

Gene panel information

Gene panel	Stroke
Version	1
Total genes	102
Activation date	Monday 29 april 2019
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ABCA1	99.79 %	600046	Tangier disease, 205400 (3), Autosomal recessive; HDL deficiency, familial, 1, 604091 (3), Autosomal dominant
ABCC6	96.83 %	603234	Pseudoxanthoma elasticum, 264800 (3), Autosomal recessive; Arterial calcification, generalized, of infancy, 2, 614473 (3), Autosomal recessive; Pseudoxanthoma elasticum, forme fruste, 177850 (3), Autosomal dominant
ACAD9	100 %	611103	Mitochondrial complex I deficiency, nuclear type 20, 611126 (3), Autosomal recessive
ACP5	99.91 %	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
ACTA2	99.81 %	102620	Smooth muscle dysfunction syndrome, 613834 (3), Autosomal dominant; Aortic aneurysm, familial thoracic 6, 611788 (3), Autosomal dominant; Moyamoya disease 5, 614042 (3)
ACVRL1	99.38 %	601284	Telangiectasia, hereditary hemorrhagic, type 2, 600376 (3), Autosomal dominant
ADA2	99.97 %	607575	Sneddon syndrome, 182410 (3), Autosomal recessive; Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688 (3), Autosomal recessive
APP	99.44 %	104760	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 (3), Autosomal dominant; Alzheimer disease 1, familial, 104300 (3), Autosomal dominant
ASS1	99.83 %	603470	Citrullinemia, 215700 (3), Autosomal recessive
ATP7A	99.79 %	300011	Occipital horn syndrome, 304150 (3), X-linked recessive; Neuronopathy, distal hereditary motor, X-linked, 300489 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive
C1R	99.9 %	613785	Ehlers-Danlos syndrome, periodontal type, 1, 130080 (3), Autosomal dominant
CACNA1A	99.72 %	601011	Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Developmental and epileptic encephalopathy 42, 617106 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant
CBS	16.71 %	613381	Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive; Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive
CCM2	99.46 %	607929	Cerebral cavernous malformations-2, 603284 (3), Autosomal dominant
CD59	99.85 %	107271	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 (3), Autosomal recessive
COG6	99.89 %	606977	Shaheen syndrome, 615328 (3), Autosomal recessive; Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
COL1A1	99.22 %	120150	Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Caffey disease, 114000 (3), Autosomal dominant; Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 (3), Autosomal dominant; Osteogenesis imperfecta, type I, 166200 (3), Autosomal dominant; {Bone mineral density variation QTL, osteoporosis}, 166710 (3), Autosomal dominant; Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant
COL3A1	99.79 %	120180	Ehlers-Danlos syndrome, vascular type, 130050 (3), Autosomal dominant; Polymicrogyria with or without vascular-type EDS, 618343 (3), Autosomal recessive
COL4A1	99.93 %	120130	?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant
COL4A2	99.69 %	120090	Brain small vessel disease 2, 614483 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3)
COL5A1	99.82 %	120215	Ehlers-Danlos syndrome, classic type, 1, 130000 (3), Autosomal dominant; Fibromuscular dysplasia, multifocal, 619329 (3), Autosomal dominant
COL5A2	99.72 %	120190	Ehlers-Danlos syndrome, classic type, 2, 130010 (3), Autosomal dominant
COQ8A	99.89 %	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
CPS1	99.76 %	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive
CST3	99.77 %	604312	{Macular degeneration, age-related, 11}, 611953 (3); Leukodystrophy, adult-onset, autosomal dominant, without amyloid angiopathy, 621214 (3), Autosomal dominant; Cerebral amyloid angiopathy, 105150 (3), Autosomal dominant
CTSA	99.77 %	613111	Galactosialidosis, 256540 (3), Autosomal recessive; Brain small vessel disease 6 with leukoencephalopathy, 621394 (3), Autosomal dominant
DYRK1B	99.76 %	604556	Abdominal obesity-metabolic syndrome 3, 615812 (3), Autosomal dominant
EFEMP2	99.7 %	604633	Cutis laxa, autosomal recessive, type IB, 614437 (3), Autosomal recessive
ENG	99.82 %	131195	Telangiectasia, hereditary hemorrhagic, type 1, 187300 (3), Autosomal dominant
ENPP1	99.81 %	173335	{Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial; Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant
ESCO2	99.95 %	609353	Juberg-Hayward syndrome, 216100 (3), Autosomal recessive; Roberts-SC phocomelia syndrome, 268300 (3), Autosomal recessive
F10	99.8 %	613872	Factor X deficiency, 227600 (3), Autosomal recessive
F13A1	99.56 %	134570	Factor XIIIa deficiency, 613225 (3), Autosomal recessive; {Myocardial infarction, protection against}, 608446 (3); {Venous thrombosis, protection against}, 188050 (3), Autosomal dominant
F2	99.82 %	176930	Hypoprothrombinemia, 613679 (3), Autosomal recessive; {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 (3), Autosomal dominant; Dysprothrombinemia, 613679 (3), Autosomal recessive; Thrombophilia 1 due to thrombin defect, 188050 (3), Autosomal dominant; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
F5	99.31 %	612309	Thrombophilia 2 due to activated protein C resistance, 188055 (3), Autosomal dominant; {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 (3), Autosomal dominant; {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055 (3), Autosomal dominant; {Budd-Chiari syndrome}, 600880 (3), Autosomal recessive; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial; Factor V deficiency, 227400 (3), Autosomal recessive
F7	99.98 %	613878	{Myocardial infarction, decreased susceptibility to}, 608446 (3); Factor VII deficiency, 227500 (3), Autosomal recessive
F8A1	11.3 %	305423	No OMIM phenotypes
FBN1	99.73 %	134797	Geleophysic dysplasia 2, 614185 (3), Autosomal dominant; Weill-Marchesani syndrome 2, dominant, 608328 (3), Autosomal dominant; Ectopia lentis, familial, 129600 (3), Autosomal dominant; MASS syndrome, 604308 (3), Autosomal dominant; Marfan lipodystrophy syndrome, 616914 (3), Autosomal dominant; Acromicric dysplasia, 102370 (3), Autosomal dominant; Marfan syndrome, 154700 (3), Autosomal dominant; Stiff skin syndrome, 184900 (3), Autosomal dominant
FGA	99.96 %	134820	Amyloidosis, hereditary systemic 2, 105200 (3), Autosomal dominant; Hypodysfibrinogenemia, congenital, 616004 (3), Autosomal dominant; Dysfibrinogenemia, congenital, 616004 (3), Autosomal dominant; Afibrinogenemia, congenital, 202400 (3), Autosomal recessive
FGB	99.88 %	134830	Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive; Dysfibrinogenemia, congenital, 616004 (3), Autosomal dominant; Afibrinogenemia, congenital, 202400 (3), Autosomal recessive
FGG	99.89 %	134850	Dysfibrinogenemia, congenital, 616004 (3), Autosomal dominant; Hypodysfibrinogenemia, 616004 (3), Autosomal dominant; Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive; Afibrinogenemia, congenital, 202400 (3), Autosomal recessive
FOXC1	99.97 %	601090	Axenfeld-Rieger syndrome, type 3, 602482 (3), Autosomal dominant; Anterior segment dysgenesis 3, multiple subtypes, 601631 (3), Autosomal dominant
GAA	99.83 %	606800	Pompe disease, late-onset, 621314 (3), Autosomal recessive; Pompe disease, infantile-onset, 232300 (3), Autosomal recessive
GATA3	99.81 %	131320	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255 (3), Autosomal dominant
GCDH	100 %	608801	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
GGCX	99.83 %	137167	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 (3), Autosomal recessive; Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 (3)
GLA	99.85 %	300644	Fabry disease, cardiac variant, 301500 (3), X-linked; Fabry disease, 301500 (3), X-linked
GUCY1A1	99.94 %	139396	Moyamoya 6 with achalasia, 615750 (3), Autosomal recessive
HBB	100 %	141900	Methemoglobinemia, beta type, 617971 (3), Autosomal dominant; Thalassemia-beta, dominant inclusion-body, 603902 (3), Autosomal dominant; Sickle cell disease, 603903 (3), Autosomal recessive; Thalassemia, beta, 613985 (3); Delta-beta thalassemia, 141749 (3), Autosomal dominant; {Malaria, resistance to}, 611162 (3); Hereditary persistence of fetal hemoglobin, 141749 (3), Autosomal dominant; Erythrocytosis, familial, 6, 617980 (3), Autosomal dominant; Heinz body anemia, 140700 (3), Autosomal dominant
HSD11B2	99.86 %	614232	Apparent mineralocorticoid excess, 218030 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
HTRA1	99.75 %	602194	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy 2, 616779 (3), Autosomal dominant; Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy 2, 600142 (3), Autosomal recessive
ITM2B	99.56 %	603904	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 (3), Autosomal dominant; Dementia, familial British, 176500 (3), Autosomal dominant; Dementia, familial Danish, 117300 (3), Autosomal dominant
IVD	99.86 %	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive
JAG1	99.94 %	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
JAK2	99.41 %	147796	{Budd-Chiari syndrome, somatic}, 600880 (3); Myelofibrosis, somatic, 254450 (3); Erythrocytosis, somatic, 133100 (3); Leukemia, acute myeloid, somatic, 601626 (3); Thrombocythemia 3, 614521 (3), Somatic mutation, Autosomal dominant; Polycythemia vera, somatic, 263300 (3)
JAM3	99.94 %	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
KNG1	99.87 %	612358	[Kininogen deficiency], 228960 (3), Autosomal recessive; Angioedema, hereditary, 6, 619363 (3), Autosomal dominant; [High molecular weight kininogen deficiency], 228960 (3), Autosomal recessive
KRIT1	99.03 %	604214	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860 (3), Autosomal dominant; Cerebral cavernous malformations-1, 116860 (3), Autosomal dominant; Cavernous malformations of CNS and retina, 116860 (3), Autosomal dominant
LMNA	99.84 %	150330	Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Restrictive dermopathy 2, 619793 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant
MFAP5	99.7 %	601103	Aortic aneurysm, familial thoracic 9, 616166 (3), Autosomal dominant
MFN2	99.82 %	608507	Lipomatosis, multiple symmetric, with or without peripheral neuropathy, 151800 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 (3), Autosomal recessive; Hereditary motor and sensory neuropathy VIA, 601152 (3), Autosomal dominant
MMACHC	99.9 %	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
MMUT	99.71 %	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
MTHFR	99.8 %	607093	{Vascular disease, susceptibility to} (3); Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive
MYH11	99.23 %	160745	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351 (3), Autosomal recessive; Aortic aneurysm, familial thoracic 4, 132900 (3), Autosomal dominant; Visceral myopathy 2, 619350 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NF1	99.83 %	613113	Watson syndrome, 193520 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Somatic mutation, Autosomal dominant; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant
NOTCH3	99.56 %	600276	Lateral meningocele syndrome, 130720 (3), Autosomal dominant; Lipodystrophy, familial partial, type 1, 608600 (3), Autosomal dominant; Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy 1, 621295 (3), Autosomal recessive; ?Myofibromatosis, infantile 2, 615293 (3), Autosomal dominant; Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy 1, 125310 (3), Autosomal recessive, Autosomal dominant
OTC	99.67 %	300461	Ornithine transcarbamylase deficiency, 311250 (3), X-linked
PCCA	99.91 %	232000	Propionicacidemia, 606054 (3), Autosomal recessive
PCCB	99.86 %	232050	Propionicacidemia, 606054 (3), Autosomal recessive
PCNT	99.91 %	605925	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive
PDCD10	99.52 %	609118	Cerebral cavernous malformations-3, 603285 (3), Autosomal dominant
PDE3A	99.9 %	123805	Hypertension and brachydactyly syndrome, 112410 (3), Autosomal dominant
PKD1	99.48 %	601313	Polycystic kidney disease 1, 173900 (3), Autosomal dominant
PLG	99.61 %	173350	Dysplasminogenemia, 217090 (3), Autosomal recessive; Angioedema, hereditary, 4, 619360 (3), Autosomal dominant; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive
PLOD1	98.95 %	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400 (3), Autosomal recessive
PLOD3	98.22 %	603066	BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394 (3), Autosomal recessive
PROC	99.58 %	612283	Thrombophilia 3 due to protein C deficiency, autosomal dominant, 176860 (3), Autosomal dominant; Thrombophilia 3 due to protein C deficiency, autosomal recessive, 612304 (3), Autosomal recessive
PROS1	99.58 %	176880	Thrombophilia 5 due to protein S deficiency, autosomal recessive, 614514 (3), Autosomal recessive; Thrombophilia 5 due to protein S deficiency, autosomal dominant, 612336 (3), Autosomal dominant
PTPN11	99.84 %	176876	Noonan syndrome 1, 163950 (3), Autosomal dominant; LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
RASA1	99.23 %	139150	Capillary malformation-arteriovenous malformation 1, 608354 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3)
SAMHD1	99.94 %	606754	?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive
SERPINE1	99.39 %	173360	Plasminogen activator inhibitor-1 deficiency, 613329 (3), Autosomal recessive, Autosomal dominant; {Transcription of plasminogen activator inhibitor, modulator of} (3)
SLC19A2	98.59 %	603941	Thiamine-responsive megaloblastic anemia syndrome, 249270 (3), Autosomal recessive
SLC2A10	99.96 %	606145	Arterial tortuosity syndrome, 208050 (3), Autosomal recessive
SMAD3	99.95 %	603109	Loeys-Dietz syndrome 3, 613795 (3), Autosomal dominant
SMAD4	99.95 %	600993	Pancreatic cancer, somatic, 260350 (3); Myhre syndrome, 139210 (3), Autosomal dominant; Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant
SMARCA1	99.78 %	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SPARC	98.85 %	182120	Osteogenesis imperfecta, type XVII, 616507 (3), Autosomal recessive
STAT1	99.66 %	600555	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162 (3), Autosomal dominant; Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 (3), Autosomal dominant; Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 (3), Autosomal recessive
STIM1	99.89 %	605921	Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant; Immunodeficiency 10, 612783 (3), Autosomal recessive
TGFB2	99.85 %	190220	Loeys-Dietz syndrome 4, 614816 (3), Autosomal dominant; Camurati-Engelmann disease 2, 606631 (3), Autosomal dominant
TGFB3	99.97 %	190230	Arrhythmogenic right ventricular dysplasia 1, 107970 (3), Autosomal dominant; Loeys-Dietz syndrome 5, 615582 (3), Autosomal dominant
TGFBR1	99.84 %	190181	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant; Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant
TGFBR2	99.91 %	190182	Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3)
THBD	99.91 %	188040	Thrombophilia 12 due to thrombomodulin defect, 614486 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 (3), Autosomal dominant
TREX1	100 %	606609	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant
TSC1	99.91 %	605284	Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Lymphangioleiomyomatosis, 606690 (3)
TSC2	99.88 %	191092	Lymphangioleiomyomatosis, somatic, 606690 (3); ?Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant
TTR	99.8 %	176300	Amyloidosis, hereditary systemic 1, 105210 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant
VHL	99.98 %	608537	Hemangioblastoma, cerebellar, somatic (3); Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); Pheochromocytoma, 171300 (3), Autosomal dominant
YY1AP1	99.56 %	607860	Grange syndrome, 602531 (3), Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2025-11-12**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

- (1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
- (2) the disorder has been placed on the map by linkage; no mutation has been found
- (3) the molecular basis for the disorder is known; a mutation has been found in the gene
- (4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.