Clinical Utility Gene Cards

Status list, May 21 2017

Disease	Status	OMIM #	Release year
15q13.3 microdeletion syndrome	Completed	612001	2014
16p12.1 microdeletion syndrome	In progress		
16p12.2 microdeletion	Completed		
16p13.11 microdeletion syndrome	Completed		2013
1q21.1 microdeletion syndrome	In progress	612474	
3-M syndrome	Completed	273750, 612921	2011
3-M syndrome - update	Completed		2013
3-methylcrotonylglycinuria	No author assigned	210200, 210210	
3-Methylglutaconic aciduria type 3	No author assigned	258501	
Aarskog-Scott syndrome (Faciogenital dysplasia)	Completed	305400	2011
Aarskog-Scott syndrome – update 2014	Completed		
Abetalipoproteinemia	Completed	200100	2012
Abetalipoproteinaemia - Update 2014	Completed		2014
Acatalasemia	No author assigned	614097	
Aceruloplasminemia	No author assigned	604290	
Achondrogenesis	No author assigned	200600, 200610, 600972	
Achondroplasia	In progress	100800	
Achromatopsia	Completed	139340, 216900, 262300, 613093	2011
Achromatopsia - UPDATE 2013	Completed		2013
Acrodermatitis enteropathica, zinc deficiency type	Completed	201100	2011
Acrodermatitis enteropathica, zinc deficiency type – update 2015	Completed	201100	
Acute hepatic porphyria	No author assigned	612740	

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Acute intermittent porphyria	No author assigned	176000	
Acute myeloid leukemia	No author assigned	601626 164040	
Acute promyelocytic leukemia	No author assigned	612376	
Adenosine monophosphate deaminase deficiency	No author assigned	102770, 612874	
Adrenocortical carcinoma (ADCC)	No author assigned	202300	
Adrenoleukodystrophy ALD /Adrenomyeloneuropathie ALD/AMN	Completed	300100, 302700	2011
Afibrinogenemia	No author assigned	202400	
Agenesis of the corpus callosum with peripheral neuronopathy ACCPN	In progress	218000	
Aicardi-Goutieres syndrome	In progress	225750 610181 610329 610333 612952	
Alagille Syndrome	Completed	118450, 610205	2013
Alexander Disease	In progress	203450	
ALG1 defective congenital disorder of glycosylation	Completed		2015
Alkaptonuria	No author assigned	203500	
Alopecia universalis congenita	No author assigned	203655	
Alpha-1-Antitrypsin deficiency	Completed	613490	2011
Alpha-mannosidosis	Completed	248500	2011
Alpha-thalassemia	No author assigned	141750	
Alport Syndrome	Completed	104200, 203780, 301050,	2011
Alport syndrome - update 2014	Completed		2014
Alstrom Syndrome	Completed	203800	2011
Alveolar rhabdomyosarcoma	Completed	268220	2011
Alzheimer disease	No author assigned	104300	
Amyloidosis	No author assigned	105200	
Androgen insensitivity syndrome (CAIS)	Re-edit required	300068, 300274, 312300	
Angelman syndrome	Completed	105830	2014
Angioneurotic edema (Angioedema)	No author assigned	106100, 610618	
Aniridia (AN)	Completed	106210	
Ankyloblepharon-Ectodermal Defects-Cleft Lip/Palate Syndrome	No author assigned	106260	
Apert syndrome	No author assigned	101200	
Arrhythmogenic right ventricular cardiomyopathy/dysplasia	Completed	107970, 600996, 602086, 602087,	2013

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		604400, 604401, 607450, 609040, 609160, 610193, 610476, 611528	
Arterial Tortuosity syndrome	Completed	208050	2013
Arthrogryposis-> Sheldon-Hall syndrome	No author assigned	601680	
Ataxia-oculomotor apraxia syndrome	No author assigned	208920	
Ataxia-Telangiectasia	No author assigned	208900	
Atelosteogenesis I	No author assigned	108720	
Atelosteogenesis type II	No author assigned	256050	
Atelosteogenesis type III	No author assigned	108721	
Atrioventricular septal defect (AVSD)	No author assigned	600309	
Atypical hemolytic uremic syndrome	In progress	235400	
Autoimmune lymphoproliferative syndrome	In progress	601859, 603909, 605233, 607271	
Autosomal dominant epidermolysis bullosa dystrophica (DBEB)	No author assigned	131750	
Autosomal dominant hidrotic ectodermal dysplasia	No author assigned	129500	
Autosomal Dominant Hyper IgE syndrome	No author assigned	147060 243700	
Autosomal dominant isolated ectopia lentis	No author assigned	129600	
Autosomal dominant keratitis	No author assigned	148190	
Autosomal dominant myotonia congenita (Thomsen disease)	In progress	160800	
Autosomal dominant or sporadic congenital neutropenia	No author assigned		
Autosomal dominant osteopetrosis	In progress		
Autosomal Dominant Polycystic Kidney Disease	In progress	173900 173910 600666 601313	
Autosomal Recessive Cone-rod dystrophy	Completed	600624 604116 605549 612775 612657 608194 613660 614500 268100 610024 610356	2015
Autosomal recessive isolated ectopia lentis	No author assigned	225100	

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Autosomal recessive juvenile Parkinson disease	No author assigned	600116 605909 606324	
Autosomal recessive limb girdle muscular dystrophy	No author assigned	253600	
Autosomal recessive osteopetrosis	In progress	259700 259710 259720 611490	
Autosomal Recessive Polycystic Kidney Disease	In progress	263200	
Axenfeld-Rieger syndrome	Completed	602482, 601631, 180500	2010
Azoospermia (non-obstructive, Y-linked spermatogenic failure)	In progress	415000	
B4GALT7-CDG, Congenital disorder of glycosylation type O	Completed		
Baller-Gerold Syndrome	In progress	218600	
Bardet-Biedl Syndrome	Completed	209900	2010
Bardet-Biedl Syndrome - UPDATE 2013	Completed		2013
Bartter syndrome, type 1-5	No author assigned	601678, 241200, 607364, 602522	
Becker Muscular Dystrophy BMD	In progress	300376	
Beckwith-Wiedemann Syndrome	Completed	130650	2013
Benign essential blepharospasm	No author assigned	606798	
Bernard-Soulier syndrome (Hemorrhagiparous thrombocytic dystrophy)	No author assigned	153670, 231200	
Best-1 related dystrophies	Completed	153700	2012
Beta-thalassemia (β-thalassemia)	In progress	141900, 604131	
Biotinidase deficiency	Completed	253260	2012
Biotinidase deficiency – update 2015	Completed	253260	
Birt-Hogg-Dube syndrome	No author assigned	135150	
Blepharophimosis, Ptosis, and Epicanthus Inversus	No author assigned	110100	
Bloom syndrome	No author assigned	210900	
Blue cone monochromatism	Completed	303700	2011
Branchiootorenal syndrome	No author assigned	113650	
Brugada Syndrome	In progress	601144, 611777, 611875, 611876, 612838, 613119, 613120, 613123	
Budd-Chiari syndrome	No author assigned	600880	

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Buschke-Ollendorff syndrome	In progress	166700	
Campomelic Dysplasia	Completed	114290 211990 602196	2012
Camurati-Engelmann disease	No author assigned	131300 606631	
Cantú syndrome	Completed		
Carbamoyl phosphate synthetase I deficiency	No author assigned	237300	
Carney complex	Completed	160980	
Carnitine Palmitoyltransferase 1A Deficiency	No author assigned	255120	
Carnitine Palmitoyltransferase II Deficiency	No author assigned	255110 600649 608836	
Cat eye syndrome (CES)	No author assigned	115470	
Catecholaminergic polymorphic ventricular tachycardia (CPVT)	Completed	604772 192605	2013
Central areolar choroidal dystrophy	No author assigned	613105	
Central core disease	Completed	117000	2011
Centronuclear and myotubular myopathies	Completed	160150	2012
Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL)	No author assigned	125310	
Cerebrotendinous xanthomatosis	In progress	213700	
CHARGE syndrome	Completed	214800	2011
CHARGE syndrome	Completed		2015
Choanal atresia and lymphedema	No author assigned	613611	
Choroideremia	Completed	303100	2013
Chronic lymphocytic leukemia (CLL)	In progress	151400	
Chronic myeloid leukemia (CML)	No author assigned	608232	
Citrullinemia	No author assigned	215700	
Cleft palate	No author assigned	119540	
Cleidocranial Dysplasia	No author assigned	119600 216330	
Coffin-Lowry Syndrome	In progress	303600	
Cohen Syndrome	No author assigned	216550	
Combined pituitary hormone deficiency	No author assigned	613038 262600 221750 262700 613986	
Common variable immunodeficiency	No author assigned	607594 240500 613943 613494 613495 613496	
Cone rod dystrophy	In progress	120970 300476 304020	

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		600624 600977 601777 602093 603649 604011 604116 604393 605549 608194 610283 610381 610478 612657 612775	
Congenital Adrenal Hyperplasia (CAH)	No author assigned	145295 201710 201750 201810 201910 202010 202110	
Congenital Bilateral Aplasia of Vas Deferens - CBAVD/CUAVD	In progress	277180	
Congenital central hypoventilation syndrome (CCHS)	No author assigned	209880	
Congenital disorder of glycosylation type Ic (ALG6-CDG)	Completed	603147	
Congenital disorder of glycosylation type Ij (CDG Ij, CDG1J)	In progress	608093	
MAN1B1 Congenital disorder of glycosylation	Completed		
Congenital dyserythropoietic anemia	No author assigned	224120	
Congenital Generalized Lipodystrophy	Completed		
Congenital hereditary endothelial dystrophy with progressive sensorineural deafness (Harboyan syndrome)	No author assigned	217400	
Congenital Lactase deficiency	In progress	223000	
Congenital nongoitrous hypothyroidism	No author assigned	275200 218700 609893 275100 225250 614450	
Congenital primary aphakia	In progress	610256	
Cornelia de Lange Syndrome	Completed	122470 300590 610759	2014
Costello Syndrome	In progress	218040	
Cowden syndrome	In progress	158350	
Craniofrontonasal syndrome - CFNS -	No author assigned	304110	
Craniosynostosis	No author assigned	123100	

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		604757	
Creutzfeldt-Jakob disease	No author assigned	123400	
Crigler-Najjar syndrome	No author assigned	218800	
Crouzon disease	No author assigned	123500	
Currarino syndrome	No author assigned	176450	
Cushing syndrome (ACTH-secreting pituitary adenoma)	No author assigned	219090	
Cystic fibrosis	In progress	219700	
Cystinosis	Completed	219800	2013
Cystinuria	Completed	220100	2011
Darier-White disease	No author assigned	124200	
Deletion 22q13 Syndrome (Phelan-McDermid Syndrome)	Completed	606232	2010
Dentatorubral-pallidoluysian atrophy	No author assigned	125370	
Dent disease	Completed		2014
Denys-Drash and Frasier Syndromes	In progress	194080 136680	
Dermatofibrosarcoma protuberans	No author assigned	607907	
Diamond-Blackfan Anemia	Completed	105650, 610629, 612527, 612528, 612561,	2011
Diamond-Blackfan Anemia - update 2013	Completed		2013
Diastrophic dysplasia	No author assigned	222600	
DiGeorge syndrome, velocardiofacial syndrome, Shprintzen syndrome, chromosome 22q11.2 deletion syndrome (22q11.2, TBX1)	Completed	188400, 192430	2010
Dilated cardiomyopathy (CMD)	Completed	613122	2012
Distal arthrogryposis type 1A	No author assigned	108120	
Donohue syndrome	No author assigned	246200	
Dopamine beta-hydroxylase (DBH) deficiency	No author assigned	223360	
Dopa-responsive dystonia (DRD)	No author assigned	128230	
DPAGT1 defective congenital disorder of glycosylation	Completed		2015
Duane Syndrome	No author assigned	126800	
Duchenne Muscular Dystrophy, DMD	In progress	310200	
Dyskeratosis congenita	Completed	127550	2011
Dystrophia Myotonica 1 - DM1	Re-edit required	160900	2008
Dystrophia Myotonica 2 - DM2	In progress	602668	
Early infantile epileptic encephalopathy-1 (EIEE1) [West syndrome]	No author assigned	308350	
Ehlers-Danlos syndrome types I-VII	Completed	130000, 130010, 130020, 130050,	2010

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		225400, 130060, 225410, 612350	
Ehlers-Danlos syndrome types I-VII - UPDATE 2012	Completed		2012
Elliptocytosis 1 and 2	No author assigned	611804	
Emery-Dreifuss Muscular Dystrophy	No author assigned	181350 310300 612998 612999	
Erythropoietic protoporphyria	No author assigned	177000 300752	
Esophageal cancer	No author assigned	133239	
Essential thrombocythemia	No author assigned	187950	
Ewing sarcoma family of tumors (ESFT)	No author assigned	612219	
Fabry disease	Completed	301500	2011
Fabry disease - update 2016	Completed		
Facioscapulohumeral muscular dystrophy (FSHD)	No author assigned	158900	
Factor II (prothrombin) deficiency	No author assigned	613679	
Factor V Deficiency	In progress	227400	
Factor VII deficiency	In progress	227500	
Factor X deficiency	In progress	227600	
Factor XI deficiency	No author assigned	612416	
Factor XIII deficiency	No author assigned	613225	
Familial adenomatous polyposis	Completed	175100	2011
Familial adenomatous polyposis – update 2014	Completed		2014
Familial amyotrophic lateral sclerosis	In progress	105400 205100 205200 205250 602099 602433 606640 608030 608031 608627 611895 612069 612577 613435	
Familial aortopathy	In progress		
Familial breast-ovarian cancer (BRCA)	In progress	114480	
Familial cold autoinflammatory syndrome-1 (FCAS1)	No author assigned	120100	
Familial erythrocytosis	Completed	133100 263400 609820	2012

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		611783	
Familial exudative vitreoretinopathy (FEVR)	In progress	133780	
Familial hemiplegic migraine	No author assigned	141500	
Familial hypobetalipoproteinaemia (APOB) - Update 2014	Completed	2014	
Familial hypomagnesemia with hypercalciuria and nephrocalcinosis (FHHNC)	In progress	248250	
Familial Parkinson disease dementia	No author assigned	125320 260540	
Familial partial lipodystrophy	Completed		
Familial platelet disorder with associated myeloid malignancies	Completed	601399	
Familial restrictive cardiomyopathy (RCM)	No author assigned	115210	
Familial Wilms tumor (Nephroblastoma)	In progress	194070 194071 194090 601363 605982	
Fanconi anemia	No author assigned	227650	
Feingold syndrome	No author assigned	164280 602585	
Fibrodysplasia ossificans progressiva	No author assigned	135100	
Focal segmental glomerulosclerosis	In progress	603278 603965 607832 612551 613237	
Fragile X mental retardation syndrome, fragile X-associated tremor/ataxia syndrome and fragile X-associated primary ovarian insufficiency	Completed	300624	2011
Fraser syndrome	In progress	219000	
Friedreich-Ataxia 1 - FRDA	In progress	229300 601992	
Frontotemporal dementia (FTD)	No author assigned	600274	
Fructose intolerance	No author assigned	229600	
Fructose-1,6-bisphosphatase deficiency	No author assigned	229700	
Galactosemia	No author assigned	230400	
GALNT3-CDG, Congenital disorder of glycosylation type O	Completed		
Gastric cancer, familial	Completed	137215 613659	2013
Gastrointestinal stromal tumor	No author assigned	606764	
GATA2-related myelodysplastic syndrome	In progress		
Gaucher disease	No author assigned	230800 230900 231000 608013	
Genetic Parkinson disease	No author assigned	168600 168601	

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		300557 556500 602404 602544 605543 606693 606852 607060 607688 610297	
Gitelman syndrome	Completed	263800	2011
Glanzmann thrombasthenia	Completed	187800	
Glutaric acidemia	No author assigned	231670	
Glutathione synthetase deficiency	No author assigned	231900 266130	
Glycine encephalopathy (GCE)	No author assigned	605899	
Glycogen storage disease type 2	No author assigned	232300	
Gonadal Dysgenesis	re-edit required	233420 400044 612965 607080 400045 613080	2008
Gorlin Syndrome	Completed	109400	2011
Gorlin Syndrome - update 2013	Completed	109400	2013
GRACILE syndrome	No author assigned	603358	
Growth hormone insensitivity syndrome (Laron syndrome)	No author assigned	262500	
Growth hormone-secreting pituitary adenoma (incl. Acromegaly)	No author assigned	102200	
Guanidinoacetate methyltransferase deficiency	No author assigned	612736	
Guillain-Barré syndrome	No author assigned	139393	
Haemochromatosis	Completed	235200	2010
Hartnup disorder	No author assigned	234500	
Hemophilia A	Completed	134500	2011
Hemophilia B	Completed	306900	2012
Hepatic Veno-Occlusive Disease with Immunodeficiency	No author assigned	235550	
Hepatocellular carcinoma (HCC) and hepatoblastoma	No author assigned	114550	
Hereditary angioedema types I, II and III	In progress	106100 610618	
Hereditary angioedema with normal C1 inhibitor	Completed		
Hereditary megaloblastic anemia-1 (Imerslund-Gräsbeck syndrome - selective vitamin B12 malabsorption with proteinuria)	No author assigned	261100	
Hereditary methemoglobinemia	No author assigned	250700	
Hereditary pancreatitis	In progress	167800	

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Hereditary recurrent fevers	Completed	142680 249100 260920 610377	2013
Hereditary spherocytosis	No author assigned	182900 270970 612653 612690	
Hereditary Thoracic Aortic Aneurysm and Dissection (TAAD)	Completed		
Hereditary thrombocythaemia	Completed	187950	
Hermansky-Pudlak Syndrome	No author assigned	203300 608233	
Heterotaxy	No author assigned	613751 306955 270100 605376 613751	
Hirschsprung disease	No author assigned	142623 600155	
Histidinemia	No author assigned	235800	
HMSN/HNPP HMSN types 1, 2, 3, 6 (CMT1,2,4, DSN, CHN, GAN, CCFDN, HNA); HNPP	Completed	118220, 118210, 118200, 609260, 162500, 145900, 302800 607677,	2010
Hodgkin disease, classical	No author assigned	236000	
Holoprosencephaly	Completed	236100	2010
Holt-Oram syndrome (HOS)	No author assigned	142900	
Homocystinuria - MTHFR - Hyperhomozysteinämie	No author assigned	236250	
Huntington disease	Completed	143100	2013
Hutchinson-Gilford progeria syndrome	In progress	176670	
Hydrolethalus syndrome 1	No author assigned	236680	
Hyperkalemic periodic paralysis (HYPP)	In progress	170500	
Hyperlipoproteinemia Type III	No author assigned	107741	
Hyperlipoproteinemia, TYPE I and TYPE V	No author assigned	207750	
Hyperlipoproteinemia, TYPE II	Completed	143890	2013
Hyperlipoproteinemia, TYPE IV	No author assigned	144600	
Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome	In progress	238970	
Hypertrophic cardiomyopathy	Completed	192600 115195 115196 115197 600858	2011
Hypertrophic cardiomyopathy – update 2017	In progress		

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Hypobetalipoproteinemia	Completed	605019	
Hypochondroplasia	No author assigned	146000	
Hypokalemic periodic paralysis (HOKPP)	In progress	170400	
Hypophosphatasia	Completed	146300, 241500, 241510	2010
Hypophosphatasia - Update	Completed	146300, 241500, 241510	2013
Idiopathic hypereosinophilic syndrome	No author assigned	607685	
Idiopathic pulmonary fibrosis (IPF)	No author assigned	178500	
Iminoglycinuria	No author assigned	242600	
Incontinentia Pigmenti	Completed	308300	
Infantile Refsum disease	No author assigned	266510	
IPEX syndrome	In progress	304790	
Isolated microphthalmia	In progress	610093 613094 613517 611040 613704 611038	
Isovaleric acidemia (IVA)	No author assigned	243500	
Johanson-Blizzard syndrome	Completed		
Joubert syndrome	Completed	213300 243910 216360 277170	2011
Joubert syndrome - update 2013	Completed		2013
Kabuki Syndrome	In progress	147920	
Kallmann syndrome	No author assigned	147950 244200 610628 612370 612702	
Kaposi sarcoma	No author assigned	148000	
Klinefelter Syndrome	No author assigned		
Klippel-Feil syndrome	No author assigned	118100	
Krabbe Disease	No author assigned	245200	
Laing distal myopathy	Completed	160500	2010
Leber amaurosis, congenital	In progress	204000 204100 604232 604393 604537 610612 611755 612712 613341	
Leber optic atrophy and dystonia	No author assigned	500001	

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Legg-Calve-Perthes disease	No author assigned	150600	
Leigh syndrome	No author assigned	256000	
Lennox-Gastaut epileptic encephalopathy	No author assigned	606369	
Lenz Microphthalmia Syndrome	No author assigned		
Leopard syndrome	No author assigned	611554	
Leri-Weill dyschondrosteosis (LWD)	In progress	127300	
Lesch-Nyhan syndrome (HPRT deficiency)	Completed	300322	2010
Lesch-Nyhan syndrome (HPRT deficiency) - UPDATE 2013	Completed		2013
Li-Fraumeni syndrome	No author assigned	151623 609265	
Lissencephaly (incl. Miller-Dieker-Syndrom)	Re-edit required	607432 300067 300215 257320 247200 611603	
Loeys-Dietz syndrome	Completed	609192 610380 610168 608967	2011
Loeys-Dietz syndrome – update 2015	In progress		
Long-chain 3-hydroxyl-CoA dehydrogenase deficiency (LCHAD)	No author assigned	609016	
Long-QT syndrome	Completed	192500 600919 603830 611818 611819 611820 612955 613485	2013
Lowe Syndrome	Completed	309000	
Lymphoproliferative Disease, X-Linked	No author assigned	300635 308240	
Lynch syndrome (MLH1, MSH2, MSH6, PMS2)	Completed	120435	2010
Lynch syndrome (MLH1, MSH2, MSH6, PMS2) - UPDATE 2012	Completed		2012
Lysinuric protein intolerance	In progress		
Lysosomal acid lipase deficiency (Wolman disease)	No author assigned	278000	
Malignant hyperthermia	Completed	145600	2011
MAN1B1 Congenital disorder of glycosylation	Completed		
Maple syrup urine disease	No author assigned	248600	
Marfan syndrome type 1 and related phenotypes [FBN1]	Completed	154700	2010
Marinesco-Sjögren syndrome	In progress	248800	
MASA syndrome (spastic paraplegia-1((SPG1)	No author assigned	303350	

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Maternally inherited diabetes-deafness syndrome	No author assigned	520000	
Mayer-Rokitansky-Küster-Hauser syndrome	Completed	277000	2011
McCune-Albright syndrome	No author assigned	174800	
McLeod Neuroacanthocytosis syndrome	No author assigned	314850	
Meckel syndrome	Completed	249000 603194 607361 611134 611561 612284	2011
Meckel syndrome - update 2016	Completed		
Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency	No author assigned	201450	
Medullary cystic kidney disease (MCKD)	No author assigned	603860	
Medullary thyroid carcinoma (MTC)	No author assigned	155240	
Medulloblastoma	No author assigned	155255	
MELAS syndrome	No author assigned	540000	
Menkes disease	Completed	309400	2011
Mental retardation, Enteropathy, Deafness, peripheral Neuropathy, Ichthyosis, and Keratoderma MEDNIK syndrome	In progress		
Merosin-deficient congenital muscular dystrophy	No author assigned	607855	
MERRF	No author assigned	545000	
Metachromatic leukodystrophy	No author assigned	250100	
Milroy disease (hereditary lymphedema type IA)	In progress	153100	
Mitochondrial DNA depletion syndrome-4A (MTDPS4A) [Alpers syndrome]	No author assigned	203700	
Maturity Onset Diabetes of the Young (MODY)	Completed	125850 125851 137920 600496 606391 606392 606394 609812 610508 612225 613370 613375	
Mowat-Wilson syndrome	Completed	235730	2011
Moyamoya disease	No author assigned	607151	
Mucolipidosis II	No author assigned	252500	
Mucolipidosis III	No author assigned	252600 252605	
Mucopolysaccharidosis Type I	No author assigned	607014 607015 607016	

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Mucopolysaccharidosis TYPE II, M. Hunter - MPS II	Completed	309900	2011
Mucopolysaccharidosis TYPE III	No author assigned	252920	
Mucopolysaccharidosis TYPE VI - MPS VI	No author assigned	253200	
Muenke syndrome	No author assigned	602849	
Multi-minicore disease	Completed	117000 255320 602771	2011
Multiple endocrine neoplasia type 1	In progress	131100	
Multiple endocrine neoplasia type 2	Completed	162300 171400	2011
Multiple exostoses (type 1 and 2)	In progress	133700	
Multiple myeloma	No author assigned	254500	
Multiple exostoses	No author assigned	133700 133701	
MUTYH-associated polyposis, autosomal recessive colorectal adenomatous polyposis	Completed	608456	2010
MUTYH-associated polyposis, autosomal recessive colorectal adenomatous polyposis - UPDATE 2012	Completed		2012
Myasthenia gravis	No author assigned	254200	
Myelofibrosis	No author assigned	254450	
Myoclonic epilepsy of Lafora	No author assigned	254780	
Naegeli-Franceschetti-Jadassohn syndrome (NFJS)	No author assigned	161000	
Nail-Patella Syndrome	In progress	161200	
Narcolepsy (including cataplexy)	No author assigned	161400 614250	
NARP syndrome (neuropathy, ataxia and retinitis pigmentosa)	No author assigned	551500	
Naxos disease: Cardiocutaneous syndrome due to cell adhesion defect	No author assigned	601214	
Nemaline myopathy	Completed	161800 256030 605355 609284 609285 610687	2012
Nemaline myopathy – update 2015	Completed		2015
Nephronophthisis	In progress	256100 602088 604387 606966 611498 613550	
Netherton syndrome	No author assigned	256500	
Neural tube defects (including spina bifida)	No author assigned	182940	
Neurofibromatosis type 1	No author assigned	162200 162210	
Neurofibromatosis type 2	In progress	101000	

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Neuronal Ceroid-Lipofuscinoses (1-10)	No author assigned	256730 204500 204200 256731 601780 610951 600143 610127	
Niemann-Pick disease	No author assigned	257200 257220 607616 607625	
Nijmegen Breakage Syndrome	In progress	251260	
Nonaka myopathy	No author assigned	605820	
Non-Hodgkin lymphoma	No author assigned	605027	
Non-Syndromic Microphthalmia Including Next-Generation Sequencing-Based Approaches	Completed		
Nonsyndromic trigonocephaly	No author assigned	190440	
Noonan syndrome	In progress	163950 609942 610733 611553 613224	
Ocular albinism	No author assigned	300500	
Oculocutaneous albinism	Completed	203100 203200 203290 606574 606952	2013
Oculopharyngeal muscular dystrophy (OPMD)	No author assigned	164300	
Ollier disease / Enchondromatosis	No author assigned	166000	
Opitz GBBB syndrome	No author assigned	145410	
Optic atrophy	No author assigned	165500 165300	
Ornithine transcarbamylase deficiency	No author assigned	311250	
Orofaciodigital syndrome I	No author assigned	311200	
Osler-Rendu-Weber disease	Re-edit required	187300	2008
Osteochondritis dissecans	No author assigned	165800	
Osteogenesis Imperfecta	Completed	166200 166210 166220 259420 259440 610682 610854 610915 610968	
Osteosarcoma (Osteogenic sarcoma)	No author assigned	259500	

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Otopalatodigital syndromes (type 1-4)	No author assigned	311300 304120	
Ovarian dysgenesis	No author assigned	233300	
Pachyonychia Congenita	In progress	167200 167210	
Pallister-Hall and Greig Cephalopolysyndactyly Syndromes	In progress	146510 175700	
Pallister-Killian syndrome (PKS)	No author assigned	601803	
Papillon-Lefevre syndrome (PALS)	No author assigned	245000	
Papillo-renal syndrome	Completed	120330	2011
Parietal foramina	In progress	168500	
Paroxysmal nocturnal hemoglobinuria	In progress	300818	
Pelizaeus-Merzbacher disease (PMD)	No author assigned	312080	
Pendred Syndrome/DFNB4	No author assigned	274600	
Peters Plus syndrome	Completed	261540	
Peutz-Jeghers syndrome	No author assigned	175200	
Pfeiffer syndrome	No author assigned	101600	
Phenylketonuria - PKU	Completed	261600	2011
Pheochromocytomas and secreting paragangliomas	No author assigned	115310 168000 171300 601650 605373	
Phosphomannomutase 2 defciency	Completed	212065	2013
Phosphomannose isomerase deficiency	Completed	602579	2013
Piebaldism	No author assigned	172800	
Plasminogen deficiency type I	No author assigned	217090	
Pleuropulmonary blastoma (PPB)	In progress	601200	
Poikiloderma with Neutropenia	Completed	604173	
Poikiloderma with tendon contractures, myopathy, and pulmonary fibrosis (POIKTMP)	In progress	615704	
Polycythemia vera	Completed	263300	2012
Pontocerebellar Hypoplasia Type 2 and Type 4	In progress	277470 612389 612390 225753	
Popliteal pterygium syndrome (PPS)	No author assigned	119500	
Porphyria cutanea tarda	No author assigned	176100	
Prader-Willi syndrome	Completed	176270	2014
Primary ciliary dyskinesia	No author assigned	244400 608644 610852 611884 612444 612518 612649 612650 613193	

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Primary congenital glaucoma	In progress		
Primary pulmonary hypertension	No author assigned	178600	
Progressive external ophthalmoplegia	No author assigned	613077 157640 258450 609283 609286 610131	
Progressive familial intrahepatic cholestasis type 1	Completed	602397	2013
Progressive familial intrahepatic cholestasis type 2	Completed	601847	2013
Progressive familial intrahepatic cholestasis type 3	Completed	602347	2013
Propionic acidemia	No author assigned	606054	
Proteus Syndrome	In progress	176920	
Proximal spinal muscular atrophy - SMA	Completed	313200 604320 271150 253550 301830 253400 253300 600175 300489	2012
Proximal spinal muscular atrophy – update 2015	Completed	313200 604320 271150 253550 301830 253400 253300 600175 300489	2015
Pseudoachondroplasia (PSACH)	No author assigned	177170	
Pseudohypoparathyroidism type Ia (PHP Ia) [Albright hereditary osteodystrophy]	Completed	103580	
Pseudoxanthoma Elasticum	In progress	177850 264800	
Pyridoxine-dependent epilepsy (EPD)	No author assigned	266100	
Pyruvate Carboxylase Deficiency	No author assigned	266150	
Rapid-Onset Dystonia Parkinsonism	No author assigned	128235	
Red cell pyruvate kinase (PK) deficiency	No author assigned	266200	
Refsum disease	No author assigned	266500	
Renal adysplasia	No author assigned	191830	
Renal cysts and diabetes syndrome	In progress	137920	
Renal tubular dysgenesis	In progress		
Retinitis pigmentosa	No author assigned	180100 180104 180105	

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		268000 300029 300455 312600 500004 600059 600105 600132 600138 600852 601414 601718 602772 604232 604393 606068 607921 608133 608380 609923 610282 610359 610599 611131 612095 612572 612943 613194 613341 613428 613464 613575 613581 613582 613617	
Retinoblastoma	Completed	180200	2010
Rett Syndrome	In progress	312750	
Rh Deficiency syndrome (RH-NULL, REGULATOR TYPE; RHN)	No author assigned	268150	
Rhizomelic chondrodysplasia punctata	No author assigned	215100	
Roberts syndrome and SC phocomelia syndrome	No author assigned	268300 269000	
Rothmund-Thomson syndrome	In progress	268400	
Rubinstein-Taybi Syndrome	Completed	180849 610543	
Russell-Silver syndrome	Completed	180860	2010
Sacral defect with anterior meningocele (SDAM)	No author assigned	600145	
Saethre-Chotzen syndrome	No author assigned	101400	
Sandhoff disease	No author assigned	268800	
Sarcosinemia	No author assigned	268900	
Schizencephaly	No author assigned	269160	

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Senior Loken Syndrome	No author assigned	266900 606996 609254 610189	
Shwachman-Diamond Syndrome	No author assigned	260400	
Sickle cell anemia	No author assigned	603903	
Sitosterolaemia	Completed		
Sjogren-Larsson syndrome	No author assigned	270200	
Smith-Lemli-Opitz Syndrome	Completed	270400	
Smith-Magenis Syndrome	In progress	182290	
Sotos Syndrome	In progress	117550	
Spinal muscular atrophy with respiratory distress - SMARD	In progress	604320	
Spinocerebellar ataxia - SCA - Spinocerebelläre Ataxie (Typ 1 bis 17)	No author assigned	183090 164400 117210 606002 606658 610743 164500 609307 612016 606937 183086 603516 607136 605361 600224 605259 608768 610245 604326 610246 607250 604432	
Split-hand/foot malformation	No author assigned	183600 246560 225300	
Squamous cell carcinoma of the head and neck (HNSCC)	No author assigned	275355	
Stargardt disease	No author assigned	248200 600110 603786	
Stickler Syndrome	No author assigned	108300 184840 604841 609508	
Succinic semialdehyde dehydrogenase, 4-hydroxybutyricaciduria	No author assigned	271980	
Syndromic microphthalmia	No author assigned		
Systemic juvenile rheumatoid arthritis	No author assigned	604302	

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Tay-Sachs disease	No author assigned	272800	
Tetrahydrobiopterin (BH4)-deficient hyperphenylalaninemia, 6-pyruvoyl-tetrahydropterin synthase deficiency	No author assigned	261640 233910 264070 261630	
Tetralogy of Fallot (TOF)	No author assigned	187500	
Thrombotic thrombocytopenic purpura (TTP)	No author assigned	274150	
Tibial muscular dystrophy (TMD)	No author assigned	600334	
Timothy syndrome	In progress	601005	
Torsion dystonia	No author assigned	128100	
Townes-Brocks syndrome	No author assigned	107480	
Transient neonatal diabetes mellitus	Completed	601410	2013
Treacher Collins Syndrome	No author assigned	154500	
Trigonocephaly (metopic craniosynostosis)	No author assigned	190440 614485	
Trimethylaminuria	Completed	602079	2011
Trimethylaminuria - update 2014	Completed		2014
Tritanopia	No author assigned	190900	
Troyer syndrome (Autosomal recessive spastic paraplegia type 20)	No author assigned	275900	
Tuberous Sclerosis Complex	Completed	191100 613254	
UMOD-related kidney disease	In progress	162000	
Unverricht-Lundborg Disease	No author assigned	254800	
Usher Syndrome	Completed	276900	2011
Usher Syndrome - update 2013	In progress	276900	2013
VACTERL (VATER) association	No author assigned	192350	
van der Woude syndrome	No author assigned	119300	
Vici syndrome	Completed		2013
von Hippel-Lindau syndrome	Completed	193300	2013
Von Willebrand disease	Completed	193400	2011
Waardenburg syndrome	No author assigned	277580 148820 193510 611584 193500 613266 613265 608890	
WAGR Syndrome	Completed	194072	2011
Waldenström macroglobulinemia	No author assigned	153600 610430	
Walker-Warburg syndrome	No author assigned	613150	
Weill-Marchesani syndrome	No author assigned	277600 608328	
Werner syndrome	Completed	277700	2012
Williams-Beuren syndrome - WBS	Completed	194050	2013

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Wilson disease	No author assigned	277900	
Wiskott-Aldrich syndrome	No author assigned	301000	
Wolf-Hirschhorn (4p-) Syndrome	Completed	194190	2010
Wolfram syndrome	Completed	222300 604928 598500	
Xeroderma Pigmentosum	Completed	194400 278700 278720 278730 278740 278750 278760 278780 278810 610651	2013
X-Linked Agammaglobulinemia	No author assigned	300755	
X-linked chronic granulomatous disease (CGD)	No author assigned	306400	
X-linked dominant chondrodysplasia punctata	No author assigned	302960	
X-linked hypohidrotic ectodermal dysplasia	No author assigned	305100	
X-linked Ichthyosis	In progress	308100	
X-linked nephrogenic diabetes insipidus (NDI)	No author assigned	304800	
X-linked retinoschisis	No author assigned	312700	
Zellweger syndrome	Completed	214100	2014

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