

CODE	PATHOLOGY	Omim	Individuals	Family	Affecteds
ACC	CONGENITA, CEREBELLAR ATAXIA		15	5	8
ACCPN	AGENESIS OF THE CORPUS CALLOSUM WITH PERIPHERAL NEUROPATHY	218000	2	1	2
ACV	CEREBELLAR VERMIS APLASIA	117360	15	2	3
ADCA	CEREBELLAR ATAXIA	117210	29	12	22
ADCA2	CEREBELLAR ATAXIA 2	302500	14	4	5
ADPE	LATERAL TEMPORAL LOBE EPILEPSY	600513	65	12	29
AE	CHILDHOOD AND JUVENILE ABSENCE EPILEPSY	607631	312	63	141
AGS	AICARDI-GOUTIERES SYNDROME	225750	148	38	53
ALD	ADRENOLEUKODYSTROPHY	300100	14	10	10
ALZ	ALZHEIMER DISEASE	104300	9	6	6
AOA	EARLY-ONSET, WITH OCULOMOTOR APRAXIA AND HYPOALBEMINURIA	208920	6	2	2
AOPC	OLIVOPONTOCEREBELLAR ATROPHY	164400	59	14	19
APS	SPINAL MUSCULAR ATROPHY, PROXIMAL	182980	60	56	5
ARCA	AUTOSOMIQUE RECESSIVE CEREBELLAR ATAXIA		216	60	98
ARSAC	SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY	270550	12	2	5
AT	ATAXIA TELANGIECTASIA	208900	426	300	306
AVED	ATAXIA, WITH SELECTIVE VITAMIN E DEFICIENCY	277460	7	1	3
B	TUBEROUS SCLEROSIS	191100	213	64	79
BFIC	BENIGN FAMILIAL INFANTILE CONVULSIONS	601764	158	21	70
BFNC	BENIGN FAMILIAL NEONATAL EPILEPSY	121200	167	24	78
BOE	BENIGN OCCIPITAL EPILEPSY	132090	13	2	6
BPE	PARTIAL SIMPLE EPILEPSY		24	7	13
BRE	BENIGN ROLANDIC EPILEPSY	117100	14	4	7
C	CONVULSIVE DISORDERS	217200	4	1	0
CDL	CEROID LIPOFUSCINOSIS NEURONAL INFANTILE	256730	9	3	3
CGS	SOTOS SYNDROME	117550	13	6	7
CLS	COFFIN-LOWRY SYNDROME	303600	64	52	55
COH1	COHEN SYNDROME	216550	5	3	2
CPD	JOUBERT SYNDROME	213300	18	4	6
D	DYSTONIA	128100	17	8	8
DAX	NEUROAXONAL DYSTROPHY	256600	5	3	4
DOOS	DOOSE SYNDROME		71	23	23
DPH	DYSPHASIA	600117	14	4	13
DWS	DANDY-WALKER SYNDROME	220200	2	2	2
DY	DYSAUTONOMIA FAMILIAL	223900	1	1	1
DYS	FAMILIAL PAROXYSMAL DYSTONIA	128230	19	9	5
DYT12	DYSTONIA 12	128235	3	1	1
EA1	EPISODIC ATAXIA 1	160120	10	8	8
ERM	EPILEPSY, WITH MENTAL RETARDATION		11	3	5
ESC	ESCOBAR SYNDROME	265000	13	3	5
FC	FEBRILE SEIZURES	121210	799	109	391
FLD	PROGRESSIVE BULBAR PARALISY OF CHILDHOOD	211500	1	1	1
FRAX	FRAGIL X SYNDROME	309550	11	7	5
FRDA	FRIEDREICH ATAXIA	229300	150	44	52
GEFS+	GENERALIZED EPILEPSY WITH FEBRILES SEIZURES PLUS	604233	2	2	1
GTS	TOURETTE SYNDROME	137580	3	1	1
HA	ALTERNATING HEMIPLEGIA OF CHILDHOOD	104290	119	36	37
HD	HUNTINGTON DISEASE	143100	20	16	17
HFE	FAMILIAL EPILEPSY HETEROGENOUS		85	16	33
HHE	HEMIPARESIS, HEMIANOPSIA, EPILEPSY		1	1	1
HLSC	DOUBLECORTINE	300121	90	20	21

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HMSN	PERIPHERAL NEUROPATHY	601152	15	8	9
HNPP	NEUROPATHY, HEREDITARY, WITH LIABILITY TO PRESSURE PALSIES	162500	7	6	6
HSP	HEREDITARY SPASTIC PARAPLEGIAS	2601/6072	1302	246	537
IGEGM	EPILEPSY IDIOPATHIC GENERALIZED	600669	165	34	74
IGEHE	EPILEPSY IDIOPATHIC GENERALIZED HETEROGENOUS		116	19	53
JME	JUVENILE MYOCLONIC EPILEPSY	606904	513	92	200
LDM	METACHROMATIC LEUKODYSTROPHY	250100	12	5	5
LEC	LEUCOENCEPHALOPATHY WITH BILATERAL ANTERIOR TEMPORAL LOBE CYSTS		15	5	7
LFR	MYOCLONIC EPILEPSY OF LAFORA	254780	60	17	18
LS	LEIGH SYNDROME	256000	6	3	3
MAR	MARDEN-WALKER SYNDROME	248700	2	1	2
MCT	MICROCEPHALY PRIMARY	251200	4	4	4
MK	MENKES DISEASE	309400	4	2	2
MLC	NCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICA	604004	127	27	30
MS	SUSCEPTIBILITY TO MULTIPLE SCLEROSIS	126200	236	38	88
MSS	MARINESCO-SJOGREN SYNDROME	248800	19	4	9
NCL	JANSKY BIELCHOWSKY DISEASE	204500	22	5	5
NF1	NEUROFIBROMATOSIS TYPE 1	162200	23	16	15
NF2	NEUROFIBROMIN	607379	1	1	1
PARK	PARKINSON DISEASE	168600	47	24	26
PARK+	PARKINSON WITH ATROPHY AND PARALYSIS		468	129	395
PDC	PYRIDOXINE DEPENDENT EPILEPSY	266100	29	7	13
PEHO	PEHO SYNDROME	260565	2	1	1
PHE	PHOTOSENSITIVE EPILEPSY	132100	25	4	11
PKAN	PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION	234200	5	1	3
PLP	PELIZAEUS-MERZBACHE DISEASE	312080	8	3	4
PME	PROGRESSIVE MYOCLONIC EPILEPSY	254800	71	23	36
PNKD1	PAROXYSMAL NONKINESIGENIC DYSKINESIA 1	118800	2	2	2
RDPM	RETARDATION OF DEVELOPMENT PSYCHOMOTEUR		39	21	22
RES	REFSUM DISEASE	266500	8	2	3
RETT	RETT SYNDROME	312750	441	115	112
RM	NON SUNDROMIC MENTAL RETARDATION		59	29	35
RMX	MENTAL RETARDATION X LINKED		6	3	3
RTB	RUBINSTEIN-TAYBI SYNDROME	180849	45	40	38
S	SCHIZOPHRENIA		84	35	36
SCA	SPINOCEREBELLAR ATAXIA	164400	21	6	10
SEP	MULTIPLE SCLEROSIS	126200	5535	2944	3191
SIN	SITUS INVERSUS VISCERUM	270100	3	1	1
SJS1	SCHWARTZ-JAMPEL SYNDROME	255800	28	12	14
SLA	AMYOTROPHIC LATERAL SCLEROSIS	105400	3442	1019	2201
SME	SEVERE MYOCLONIC EPILEPSY OF INFANCY	607208	361	109	130
STW	STUVE WIEDEMANN SYNDROME	601559	1	1	1
TRB	TREMOR HEREDITARY ESSENTIAL	190300	206	63	126
TSD	TAY-SACHS DISEASE	272800	10	2	2
TTR	TRANSTHYRETIN	176300	1	1	1
UE	UNDEFINED EPILEPSY		76	32	47
WAS	WISKOTT-ALDRICH SYNDROME	3011000	27	24	25
WEST	INFANTILE SPASM SYNDROME	308350	20	7	9