
603133	Dislocated Elbows, Bowed Tibias, Scoliosis, Deafness, Cataract, Microcephaly, and Mental Retardation	Le
603165	Dermatitis, Atopic	O U
603194	Meckel Syndrome, Type 2	A T
603266	Diabetes Mellitus, Insulin-Dependent, 17	P
603278	Focal Segmental Glomerulosclerosis 1	B U
603438	Radioulnar Synostosis With Microcephaly, Short Stature, Scoliosis, And Mental Retardation	P
603513	Cerebral Palsy, Spastic, Symmetric, Autosomal Recessive	O
603546	Spondyloepimetaphyseal Dysplasia with Multiple Dislocations	I Le
603553	Hemophagocytic Lymphohistiocytosis, Familial, 2	O
603554	Omenn Syndrome	Mo
603629	Deafness, Autosomal Recessive 21	Le
603642	Atrial Septal Defect, Secundum, with Various Cardiac and Noncardiac Defects	Le
603650	Bardet-Biedl Syndrome 5	K Sa
603671	Acromelic Frontonasal Dysostosis	U
603678	Deafness, Autosomal Recessive 14	Le
603720	Deafness, Autosomal Recessive 16	P Sy
603802	Microcephaly with Simplified Gyral Pattern	O U
603813	Hypercholesterolemia, Autosomal Recessive	Sy
603896	Leukoencephalopathy with Vanishing White Matter	A
603903	Sickle Cell Anemia	A B C K O P Sa Su U Y
603933	Diabetic Nephropathy, Susceptibility to	B K O Q Sa U
604201	Hepatic Fibrosis, Severe, Susceptibility to, due to Schistosoma Mansoni Infection	Su
604228	Partial Albinism and Immunodeficiency Syndrome	Sa
604232	Leber Congenital Amaurosis, Type III	Sa
604320	Spinal Muscular Atrophy with Respiratory Distress 1	Le
604321	Microcephaly, Primary Autosomal Recessive, 4	Mo
604363	Myoclonic Epilepsy, Congenital Deafness, Macular Dystrophy, and Psychiatric Disorders	Le
604370	Ovarian Cancer, Epithelial	O Q
604498	Amegakaryocytic Thrombocytopenia, Congenital	Sa U
604563	Charcot-Marie-Tooth Disease, Type 4B2	Mo T
604571	Bare Lymphocyte Syndrome, Type I	Mo
604801	Muscular Dystrophy, Congenital, 1B	U
605027	Lymphoma, Non-Hodgkin, Familial	B O U
605074	Renal Cell Carcinoma, Papillary	B Eg
605156	Multicentric Osteolysis, Nodulosis, And Arthropathy	Sa
605203	Multiple Pterygium Syndrome, Aslan Type	Q
605225	Inflammatory Bowel Disease 7	I
605316	Deafness, Congenital Neurosensory, Autosomal Recessive 10	P
605552	Abdominal Obesity-Metabolic Syndrome	I O
605588	Charcot-Marie-Tooth Disease, Axonal, Type 2B1	A Mo
605685	Cutis Verticis Gyrata, Retinitis Pigmentosa, and Sensorineural Deafness	Le
605726	Neuropathy, Distal Hereditary Motor, Jerash Type	J
605818	Deafness, Autosomal Recessive 27	U
605822	Spondyloocular Syndrome, Autosomal Recessive	I
605899	Glycine Encephalopathy	O P
606054	Propionic Acidemia	B O Sa U
606072	Rippling Muscle Disease	O
606176	Diabetes Mellitus, Permanent Neonatal	O
606220	Mental Retardation, Short Stature, Facial Anomalies, and Joint Dislocations	Le
606324	Parkinson Disease 7, Autosomal Recessive Early-Onset	Le
606369	Epileptic Encephalopathy, Lennox-Gastaut Type	O Sa
606438	Huntington Disease-Like 2	Mo Sa
606527	Megarbane Syndrome	I Le
606545	Ichthyosis, Lamellar, 5	U
606612	Muscular Dystrophy, Congenital, 1C	A T
606693	Parkinson Disease 9	J
606744	Seckel Syndrome 2	I
606788	Anorexia Nervosa, Susceptibility to, 1	O U
606812	Fumarase Deficiency	U

OMIM #	Name	Country*
606824	Glucose/Galactose Malabsorption	I
606854	Polymicrogyria, Bilateral Frontoparietal	P U
606893	Vascular Malformation, Primary Intraosseous	Mo O
606937	Spinocerebellar Ataxia, Autosomal Recessive 5	Le
606943	Usher Syndrome, Type IG	J P T
607014	Hurler Syndrome	Mo O
607039	Deafness, Autosomal Recessive 22	P
607084	Deafness, Autosomal Recessive 31	J P
607088	Spinal Muscular Atrophy, Distal, Autosomal Recessive	Le U
607095	Anauxetic Dysplasia	J
607101	Deafness, Autosomal Recessive 30	I
607131	Macrocephaly with Multiple Epiphyseal Dysplasia and Distinctive Facies	O U
607154	Allergic Rhinitis	B O Sa
607155	Muscular Dystrophy, Limb-Girdle, Type 2I	T
607239	Deafness, Autosomal Recessive 33	J
607250	Spinocerebellar Ataxia, Autosomal Recessive, with Axonal Neuropathy	Sa
607364	Bartter Syndrome, Type 3	K P Sa U
607398	Glucocorticoid Deficiency 2	U Y
607411	Patent Ductus Arteriosus	U
607432	Lissencephaly I	O Sa U
607473	Vitamin K-Dependent Clotting Factors, Combined Deficiency of, 2	Le
607483	Basal Ganglia Disease, Biotin-Responsive	O Sy Y
607499	Bulimia Nervosa, Susceptibility to, 1	Mo O
607572	Leprosy, Susceptibility to, 2	U
607584	Spastic Paraplegia 24, Autosomal Recessive	Sa
607608	Sphingomyelin Phosphodiesterase 1, Acid Lysosomal	A Mo Sa T
607624	Griscelli Syndrome, Type 2	P
607625	Niemann-Pick Disease, Type C2	A
607626	Ichthyosis, Leukocyte Vacuoles, Alopecia, and Sclerosing Cholangitis	Mo
607665	Tubulointerstitial Nephritis with Uveitis	J Mo T
607694	Leukodystrophy with Oligodontia	Le Sy
607731	Charcot-Marie-Tooth Disease, Axonal, Type 2H	T
607765	Cholestasis, Progressive Familial Intrahepatic 4	Sa
607812	Craniolenticulosutural Dysplasia	Sa
607831	Charcot-Marie-Tooth Disease, Axonal, Type 2K	Mo
607847	Neutropenia, Nonimmune Chronic Idiopathic, of Adults	J O
607907	Dermatofibrosarcoma Protuberans	O Sa T
608027	Cerebellar Atrophy with Progressive Microcephaly	O
608091	Joubert Syndrome 2	U
608097	Heterotopia, Periventricular, Autosomal Recessive	Y
608099	Muscular Dystrophy, Limb-Girdle, Type 2D	T
608115	Ovarian Hyperstimulation Syndrome	Mo
608154	Lipodystrophy, Generalized, with Mental Retardation, Deafness, Short Stature, and Slender Bones	O
608156	Nablus Mask-Like Facial Syndrome	P
608207	Kala-Azar, Susceptibility to	Su
608232	Leukemia, Chronic Myeloid	O Q
608358	Myopathy, Myosin Storage	Sa
608395	Karak Syndrome	J
608443	Mental Retardation, Autosomal Recessive, 3	P
608446	Myocardial Infarction, Susceptibility to, 1	?
608509	Alopecia Universalis Congenita, XY Gonadal Dysgenesis, and Laryngomalacia	J
608585	Brachial Palsy, Familial Congenital	Eg
608594	Lipodystrophy, Congenital Generalized, Type 1	A Eg Mo O P T U
608637	Spondyloepiphyseal Dysplasia, Omani Type	O
608653	Deafness, Autosomal Recessive 32	T
608681	Spondylocostal Dysostosis, Autosomal Recessive 2	Le
608710	Wegener Granulomatosis	B O
608728	Spondyloepimetaphyseal Dysplasia, Matrilin-3 Related	?
608808	Transposition of the Great Arteries, Dextro-Looped	Le O
608836	Carnitine Palmitoyltransferase II Deficiency, Lethal Neonatal	B Mo O
608911	Choanal Atresia, Posterior	Eg Le O P Sa Sy U Y
608931	Myasthenic Syndrome, Congenital, Associated with Acetylcholine Receptor Deficiency	I
608980	Bifid Nose, Renal Agenesis, and Anorectal Malformations	Eg U
609047	Skeletal Dysplasia, Rhizomelic, with Retinitis Pigmentosa	Le
609166	Branchiogenic-Deafness Syndrome	Le
609192	Loeys-Dietz Syndrome, Type 1A	Le
609222	Cephalocele, Atretic	O
609311	Charcot-Marie-Tooth Disease, Type 4H	A Le
609465	Al-Gazali Syndrome	P Su U
609654	Short Stature and Facioauriculothoracic Malformations	Le
610006	2-@Methylbutyryl-CoA Dehydrogenase Deficiency	Er So
610093	Microphthalmia, Isolated 2	Q
610685	Split-Hand/Foot Malformation with Long Bone Deficiency 2	O U

*A: Algeria, B: Bahrain, C: Comoros, D: Djibouti, Eg: Egypt, Er: Eritrea, I: Iraq, J: Jordan, K: Kuwait, Le: Lebanon, Li: Libya, Ma: Mauritania, Mo: Morocco, O: Oman, P: Palestine, Q: Qatar, Sa: Saudi Arabia, So: Somalia, Su: Sudan, Sy: Syria, T: Tunisia, U: United Arab Emirates, Y: Yemen, ?: country not specified.