Supporing Information Table 3. **Network characteristics of disease genes.**
Class of disease gene is assigned if all the implicated diseases are of same class, otherwise, it is assigned "Grey."

Entrez ID	Symbol	Disorder class	Size (s)	Degree (k)	Number of classes associated	Implicated diseases (Disease ID) [comma-deliminated]
2	A2M	Neurological	1	11	1	Alzheimer_disease (98)
10	NAT2	Metabolic	1	0	1	Slow_acetylation (1403)
12	SERPINA3	Grey	2	0	1	Alpha-1-antichymotrypsin_deficiency (87), Cerebrovascular_disease,_occlusive (294)
15	AANAT	Psychiatric	1	0	1	Delayed_sleep_phase_syndrome (411)
18	ABAT	Metabolic	1	0	1	GABA-transaminase_deficiency (564)
19	ABCA1	Grey	4	7	3	Cerebral_amyloid_angiopathy (291), Coronary_artery_disease (365), HDL_cholesterol_level_QTL (644), Tangier_disease (1473)
21	ABCA3	Respiratory	1	1	1	Surfactant_deficiency (1464)
22	ABCB7	Hematological	1	8	1	Anemia (110)
24	ABCA4	Ophthamological	5	36	1	Cone_dystrophy (352), Fundus_albipunctatus (562), Macular_dystrophy (2937), Retinitis_pigmentosa (1316), Stargardt_disease (1439)
25	ABL1	Cancer	1	36	1	Leukemia (886)
28	ABO	Hematological	1	22	1	Blood_group (212)
33	ACADL	Metabolic	1	2	1	Acyl-CoA_dehydrogenase,_deficiency_of (44)
34	ACADM	Metabolic	1	2	1	Acyl-CoA_dehydrogenase,_deficiency_of (44)
35	ACADS	Metabolic	1	2	1	Acyl-CoA_dehydrogenase,_deficiency_of (44)
36	ACADSB	Metabolic	1	0	1	2-methylbutyrylglycinuria (4)
37	ACADVL	Metabolic	1	0	1	VLCAD_deficiency (1570)
38	ACAT1	Metabolic	1	0	1	Alpha-methylacetoacetic_aciduria (90)
43	ACHE	Hematological	1	22	1	Blood_group (212)
58	ACTA1	Muscular	1	9	1	Myopathy (1059)
70	ACTC	Cardiovascular	1	24	1	Cardiomyopathy (268)
71	ACTG1	Ear,Nose,Throat	1	40	1	Deafness (406)
81	ACTN4	Renal	1	2	1	Glomerulosclerosis (596)
89	ACTN3	Muscular	2	0	1	Alpha-actinin-3_deficiency (88), Elite_sprint_athletic_performance (471)
91	ACVR1B	Cancer	1	8	1	Pancreatic_cancer (1178)
93	ACVR2B	Developmental	1	1	1	Left-right_axis_malformations (874)
94	ACVRL1	Cardiovascular	1	1	1	Hereditary_hemorrhagic_telangiectasia (669)
100	ADA	Immunological	2	7	1	Adenosine_deaminase_deficiency (49), Severe_combined_immunodeficiency (1383)
103	ADAR	Dermatological	1	0	1	Dyschromatosis (455)
118	ADD1	Cardiovascular	1	11	1	Hypertension (752)
152	ADRA2C	Cardiovascular	1	1	1	Congestive_heart_failure (355)
153	ADRB1	Cardiovascular	2	1	1	Congestive_heart_failure (355), Resting_heart_rate (1312)
154	ADRB2	Grey	3	32	2	Asthma (153), Beta-2-adrenoreceptor_agonist,_reduced_response_to (198), Obesity (1126)
155	ADRB3	Nutritional	1	20	1	Obesity (1126)
158	ADSL	Metabolic	1	0	1	Adenylosuccinase_deficiency (50)
174	AFP	Hematological	1	0	1	Hereditary_persistence_of_fetal_hemoglobin (2669)
175	AGA	Metabolic	1	0	1	Aspartylglucosaminuria (150)
176	AGC1	Skeletal	1	4	1	Spondyloepiphyseal_dysplasia (1435)
178	AGL	Metabolic	1	8	1	Glycogen_storage_disease (610)
181	AGRP	Nutritional	2	20	1	Leanness,_inherited (872), Obesity (1126)
182	JAG1	Grey	3	42	3	Alagille_syndrome (71), Deafness (406), Tetralogy_of_Fallot (1483)
183	AGT	Grey	3	15	2	Hypertension (752), Preeclampsia (1257), Renal_tubular_dysgenesis (9308)
185	AGTR1	Grey	2	13	2	Hypertension (752), Renal_tubular_dysgenesis (9308)
186	AGTR2	Neurological	1	23	1	Mental_retardation (990)
189	AGXT	Metabolic	1	1	1	Hyperoxaluria (742)

190	NR0B1	Endocrine	2	3	1	Adrenal_hyperplasia,_congenital (53), Dosage-sensitive_sex_reversal (443)
191	AHCY	Metabolic	1	1	1	Hypermethioninemia (739)
203	AK1	Hematological	1	9	1	Hemolytic_anemia (660)
208	AKT2	Endocrine	1	26	1	Diabetes_mellitus (427)
210	ALAD	Metabolic	2	5	1	Lead_poisoning (871), Porphyria (1249)
212	ALAS2	Hematological	1	8	1	Anemia (110)
213	ALB	Hematological	2	0	1	Analbuminemia (105), Dysalbuminemic_hyperthyroxinemia (453)
215	ABCD1	Grey	2	5	2	Adrenoleukodystrophy (56), Adrenomyeloneuropathy (57)
217	ALDH2	Metabolic	1	0	1	Alcohol_intolerance (2074)
224	ALDH3A2	Metabolic	1	0	1	Sjogren-Larsson_syndrome (1400)
226	ALDOA	Metabolic	1	0	1	Aldolase_A_deficiency (76)
229	ALDOB	Metabolic	1	0	1	Fructose_intolerance (556)
240	ALOX5	Grey	2	12	2	Asthma (153), Atherosclerosis (163)
241	ALOX5AP	Cardiovascular	2	10	1	Myocardial_infarction (1054), Stroke (1454)
242	ALOX12B	Dermatological	1	2	1	Ichthyosiform_erythroderma (793)
249	ALPL	Grey	2	0	2	Hypophosphatasia (782), Odontohypophosphatasia (1136)
265	AMELX	Bone	1	3	1	Amelogenesis_imperfecta (99)
268	AMH	Developmental	1	1	1	Persistent_Mullerian_duct_syndrome (1212)
269	AMHR2	Developmental	1	1	1	Persistent_Mullerian_duct_syndrome (1212)
270	AMPD1	Muscular	1	0	1	Myoadenylate_deaminase_deficiency (1053)
272	AMPD3	Hematological	1	0	1	AMP_deaminase_deficiency (101)
275	AMT	Metabolic	1	3	1	Glycine_encephalopathy (608)
286	ANK1	Hematological	1	4	1	Spherocytosis (1423)
287	ANK2	Cardiovascular	1	6	1	Long_QT_syndrome (912)
291	SLC25A4	Ophthamological	1	2	1	Progressive_external_ophthalmoplegia_with_mitochondrial_DNA_deletions (1265)
323	APBB2	Neurological	1	11	1	Alzheimer_disease (98)
324	APC	Cancer	7	42	1	Adenoma,_periampullary (46), Adenomas (47), Colon_cancer (346), Desmoid_disease,_hereditary (425), Gardner_syndrome (571), Gastric_cancer (572), Turcot_syndrome (1536)
326	AIRE	Immunological	1	3	1	Autoimmune_disease (174)
335	APOA1	Grey	5	17	3	Amyloidosis (103), Apolipoprotein_deficiency (136), Corneal_dystrophy (362), Hypertriglyceridemia (757), Hypoalphalipoproteinemia (762)
336	APOA2	Metabolic	2	10	1	Apolipoprotein_deficiency (136), Hypercholesterolemia (724)
338	APOB	Metabolic	3	8	1	Abetalipoproteinemia (17), Hypercholesterolemia (724), Hypobetalipoproteinemia (763)
344	APOC2	Metabolic	1	1	1	Hyperlipoproteinemia (737)
345	APOC3	Metabolic	1	3	1	Apolipoprotein_deficiency (136)
348	APOE	Grey	4	20	4	Alzheimer_disease (98), Hyperlipoproteinemia (737), Myocardial_infarction (1054), Sea-blue_histiocyte_disease (1366)
350	APOH	Metabolic	1	3	1	Apolipoprotein_deficiency (136)
351	APP	Grey	3	24	2	Alzheimer_disease (98), Amyloidosis (103), Schizophrenia (1359)
353	APRT	Metabolic	1	1	1	Urolithiasise (1550)
355	FAS	Grey	2	5	2	Autoimmune_disease (174), Squamous_cell_carcinoma (1437)
356	FASLG	Immunological	1	3	1	Systemic_lupus_erythematosus (1471)
358	AQP1	Grey	2	22	1	Aquaporin-1_deficiency (138), Blood_group (212)
359	AQP2	Endocrine	1	2	1	Diabetes_insipidus (2427)
360	AQP3	Hematological	1	22	1	Blood_group (212)
367	AR	Grey	5	33	3	Androgen_insensitivity (109), Breast_cancer (228), Perineal_hypospadias (1205), Prostate_cancer (1272), Spinal_muscular_atrophy (1426)
368	ABCC6	Connective_tissue_disorder	1	0	1	Pseudoxanthoma_elasticum (1285)
383	ARG1	Metabolic	1	1	1	Argininemia (140)

401	PHOX2A	Ophthamological	1	1	1	Fibrosis (538)
405	ARNT	Cancer	1	36	1	Leukemia (886)
410	ARSA	Neurological	1	1	1	Metachromatic_leukodystrophy (996)
411	ARSB	Metabolic	1	0	1	Maroteaux-Lamy_syndrome (952)
412	STS	Grey	2	6	2	Ichthyosis (794), Placental_steroid_sulfatase_deficiency (5233)
415	ARSE	Connective_tissue_disorder	1	2	1	Chondrodysplasia_punctata (315)
420	DO	Hematological	1	22	1	Blood_group (212)
427	ASAH1	Metabolic	1	0	1	Farber_lipogranulomatosis (526)
435	ASL	Metabolic	1	1	1	Argininemia (140)
443	ASPA	Metabolic	1	0	1	Canavan_disease (258)
445	ASS	Metabolic	1	1	1	Citrullinemia (335)
462	SERPINC1	Hematological	1	0	1	Antithrombin_III_deficiency (127)
463	ATBF1	Cancer	1	11	1	Prostate_cancer (1272)
471	ATIC	Metabolic	1	0	1	AICA-ribosiduria_due_to_ATIC_deficiency (69)
472	ATM	Grey	4	28	2	Ataxia-telangiectasia (157), Breast_cancer (228), Lymphoma (925), T-cell_lymphoblastic_leukemia (1478)
477	ATP1A2	Neurological	2	3	1	Alternating_hemiplegia_of_childhood (96), Migraine (1013)
478	ATP1A3	Neurological	1	5	1	Dystonia (462)
486	FXYD2	Renal	1	2	1	Hypomagnesemia (778)
487	ATP2A1	Muscular	1	0	1	Brody_myopathy (231)
488	ATP2A2	Dermatological	2	0	1	Acrokeratosis_verruciformis (36), Darier_disease (404)
491	ATP2B2	Ear,Nose,Throat	1	40	1	Deafness (406)
525	ATP6V1B1	Renal	1	4	1	Renal_tubular_acidosis (7308)
538	ATP7A	Grey	3	2	3	Cutis_laxa (395), Menkes_disease (988), Occipital_horn_syndrome (1129)
540	ATP7B	Metabolic	1	0	1	Wilson_disease (1596)
545	ATR	Developmental	1	0	1	Seckel_syndrome (1369)
546	ATRX	Grey	5	0	2	Alpha-thalassemia/mental_retardation_syndrome (92), Chudley-Lowry_syndrome (328), Juberg-Marsidi_syndrome (832), Smith-Fineman-Myers_syndrome (1409), Sutherland-Haan_syndrome-like (1465)
549	AUH	Metabolic	1	0	1	3-methylglutaconic_aciduria (8)
551	AVP	Endocrine	1	2	1	Diabetes_insipidus (2427)
554	AVPR2	Grey	2	2	2	Diabetes_insipidus (2427), Nephrogenic_syndrome_of_inappropriate_antidiuresis (1080)
570	BAAT	Gastrointestinal	1	2	1	Hypercholanemia (723)
580	BARD1	Cancer	1	18	1	Breast_cancer (228)
581	BAX	Cancer	2	34	1	Colon_cancer (346), T-cell_lymphoblastic_leukemia (1478)
582	BBS1	multiple	1	7	1	Bardet-Biedl_syndrome (183)
583	BBS2	multiple	1	7	1	Bardet-Biedl_syndrome (183)
585	BBS4	multiple	1	7	1	Bardet-Biedl_syndrome (183)
590	BCHE	Unclassified	1	0	0	Apnea,_postanesthetic (134)
593	BCKDHA	Metabolic	1	3	1	Maple_syrup_urine_disease (948)
594	BCKDHB	Metabolic	1	3	1	Maple_syrup_urine_disease (948)
595	CCND1	Cancer	3	70	1	Colon_cancer (346), Leukemia (886), von_Hippel-Lindau_syndrome (1572)
596	BCL2	Cancer	1	36	1	Leukemia (886)
605	BCL7A	Cancer	1	0	1	B-cell_non-Hodgkin_lymphoma,_high-grade (190)
606	BCL8	Cancer	1	9	1	Lymphoma (925)
611	OPN1SW	Ophthamological	1	2	1	Colorblindness (348)
613	BCR	Cancer	1	36	1	Leukemia (886)
617	BCS1L	Grey	3	14	3	GRACILE_syndrome (622), Leigh_syndrome (877), Mitochondrial_complex_deficiency (1016)
627	BDNF	Grey	3	6	3	Central_hypoventilation_syndrome (287), Memory_impairment (984), Obsessive-compulsive_disorder (1128)
641	BLM	Cancer	1	0	1	Bloom_syndrome (213)
642	BLMH	Neurological	1	11	1	Alzheimer_disease (98)

657	BMPR1A	Cancer	2	2	1	Cowden_disease (372), Polyposis (1245)
658	BMPR1B	Skeletal	1	4	1	Brachydactyly (224)
659	BMPR2	Cardiovascular	1	0	1	Pulmonary_hypertension,_familial_primary (5291)
668	FOXL2	Grey	2	1	2	Blepharophimosis,_epicanthus_inversus,_and_ptosis (210), Premature_ovarian_failure (1260)
669	BPGM	Hematological	1	9	1	Hemolytic_anemia (660)
672	BRCA1	Cancer	3	23	1	Breast_cancer (228), Ovarian_cancer (1170), Papillary_serous_carcinoma_of_the_peritoneum (1183)
673	BRAF	Cancer	4	41	1	Adenocarcinoma (45), Colon_cancer (346), Melanoma (978), Nonsmall_cell_lung_cancer (1115)
675	BRCA2	Grey	5	45	2	Breast_cancer (228), Fanconi_anemia (523), Pancreatic_cancer (1178), Prostate_cancer (1272), Wilms_tumor (1595)
682	BSG	Hematological	1	22	1	Blood_group (212)
686	BTD	Metabolic	1	0	1	Biotinidase_deficiency (203)
695	BTK	Grey	2	3	2	Agammaglobulinemia (63), XLA_and_isolated_growth_hormone_deficiency (1611)
699	BUB1	Cancer	1	33	1	Colon_cancer (346)
701	BUB1B	Cancer	1	33	1	Colon_cancer (346)
710	SERPING1	Immunological	1	1	1	Angioedema (114)
712	C1QA	Immunological	1	12	1	Complementary_component_deficiency (240)
713	C1QB	Immunological	1	12	1	Complementary_component_deficiency (240)
714	C1QG	Immunological	1	12	1	Complementary_component_deficiency (240)
716	C1S	Immunological	1	12	1	Complementary_component_deficiency (240)
717	C2	Immunological	1	12	1	Complementary_component_deficiency (240)
718	C3	Immunological	1	12	1	Complementary_component_deficiency (240)
720	C4A	Immunological	1	12	1	Complementary_component_deficiency (240)
721	C4B	Immunological	1	12	1	Complementary_component_deficiency (240)
729	C6	Immunological	2	13	1	Complementary_component_deficiency (240), Combined_immunodeficiency (4350)
730	C7	Immunological	1	12	1	Complementary_component_deficiency (240)
732	C8B	Immunological	1	12	1	Complementary_component_deficiency (240)
735	C9	Immunological	1	12	1	Complementary_component_deficiency (240)
760	CA2	Renal	1	4	1	Renal_tubular_acidosis (7308)
762	CA4	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
773	CACNA1A	Neurological	4	14	1	Cerebellar_ataxia (289), Episodic_ataxia (497), Hemiplegic_migraine,_familial (656), Spinocereballar_ataxia (1428)
775	CACNA1C	multiple	1	0	1	Timothy_syndrome (1509)
778	CACNA1F	Ophthamological	1	4	1	Night_blindness (1110)
779	CACNA1S	Grey	3	3	3	Hypokalemic_periodic_paralysis (776), Malignant_hyperthermia_susceptibility (942), Thyrotoxic_periodic_paralysis (1504)
785	CACNB4	Neurological	2	20	1	Ataxia (154), Epilepsy (495)
788	SLC25A20	Metabolic	1	0	1	Carnitine-acylcarnitine_translocase_deficiency (271)
796	CALCA	Bone	1	5	1	Osteoporosis (1163)
799	CALCR	Bone	1	5	1	Osteoporosis (1163)
825	CAPN3	Muscular	1	17	1	Muscular_dystrophy (1040)
841	CASP8	Grey	2	10	2	Autoimmune_disease (174), Hepatic_adenoma (668)
843	CASP10	Grey	3	12	2	Autoimmune_disease (174), Gastric_cancer (572), Non-Hodgkin_lymphoma (1114)
845	CASQ2	Cardiovascular	1	2	1	Ventricular_tachycardia (1558)
846	CASR	Endocrine	3	2	1	Hyperparathyroidism (743), Hypocalcemia (764), Hypocalciuric_hypercalcemia (765)
847	CAT	Hematological	1	0	1	Acatalasemia (21)
859	CAV3	Grey	5	44	3	Cardiomyopathy (268), Creatine_phosphokinase (385), Muscular_dystrophy (1040), Myopathy (1059), Rippling_muscle_disease (1333)
860	RUNX2	Skeletal	2	0	1	Cleidocranial_dysplasia (337), Dental_anomalies,_isolated (414)
861	RUNX1	Grey	3	46	3	Leukemia (886), Platelet_defect/deficiency (1237), Rheumatoid_arthritis (1324)
865	CBFB	Cancer	1	3	1	Myelogenous_leukemia (1047)

875	CBS	Grey	2	9	2	Homocystinuria (698), Thrombophilia (1497)
889	KRIT1	Grey	3	2	2	Cavernous_malformations_of_CNS_and_retina (279), Cerebral_cavernous_malformations (4291), Hyperkeratotic_cutaneous_capillary-venous_malformations_associated_with_cerebral_capillary_malformations (734)
915	CD3D	Immunological	1	7	1	Severe_combined_immunodeficiency (1383)
916	CD3E	Immunological	1	5	1	Immunodeficiency (802)
917	CD3G	Immunological	1	5	1	Immunodeficiency (802)
925	CD8A	Immunological	1	0	1	CD8_deficiency,_familial (284)
948	CD36	Grey	2	7	2	Malaria (940), Platelet_defect/deficiency (1237)
958	CD40	Immunological	1	5	1	Immunodeficiency (802)
959	CD40LG	Immunological	1	5	1	Immunodeficiency (802)
960	CD44	Hematological	1	22	1	Blood_group (212)
966	CD59	Immunological	1	0	1	CD59_deficiency (283)
977	CD151	Renal	1	0	1	Nephropathy (1085)
999	CDH1	Grey	5	33	2	Breast_cancer (228), Endometrial_carcinoma (479), Gastric_cancer (572), Listeria_monocytogenes (909), Ovarian_cancer (1170)
1001	CDH3	Dermatological	1	3	1	Hypotrichosis (788)
1019	CDK4	Cancer	1	4	1	Melanoma (978)
1028	CDKN1C	multiple	1	3	1	Beckwith-Wiedemann_syndrome (194)
1029	CDKN2A	Cancer	4	11	1	Li_Fraumeni_syndrome (897), Melanoma (978), Orolaryngeal_cancer (1148), Pancreatic_cancer (1178)
1041	CDSN	Dermatological	1	3	1	Hypotrichosis (788)
1050	CEBPA	Cancer	1	36	1	Leukemia (886)
1053	CEBPE	Immunological	1	0	1	Specific_granule_deficiency (3419)
1071	CETP	Grey	3	1	2	CETP_deficiency (299), Hyperalphalipoproteinemia (716), Longevity (911)
1075	CTSC	Grey	3	0	2	Haim-Munk_syndrome (634), Papillon-Lefevre_syndrome (1184), Periodontitis (1207)
1080	CFTR	Grey	5	2	2	Congenital_bilateral_absence_of_vas_deferens (2354), Cystic_fibrosis (399), Hypertrypsinemia (758), Pancreatitis (1179), Sweat_chloride_elevation_without_CF (1466)
1103	CHAT	Muscular	1	6	1	Myasthenic_syndrome (1042)
1118	CHIT1	Metabolic	1	0	1	Chitotriosidase_deficiency (307)
1121	CHM	Ophthamological	1	0	1	Choroidal_dystrophy (323)
1130	LYST	multiple	1	0	1	Chediak-Higashi_syndrome (304)
1134	CHRNA1	Muscular	1	6	1	Myasthenic_syndrome (1042)
1137	CHRNA4	Grey	2	18	2	Epilepsy (495), Nicotine_addiction (1106)
1140	CHRNB1	Muscular	1	6	1	Myasthenic_syndrome (1042)
1141	CHRNB2	Neurological	1	16	1	Epilepsy (495)
1144	CHRND	Muscular	1	6	1	Myasthenic_syndrome (1042)
1145	CHRNE	Muscular	1	6	1	Myasthenic_syndrome (1042)
1161	ERCC8	multiple	1	1	1	Cockayne_syndrome (339)
1180	CLCN1	Muscular	1	1	1	Myotonia_congenita (1062)
1181	CLCN2	Neurological	1	16	1	Epilepsy (495)
1184	CLCN5	Grey	4	3	2	Dent_disease (416), Hypophosphatemia (783), Nephrolithiasis (1081), Proteinuria (1274)
1186	CLCN7	Bone	1	3	1	Osteopetrosis (1161)
1187	CLCNKA	multiple	1	4	1	Bartter_syndrome (187)
1188	CLCNKB	multiple	1	4	1	Bartter_syndrome (187)
1200	TPP1	Neurological	1	5	1	Ceroid-lipofuscinosis (296)
1201	CLN3	Neurological	1	5	1	Ceroid-lipofuscinosis (296)
1203	CLN5	Neurological	1	5	1	Ceroid-lipofuscinosis (296)
1231	CCR2	Immunological	1	3	1	HIV (684)
1234	CCR5	Immunological	1	3	1	HIV (684)
1244	ABCC2	Metabolic	1	0	1	Dubin-Johnson_syndrome (450)

1258	CNGB1	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
1259	CNGA1	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
1261	CNGA3	Ophthamological	1	2	1	Achromatopsia (29)
1277	COL1A1	Grey	5	12	2	Caffey_disease (253), Dissection_of_cervical_arteries (439), Ehlers-Danlos_syndrome (470), Osteogenesis_imperfecta (1156), Osteoporosis (1163)
1278	COL1A2	Grey	4	13	2	Ehlers-Danlos_syndrome (470), Marfan_syndrome (950), Osteogenesis_imperfecta (1156), Osteoporosis (1163)
1280	COL2A1	Grey	6	5	5	Achondrogenesis-hypochondrogenesis,_type_II (26), Kniest_dysplasia (854), Osteoarthritis (1154), SMED_Strudwick_type (1408), Stickler_syndrome (1447), Wagner_syndrome (1575)
1281	COL3A1	Grey	2	8	1	Aneurysm,_familial_arterial (2112), Ehlers-Danlos_syndrome (470)
1282	COL4A1	Neurological	1	0	1	Porencephaly (1247)
1285	COL4A3	Renal	1	2	1	Alport_syndrome (94)
1286	COL4A4	Renal	2	2	1	Alport_syndrome (94), Hematuria,_familial_benign (653)
1287	COL4A5	Renal	1	2	1	Alport_syndrome (94)
1288	COL4A6	Cancer	1	1	1	Leiomyomatosis (878)
1289	COL5A1	Connective_tissue_disorder	1	8	1	Ehlers-Danlos_syndrome (470)
1290	COL5A2	Connective_tissue_disorder	1	8	1	Ehlers-Danlos_syndrome (470)
1291	COL6A1	Muscular	2	2	1	Bethlem_myopathy (200), Ullrich_congenital_muscular_dystrophy (1542)
1292	COL6A2	Muscular	2	2	1	Bethlem_myopathy (200), Ullrich_congenital_muscular_dystrophy (1542)
1293	COL6A3	Muscular	2	2	1	Bethlem_myopathy (200), Ullrich_congenital_muscular_dystrophy (1542)
1294	COL7A1	Dermatological	4	10	1	EBD (465), Epidermolysis_bullosa (493), Toenail_dystrophy,_isolated (1510), Transient_bullous_of_the_newborn (1518)
1296	COL8A2	Ophthamological	2	6	1	Corneal_dystrophy (362), Fuchs_endothelial_corneal_dystrophy (558)
1297	COL9A1	Bone	1	5	1	Epiphyseal_dysplasia (496)
1298	COL9A2	Grey	2	5	2	Epiphyseal_dysplasia (496), Intervertebral_disc_disease (817)
1299	COL9A3	Grey	2	5	2	Epiphyseal_dysplasia (496), Intervertebral_disc_disease (817)
1300	COL10A1	Grey	2	2	2	Metaphyseal_chondrodysplasia (997), Spondylometaphyseal_dysplasia (1436)
1301	COL11A1	multiple	2	2	1	Marshall_syndrome (953), Stickler_syndrome (1447)
1302	COL11A2	Grey	4	42	4	Deafness (406), OSMED_syndrome (1151), Stickler_syndrome (1447), Weissenbacher-Zweymuller_syndrome (1586)
1308	COL17A1	Dermatological	1	10	1	Epidermolysis_bullosa (493)
1311	COMP	Grey	2	5	2	Epiphyseal_dysplasia (496), Pseudoachondroplasia (1278)
1312	COMT	Psychiatric	1	8	1	Schizophrenia (1359)
1316	KLF6	Cancer	2	20	1	Gastric_cancer (572), Prostate_cancer (1272)
1326	MAP3K8	Cancer	1	3	1	Lung_cancer (918)
1352	COX10	Neurological	1	1	1	Encephalopathy (476)
1355	COX15	Grey	2	35	2	Cardiomyopathy (268), Leigh_syndrome (877)
1356	CP	Grey	3	1	3	Cerebellar_ataxia (289), Hemosiderosis,_systemic,_due_to_aceruloplasminemia (665), Hypoceruloplasminemia (766)
1369	CPN1	Hematological	1	0	1	Carboxypeptidase_N_deficiency (264)
1371	CPOX	Metabolic	2	0	1	Coproporphyria (361), Harderoporphyrinuria (639)
1373	CPS1	Metabolic	1	0	1	Carbamoylphosphate_synthetase_I_deficiency (262)
1374	CPT1A	Metabolic	1	1	1	CPT_deficiency,_hepatic (374)
1376	CPT2	Grey	2	10	2	CPT_deficiency,_hepatic (374), Myopathy (1059)
1378	CR1	Hematological	1	22	1	Blood_group (212)
1387	CREBBP	multiple	1	1	1	Rubenstein-Taybi_syndrome (1341)
1406	CRX	Ophthamological	3	33	1	Cone_dystrophy (352), Leber_congenital_amaurosis (873), Retinitis_pigmentosa (1316)
1409	CRYAA	Ophthamological	1	14	1	Cataract (277)
1410	CRYAB	Grey	2	23	2	Cataract (277), Myopathy (1059)

1411	CRYBA1	Ophthamological	1	14	1	Cataract (277)
1414	CRYBB1	Ophthamological	1	14	1	Cataract (277)
1415	CRYBB2	Ophthamological	1	14	1	Cataract (277)
1420	CRYGC	Ophthamological	1	14	1	Cataract (277)
1421	CRYGD	Ophthamological	1	14	1	Cataract (277)
1428	CRYM	Ear,Nose,Throat	1	40	1	Deafness (406)
1436	CSF1R	Cancer	1	3	1	Myelogenous_leukemia (1047)
1439	CSF2RB	Respiratory	1	3	1	Pulmonary_fibrosis (1291)
1471	CST3	Neurological	1	1	1	Cerebral_amyloid_angiopathy (291)
1476	CSTB	Neurological	1	16	1	Epilepsy (495)
1482	NKX2-5	Cardiovascular	3	8	1	Atrial_fibrillation (166), Atrioventricular_block (168), Tetralogy_of_Fallot (1483)
1491	CTH	Metabolic	2	0	1	Cystathioninuria (398), Homocysteine_plasma_level (697)
1493	CTLA4	Endocrine	2	7	1	Graves_disease (624), Hypothyroidism (787)
1497	CTNS	Renal	1	0	1	Cystinosis (400)
1499	CTNNB1	Cancer	4	42	1	Colon_cancer (346), Hepatic_adenoma (668), Ovarian_cancer (1170), Pilomatricoma (1230)
1513	CTSK	Skeletal	1	0	1	Pycnodysostosis (1293)
1524	CX3CR1	Immunological	1	0	1	Rapid_progression_to_AIDS_from_HIV1_infection (1304)
1528	CYB5	Hematological	1	3	1	Methemoglobinemia (999)
1535	CYBA	Immunological	1	3	1	Chronic_granulomatous_disease (327)
1536	CYBB	Immunological	1	3	1	Chronic_granulomatous_disease (327)
1540	CYLD	Dermatological	1	0	1	Cylindromatosis,_familial (397)
1545	CYP1B1	Grey	2	4	2	Glaucoma (589), Peters_anomaly (1213)
1548	CYP2A6	Grey	2	2	2	Coumarin_resistance (370), Nicotine_addiction (1106)
1557	CYP2C19	Metabolic	3	0	1	Mephenytoin_poor_metabolizer (991), Opremazole_poor_metabolizer (1143), Proguanil_poor_metabolizer (1266)
1559	CYP2C9	Grey	2	2	1	Tolbutamide_poor_metabolizer (1511), Warfarin_resistance/sensitivity (1580)
1565	CYP2D6	Metabolic	2	0	1	Codeine_sensitivity (340), Debrisoquine_sensitivity (407)
1577	CYP3A5	Cardiovascular	1	11	1	Hypertension (752)
1583	CYP11A1	Endocrine	1	1	1	Lipoid_adrenal_hyperplasia (903)
1584	CYP11B1	Endocrine	2	3	1	Adrenal_hyperplasia,_congenital (53), Aldosteronism (78)
1585	CYP11B2	Grey	3	0	3	Aldosterone_to_renin_ratio_raised (77), Hypoaldosteronism (761), Low_renin_hypertension (915)
1586	CYP17A1	Endocrine	1	0	1	17,20-lyase_deficiency (1)
1588	CYP19A1	Grey	2	0	2	Aromatase_deficiency (142), Virilization (1563)
1589	CYP21A2	Endocrine	2	3	1	Adrenal_hyperplasia,_congenital (53), Hyperandrogenism (718)
1593	CYP27A1	Metabolic	1	0	1	Cerebrotendinous_xanthomatosis (293)
1594	CYP27B1	Bone	1	0	1	Pseudovitamin_D_deficiency_rickets_1 (1284)
1603	DAD1	Unclassified	1	0	0	Temperature-sensitive_apoptosis,_cellular (1480)
1604	DAF	Hematological	1	22	1	Blood_group (212)
1618	DAZL	Renal	1	0	1	Spermatogenic_failure (1422)
1621	DBH	Grey	2	10	2	Dopamine_beta-hydroxylase_deficiency (441), Parkinson_disease (1192)
1629	DBT	Metabolic	1	3	1	Maple_syrup_urine_disease (948)
1630	DCC	Cancer	1	33	1	Colon_cancer (346)
1636	ACE	Grey	6	48	5	Alzheimer_disease (98), Angiotensin_I-converting_enzyme (117), Diabetes_mellitus (427), Myocardial_infarction (1054), Renal_tubular_dysgenesis (9308), SARS,_progression_of (1352)
1639	DCTN1	Neurological	2	5	1	Amyotrophic_lateral_sclerosis (104), Lower_motor_neuron_disease,_progressive,_without_sensory_symptoms (913)
1641	DCX	Neurological	2	3	1	Lissencephaly (908), Subcortical_laminar_heterotopia (1456)
1643	DDB2	Dermatological	1	7	1	Xeroderma_pigmentosum (1608)
1644	DDC	Metabolic	1	0	1	Aromatic_L-amino_acid_decarboxylase_deficiency (143)
1646	AKR1C2	Nutritional	1	20	1	Obesity (1126)
1649	DDIT3	Cancer	1	0	1	Myxoid_liposarcoma (1065)

1674	DES	Grey	2	31	2	Cardiomyopathy (268), Myopathy (1059)
1678	TIMM8A	Grey	3	40	2	Deafness (406), Jensen_syndrome (829), Mohr-Tranebjaerg_syndrome (1022)
1687	DFNA5	Ear,Nose,Throat	1	40	1	Deafness (406)
1690	COCH	Grey	2	40	2	Deafness (406), Meniere_disease (985)
1716	DGUOK	multiple	1	1	1	Mitochondrial_DNA_depletion_syndrome (5016)
1717	DHCR7	multiple	1	0	1	Smith-Lemli-Opitz_syndrome (1410)
1718	DHCR24	Metabolic	1	0	1	Desmosterolosis (426)
1727	CYB5R3	Hematological	1	3	1	Methemoglobinemia (999)
1728	NQO1	Grey	2	36	1	Benzene_toxicity (195), Leukemia (886)
1729	DIAPH1	Ear,Nose,Throat	1	40	1	Deafness (406)
1730	DIAPH2	Renal	1	1	1	Premature_ovarian_failure (1260)
1736	DKC1	Grey	2	1	2	Dyskeratosis (458), Hoyeraal-Hreidarsson_syndrome (702)
1741	DLG3	Neurological	1	23	1	Mental_retardation (990)
1747	DLX3	Grey	2	3	2	Amelogenesis_imperfecta (99), Trichodontoosseous_syndrome (1522)
1755	DMBT1	Cancer	1	4	1	Glioblastoma (590)
1756	DMD	Grey	3	25	2	Becker_muscular_dystrophy (193), Cardiomyopathy (268), Duchenne_muscular_dystrophy (451)
1760	DMPK	Muscular	1	1	1	Myotonic_dystrophy (1063)
1767	DNAH5	Grey	2	2	2	Ciliary_dyskinesia (331), Kartagener_syndrome (840)
1773	DNASE1	Immunological	1	3	1	Systemic_lupus_erythematosus (1471)
1785	DNM2	Neurological	1	17	1	Charcot-Marie-Tooth_disease (301)
1789	DNMT3B	multiple	1	0	1	Immunodeficiency-centromeric_instability-facial_anomalies_syndrome (801)
1798	DPAGT1	Metabolic	1	12	1	Congenital_disorder_of_glycosylation (354)
1806	DPYD	Metabolic	2	0	1	Fluorouracil_toxicity,_sensitivity_to (544), Thymine-uraciluria (1502)
1807	DPYS	Metabolic	1	0	1	Dihydropyrimidinuria (433)
1811	SLC26A3	Gastrointestinal	1	0	1	Chloride_diarrhea,_congenital,_Finnish_type (308)
1813	DRD2	Neurological	1	5	1	Dystonia (462)
1815	DRD4	Psychiatric	1	0	1	Autonomic_nervous_system_dysfunction (175)
1816	DRD5	Grey	3	5	3	Attention-deficit_hyperactivity_disorder (171), Blepharospasm (211), Dystonia (462)
1822	ATN1	Neurological	1	0	1	Dentatorubro-pallidoluysian_atrophy (415)
1828	DSG1	Dermatological	1	3	1	Keratosis_palmoplantaria_striata (847)
1832	DSP	Grey	5	15	3	Arrhythmogenic_right_ventricular_dysplasia (144), Dilated_cardiomyopathy_with_woolly_hair_and_keratoderma (434), Epidermolysis_bullosa (493), Keratosis_palmoplantaria_striata (847), Skin_fragility-woolly_hair_syndrome (1401)
1834	DSPP	Grey	3	40	2	Deafness (406), Dentin_dysplasia,_type_II (417), Dentinogenesis_imperfecta,_Shields_type (418)
1836	SLC26A2	Grey	4	6	3	Achondrogenesis_Ib (27), Atelosteogenesis (160), Diastrophic_dysplasia (430), Epiphyseal_dysplasia (496)
1837	DTNA	Cardiovascular	1	0	1	Left_ventricular_noncompaction (875)
1861	TOR1A	Neurological	1	5	1	Dystonia (462)
1889	ECE1	Grey	2	17	2	Hirschsprung_disease (681), Hypertension (752)
1890	ECGF1	multiple	1	0	1	Myoneurogastrointestinal_encephalomyopathy_syndrome (1058)
1893	ECM1	Metabolic	1	0	1	Lipoid_proteinosis (2903)
1896	EDA	Dermatological	1	7	1	Ectodermal_dysplasia (466)
1908	EDN3	Grey	3	7	3	Central_hypoventilation_syndrome (287), Hirschsprung_disease (681), Shah-Waardenburg_syndrome (1386)
1909	EDNRA	Neurological	1	3	1	Migraine (1013)
1910	EDNRB	Grey	3	7	2	ABCD_syndrome (15), Hirschsprung_disease (681), Waardenburg-Shah_syndrome (1573)
1947	EFNB1	Skeletal	1	0	1	Craniofrontonasal_dysplasia (380)
1956	EGFR	Cancer	2	4	1	Adenocarcinoma (45), Nonsmall_cell_lung_cancer (1115)
1959	EGR2	Grey	3	22	2	Charcot-Marie-Tooth_disease (301), Dejerine-Sottas_disease (410), Neuropathy (1099)
1967	EIF2B1	Neurological	1	4	1	Leukoencephalopathy_with_vanishing_white_matter (891)
1991	ELA2	Hematological	2	3	1	Hematopoiesis,_cyclic (652), Neutropenia (1101)
2006	ELN	Grey	3	2	3	Cutis_laxa (395), Supravalvar_aortic_stenosis (1463), Williams-Beuren_syndrome (1594)

2010	EMD	Muscular	1	1	1	Emery-Dreifuss_muscular_dystrophy (474)
2018	EMX2	Neurological	1	0	1	Schizencephaly (1357)
2022	ENG	Cardiovascular	1	1	1	Hereditary_hemorrhagic_telangiectasia (669)
2027	ENO3	Metabolic	1	0	1	Enolase_deficiency (485)
2033	EP300	Grey	2	34	2	Colon_cancer (346), Rubenstein-Taybi_syndrome (1341)
2035	EPB41	Hematological	1	3	1	Elliptocytosis (472)
2038	EPB42	Hematological	1	4	1	Spherocytosis (1423)
2048	EPHB2	Cancer	1	11	1	Prostate_cancer (1272)
2052	EPHX1	Grey	2	4	2	Hypercholanemia (723), Preeclampsia (1257)
2053	EPHX2	Metabolic	1	7	1	Hypercholesterolemia (724)
2055	CLN8	Neurological	1	5	1	Ceroid-lipofuscinosis (296)
2057	EPOR	Hematological	1	1	1	Erythrocytosis (505)
2064	ERBB2	Cancer	4	21	1	Adenocarcinoma (45), Gastric_cancer (572), Glioblastoma (590), Ovarian_cancer (1170)
2068	ERCC2	Grey	3	10	2	Cerebrooculofacioskeletal_syndrome (292), Trichothiodystrophy (1525), Xeroderma_pigmentosum (1608)
2070	EYA4	Grey	2	64	2	Cardiomyopathy (268), Deafness (406)
2071	ERCC3	Dermatological	2	9	1	Trichothiodystrophy (1525), Xeroderma_pigmentosum (1608)
2072	ERCC4	Dermatological	1	7	1	Xeroderma_pigmentosum (1608)
2073	ERCC5	Grey	2	8	2	Cerebrooculofacioskeletal_syndrome (292), Xeroderma_pigmentosum (1608)
2074	ERCC6	multiple	3	3	1	Cerebrooculofacioskeletal_syndrome (292), Cockayne_syndrome (339), De_Sanctis-Cacchione_syndrome (424)
2099	ESR1	Grey	3	4	3	Estrogen_resistance (509), HDL_cholesterol_level_QTL (644), Migraine (1013)
2108	ETFA	Metabolic	1	3	1	Glutaricaciduria (603)
2109	ETFB	Metabolic	1	3	1	Glutaricaciduria (603)
2110	ETFDH	Metabolic	1	3	1	Glutaricaciduria (603)
2121	EVC	Skeletal	2	1	1	Ellis-van_Creveld_syndrome (473), Weyers_acrodental_dysostosis (1590)
2130	EWSR1	Cancer	2	2	1	Chondrosarcoma (316), Ewing_sarcoma (511)
2131	EXT1	Grey	2	3	2	Chondrosarcoma (316), Exostoses (514)
2132	EXT2	Bone	1	1	1	Exostoses (514)
2138	EYA1	Grey	2	3	2	Anterior_segment_anomalies_and_cataract (126), Branchiootic_syndrome (226)
2147	F2	Hematological	3	0	1	Dysprothrombinemia (460), Hyperprothrombinemia (751), Hypoprothrombinemia (786)
2153	F5	Hematological	3	12	1	Hemorrhagic_diathesis (664), Thrombocytopenia (1494), Thrombophilia (1497)
2155	F7	Grey	2	16	2	Factor_x_deficiency (520), Myocardial_infarction (1054)
2157	F8	Hematological	1	1	1	Hemophilia (663)
2158	F9	Hematological	2	3	1	Hemophilia (663), Warfarin_resistance/sensitivity (1580)
2159	F10	Hematological	1	7	1	Factor_x_deficiency (520)
2160	F11	Hematological	1	7	1	Factor_x_deficiency (520)
2161	F12	Hematological	1	7	1	Factor_x_deficiency (520)
2162	F13A1	Hematological	1	7	1	Factor_x_deficiency (520)
2165	F13B	Hematological	1	7	1	Factor_x_deficiency (520)
2166	FAAH	Psychiatric	1	0	1	Drug_addiction (447)
2175	FANCA	multiple	1	10	1	Fanconi_anemia (523)
2176	FANCC	multiple	1	10	1	Fanconi_anemia (523)
2177	FANCD2	multiple	1	10	1	Fanconi_anemia (523)
2178	FANCE	multiple	1	10	1	Fanconi_anemia (523)
2182	ACSL4	Neurological	1	23	1	Mental_retardation (990)
2184	FAH	Metabolic	1	2	1	Tyrosinemia (1540)
2187	FANCB	multiple	1	10	1	Fanconi_anemia (523)
2188	FANCF	multiple	1	10	1	Fanconi_anemia (523)
2189	FANCG	multiple	1	10	1	Fanconi_anemia (523)
2192	FBLN1	Skeletal	1	1	1	Synpolydactyly (1470)

2200	FBN1	Grey	6	3	4	Aortic_aneurysm (130), Ectopia (467), Marfan_syndrome (950), MASS_syndrome (957), Shprintzen-Goldberg_syndrome (1388), Weill-Marchesani_syndrome (1585)
2201	FBN2	Connective_tissue_disorder	1	0	1	Contractural_arachnodactyly,_congenital (358)
2202	EFEMP1	Ophthamological	1	0	1	Doyne_honeycomb_degeneration_of_retina (446)
2203	FBP1	Metabolic	1	0	1	Fructose-bisphosphatase_deficiency (555)
2206	MS4A2	Respiratory	1	12	1	Asthma (153)
2209	FCGR1A	Immunological	1	0	1	IgG_receptor_I,_phagocytic,_familial_deficiency_of (799)
2212	FCGR2A	Connective_tissue_disorder	1	0	1	Lupus_erythematosus (919)
2213	FCGR2B	Cancer	1	9	1	Lymphoma (925)
2214	FCGR3A	Grey	2	4	2	Neutropenia (1101), Viral_infection (1562)
2218	FCMD	Grey	2	18	2	Muscular_dystrophy (1040), Walker-Warburg_syndrome (1578)
2235	FECH	Metabolic	1	0	1	Protoporphyria (1276)
2243	FGA	Grey	3	7	2	Afibrinogenemia (61), Amyloidosis (103), Dysfibrinogenemia (457)
2244	FGB	Hematological	3	9	1	Afibrinogenemia (61), Dysfibrinogenemia (457), Thrombophilia (1497)
2245	FGD1	Grey	2	23	2	Aarskog-Scott_syndrome (13), Mental_retardation (990)
2255	FGF10	Gastrointestinal	1	0	1	Aplasia_of_lacrimal_and_salivary_glands (132)
2259	FGF14	Neurological	1	12	1	Spinocereballar_ataxia (1428)
2260	FGFR1	Grey	3	2	2	Jackson-Weiss_syndrome (827), Kallmann_syndrome (836), Pfeiffer_syndrome (1215)
2261	FGFR3	Grey	8	37	2	Achondroplasia (28), Bladder_cancer (207), Cervical_carcinoma (298), Colon_cancer (346), Crouzon_syndrome (391), Hypochondroplasia (767), Muenke_syndrome (1034), Thanatophoric_dysplasia,_types_I_and_II (1490)
2263	FGFR2	Grey	9	13	5	Apert_syndrome (131), Beare-Stevenson_cutis_gyrata_syndrome (192), Craniofacial-skeletal-dermatologic_dysplasia (379), Craniosynostosis (383), Crouzon_syndrome (391), Gastric_cancer (572), Jackson-Weiss_syndrome (827), Pfeiffer_syndrome (1215), Saethre-Chotzen_syndrome (1344)
2264	FGFR4	Cancer	1	1	1	Cancer_susceptibility (259)
2266	FGG	Hematological	3	9	1	Dysfibrinogenemia (457), Hypofibrinogenemia (770), Thrombophilia (1497)
2271	FH	Grey	3	1	2	Fumarase_deficiency (561), Leiomyomatosis (878), Multiple_cutaneous_and_uterine_leiomyomata (3037)
2289	FKBP5	Psychiatric	1	0	1	Major_depressive_disorder (939)
2296	FOXC1	Grey	5	4	2	Anterior_segment_anomalies_and_cataract (126), Axenfeld_anomaly (177), Iridogoniodysgenesis (822), Iris_hypoplasia_and_glaucoma (823), Rieger_syndrome (1331)
2301	FOXE3	Ophthamological	1	3	1	Anterior_segment_anomalies_and_cataract (126)
2303	FOXC2	Grey	2	1	2	Lymphedema (921), Yellow_nail_syndrome (1613)
2304	FOXE1	Endocrine	1	0	1	Bamforth-Lazarus_syndrome (180)
2308	FOXO1A	Cancer	1	3	1	Rhabdomyosarcoma (1323)
2316	FLNA	Grey	4	0	3	Frontometaphyseal_dysplasia (554), Heterotopia (676), Melnick-Needles_syndrome (981), Otopalatodigital_syndrome (1168)
2317	FLNB	Grey	3	1	2	Atelosteogenesis (160), Larson_syndrome (865), Spondylocarpotarsal_synostosis_syndrome (1432)
2322	FLT3	Cancer	1	36	1	Leukemia (886)
2324	FLT4	Grey	2	2	2	Hemangioma (651), Lymphedema (921)
2328	FMO3	Metabolic	1	0	1	Fish-odor_syndrome (541)
2332	FMR1	Neurological	1	0	1	Fragile_X_syndrome (550)
2334	AFF2	Neurological	1	23	1	Mental_retardation (990)
2395	FXN	Neurological	1	0	1	Friedreich_ataxia (553)
2487	FRZB	Connective_tissue_disorder	1	3	1	Osteoarthritis (1154)
2488	FSHB	Endocrine	1	0	1	Follicle-stimulating_hormone_deficiency,_isolated (546)
2489	FSHMD1A	Muscular	1	0	1	Facioscapulohumeral_muscular_dystrophy (519)
2492	FSHR	Grey	4	1	2	Ovarian_dysgenesis (3171), Ovarian_hyperstimulation_syndrome (5170), Ovarian_sex_cord_tumors (7170), Twinning,_dizygotic (1538)

2495	FTH1	Hematological	1	1	1	Iron_overload/deficiency (824)
2512	FTL	Grey	2	1	2	Basal_ganglia_disease (4188), Hyperferritinemia-cataract_syndrome (727)
2516	NR5A1	Grey	2	0	1	Adrenocortical_insufficiency (2055), Sex_reversal (1384)
2517	FUCA1	Metabolic	1	0	1	Fucosidosis (559)
2523	FUT1	Hematological	1	1	1	Bombay_phenotype (216)
2524	FUT2	Grey	2	1	2	Bombay_phenotype (216), Norwalk_virus_infection,_resistance_to (1119)
2525	FUT3	Hematological	1	22	1	Blood_group (212)
2528	FUT6	Metabolic	1	0	1	Fucosyltransferase-6_deficiency (560)
2538	G6PC	Metabolic	1	8	1	Glycogen_storage_disease (610)
2539	G6PD	Grey	3	9	2	Favism (528), G6PD_deficiency (563), Hemolytic_anemia (660)
2542	SLC37A4	Metabolic	1	8	1	Glycogen_storage_disease (610)
2548	GAA	Metabolic	1	8	1	Glycogen_storage_disease (610)
2554	GABRA1	Neurological	1	16	1	Epilepsy (495)
2555	GABRA2	Unclassified	1	0	0	Alcoholism (75)
2562	GABRB3	Psychiatric	1	1	1	Insomnia (812)
2566	GABRG2	Neurological	2	18	1	Epilepsy (495), Myoclonic_epilepsy (1055)
2581	GALC	Neurological	1	0	1	Krabbe_disease (856)
2582	GALE	Metabolic	1	0	1	Galactose_epimerase_deficiency (566)
2584	GALK1	Metabolic	1	0	1	Galactokinase_deficiency (565)
2588	GALNS	Metabolic	1	6	1	Mucopolysaccharidosis (1033)
2591	GALNT3	Bone	1	1	1	Calcinosis,_tumoral (254)
2592	GALT	Metabolic	1	0	1	Galactosemia (567)
2593	GAMT	Metabolic	1	0	1	GAMT_deficiency (570)
2617	GARS	Grey	2	23	2	Charcot-Marie-Tooth_disease (301), Spinal_muscular_atrophy (1426)
2623	GATA1	Grey	3	36	2	Dyserythropoietic_anemia (456), Leukemia (886), Macrothrombocytopenia (936)
2625	GATA3	Endocrine	1	0	1	Hypoparathyroidism (781)
2626	GATA4	Cardiovascular	1	4	1	Atrial_fibrillation (166)
2628	GATM	Metabolic	1	0	1	AGAT_deficiency (64)
2629	GBA	Metabolic	1	1	1	Gaucher_disease (575)
2632	GBE1	Metabolic	1	8	1	Glycogen_storage_disease (610)
2638	GC	Endocrine	1	1	1	Graves_disease (624)
2639	GCDH	Metabolic	1	3	1	Glutaricaciduria (603)
2642	GCGR	Endocrine	1	26	1	Diabetes_mellitus (427)
2643	GCH1	Neurological	1	5	1	Dystonia (462)
2645	GCK	Grey	3	27	2	Diabetes_mellitus (427), Hyperinsulinism (731), MODY (1020)
2651	GCNT2	Hematological	2	22	1	Adult_i_phenotype (58), Blood_group (212)
2652	OPN1MW	Ophthamological	2	2	1	Blue-cone_monochromacy (214), Colorblindness (348)
2653	GCSH	Metabolic	1	3	1	Glycine_encephalopathy (608)
2654	GCSL	Grey	2	14	2	Leigh_syndrome (877), Maple_syrup_urine_disease (948)
2660	GDF8	Muscular	1	1	1	Muscle_hypertrophy (1039)
2664	GDI1	Neurological	1	23	1	Mental_retardation (990)
2668	GDNF	Grey	2	7	2	Central_hypoventilation_syndrome (287), Hirschsprung_disease (681)
2670	GFAP	multiple	1	1	1	Alexander_disease (79)
2672	GFI1	Hematological	1	3	1	Neutropenia (1101)
2677	GGCX	Hematological	1	1	1	Vitamin_K-dependent_coagulation_defect (1565)
2683	B4GALT1	Metabolic	1	12	1	Congenital_disorder_of_glycosylation (354)
2688	GH1	Endocrine	1	0	1	Isolated_growth_hormone_deficiency (825)
2690	GHR	Skeletal	2	2	1	Laron_dwarfism (863), Short_stature (1387)
2692	GHRHR	Endocrine	1	1	1	Growth_hormone (628)
2694	GIF	Hematological	1	0	1	Intrinsic_factor_deficiency (820)

2697	GJA1	Grey	4	2	2	Atrioventricular_block (168), Hypoplastic_left_heart_syndrome (2785), Oculodentodigital_dysplasia (1132), Syndactyly (1468)
2700	GJA3	Ophthamological	1	14	1	Cataract (277)
2703	GJA8	Ophthamological	1	14	1	Cataract (277)
2705	GJB1	Neurological	1	17	1	Charcot-Marie-Tooth_disease (301)
2706	GJB2	Grey	6	41	3	Bart-Pumphrey_syndrome (186), Deafness (406), Hystrix-like_ichthyosis_with_deafness (792), Keratitis-ichthyosis-deafness_syndrome (843), Keratoderma,_palmoplantar,_with_deafness (845), Vohwinkel_syndrome (1571)
2707	GJB3	Grey	2	42	2	Deafness (406), Erythrokeratoderma (507)
2710	GK	Metabolic	1	0	1	Glycerol_kinase_deficiency (607)
2717	GLA	Metabolic	1	0	1	Fabry_disease (518)
2719	GPC3	Grey	2	3	2	Simpson-Golabi-Behmel_syndrome (1397), Wilms_tumor (1595)
2720	GLB1	Metabolic	2	8	1	GM-gangliosidosis (614), Mucopolysaccharidosis (1033)
2729	GCLC	Hematological	1	9	1	Hemolytic_anemia (660)
2730	GCLM	Cardiovascular	1	9	1	Myocardial_infarction (1054)
2731	GLDC	Metabolic	1	3	1	Glycine_encephalopathy (608)
2736	GLI2	multiple	1	0	1	Pituitary_anomalies_with_holoprosencephaly-like_features (3232)
2737	GLI3	Grey	4	0	2	Acrocallosal_syndrome (32), Greig_cephalopolysyndactyly_syndrome (626), Pallister-Hall_syndrome (1175), Polydactyly (1243)
2739	GLO1	Psychiatric	1	3	1	Autism (173)
2741	GLRA1	Neurological	2	1	1	Hyperekplexia (725), Startle_disease (1440)
2743	GLRB	Neurological	1	1	1	Hyperekplexia (725)
2746	GLUD1	Metabolic	1	0	1	Hyperinsulinism-hyperammonemia_syndrome (732)
2760	GM2A	Metabolic	1	2	1	GM-gangliosidosis (614)
2771	GNAI2	Grey	2	4	2	Pituitary_ACTH-secreting_adenoma (1232), Ventricular_tachycardia (1558)
2778	GNAS	Grey	6	3	4	Acromegaly (38), McCune-Albright_syndrome (963), Osseous_heteroplasia (1152), Pituitary_ACTH-secreting_adenoma (1232), Pseudohypoparathyroidism (1282), Somatotrophinoma (1415)
2779	GNAT1	Ophthamological	1	4	1	Night_blindness (1110)
2780	GNAT2	Ophthamological	1	2	1	Achromatopsia (29)
2784	GNB3	Cardiovascular	1	11	1	Hypertension (752)
2798	GNRHR	Endocrine	2	3	1	Fertile_eunuch_syndrome (532), Hypogonadotropic_hypogonadism (774)
2799	GNS	Metabolic	1	6	1	Mucopolysaccharidosis (1033)
2811	GP1BA	Hematological	1	2	1	Bernard-Soulier_syndrome (196)
2812	GP1BB	Hematological	2	2	1	Bernard-Soulier_syndrome (196), Giant_platelet_disorder,_isolated (584)
2815	GP9	Hematological	1	2	1	Bernard-Soulier_syndrome (196)
2820	GPD2	Endocrine	1	26	1	Diabetes_mellitus (427)
2821	GPI	Hematological	1	9	1	Hemolytic_anemia (660)
2908	NR3C1	Endocrine	1	0	1	Cortisol_resistance (367)
2934	GSN	Neurological	1	5	1	Amyloidosis (103)
2937	GSS	Grey	2	9	2	Glutathione_synthetase_deficiency (604), Hemolytic_anemia (660)
2956	MSH6	Cancer	4	40	1	Cancer_susceptibility (259), Colon_cancer (346), Endometrial_carcinoma (479), Ovarian_cancer (1170)
2978	GUCA1A	Ophthamological	1	6	1	Cone_dystrophy (352)
2989	GULOP	Nutritional	1	0	1	Scurvy (1365)
2990	GUSB	Metabolic	1	6	1	Mucopolysaccharidosis (1033)
2993	GYPA	Hematological	1	22	1	Blood_group (212)
2994	GYPB	Hematological	1	22	1	Blood_group (212)
2995	GYPC	Grey	2	26	2	Blood_group (212), Malaria (940)
2998	GYS2	Metabolic	1	8	1	Glycogen_storage_disease (610)
3000	GUCY2D	Ophthamological	2	9	1	Cone_dystrophy (352), Leber_congenital_amaurosis (873)
3028	HADH2	Metabolic	1	0	1	2-methyl-3-hydroxybutyryl-CoA_dehydrogenase_deficiency (3)

3030	HADHA	Metabolic	4	1	1	Fatty_liver,_acute,_of_pregnancy (527), HELLP_syndrome (649), LCHAD_deficiency (870), Trifunctional_protein_deficiency (1526)
3032	HADHB	Metabolic	1	1	1	Trifunctional_protein_deficiency (1526)
3033	HADHSC	Metabolic	1	0	1	3-hydroxyacyl-CoA_dehydrogenase_deficiency (6)
3034	HAL	Metabolic	1	0	1	Histidinemia (682)
3039	HBA1	Hematological	4	6	1	Erythremias (504), Heinz_body_anemia (648), Methemoglobinemia (999), Thalassemias (1486)
3040	HBA2	Hematological	5	5	1	Erythrocytosis (505), Heinz_body_anemia (648), Hemoglobin_H_disease (659), Hypochromic_microcytic_anemia (768), Thalassemias (1486)
3043	HBB	Hematological	6	8	1	Erythremias (504), Heinz_body_anemia (648), HPFH (703), Methemoglobinemia (999), Sickle_cell_anemia (1393), Thalassemias (1486)
3045	HBD	Hematological	1	4	1	Thalassemias (1486)
3047	HBG1	Hematological	1	2	1	HPFH (703)
3048	HBG2	Hematological	1	2	1	HPFH (703)
3053	SERPIND1	Hematological	1	8	1	Thrombophilia (1497)
3060	HCRT	Psychiatric	1	0	1	Narcolepsy (1073)
3064	HD	Neurological	1	3	1	Huntington_disease (708)
3073	HEXA	Metabolic	3	2	1	GM-gangliosidosis (614), Hex_A_pseudodeficiency (677), Tay-Sachs_disease (1477)
3074	HEXB	Grey	2	6	2	Sandhoff_disease,_infantile,_juvenile,_and_adult_forms (1347), Spinal_muscular_atrophy (1426)
3075	CFH	Grey	4	11	3	Factor_x_deficiency (520), Hemolytic-uremic_syndrome (661), Macular_degeneration (937), Nephropathy-hypertension (1084)
3077	HFE	Metabolic	2	9	1	Hemochromatosis (657), Porphyria (1249)
3081	HGD	Metabolic	1	0	1	Alkaptonuria (80)
3098	HK1	Hematological	1	9	1	Hemolytic_anemia (660)
3106	HLA-B	Grey	3	0	3	Abacavir_hypersensitivity (14), Ankylosing_spoldylitis (121), Stevens-Johnson_syndrome,_carbamazepine-induced (1446)
3110	HLXB9	Skeletal	1	0	1	Currarino_syndrome (394)
3115	HLA-DPB1	Unclassified	1	0	0	Beryllium_disease,_chronic (197)
3119	HLA-DQB1	Neurological	1	1	1	Creutzfeldt-Jakob_disease (388)
3123	HLA-DRB1	Immunological	1	2	1	Sarcoidosis (1349)
3141	HLCS	Metabolic	1	0	1	Holocarboxylase_synthetase_deficiency (688)
3145	HMBS	Metabolic	1	5	1	Porphyria (1249)
3155	HMGCL	Metabolic	1	1	1	HMG-CoA_deficiency (686)
3156	HMGCR	Metabolic	1	0	1	Statins (1442)
3158	HMGCS2	Metabolic	1	1	1	HMG-CoA_deficiency (686)
3162	HMOX1	Metabolic	1	0	1	Heme_oxygenase-1_deficiency (654)
3172	HNF4A	Endocrine	2	27	1	Diabetes_mellitus (427), MODY (1020)
3176	HNMT	Respiratory	1	12	1	Asthma (153)
3198	HOXA1	Neurological	2	0	1	Athabaskan_brainstem_dysgenesis_syndrome (162), Bosley-Salih-Alorainy_syndrome (220)
3207	HOXA11	multiple	1	0	1	Radioulnar_synostosis_with_amegakaryocytic_thrombocytopenia (1301)
3209	HOXA13	multiple	2	0	1	Guttmacher_syndrome (630), Hand-foot-uterus_syndrome (638)
3233	HOXD4	Cancer	1	36	1	Leukemia (886)
3236	HOXD10	Grey	2	17	2	Charcot-Marie-Tooth_disease (301), Vertical_talus (1559)
3239	HOXD13	Skeletal	2	5	1	Brachydactyly (224), Synpolydactyly (1470)
3240	HP	Hematological	2	0	1	Anhaptoglobinemia (118), Hypohaptoglobinemia (775)
3242	HPD	Metabolic	1	2	1	Tyrosinemia (1540)
3251	HPRT1	Metabolic	2	0	1	HPRT-related_gout (704), Lesch-Nyhan_syndrome, (884)
3257	HPS1	multiple	1	6	1	Hermansky-Pudlak_syndrome (670)
3265	HRAS	Grey	3	13	2	Bladder_cancer (207), Costello_syndrome (369), Thyroid_carcinoma (1503)
3273	HRG	Hematological	1	8	1	Thrombophilia (1497)
3284	HSD3B2	Metabolic	1	0	1	3-beta-hydroxysteroid_dehydrogenase,_type_II,_deficiency (5)

3290	HSD11B1	Metabolic	1	1	1	Cortisone_reductase_deficiency (368)
3291	HSD11B2	Grey	2	11	2	Apparent_mineralocorticoid_excess,_hypertension_due_to (137), Hypertension (752)
3293	HSD17B3	Developmental	1	1	1	Pseudohermaphroditism,_male (1279)
3295	HSD17B4	Metabolic	1	0	1	D-bifunctional_protein_deficiency (405)
3299	HSF4	Ophthamological	1	14	1	Cataract (277)
3315	HSPB1	Neurological	2	21	1	Charcot-Marie-Tooth_disease (301), Neuropathy (1099)
3329	HSPD1	Neurological	1	8	1	Spastic_ataxia/paraplegia (1418)
3339	HSPG2	Grey	2	0	2	Dyssegmental_dysplasia,_Silverman-Handmaker_type (461), Schwartz-Jampel_syndrome,_type_1 (1361)
3356	HTR2A	Grey	5	10	2	Alcohol_dependence (74), Anorexia_nervosa (124), Obsessive-compulsive_disorder (1128), Schizophrenia (1359), Seasonal_affective_disorder (1367)
3373	HYAL1	Metabolic	1	6	1	Mucopolysaccharidosis (1033)
3383	ICAM1	Immunological	1	4	1	Malaria (940)
3386	ICAM4	Hematological	1	22	1	Blood_group (212)
3423	IDS	Metabolic	1	6	1	Mucopolysaccharidosis (1033)
3425	IDUA	Metabolic	1	6	1	Mucopolysaccharidosis (1033)
3426	IF	Immunological	1	12	1	Complementary_component_deficiency (240)
3458	IFNG	Grey	4	7	4	AIDS (70), Aplastic_anemia (133), Tuberous_sclerosis (1534), Tuberculosis (1533)
3459	IFNGR1	Grey	4	5	2	BCG_infection (191), H._pylori_infection (705), Mycobacterial_infection (1043), Tuberculosis (1533)
3460	IFNGR2	Immunological	1	4	1	Mycobacterial_infection (1043)
3476	IGBP1	Neurological	1	0	1	Corpus_callosum,_agenesis_of,_with_mental_retardation,_ocular_coloboma_and_micrognathia (366)
3479	IGF1	Developmental	1	0	1	Growth_retardation (2628)
3480	IGF1R	Developmental	1	1	1	Intrauterine_and_postnatal_growth_retardation (819)
3481	IGF2	Developmental	1	1	1	Intrauterine_and_postnatal_growth_retardation (819)
3482	IGF2R	Cancer	1	7	1	Hepatic_adenoma (668)
3483	IGFALS	Endocrine	1	0	1	Acid-labile_subunit,_deficiency_of (30)
3501	IGHG2	Immunological	1	0	1	IgG2_deficiency (798)
3507	IGHM	Hematological	1	3	1	Agammaglobulinemia (63)
3508	IGHMBP2	Muscular	1	6	1	Spinal_muscular_atrophy (1426)
3514	IGKC	Immunological	1	0	1	Kappa_light_chain_deficiency (839)
3543	IGLL1	Hematological	1	3	1	Agammaglobulinemia (63)
3549	IHH	Skeletal	2	4	1	Acrocapitofemoral_dysplasia (33), Brachydactyly (224)
3553	IL1B	Cancer	1	9	1	Gastric_cancer (572)
3557	IL1RN	Cancer	1	9	1	Gastric_cancer (572)
3559	IL2RA	Immunological	1	0	1	Interleukin-2_receptor,_alpha_chain,_deficiency_of (816)
3561	IL2RG	Immunological	2	8	1	Combined_immunodeficiency (4350), Severe_combined_immunodeficiency (1383)
3566	IL4R	Immunological	1	4	1	Atopy (164)
3569	IL6	Immunological	1	0	1	Kaposi_sarcoma (838)
3575	IL7R	Immunological	1	7	1	Severe_combined_immunodeficiency (1383)
3586	IL10	Grey	3	10	2	Graft-versus-host_disease (623), HIV (684), Rheumatoid_arthritis (1324)
3593	IL12B	Respiratory	1	12	1	Asthma (153)
3594	IL12RB1	Immunological	1	4	1	Mycobacterial_infection (1043)
3596	IL13	Grey	2	12	2	Allergic_rhinitis (83), Asthma (153)
3614	IMPDH1	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
3621	ING1	Cancer	1	2	1	Squamous_cell_carcinoma (1437)
3630	INS	Endocrine	2	5	1	Hyperproinsulinemia (748), MODY (1020)
3640	INSL3	Renal	1	1	1	Cryptorchidism (392)
3643	INSR	Grey	3	26	3	Diabetes_mellitus (427), Leprechaunism (881), Rabson-Mendenhall_syndrome (1298)
3651	IPF1	Grey	3	27	2	Diabetes_mellitus (427), MODY (1020), Pancreatic_agenesis (3178)
3655	ITGA6	Dermatological	1	10	1	Epidermolysis_bullosa (493)

3659	IRF1	Grey	5	15	3	Gastric_cancer (572), Macrocytic_anemia (935), Myelodysplastic_syndrome (1045), Myelogenous_leukemia (1047), Nonsmall_cell_lung_cancer (1115)
3662	IRF4	Cancer	1	1	1	Multiple_myeloma (9037)
3664	IRF6	Grey	3	0	3	Orofacial_cleft (1147), Popliteal_pterygium_syndrome (1246), van_der_Woude_syndrome (3554)
3667	IRS1	Endocrine	1	26	1	Diabetes_mellitus (427)
3674	ITGA2B	Hematological	1	1	1	Glanzmann_thrombasthenia (588)
3679	ITGA7	Muscular	1	9	1	Myopathy (1059)
3689	ITGB2	Immunological	1	0	1	Leukocyte_adhesion_deficiency (889)
3690	ITGB3	Hematological	1	1	1	Glanzmann_thrombasthenia (588)
3691	ITGB4	Dermatological	1	10	1	Epidermolysis_bullosa (493)
3700	ITIH4	Metabolic	1	7	1	Hypercholesterolemia (724)
3704	ITPA	Metabolic	1	0	1	Inosine_triphosphatase_deficiency (810)
3712	IVD	Metabolic	1	0	1	Isovaleric_acidemia (826)
3717	JAK2	Hematological	3	2	1	Myelofibrosis,_idiopathic (1046), Polycythemia (1242), Thrombocythemia (1493)
3718	JAK3	Immunological	1	0	1	SCID (1362)
3728	JUP	multiple	1	0	1	Naxos_disease (1077)
3730	KAL1	multiple	1	1	1	Kallmann_syndrome (836)
3736	KCNA1	Neurological	1	1	1	Episodic_ataxia (497)
3753	KCNE1	Grey	2	6	2	Jervell_and_Lange-Nielsen_syndrome (830), Long_QT_syndrome (912)
3757	KCNH2	Cardiovascular	2	6	1	Acquired_long_QT_syndrome (31), Long_QT_syndrome (912)
3758	KCNJ1	multiple	1	4	1	Bartter_syndrome (187)
3759	KCNJ2	Cardiovascular	1	6	1	Long_QT_syndrome (912)
3767	KCNJ11	Grey	2	26	2	Diabetes_mellitus (427), Persistent_hyperinsulinemic_hypoglycemia_of_infancy (3212)
3778	KCNMA1	Neurological	1	1	1	Generalized_epilepsy (578)
3779	KCNMB1	Cardiovascular	1	11	1	Hypertension (752)
3784	KCNQ1	Grey	3	9	2	Atrial_fibrillation (166), Jervell_and_Lange-Nielsen_syndrome (830), Long_QT_syndrome (912)
3785	KCNQ2	Neurological	2	16	1	Epilepsy (495), Myokymia_with_neonatal_epilepsy (1057)
3786	KCNQ3	Neurological	1	16	1	Epilepsy (495)
3791	KDR	Cancer	1	1	1	Hemangioma (651)
3792	KEL	Hematological	1	22	1	Blood_group (212)
3795	KHK	Metabolic	1	0	1	Fructosuria (557)
3798	KIF5A	Neurological	1	8	1	Spastic_ataxia/paraplegia (1418)
3811	KIR3DL1	Immunological	1	2	1	AIDS (70)
3815	KIT	Grey	6	38	3	Gastrointestinal_stromal_tumor (574), Germ_cell_tumor (580), Leukemia (886), Mast_cell_leukemia (958), Mastocytosis_with_associated_hematologic_disorder (959), Piebaldism (1226)
3816	KLK1	Renal	1	0	1	Kallikrein,_decreased_urinary_activity_of (835)
3818	KLKB1	Hematological	1	0	1	Prekallikrein_deficiency (1259)
3827	KNG1	Hematological	3	0	1	Fitzgerald_factor_deficiency (542), High-molecular-weight_kininogen_deficiency (679), Kininogen_deficiency (851)
3845	KRAS	Cancer	6	65	1	Bladder_cancer (207), Breast_cancer (228), Leukemia (886), Lung_cancer (918), Pancreatic_cancer (1178), Stomach_cancer (1449)
3848	KRT1	Dermatological	5	10	1	Cyclic_ichthyosis_with_epidermolytic_hyperkeratosis (396), Epidermolytic_hyperkeratosis (494), Ichthyosis (794), Keratosis_palmoplantaria_striata (847), Unna-Thost_disease,_nonepidermolytic (1545)
3849	KRT2A	Dermatological	1	6	1	Ichthyosis (794)
3850	KRT3	Ophthamological	1	1	1	Meesmann_corneal_dystrophy (971)
3851	KRT4	Connective_tissue_disorder	1	1	1	White_sponge_nevus (1592)
3852	KRT5	Dermatological	1	10	1	Epidermolysis_bullosa (493)
3853	KRT6A	Dermatological	1	3	1	Pachyonychia_congenita (1172)
3854	KRT6B	Dermatological	1	3	1	Pachyonychia_congenita (1172)

3856	KRT8	Gastrointestinal	1	2	1	Cirrhosis (334)
3857	KRT9	Dermatological	1	2	1	Epidermolytic_hyperkeratosis (494)
3858	KRT10	Dermatological	3	7	1	Epidermolytic_hyperkeratosis (494), Ichthyosis (794), Nevus,_epidermal,_epidermolytic_hyperkeratotic_type (1104)
3859	KRT12	Ophthamological	1	1	1	Meesmann_corneal_dystrophy (971)
3860	KRT13	Connective_tissue_disorder	1	1	1	White_sponge_nevus (1592)
3861	KRT14	Dermatological	1	10	1	Epidermolysis_bullosa (493)
3868	KRT16	Dermatological	2	3	1	Pachyonychia_congenita (1172), Palmoplantar_keratoderma (1176)
3872	KRT17	Dermatological	2	3	1	Pachyonychia_congenita (1172), Steatocystoma_multiplex (1444)
3875	KRT18	Gastrointestinal	1	2	1	Cirrhosis (334)
3887	KRTHB1	Dermatological	1	1	1	Monilethrix (1024)
3892	KRTHB6	Dermatological	1	1	1	Monilethrix (1024)
3897	L1CAM	Grey	3	0	2	CRASH_syndrome (384), Hydrocephalus (712), MASA_syndrome (955)
3908	LAMA2	Muscular	1	17	1	Muscular_dystrophy (1040)
3909	LAMA3	Grey	2	10	2	Epidermolysis_bullosa (493), Laryngoonychocutaneous_syndrome (868)
3913	LAMB2	Ophthamological	1	0	1	Microcoria-congenital_nephrosis_syndrome (1009)
3914	LAMB3	Dermatological	1	10	1	Epidermolysis_bullosa (493)
3918	LAMC2	Dermatological	1	10	1	Epidermolysis_bullosa (493)
3920	LAMP2	Metabolic	1	8	1	Glycogen_storage_disease (610)
3930	LBR	Grey	2	0	2	Greenberg_dysplasia (625), Pelger-Huet_anomaly (1199)
3931	LCAT	Metabolic	2	0	1	Fish-eye_disease (540), Norum_disease (1118)
3938	LCT	Metabolic	1	1	1	Hypolactasia,_adult_type (777)
3939	LDHA	Metabolic	1	0	1	Exertional_myoglobinuria_due_to_deficiency_of_LDH-A (512)
3945	LDHB	Metabolic	1	0	1	Lactate_dehydrogenase-B_deficiency (858)
3949	LDLR	Metabolic	1	7	1	Hypercholesterolemia (724)
3952	LEP	Nutritional	1	20	1	Obesity (1126)
3953	LEPR	Nutritional	1	20	1	Obesity (1126)
3957	LGALS2	Cardiovascular	1	9	1	Myocardial_infarction (1054)
3972	LHB	Endocrine	1	0	1	Hypogonadism,_hypergonadotropic (773)
3973	LHCGR	Grey	5	4	4	Hypogonadotropic_hypogonadism (774), Leydig_cell_adenoma (894), Micropenis (1011), Precocious_puberty,_male (1256), Pseudohermaphroditism,_male (1279)
3977	LIFR	multiple	1	0	1	Stuve-Wiedemann_syndrome/Schwartz-Jampel_type_2_syndrome (1455)
3978	LIG1	multiple	1	0	1	DNA_ligase_I_deficiency (2440)
3981	LIG4	Grey	2	1	2	LIG4_syndrome (899), Multiple_myeloma (9037)
3982	LIM2	Ophthamological	1	14	1	Cataract (277)
3988	LIPA	Metabolic	2	0	1	Cholesteryl_ester_storage_disease (313), Wolman_disease (1604)
3990	LIPC	Metabolic	1	0	1	Hepatic_lipase_deficiency (4666)
3998	LMAN1	Hematological	1	0	1	Combined_factor_V_and_VIII_deficiency (350)
4000	LMNA	Grey	3	29	3	Cardiomyopathy (268), Emery-Dreifuss_muscular_dystrophy (474), Lipodystrophy (902)
4010	LMX1B	multiple	1	0	1	Nail-patella_syndrome (1070)
4014	LOR	Grey	2	3	2	Erythrokeratoderma (507), Vohwinkel_syndrome (1571)
4018	LPA	Metabolic	1	0	1	LPA_deficiency,_congenital (916)
4023	LPL	Metabolic	3	0	1	Chylomicronemia_syndrome,_familial (329), Combined_hyperlipemia (2350), Lipoprotein_lipase_deficiency (906)
4026	LPP	Cancer	2	38	1	Leukemia (886), Lipoma (904)
4041	LRP5	Grey	7	10	2	Bone_mineral_density_variability (217), Exudative_vitreoretinopathy (515), Hyperostosis,_endosteal (741), Osteopetrosis (1161), Osteoporosis (1163), Osteoporosis-pseudoglioma_syndrome (1164), van_Buchem_disease (1554)
4049	LTA	Cardiovascular	1	9	1	Myocardial_infarction (1054)
4059	LU	Hematological	1	22	1	Blood_group (212)

4068	SH2D1A	Cancer	1	9	1	Lymphoma (925)
4069	LYZ	Neurological	1	5	1	Amyloidosis (103)
4070	TACSTD2	Ophthamological	1	6	1	Corneal_dystrophy (362)
4089	SMAD4	Cancer	3	9	1	Juvenile_polyposis/hereditary_hemorrhagic_telangiectasia_syndrome (833), Pancreatic_cancer (1178), Polyposis (1245)
4125	MAN2B1	Metabolic	1	1	1	Mannosidosis (947)
4126	MANBA	Metabolic	1	1	1	Mannosidosis (947)
4128	MAOA	Unclassified	1	0	0	Brunner_syndrome (235)
4137	MAPT	Neurological	4	5	1	Dementia (412), Pallidopontonigral_degeneration (1174), Supranuclear_palsy (1462), Tauopathy_and_respiratory_failure (1476)
4143	MAT1A	Metabolic	2	1	1	Hypermethioninemia (739), Methionine_adenosyltransferase_deficiency,_autosomal_recessive (1001)
4148	MATN3	Grey	3	12	3	Epiphyseal_dysplasia (496), Osteoarthritis (1154), Spondyloepiphyseal_dysplasia (1435)
4153	MBL2	Immunological	2	0	1	Chronic_infections,_due_to_opsonin_defect (2327), Meningococcal_disease (987)
4157	MC1R	Grey	4	2	2	Analgesia_from_kappa-opioid_receptor_agonist,_female-specific (107), Ocular_albinism (1130), Red_hair/fair_skin (1306), UV-induced_skin_damage (1553)
4158	MC2R	Endocrine	1	1	1	Glucocorticoid_deficiency (598)
4159	MC3R	Nutritional	1	20	1	Obesity (1126)
4160	MC4R	Nutritional	1	20	1	Obesity (1126)
4163	MCC	Cancer	1	33	1	Colon_cancer (346)
4166	CHST6	Ophthamological	1	4	1	Macular_dystrophy (2937)
4175	MCM6	Grey	2	1	2	Hypolactasia,_adult_type (777), Tall_stature (1472)
4193	MDM2	Cancer	1	0	1	Accelerated_tumor_formation (22)
4197	MDS1	Muscular	1	2	1	Myelodysplastic_syndrome (1045)
4200	ME2	Neurological	1	16	1	Epilepsy (495)
4204	MECP2	Grey	5	27	3	Angelman_syndrome (113), Autism (173), Mental_retardation (990), PPM-X_syndrome (1253), Rett_syndrome (1321)
4205	MEF2A	Cardiovascular	1	5	1	Coronary_artery_disease (365)
4210	MEFV	Immunological	1	0	1	Familial_Mediterranean_fever (522)
4221	MEN1	Grey	8	5	2	Adrenal_adenoma (2053), Angiofibroma,_sporadic (115), Carcinoid_tumor_of_lung (2265), Hyperparathyroidism (743), Lipoma (904), Multiple_endocrine_neoplasia (1037), Parathyroid_adenoma (1189), Prolactinoma,_hyperparathyroidism,_carcinoid_syndrome (1267)
4233	MET	Cancer	2	13	1	Hepatic_adenoma (668), Renal_cell_carcinoma (1308)
4247	MGAT2	Metabolic	1	2	1	Carbohydrate-deficient_glycoprotein_syndrome (263)
4256	MGP	multiple	1	0	1	Keutel_syndrome (849)
4261	CIITA	Grey	3	12	3	Bare_lymphocyte_syndrome (184), Multiple_sclerosis (9038), Rheumatoid_arthritis (1324)
4281	MID1	multiple	1	0	1	Opitz_G_syndrome (1142)
4282	MIF	Connective_tissue_disorder	1	7	1	Rheumatoid_arthritis (1324)
4284	MIP	Ophthamological	1	14	1	Cataract (277)
4286	MITF	multiple	2	3	1	Tietz_syndrome (1508), Waardenburg_syndrome (1574)
4287	ATXN3	Neurological	1	0	1	Machado-Joseph_disease (933)
4292	MLH1	Cancer	4	33	1	Cafe-au-lait_spots (252), Colon_cancer (346), Muir-Torre_syndrome (1035), Turcot_syndrome (1536)
4306	NR3C2	Grey	2	16	2	Hypertension (752), Pseudohypoaldosteronism (1281)
4312	MMP1	Respiratory	1	0	1	COPD,_rate_of_decline_of_lung_function_in (360)
4313	MMP2	Bone	1	1	1	Osteolysis (1157)
4314	MMP3	Cardiovascular	1	5	1	Coronary_artery_disease (365)
4329	ALDH6A1	Metabolic	1	0	1	Methylmalonate_semialdehyde_dehydrogenase_deficiency (1003)
4330	MN1	Cancer	1	3	1	Meningioma (986)
4337	MOCS1	Metabolic	1	2	1	Molybdenum_cofactor_deficiency (1023)
4338	MOCS2	Metabolic	1	2	1	Molybdenum_cofactor_deficiency (1023)

4351	MPI	Metabolic	1	2	1	Carbohydrate-deficient_glycoprotein_syndrome (263)
4352	MPL	Hematological	1	3	1	Thrombocytopenia (1494)
4353	MPO	Grey	2	11	2	Alzheimer_disease (98), Myeloperoxidase_deficiency (1051)
4359	MPZ	Grey	4	22	2	Charcot-Marie-Tooth_disease (301), Dejerine-Sottas_disease (410), Neuropathy (1099), Roussy-Levy_syndrome (1339)
4361	MRE11A	Immunological	1	1	1	Ataxia-telangiectasia (157)
4436	MSH2	Cancer	8	48	1	Cafe-au-lait_spots (252), Colon_cancer (346), Glioblastoma (590), Lymphoma (925), Lynch_cancer_family_syndrome_II (930), Muir-Torre_syndrome (1035), Neurofibromatosis (1097), Ovarian_cancer (1170)
4437	MSH3	Cancer	1	3	1	Endometrial_carcinoma (479)
4481	MSR1	Cancer	1	11	1	Prostate_cancer (1272)
4487	MSX1	Grey	3	3	2	Cleft_palate (336), Hypodontia (769), Witkop_syndrome (1599)
4488	MSX2	Skeletal	2	2	1	Craniosynostosis (383), Parietal_foramina (1190)
4522	MTHFD1	Developmental	2	2	1	Neural_tube_defects,_maternal_risk_of (1090), Spina_bifida (1425)
4524	MTHFR	Metabolic	1	1	1	Homocystinuria (698)
4534	MTM1	Muscular	1	0	1	Myotubular_myopathy (1064)
4547	MTP	Metabolic	1	1	1	Abetalipoproteinemia (17)
4548	MTR	Grey	3	2	3	Down_syndrome (445), Methylcobalamin_deficiency,_cblG_type (1002), Spina_bifida (1425)
4552	MTRR	Grey	2	2	2	Homocystinuria-megaloblastic_anemia,_cbl_E_type (699), Spina_bifida (1425)
4591	TRIM37	multiple	1	0	1	Mulibrey_nanism (1036)
4594	MUT	Metabolic	1	2	1	Methylmalonic_aciduria (1004)
4595	MUTYH	Cancer	3	42	1	Adenomas (47), Colon_cancer (346), Gastric_cancer (572)
4598	MVK	Grey	2	0	2	Hyper-IgD_syndrome (728), Mevalonicaciduria (1005)
4601	MXI1	Cancer	2	11	1	Neurofibrosarcoma (1098), Prostate_cancer (1272)
4607	MYBPC3	Cardiovascular	1	24	1	Cardiomyopathy (268)
4609	MYC	Cancer	1	0	1	Burkitt_lymphoma (237)
4613	MYCN	multiple	1	0	1	Feingold_syndrome (531)
4618	MYF6	Muscular	2	10	1	Becker_muscular_dystrophy (193), Myopathy (1059)
4620	MYH2	Muscular	1	2	1	Inclusion_body_myopathy (805)
4624	MYH6	Cardiovascular	2	28	1	Atrial_fibrillation (166), Cardiomyopathy (268)
4625	MYH7	Grey	3	32	2	Cardiomyopathy (268), Central_core_disease (2287), Myopathy (1059)
4626	MYH8	multiple	2	1	1	Carney_complex (269), Trismus-pseudocomptodactyly_syndrome (1528)
4627	MYH9	Grey	5	40	3	Deafness (406), Epstein_syndrome (500), Fechtner_syndrome (530), May-Hegglin_anomaly (961), Sebastian_syndrome (1368)
4633	MYL2	Cardiovascular	1	24	1	Cardiomyopathy (268)
4634	MYL3	Cardiovascular	1	24	1	Cardiomyopathy (268)
4640	MYO1A	Ear,Nose,Throat	1	40	1	Deafness (406)
4644	MYO5A	Dermatological	1	2	1	Griscelli_syndrome (627)
4646	MYO6	Ear,Nose,Throat	1	40	1	Deafness (406)
4647	MYO7A	Grey	2	44	2	Deafness (406), Usher_syndrome (1551)
4653	MYOC	Ophthamological	1	3	1	Glaucoma (589)
4668	NAGA	Metabolic	2	0	1	Kanzaki_disease (837), Schindler_disease (1355)
4669	NAGLU	Metabolic	1	1	1	Sanfilippo_syndrome (1348)
4683	NBN	Grey	2	36	2	Leukemia (886), Nijmegen_breakage_syndrome (1111)
4687	NCF1	Immunological	1	3	1	Chronic_granulomatous_disease (327)
4688	NCF2	Immunological	1	3	1	Chronic_granulomatous_disease (327)
4692	NDN	multiple	1	1	1	Prader-Willi_syndrome (1254)
4693	NDP	Grey	3	2	2	Coats_disease (338), Exudative_vitreoretinopathy (515), Norrie_disease (1117)
4703	NEB	Muscular	1	3	1	Nemaline_myopathy (1078)
4719	NDUFS1	multiple	1	6	1	Mitochondrial_complex_deficiency (1016)

4720	NDUFS2	multiple	1	6	1	Mitochondrial_complex_deficiency (1016)
4722	NDUFS3	Neurological	1	11	1	Leigh_syndrome (877)
4723	NDUFV1	Grey	3	15	2	Alexander_disease (79), Leigh_syndrome (877), Mitochondrial_complex_deficiency (1016)
4724	NDUFS4	Grey	2	14	2	Leigh_syndrome (877), Mitochondrial_complex_deficiency (1016)
4726	NDUFS6	multiple	1	1	1	Complex_mitochondrial_respiratory_chain,_deficiency_of (351)
4728	NDUFS8	Neurological	1	11	1	Leigh_syndrome (877)
4729	NDUFV2	Neurological	1	10	1	Parkinson_disease (1192)
4744	NEFH	Neurological	1	5	1	Amyotrophic_lateral_sclerosis (104)
4747	NEFL	Neurological	1	17	1	Charcot-Marie-Tooth_disease (301)
4758	NEU1	Metabolic	1	1	1	Sialidosis (1391)
4760	NEUROD1	Endocrine	1	26	1	Diabetes_mellitus (427)
4763	NF1	Cancer	5	38	1	Leukemia (886), Neurofibromatosis (1095), Neurofibromatosis-Noonan_syndrome (1096), Neurofibromatosis (1097), Watson_syndrome (1581)
4771	NF2	Cancer	3	5	1	Meningioma (986), Neurofibromatosis (1097), Schwannomatosis (1360)
4792	NFKBIA	Dermatological	1	7	1	Ectodermal_dysplasia (466)
4795	NFKBIL1	Connective_tissue_disorder	1	7	1	Rheumatoid_arthritis (1324)
4803	NGFB	Neurological	1	7	1	Neuropathy (1099)
4810	NHS	Ophthamological	1	0	1	Nance-Horan_syndrome (1071)
4830	NME1	Cancer	1	1	1	Neuroblastoma (1091)
4838	NODAL	Developmental	1	1	1	Situs_ambiguus (1399)
4842	NOS1	Developmental	1	0	1	Pyloric_stenosis,_infantile_hypertrophic (1294)
4843	NOS2A	Immunological	1	4	1	Malaria (940)
4846	NOS3	Grey	4	22	2	Alzheimer_disease (98), Coronary_spasms (2365), Hypertension (752), Placental_abruption (1233)
4854	NOTCH3	Cardiovascular	1	0	1	Cerebral_arteriopathy_with_subcortical_infarcts_and_leukoencephalopathy (2291)
4860	NP	Immunological	1	0	1	Nucleoside_phosphorylase_deficiency,_immunodeficiency_due_to (1122)
4864	NPC1	Metabolic	1	2	1	Niemann-Pick_disease (1107)
4867	NPHP1	Grey	3	5	2	Joubert_syndrome (831), Nephronophthisis (1082), Senior-Loken_syndrome (1375)
4868	NPHS1	Renal	1	1	1	Nephrotic_syndrome (1087)
4869	NPM1	Cancer	1	36	1	Leukemia (886)
4882	NPR2	Skeletal	1	1	1	Acromesomelic_dysplasia (39)
4891	SLC11A2	Hematological	1	8	1	Anemia (110)
4893	NRAS	Cancer	1	33	1	Colon_cancer (346)
4901	NRL	Ophthamological	2	32	1	Retinal_cone_dsytrophy (1315), Retinitis_pigmentosa (1316)
4902	NRTN	Gastrointestinal	1	6	1	Hirschsprung_disease (681)
4914	NTRK1	Grey	2	1	2	Insensitivity_to_pain (811), Medullary_thyroid_carcinoma (2969)
4915	NTRK2	Nutritional	1	20	1	Obesity (1126)
4920	ROR2	Grey	2	4	2	Brachydactyly (224), Robinow_syndrome,_autosomal_recessive (1335)
4926	NUMA1	Cancer	1	36	1	Leukemia (886)
4929	NR4A2	Neurological	1	10	1	Parkinson_disease (1192)
4935	GPR143	Ophthamological	1	2	1	Ocular_albinism (1130)
4938	OAS1	Immunological	1	1	1	Viral_infection (1562)
4942	OAT	Metabolic	1	0	1	Gyrate_atrophy_of_choroid_and_retina_with_ornithinemia,_B6_responsive_or_unresponsive (632)
4948	OCA2	Dermatological	1	2	1	Albinism (72)
4952	OCRL	Grey	2	1	2	Dent_disease (416), Lowe_syndrome (914)
4953	ODC1	Cancer	1	33	1	Colon_cancer (346)
4968	OGG1	Cancer	1	6	1	Renal_cell_carcinoma (1308)
4973	OLR1	Cardiovascular	1	9	1	Myocardial_infarction (1054)
4976	OPA1	Ophthamological	2	4	1	Glaucoma (589), Optic_atrophy (1144)
4978	OPCML	Cancer	1	0	1	Epithelial_ovarian_cancer,_somatic (498)

4982	TNFRSF11B	Bone	1	2	1	Paget_disease (1173)
4983	OPHN1	Neurological	1	23	1	Mental_retardation (990)
4988	OPRM1	Neurological	1	0	1	Response_to_morphine-6-glucuronide (1311)
4990	SIX6	Ophthamological	1	2	1	Microphthalmia (1012)
5002	SLC22A18	Cancer	3	23	1	Breast_cancer (228), Lung_cancer (918), Rhabdomyosarcoma (1323)
5009	OTC	Metabolic	1	0	1	Ornithine_transcarbamylase_deficiency (1146)
5019	OXCT1	Metabolic	1	0	1	Ketoacidosis (848)
5023	P2RX1	Hematological	1	1	1	Bleeding_disorder (209)
5027	P2RX7	Cancer	1	36	1	Leukemia (886)
5048	PAFAH1B1	Neurological	2	3	1	Lissencephaly (908), Subcortical_laminar_heterotopia (1456)
5053	PAH	Metabolic	2	3	1	Hyperphenylalaninemia (746), Phenylketonuria (1216)
5063	PAK3	Neurological	1	23	1	Mental_retardation (990)
5071	PARK2	Grey	3	13	3	Adenocarcinoma (45), Leprosy (882), Parkinson_disease (1192)
5076	PAX2	Grey	2	0	2	Optic_nerve_coloboma_with_renal_disease (3144), Renal_hypoplasia,_isolated (5308)
5077	PAX3	Grey	3	6	2	Craniofacial-deafness-hand_syndrome (378), Rhabdomyosarcoma (1323), Waardenburg_syndrome (1574)
5080	PAX6	Grey	10	17	3	Aniridia,_type_II (119), Cataract (277), Coloboma,_ocular (345), Ectopia (467), Eye_anomalies (516), Foveal_hypoplasia (548), Keratitis (842), Morning_glory_disc_anomaly (1026), Optic_nerve_hypoplasia/aplasia (5144), Peters_anomaly (1213)
5081	PAX7	Cancer	1	3	1	Rhabdomyosarcoma (1323)
5083	PAX9	Skeletal	2	1	1	Hypodontia (769), Oligodontia (1139)
5091	PC	Metabolic	1	2	1	Pyruvate_dehydrogenase_deficiency (1297)
5092	PCBD1	Metabolic	1	1	1	Hyperphenylalaninemia (746)
5095	PCCA	Metabolic	1	1	1	Propionicacidemia (1271)
5096	PCCB	Metabolic	1	1	1	Propionicacidemia (1271)
5108	PCM1	Cancer	1	10	1	Thyroid_carcinoma (1503)
5122	PCSK1	Nutritional	1	20	1	Obesity (1126)
5133	PDCD1	Immunological	1	3	1	Systemic_lupus_erythematosus (1471)
5144	PDE4D	Cardiovascular	1	1	1	Stroke (1454)
5145	PDE6A	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
5155	PDGFB	Cancer	3	3	1	Dermatofibrosarcoma_protuberans (422), Giant-cell_fibroblastoma (583), Meningioma (986)
5156	PDGFRA	Grey	2	1	2	Gastrointestinal_stromal_tumor (574), Hypereosinophilic_syndrome (726)
5157	PDGFRL	Cancer	2	38	1	Colon_cancer (346), Hepatic_adenoma (668)
5158	PDE6B	Ophthamological	2	32	1	Night_blindness (1110), Retinitis_pigmentosa (1316)
5159	PDGFRB	Cancer	2	0	1	Myelomonocytic_leukemia,_chronic (1050), Myeloproliferative_disorder (1052)
5160	PDHA1	Grey	2	13	2	Leigh_syndrome (877), Pyruvate_dehydrogenase_deficiency (1297)
5162	PDHB	Metabolic	1	2	1	Pyruvate_dehydrogenase_deficiency (1297)
5167	ENPP1	Grey	3	45	3	Diabetes_mellitus (427), Obesity (1126), Ossification_of_the_posterior_longitudinal_spinal_ligaments (1153)
5172	SLC26A4	Ear,Nose,Throat	3	40	1	Deafness (406), Enlarged_vestibular_aqueduct (484), Pendred_syndrome (1204)
5184	PEPD	Connective_tissue_disorder	1	0	1	Prolidase_deficiency (1268)
5189	PEX1	Grey	3	13	2	Adrenoleukodystrophy (56), Refsum_disease (1307), Zellweger_syndrome (1615)
5190	PEX6	multiple	1	1	1	Peroxisomal_biogenesis_disorder (1210)
5191	PEX7	Grey	2	5	2	Refsum_disease (1307), Rhizomelic_chondrodysplasia_punctata (1325)
5192	PEX10	Grey	2	11	2	Adrenoleukodystrophy (56), Zellweger_syndrome (1615)
5193	PEX12	multiple	1	1	1	Peroxisomal_biogenesis_disorder (1210)
5194	PEX13	Grey	2	11	2	Adrenoleukodystrophy (56), Zellweger_syndrome (1615)
5195	PEX14	multiple	1	10	1	Zellweger_syndrome (1615)
5199	PFC	Immunological	1	0	1	Properdin_deficiency (1270)
5205	ATP8B1	Gastrointestinal	1	3	1	Cholestasis (310)
5213	PFKM	Metabolic	1	8	1	Glycogen_storage_disease (610)

5224	PGAM2	Muscular	1	9	1	Myopathy (1059)
5230	PGK1	Grey	2	9	2	Hemolytic_anemia (660), Myoglobinuria/hemolysis_due_to_PGK_deficiency (1056)
5243	ABCB1	Unclassified	1	0	0	Colchicine_resistance (343)
5244	ABCB4	Gastrointestinal	2	3	1	Cholelithiasis (309), Cholestasis (310)
5245	PHB	Cancer	1	18	1	Breast_cancer (228)
5251	PHEX	Metabolic	1	1	1	Hypophosphatemia (783)
5255	PHKA1	Muscular	1	1	1	Muscle_hypertrophy (1039)
5256	PHKA2	Metabolic	1	1	1	Glycogenosis (609)
5257	PHKB	Metabolic	1	0	1	Phosphorylase_kinase_deficiency_of_liver_and_muscle,_autosomal_recessive (1222)
5261	PHKG2	Metabolic	1	1	1	Glycogenosis (609)
5264	PHYH	Neurological	1	4	1	Refsum_disease (1307)
5265	SERPINA1	Grey	2	1	2	Emphysema (475), Hemorrhagic_diathesis (664)
5274	SERPINI1	Neurological	1	1	1	Encephalopathy (476)
5277	PIGA	Hematological	1	0	1	Paroxysmal_nocturnal_hemoglobinuria (3195)
5290	PIK3CA	Cancer	3	51	1	Breast_cancer (228), Colon_cancer (346), Ovarian_cancer (1170)
5308	PITX2	Grey	3	1	2	Iridogoniodysgenesis (822), Rieger_syndrome (1331), Ring_dermoid_of_cornea (1332)
5309	PITX3	Ophthamological	2	17	1	Anterior_segment_anomalies_and_cataract (126), Cataract (277)
5310	PKD1	Renal	1	3	1	Polycystic_kidney_disease (1241)
5311	PKD2	Renal	1	3	1	Polycystic_kidney_disease (1241)
5313	PKLR	Hematological	1	8	1	Anemia (110)
5314	PKHD1	Renal	1	3	1	Polycystic_kidney_disease (1241)
5317	PKP1	Dermatological	1	7	1	Ectodermal_dysplasia (466)
5318	PKP2	Cardiovascular	1	2	1	Arrhythmogenic_right_ventricular_dysplasia (144)
5320	PLA2G2A	Cancer	1	33	1	Colon_cancer (346)
5324	PLAG1	Cancer	1	2	1	Adenomas (47)
5328	PLAU	Neurological	1	11	1	Alzheimer_disease (98)
5339	PLEC1	Grey	2	27	2	Epidermolysis_bullosa (493), Muscular_dystrophy (1040)
5340	PLG	Grey	2	0	2	Conjunctivitis,_ligneous (356), Plasminogen_deficiency (1235)
5345	SERPINF2	Hematological	1	0	1	Plasmin_inhibitor_deficiency (1234)
5350	PLN	Cardiovascular	1	24	1	Cardiomyopathy (268)
5351	PLOD1	Connective_tissue_disorder	2	8	1	Ehlers-Danlos_syndrome (470), Nevo_syndrome (1103)
5352	PLOD2	Unclassified	1	0	0	Bruck_syndrome (233)
5354	PLP1	Neurological	2	8	1	Pelizaeus-Merzbacher_disease (1200), Spastic_ataxia/paraplegia (1418)
5371	PML	Cancer	1	36	1	Leukemia (886)
5373	PMM2	Metabolic	1	2	1	Carbohydrate-deficient_glycoprotein_syndrome (263)
5376	PMP22	Grey	4	22	2	Charcot-Marie-Tooth_disease (301), Dejerine-Sottas_disease (410), Neuropathy (1099), Roussy-Levy_syndrome (1339)
5378	PMS1	Cancer	1	33	1	Colon_cancer (346)
5395	PMS2	Grey	3	33	2	Colon_cancer (346), Neuroectodermal_tumors (1093), Turcot_syndrome (1536)
5428	POLG	Grey	3	2	2	Alpers_syndrome (86), Progressive_external_ophthalmoplegia_with_mitochondrial_DNA_deletions (1265), Sensory_ataxic_neuropathy,_dysarthria,_and_ophthalmoparesis (1376)
5429	POLH	Dermatological	1	7	1	Xeroderma_pigmentosum (1608)
5443	POMC	Nutritional	1	20	1	Obesity (1126)
5444	PON1	Cardiovascular	1	5	1	Coronary_artery_disease (365)
5445	PON2	Cardiovascular	1	5	1	Coronary_artery_disease (365)
5447	POR	Grey	3	3	2	Adrenal_hyperplasia,_congenital (53), Antley-Bixler_syndrome (128), Disordered_steroidogenesis,_isolated (438)
5449	POU1F1	Endocrine	1	3	1	Pituitary_hormone_deficiency (5232)
5456	POU3F4	Ear,Nose,Throat	1	40	1	Deafness (406)

5459	POU4F3	Ear,Nose,Throat	1	40	1	Deafness (406)
5465	PPARA	Metabolic	1	0	1	Hyperapobetalipoproteinemia (719)
5468	PPARG	Grey	5	55	4	Diabetes_mellitus (427), Glioblastoma (590), Insulin_resistance (814), Lipodystrophy (902), Obesity (1126)
5476	PPGB	Metabolic	1	0	1	Galactosialidosis (568)
5498	PPOX	Metabolic	1	5	1	Porphyria (1249)
5506	PPP1R3A	Metabolic	1	2	1	Insulin_resistance (814)
5519	PPP2R1B	Cancer	1	3	1	Lung_cancer (918)
5521	PPP2R2B	Neurological	1	12	1	Spinocereballar_ataxia (1428)
5538	PPT1	Neurological	2	5	1	Ceroid_lipofuscinosis (295), Ceroid-lipofuscinosis (296)
5546	PRCC	Cancer	1	6	1	Renal_cell_carcinoma (1308)
5551	PRF1	Hematological	1	1	1	Hemophagocytic_lymphohistiocytosis (662)
5573	PRKAR1A	Grey	5	11	3	Adrenocortical_carcinoma (54), Carney_complex (269), Myxoma,_intracardiac (1066), Pigmented_adrenocortical_disease,_primary_isolated (3229), Thyroid_carcinoma (1503)
5578	PRKCA	Cancer	1	0	1	Pituitary_tumor,_invasive (7232)
5582	PRKCG	Neurological	1	12	1	Spinocereballar_ataxia (1428)
5589	PRKCSH	Gastrointestinal	1	1	1	Polycystic_liver_disease (3241)
5621	PRNP	Grey	5	5	2	Creutzfeldt-Jakob_disease (388), Gerstmann-Straussler_disease (581), Huntington_disease (708), Insomnia (812), Prion_disease_with_protracted_course (1263)
5624	PROC	Hematological	1	8	1	Thrombophilia (1497)
5625	PRODH	Grey	2	9	2	Hyperprolinemia (749), Schizophrenia (1359)
5626	PROP1	Endocrine	1	3	1	Pituitary_hormone_deficiency (5232)
5627	PROS1	Hematological	1	0	1	Protein_S_deficiency (1273)
5630	PRPH	Neurological	1	5	1	Amyotrophic_lateral_sclerosis (104)
5631	PRPS1	Metabolic	1	0	1	Phosphoribosyl_pyrophosphate_synthetase-related_gout (1221)
5644	PRSS1	Gastrointestinal	1	2	1	Pancreatitis (1179)
5649	RELN	Neurological	1	3	1	Lissencephaly (908)
5651	PRSS7	Gastrointestinal	1	0	1	Enterokinase_deficiency (487)
5660	PSAP	Grey	3	2	2	Combined_SAP_deficiency (8350), Gaucher_disease (575), Metachromatic_leukodystrophy (996)
5663	PSEN1	Neurological	3	16	1	Alzheimer_disease (98), Dementia (412), Pick_disease (1225)
5664	PSEN2	Neurological	1	11	1	Alzheimer_disease (98)
5723	PSPH	Metabolic	1	0	1	Phosphoserine_phosphatase_deficiency (1223)
5726	TAS2R38	Ear,Nose,Throat	1	0	1	Phenylthiocarbamide_tasting (1217)
5727	PTCH	Grey	3	7	3	Basal_cell_carcinoma (188), Basal_cell_nevus_syndrome (2188), Holoprosencephaly (689)
5728	PTEN	Grey	9	28	2	Bannayan-Riley-Ruvalcaba_syndrome (182), Cowden_disease (372), Endometrial_carcinoma (479), Lhermitte-Duclos_syndrome (895), Meningioma (986), Oligodendroglioma (1138), Prostate_cancer (1272), Thyroid_carcinoma (1503), VATER_association_with_hydrocephalus (1555)
5729	PTGDR	Respiratory	1	12	1	Asthma (153)
5740	PTGIS	Cardiovascular	1	11	1	Hypertension (752)
5741	PTH	Endocrine	1	1	1	Hypoparathyroidism (779)
5745	PTHR1	Grey	2	2	2	Enchondromatosis (477), Metaphyseal_chondrodysplasia (997)
5770	PTPN1	Metabolic	1	2	1	Insulin_resistance (814)
5781	PTPN11	Grey	3	36	3	Leopard_syndrome (880), Leukemia (886), Noonan_syndrome (1116)
5782	PTPN12	Cancer	1	33	1	Colon_cancer (346)
5788	PTPRC	Grey	2	8	2	Multiple_sclerosis (9038), Severe_combined_immunodeficiency (1383)
5795	PTPRJ	Cancer	1	33	1	Colon_cancer (346)
5805	PTS	Metabolic	1	2	1	Phenylketonuria (1216)
5818	PVRL1	Grey	3	9	3	Cleft_palate (336), Ectodermal_dysplasia (466), Zlotogora-Ogur_syndrome (1617)
5824	PEX19	multiple	1	10	1	Zellweger_syndrome (1615)
5825	ABCD3	multiple	1	10	1	Zellweger_syndrome (1615)
5828	PXMP3	Grey	2	12	2	Refsum_disease (1307), Zellweger_syndrome (1615)

5830	PEX5	Grey	2	11	2	Adrenoleukodystrophy (56), Zellweger_syndrome (1615)
5832	ALDH18A1	Metabolic	1	0	1	Hyperammonemia (717)
5836	PYGL	Metabolic	1	8	1	Glycogen_storage_disease (610)
5837	PYGM	Metabolic	1	0	1	McArdle_disease (962)
5860	QDPR	Metabolic	1	2	1	Phenylketonuria (1216)
5873	RAB27A	Dermatological	1	2	1	Griscelli_syndrome (627)
5880	RAC2	Immunological	1	0	1	Neutrophil_immunodeficiency_syndrome (1102)
5888	RAD51	Cancer	1	18	1	Breast_cancer (228)
5896	RAG1	Immunological	2	7	1	Omenn_syndrome (1141), Severe_combined_immunodeficiency (1383)
5897	RAG2	Immunological	2	7	1	Omenn_syndrome (1141), Severe_combined_immunodeficiency (1383)
5910	RAP1GDS1	Cancer	1	9	1	Lymphoma (925)
5913	RAPSN	Muscular	1	6	1	Myasthenic_syndrome (1042)
5921	RASA1	Grey	3	3	3	Basal_cell_carcinoma (188), Capillary_malformations (261), Parkes_Weber_syndrome (1191)
5925	RB1	Cancer	2	3	1	Bladder_cancer (207), Retinoblastoma (1317)
5932	RBBP8	Cancer	1	8	1	Pancreatic_cancer (1178)
5950	RBP4	Ophthamological	1	0	1	Retinol_binding_protein,_deficiency_of (1318)
5956	OPN1LW	Ophthamological	2	2	1	Blue-cone_monochromacy (214), Colorblindness (348)
5959	RDH5	Ophthamological	1	2	1	Fundus_albipunctatus (562)
5961	RDS	Ophthamological	4	32	1	Butterfly_dystrophy,_retinal (239), Foveomacular_dystrophy,_adult-onset,_with_choroidal_neovascularization (549), Macular_dystrophy (2937), Retinitis_pigmentosa (1316)
5972	REN	Grey	2	3	2	Hyperproreninemia (750), Renal_tubular_dysgenesis (9308)
5979	RET	Grey	5	9	3	Central_hypoventilation_syndrome (287), Colonic_aganglionosis,_total,_with_small_bowel_involvement (347), Hirschsprung_disease (681), Medullary_thyroid_carcinoma (2969), Multiple_endocrine_neoplasia (1037)
5993	RFX5	Immunological	1	4	1	Bare_lymphocyte_syndrome (184)
5994	RFXAP	Immunological	1	4	1	Bare_lymphocyte_syndrome (184)
5995	RGR	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
6005	RHAG	Hematological	2	8	1	Anemia (110), Rh-mod_syndrome (1326)
6006	RHCE	Hematological	2	23	1	Blood_group (212), Rh-negative_blood_type (1327)
6007	RHD	Hematological	1	1	1	Rh-negative_blood_type (1327)
6010	RHO	Ophthamological	2	32	1	Night_blindness (1110), Retinitis_pigmentosa (1316)
6011	GRK1	Ophthamological	1	1	1	Oguchi_disease (1137)
6017	RLBP1	Ophthamological	4	30	1	Bothnia_retinal_dystrophy (221), Fundus_albipunctatus (562), Newfoundland_rod-cone_dystrophy (1105), Retinitis_pigmentosa (1316)
6023	RMRP	Grey	2	2	2	Cartilage-hair_hypoplasia (276), Metaphyseal_chondrodysplasia (997)
6041	RNASEL	Cancer	1	11	1	Prostate_cancer (1272)
6049	RNF6	Cancer	1	3	1	Esophageal_cancer (508)
6094	ROM1	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
6100	RP9	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
6101	RP1	Grey	2	32	2	Hypertriglyceridemia (757), Retinitis_pigmentosa (1316)
6102	RP2	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
6103	RPGR	Ophthamological	3	36	1	Cone_dystrophy (352), Macular_degeneration (937), Retinitis_pigmentosa (1316)
6121	RPE65	Ophthamological	2	32	1	Leber_congenital_amaurosis (873), Retinitis_pigmentosa (1316)
6197	RPS6KA3	Grey	2	23	2	Coffin-Lowry_syndrome (341), Mental_retardation (990)
6223	RPS19	Hematological	1	8	1	Anemia (110)
6247	RS1	Ophthamological	1	0	1	Retinoschisis (1320)
6261	RYR1	Grey	2	2	2	Central_core_disease (2287), Malignant_hyperthermia_susceptibility (942)
6262	RYR2	Cardiovascular	2	4	1	Arrhythmogenic_right_ventricular_dysplasia (144), Ventricular_tachycardia (1558)
6295	SAG	Ophthamological	1	1	1	Oguchi_disease (1137)
6299	SALL1	multiple	1	0	1	Townes-Brocks_syndrome (1514)
6303	SAT	Dermatological	1	3	1	Keratosis_palmoplantaria_striata (847)

6309	SC5DL	Metabolic	1	0	1	Lathosterolosis (869)
6310	ATXN1	Neurological	1	12	1	Spinocereballar_ataxia (1428)
6311	ATXN2	Neurological	1	12	1	Spinocereballar_ataxia (1428)
6314	ATXN7	Neurological	1	12	1	Spinocereballar_ataxia (1428)
6315	KLHL1AS	Neurological	1	12	1	Spinocereballar_ataxia (1428)
6323	SCN1A	Neurological	1	16	1	Epilepsy (495)
6324	SCN1B	Neurological	1	1	1	Generalized_epilepsy (578)
6325	SCN2A1	Neurological	1	0	1	Seizures (1371)
6329	SCN4A	Grey	6	9	3	Cramps,_potassium-aggravated (376), Hyperkalemic_periodic_paralysis (733), Hypokalemic_periodic_paralysis (776), Myasthenic_syndrome (1042), Myotonia_congenita (1062), Paramyotonia_congenita (1188)
6331	SCN5A	Cardiovascular	5	6	1	Brugada_syndrome (234), Heart_block (647), Long_QT_syndrome (912), Sick_sinus_syndrome (1394), Ventricular_fibrillation,_idiopathic (3558)
6335	SCN9A	Neurological	1	0	1	Erythermalgia (502)
6337	SCNN1A	Endocrine	1	5	1	Pseudohypoaldosteronism (1281)
6338	SCNN1B	Grey	2	5	2	Liddle_syndrome (896), Pseudohypoaldosteronism (1281)
6340	SCNN1G	Grey	2	5	2	Liddle_syndrome (896), Pseudohypoaldosteronism (1281)
6341	SCO1	Gastrointestinal	1	0	1	Hepatic_failure,_early_onset,_and_neurologic_disorder (666)
6352	CCL5	Immunological	1	3	1	HIV (684)
6387	CXCL12	Immunological	1	2	1	AIDS (70)
6389	SDHA	Grey	2	14	2	Leigh_syndrome (877), Mitochondrial_complex_deficiency (1016)
6390	SDHB	Cancer	2	3	1	Paragangliomas (1186), Pheochromocytoma (1218)
6391	SDHC	Cancer	1	2	1	Paragangliomas (1186)
6392	SDHD	Cancer	4	3	1	Carcinoid_tumors,_intestinal (265), Merkel_cell_carcinoma (992), Paragangliomas (1186), Pheochromocytoma (1218)
6399	TRAPPC2	Skeletal	1	4	1	Spondyloepiphyseal_dysplasia (1435)
6403	SELP	Immunological	1	4	1	Atopy (164)
6435	SFTPA1	Respiratory	1	3	1	Pulmonary_fibrosis (1291)
6439	SFTPB	Respiratory	1	3	1	Pulmonary_fibrosis (1291)
6440	SFTPC	Respiratory	3	4	1	Pneumonitis,_desquamative_interstitial (1238), Pulmonary_fibrosis (1291), Surfactant_deficiency (1464)
6442	SGCA	Muscular	1	17	1	Muscular_dystrophy (1040)
6443	SGCB	Muscular	1	17	1	Muscular_dystrophy (1040)
6444	SGCD	Grey	2	38	2	Cardiomyopathy (268), Muscular_dystrophy (1040)
6445	SGCG	Muscular	1	17	1	Muscular_dystrophy (1040)
6448	SGSH	Metabolic	1	1	1	Sanfilippo_syndrome (1348)
6452	SH3BP2	Unclassified	1	0	0	Cherubism (305)
6468	FBXW4	Skeletal	1	1	1	Split-hand/foot_malformation (1430)
6469	SHH	Grey	3	5	3	Coloboma,_ocular (345), Holoprosencephaly (689), Solitary_median_maxillary_central_incisor (1414)
6473	SHOX	Skeletal	3	2	1	Langer_mesomelic_dysplasia (862), Leri-Weill_dyschondrosteosis (883), Short_stature (1387)
6476	SI	Metabolic	1	0	1	Sucrose_intolerance (1458)
6492	SIM1	Nutritional	1	20	1	Obesity (1126)
6496	SIX3	Developmental	1	4	1	Holoprosencephaly (689)
6513	SLC2A1	Metabolic	1	0	1	Glucose_transport_defect,_blood-brain_barrier (600)
6514	SLC2A2	Grey	2	26	2	Diabetes_mellitus (427), Fanconi-Bickel_syndrome (524)
6517	SLC2A4	Endocrine	1	26	1	Diabetes_mellitus (427)
6519	SLC3A1	Grey	2	1	2	Cystinuria (401), Homozygous_2p16_deletion_syndrome (701)
6521	SLC4A1	Grey	5	40	2	Blood_group (212), Elliptocytosis (472), Hemolytic_anemia (660), Renal_tubular_acidosis (7308), Spherocytosis (1423)
6523	SLC5A1	Metabolic	1	0	1	Glucose/galactose_malabsorption (599)
6524	SLC5A2	Renal	1	0	1	Renal_glucosuria (3308)
6528	SLC5A5	Endocrine	1	6	1	Hypothyroidism (787)

6530	SLC6A2	Cardiovascular	1	0	1	Orthostatic_intolerance (1150)
6532	SLC6A4	Psychiatric	2	2	1	Anxiety-related_personality_traits (129), Obsessive-compulsive_disorder (1128)
6535	SLC6A8	Neurological	2	23	1	Creatine_deficiency_syndrome,_X-linked (2385), Mental_retardation (990)
6555	SLC10A2	Gastrointestinal	1	0	1	Bile_acid_malabsorption,_primary (202)
6556	SLC11A1	Immunological	1	4	1	Mycobacterial_infection (1043)
6557	SLC12A1	multiple	1	4	1	Bartter_syndrome (187)
6559	SLC12A3	Renal	1	0	1	Gitelman_syndrome (587)
6563	SLC14A1	Hematological	1	22	1	Blood_group (212)
6567	SLC16A2	Neurological	1	0	1	Allan-Herndon-Dudley_syndrome (82)
6569	SLC34A1	Grey	2	6	2	Osteoporosis (1163), Urolithiasise (1550)
6583	SLC22A4	Connective_tissue_disorder	1	7	1	Rheumatoid_arthritis (1324)
6584	SLC22A5	Metabolic	1	0	1	Carnitine_deficiency (272)
6591	SNAI2	multiple	1	3	1	Waardenburg_syndrome (1574)
6598	SMARCB1	Cancer	1	0	1	Rhabdoid_tumors (1322)
6606	SMN1	Muscular	1	6	1	Spinal_muscular_atrophy (1426)
6608	SMO	Cancer	1	3	1	Basal_cell_carcinoma (188)
6609	SMPD1	Metabolic	1	2	1	Niemann-Pick_disease (1107)
6611	SMS	Neurological	1	23	1	Mental_retardation (990)
6620	SNCB	Neurological	1	5	1	Dementia (412)
6622	SNCA	Neurological	2	15	1	Dementia (412), Parkinson_disease (1192)
6638	SNRPN	multiple	1	1	1	Prader-Willi_syndrome (1254)
6647	SOD1	Neurological	1	5	1	Amyotrophic_lateral_sclerosis (104)
6649	SOD3	Unclassified	1	0	0	Superoxide_dismutase,_elevated_extracellular (1461)
6654	SOS1	Connective_tissue_disorder	1	1	1	Fibromatosisl (537)
6657	SOX2	Ophthamological	1	0	1	Anophthalmia (122)
6658	SOX3	Grey	2	23	2	Infundibular_hypoplasia_and_hypopituitarism (809), Mental_retardation (990)
6662	SOX9	Skeletal	2	0	1	Acampomelic_campolelic_dysplasia (18), Campomelic_dysplasia (255)
6663	SOX10	Grey	3	1	2	PCWH (1198), Waardenburg-Shah_syndrome (1573), Yemenite_deaf-blind_hypopigmentation_syndrome (1614)
6683	SPAST	Neurological	1	8	1	Spastic_ataxia/paraplegia (1418)
6687	SPG7	Neurological	1	8	1	Spastic_ataxia/paraplegia (1418)
6690	SPINK1	Gastrointestinal	3	2	1	Fibrocalculous_pancreatic_diabetes (535), Pancreatitis (1179), Tropical_calcific_pancreatitis (1529)
6697	SPR	Metabolic	1	0	1	Sepiapterin_reductase_deficiency (1377)
6708	SPTA1	Hematological	3	5	1	Elliptocytosis (472), Pyropoikilocytosis (1296), Spherocytosis (1423)
6710	SPTB	Hematological	3	13	1	Anemia (110), Elliptocytosis (472), Spherocytosis (1423)
6714	SRC	Cancer	1	33	1	Colon_cancer (346)
6716	SRD5A2	Unclassified	1	0	0	Pseudovaginal_perineoscrotal_hypospadias (1283)
6736	SRY	Endocrine	1	1	1	Gonadal_dysgenesis (619)
6755	SSTR5	Endocrine	1	1	1	Acromegaly (38)
6756	SSX1	Cancer	1	1	1	Sarcoma,_synovial (1350)
6757	SSX2	Cancer	1	1	1	Sarcoma,_synovial (1350)
6770	STAR	Endocrine	1	1	1	Lipoid_adrenal_hyperplasia (903)
6772	STAT1	Grey	2	4	1	Mycobacterial_infection (1043), STAT1_deficiency (1441)
6777	STAT5B	Grey	2	37	2	Growth_hormone (628), Leukemia (886)
6785	ELOVL4	Ophthamological	2	4	1	Macular_dystrophy (2937), Stargardt_disease (1439)
6792	CDKL5	Neurological	1	1	1	Rett_syndrome (1321)
6794	STK11	Cancer	3	11	1	Melanoma (978), Pancreatic_cancer (1178), Peutz-Jeghers_syndrome (1214)
6821	SUOX	Metabolic	1	0	1	Sulfite_oxidase_deficiency (1460)
6833	ABCC8	Grey	2	26	2	Diabetes_mellitus (427), Hypoglycemia (772)

6834	SURF1	Neurological	1	11	1	Leigh_syndrome (877)
6853	SYN1	Neurological	1	16	1	Epilepsy (495)
6854	SYN2	Psychiatric	1	8	1	Schizophrenia (1359)
6886	TAL1	Cancer	1	36	1	Leukemia (886)
6887	TAL2	Cancer	1	36	1	Leukemia (886)
6888	TALDO1	Metabolic	1	0	1	Transaldolase_deficiency (1515)
6891	TAP2	Immunological	2	4	1	Bare_lymphocyte_syndrome (184), Wegener_granulomatosis (1583)
6892	TAPBP	Immunological	1	4	1	Bare_lymphocyte_syndrome (184)
6898	TAT	Metabolic	1	2	1	Tyrosinemia (1540)
6899	TBX1	Grey	3	0	2	Conotruncal_anomaly_face_syndrome (357), DiGeorge_syndrome (432), Velocardiofacial_syndrome (1556)
6901	TAZ	Grey	3	24	2	Barth_syndrome (185), Cardiomyopathy (268), Noncompaction_of_left_ventricular_myocardium (1113)
6905	TBCE	Grey	2	0	2	Hypoparathyroidism-retardation-dysmorphism_syndrome (780), Kenny-Caffey_syndrome-1 (841)
6906	SERPINA7	Hematological	1	0	1	Thyroxine-binding_globulin_deficiency (1506)
6908	TBP	Neurological	3	25	1	Huntington_disease (708), Parkinson_disease (1192), Spinocereballar_ataxia (1428)
6910	TBX5	Developmental	1	0	1	Holt-Oram_syndrome (696)
6915	TBXA2R	Hematological	1	1	1	Bleeding_disorder (209)
6926	TBX3	multiple	1	0	1	Ulnar-mammary_syndrome (1543)
6927	TCF1	Grey	3	34	2	Diabetes_mellitus (427), Hepatic_adenoma (668), MODY (1020)
6928	TCF2	Grey	3	27	2	Diabetes_mellitus (427), Glomerulocystic_kidney_disease,_hypoplastic (594), MODY (1020)
6948	TCN2	Hematological	1	0	1	Transcobalamin_II_deficiency (1516)
6949	TCOF1	Developmental	1	0	1	Treacher_Collins_mandibulofacial_dysostosis (1520)
6955	TRA@	Cancer	1	36	1	Leukemia (886)
6997	TDGF1	Neurological	1	0	1	Forebrain_defects (547)
7007	TECTA	Ear,Nose,Throat	1	40	1	Deafness (406)
7010	TEK	Cardiovascular	1	1	1	Venous_thrombosis (1557)
7012	TERC	Grey	2	3	2	Aplastic_anemia (133), Dyskeratosis (458)
7015	TERT	Hematological	1	2	1	Aplastic_anemia (133)
7018	TF	Hematological	2	1	1	Atransferrinemia (165), Iron_overload/deficiency (824)
7021	TFAP2B	multiple	1	0	1	Char_syndrome (303)
7030	TFE3	Cancer	1	6	1	Renal_cell_carcinoma (1308)
7036	TFR2	Metabolic	1	4	1	Hemochromatosis (657)
7038	TG	Endocrine	3	7	1	Autoimmune_thyroid_disease (2174), Goiter (617), Hypothyroidism (787)
7040	TGFB1	Skeletal	1	0	1	Camurati-Engelmann_disease (257)
7044	LEFTY2	Developmental	1	1	1	Left-right_axis_malformations (874)
7045	TGFBI	Ophthamological	1	6	1	Corneal_dystrophy (362)
7046	TGFBR1	Connective_tissue_disorder	1	1	1	Loeys-Dietz_syndrome (910)
7048	TGFBR2	Grey	3	37	2	Colon_cancer (346), Esophageal_cancer (508), Loeys-Dietz_syndrome (910)
7050	TGIF	Developmental	1	4	1	Holoprosencephaly (689)
7051	TGM1	Dermatological	3	8	1	Ichthyosiform_erythroderma (793), Ichthyosis (794), Self-healing_collodion_baby (1373)
7054	TH	Neurological	1	0	1	Segawa_syndrome (1370)
7056	THBD	Grey	2	17	2	Myocardial_infarction (1054), Thrombophilia (1497)
7066	THPO	Hematological	1	1	1	Thrombocythemia (1493)
7067	THRA	Cancer	1	2	1	Pituitary_ACTH-secreting_adenoma (1232)
7068	THRB	Endocrine	1	1	1	Thyroid_hormone_resistance (3503)
7078	TIMP3	Ophthamological	1	0	1	Sorsby_fundus_dystrophy (1416)
7080	TITF1	Grey	2	0	2	Chorea,_hereditary_benign (318), Choreoathetosis,_hypothyroidism,_and_respiratory_distress (320)
7084	TK2	Muscular	1	0	1	Mitochondrial_DNA_depletion_myopathy (3016)
7086	TKT	Metabolic	1	0	1	Wernicke-Korsakoff_syndrome (1589)
7099	TLR4	Immunological	1	0	1	Endotoxin_hyporesponsiveness (481)

7100	TLR5	Immunological	1	0	1	Legionaire_disease (876)
7102	TSPAN7	Neurological	1	23	1	Mental_retardation (990)
7124	TNF	Grey	5	25	3	Asthma (153), Dementia (412), Malaria (940), Migraine (1013), Sepsis (1378)
7132	TNFRSF1A	Immunological	1	0	1	Periodic_fever,_familial (1206)
7134	TNNC1	Cardiovascular	1	24	1	Cardiomyopathy (268)
7136	TNNI2	Developmental	1	2	1	Arthrogryposis (146)
7137	TNNI3	Cardiovascular	1	24	1	Cardiomyopathy (268)
7138	TNNT1	Muscular	1	3	1	Nemaline_myopathy (1078)
7139	TNNT2	Cardiovascular	1	24	1	Cardiomyopathy (268)
7140	TNNT3	Developmental	1	2	1	Arthrogryposis (146)
7148	TNXB	Connective_tissue_disorder	1	8	1	Ehlers-Danlos_syndrome (470)
7150	TOP1	Metabolic	1	1	1	DNA_topoisomerase (440)
7153	TOP2A	Metabolic	1	1	1	DNA_topoisomerase (440)
7157	TP53	Cancer	11	70	1	Adrenal_cortical_carcinoma (2054), Breast_cancer (228), Colon_cancer (346), Hepatic_adenoma (668), Histiocytoma (683), Li-Fraumeni_syndrome (898), Multiple_malignancy_syndrome (5037), Nasopharyngeal_carcinoma (1074), Osteosarcoma (1166), Pancreatic_cancer (1178), Thyroid_carcinoma (1503)
7167	TPI1	Hematological	1	9	1	Hemolytic_anemia (660)
7168	TPM1	Cardiovascular	1	24	1	Cardiomyopathy (268)
7169	TPM2	Grey	2	5	2	Arthrogryposis (146), Nemaline_myopathy (1078)
7170	TPM3	Muscular	1	3	1	Nemaline_myopathy (1078)
7172	TPMT	Metabolic	1	0	1	6-mercaptopurine_sensitivity (12)
7173	TPO	Endocrine	4	2	1	Goiter (617), Hyperthyroidism (755), Thyroid_hormone_resistance (3503), Total_iodide_organification_defect (3512)
7201	TRHR	Endocrine	1	0	1	Thyrotropin-releasing_hormone_deficiency (1505)
7225	TRPC6	Renal	1	2	1	Glomerulosclerosis (596)
7227	TRPS1	Developmental	1	0	1	Trichorhinophalangeal_syndrome (1524)
7248	TSC1	Grey	3	2	3	Focal_cortical_dysplasia,_Taylor_balloon_cell_type (545), Lymphangioleiomyomatosis (920), Tuberous_sclerosis (1534)
7249	TSC2	Grey	2	2	2	Lymphangioleiomyomatosis (920), Tuberous_sclerosis (1534)
7251	TSG101	Cancer	1	18	1	Breast_cancer (228)
7252	TSHB	Endocrine	1	6	1	Hypothyroidism (787)
7253	TSHR	Grey	3	16	2	Hyperthroidism (754), Hypothyroidism (787), Thyroid_carcinoma (1503)
7259	TSPYL1	Unclassified	1	0	0	Sudden_infant_death_with_dysgenesis_of_the_testes_syndrome (1459)
7273	TTN	Grey	2	38	2	Cardiomyopathy (268), Muscular_dystrophy (1040)
7274	TTPA	Neurological	1	4	1	Ataxia (154)
7276	TTR	Grey	4	5	2	Amyloid_neuropathy (102), Amyloidosis (103), Carpal_tunnel_syndrome,_familial (275), Dystransthyretinemic_hyperthyroxinemia (463)
7287	TULP1	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
7291	TWIST1	Developmental	1	1	1	Saethre-Chotzen_syndrome (1344)
7292	TNFSF4	Cardiovascular	1	9	1	Myocardial_infarction (1054)
7299	TYR	Grey	2	5	2	Albinism (72), Waardenburg_syndrome (1574)
7305	TYROBP	Bone	1	1	1	Nasu-Hakola_disease (1075)
7306	TYRP1	Dermatological	1	2	1	Albinism (72)
7337	UBE3A	Developmental	1	1	1	Angelman_syndrome (113)
7345	UCHL1	Neurological	1	10	1	Parkinson_disease (1192)
7350	UCP1	Nutritional	1	20	1	Obesity (1126)
7351	UCP2	Nutritional	1	20	1	Obesity (1126)
7352	UCP3	Nutritional	1	20	1	Obesity (1126)

7356	SCGB1A1	Respiratory	1	12	1	Asthma (153)
7369	UMOD	Renal	2	0	1	Hyperuricemic_nephropathy (759), Medullary_cystic_kidney_disease (969)
7372	UMPS	Metabolic	1	0	1	Oroticaciduria (1149)
7374	UNG	Immunological	1	5	1	Immunodeficiency (802)
7381	UQCRB	multiple	1	6	1	Mitochondrial_complex_deficiency (1016)
7389	UROD	Metabolic	1	5	1	Porphyria (1249)
7390	UROS	Metabolic	1	5	1	Porphyria (1249)
7391	USF1	Metabolic	1	0	1	Hyperlipidemia (736)
7399	USH2A	Grey	2	36	2	Retinitis_pigmentosa (1316), Usher_syndrome (1551)
7401	USH3A	multiple	1	7	1	Usher_syndrome (1551)
7415	VCP	Muscular	1	2	1	Inclusion_body_myopathy (805)
7421	VDR	Bone	1	1	1	Rickets (1330)
7422	VEGF	Endocrine	1	26	1	Diabetes_mellitus (427)
7428	VHL	Grey	5	10	2	Hemangioblastoma,_cerebellar (650), Pheochromocytoma (1218), Polycythemia (1242), Renal_cell_carcinoma (1308), von_Hippel-Lindau_syndrome (1572)
7436	VLDLR	Neurological	1	0	1	Cerebellar_hypoplasia,_VLDLR-associated (4289)
7439	VMD2	Ophthamological	3	4	1	Macular_dystrophy (2937), Maculopathy,_bull's-eye (938), Vitelliform_macular_dystrophy (1566)
7450	VWF	Hematological	1	0	1	von_Willebrand_disease (3572)
7454	WAS	Grey	3	6	2	Neutropenia (1101), Thrombocytopenia (1494), Wiskott-Aldrich_syndrome (1598)
7466	WFS1	Grey	2	0	2	Hearing_loss,_low-frequency_sensorineural (646), Wolfram_syndrome (1603)
7473	WNT3	multiple	1	0	1	Tetra-amelia,_autosomal_recessive (1482)
7486	WRN	multiple	1	0	1	Werner_syndrome (1588)
7490	WT1	Grey	5	3	3	Denys-Drash_syndrome (420), Frasier_syndrome (552), Mesangial_sclerosis (993), WAGR_syndrome (1576), Wilms_tumor (1595)
7494	XBP1	Psychiatric	1	0	1	Bipolar_disorder (204)
7498	XDH	Metabolic	1	0	1	Xanthinuria,_type_I (1607)
7499	XG	Hematological	1	22	1	Blood_group (212)
7503	XIST	Unclassified	1	0	0	X-inactivation,_familial_skewed (1610)
7504	XK	Hematological	1	0	1	McLeod_syndrome (965)
7507	XPA	Dermatological	1	7	1	Xeroderma_pigmentosum (1608)
7508	XPC	Dermatological	1	7	1	Xeroderma_pigmentosum (1608)
7512	XPNPEP2	Immunological	1	1	1	Angioedema (114)
7517	XRCC3	Cancer	2	22	1	Breast_cancer (228), Melanoma (978)
7531	YWHAE	multiple	1	0	1	Miller-Dieker_lissencephaly (1014)
7535	ZAP70	Immunological	1	0	1	Selective_T-cell_defect (1372)
7546	ZIC2	Developmental	1	4	1	Holoprosencephaly (689)
7547	ZIC3	multiple	1	1	1	Heterotaxy (675)
7555	ZNF9	Muscular	1	1	1	Myotonic_dystrophy (1063)
7592	ZNF41	Neurological	1	23	1	Mental_retardation (990)
7704	ZBTB16	Cancer	1	36	1	Leukemia (886)
7750	ZNF198	Cancer	1	0	1	Stem-cell_leukemia/lymphoma_syndrome (1445)
7809	BSND	multiple	1	4	1	Bartter_syndrome (187)
7827	NPHS2	Renal	1	1	1	Nephrotic_syndrome (1087)
7840	ALMS1	Neurological	1	0	1	(null) (95)
7841	GCS1	Metabolic	1	0	1	Glucosidase_I_deficiency (601)
7849	PAX8	Endocrine	1	6	1	Hypothyroidism (787)
7852	CXCR4	Immunological	2	0	1	Myelokathexis,_isolated (1049), WHIM_syndrome (1591)
7873	ARMET	Cancer	1	8	1	Pancreatic_cancer (1178)
7879	RAB7	Neurological	1	17	1	Charcot-Marie-Tooth_disease (301)
7915	ALDH5A1	Metabolic	1	0	1	Succinic_semialdehyde_dehydrogenase_deficiency (1457)

7941	PLA2G7	Grey	3	19	3	Asthma (153), Atopy (164), Platelet_defect/deficiency (1237)
7957	EPM2A	Neurological	1	16	1	Epilepsy (495)
8013	NR4A3	Cancer	1	2	1	Chondrosarcoma (316)
8021	NUP214	Cancer	1	36	1	Leukemia (886)
8022	LHX3	Endocrine	1	3	1	Pituitary_hormone_deficiency (5232)
8028	MLLT10	Cancer	1	36	1	Leukemia (886)
8029	CUBN	Hematological	1	1	1	Megaloblastic_anemia (975)
8031	NCOA4	Cancer	1	10	1	Thyroid_carcinoma (1503)
8048	CSRP3	Cardiovascular	1	24	1	Cardiomyopathy (268)
8050	PDHX	Metabolic	1	0	1	Lacticacidemia_due_to_PDX1_deficiency (859)
8074	FGF23	Bone	2	1	1	Calcinosis,_tumoral (254), Hypophosphatemic_rickets (784)
8086	AAAS	multiple	1	0	1	Achalasia-addisonianism-alacrimia_syndrome (24)
8091	HMGA2	Cancer	3	2	1	Lipoma (904), Salivary_adenoma (1345), Uterine_leiomyoma (1552)
8106	PABPN1	Muscular	1	0	1	Oculopharyngeal_muscular_dystorphy (1135)
8132	PKDTS	Renal	1	3	1	Polycystic_kidney_disease (1241)
8139	GAN	Neurological	1	0	1	Giant_axonal_neuropathy (582)
8144	FIMG1	Muscular	1	0	1	Myasthenia (1041)
8195	MKKS	Grey	2	7	2	Bardet-Biedl_syndrome (183), McKusick-Kaufman_syndrome (964)
8200	GDF5	Skeletal	4	5	1	Acromesomelic_dysplasia (39), Brachydactyly (224), Chondrodysplasia,_Grebe_type (2315), Fibular_hypoplasia_and_complex_brachydactyly (539)
8242	SMCX	Neurological	1	23	1	Mental_retardation (990)
8287	USP9Y	Endocrine	1	1	1	Azoospermia (178)
8288	EPX	Hematological	1	0	1	Eosinophil_peroxidase_deficiency (491)
8291	DYSF	Muscular	3	25	1	Miyoshi_myopathy (1018), Muscular_dystrophy (1040), Myopathy (1059)
8292	COLQ	Neurological	1	0	1	Endplate_acetylcholinesterase_deficiency (482)
8301	PICALM	Cancer	1	36	1	Leukemia (886)
8312	AXIN1	Cancer	1	7	1	Hepatic_adenoma (668)
8313	AXIN2	Cancer	2	33	1	Colon_cancer (346), Oligodontia-colorectal_cancer_syndrome (1140)
8322	FZD4	Ophthamological	1	2	1	Exudative_vitreoretinopathy (515)
8379	MAD1L1	Cancer	2	20	1	Lymphoma (925), Prostate_cancer (1272)
8419	BFSP2	Ophthamological	1	14	1	Cataract (277)
8431	NR0B2	Nutritional	1	20	1	Obesity (1126)
8438	RAD54L	Cancer	3	54	1	Breast_cancer (228), Colon_cancer (346), Lymphoma (925)
8443	GNPAT	Connective_tissue_disorder	1	2	1	Chondrodysplasia_punctata (315)
8456	FOXN1	Immunological	1	0	1	T-cell_immunodeficiency,_congenital_alopecia,_and_nail_dystrophy (3478)
8481	OFD1	Skeletal	1	0	1	Oral-facial-digital_syndrome (1145)
8483	CILP	Skeletal	1	0	1	Lumbar_disc_disease (917)
8492	PRSS12	Neurological	1	23	1	Mental_retardation (990)
8493	PPM1D	Cancer	1	18	1	Breast_cancer (228)
8504	PEX3	multiple	1	10	1	Zellweger_syndrome (1615)
8517	IKBKG	Dermatological	2	7	1	Ectodermal_dysplasia (466), Incontinentia_pigmenti (806)
8518	IKBKAP	Neurological	1	0	1	Dysautonomia (454)
8540	AGPS	multiple	1	1	1	Rhizomelic_chondrodysplasia_punctata (1325)
8546	AP3B1	multiple	1	6	1	Hermansky-Pudlak_syndrome (670)
8557	TCAP	Grey	2	38	2	Cardiomyopathy (268), Muscular_dystrophy (1040)
8572	PDLIM4	Bone	1	0	1	Osteoporosis (1165)
8625	RFXANK	Immunological	1	0	1	MHC_class_II_deficiency (1006)
8626	TP73L	Grey	6	1	2	ADULT_syndrome (59), EEC_syndrome (468), Hay-Wells_syndrome (643), Limb-mammary_syndrome (901), Rapp-Hodgkin_syndrome (1305), Split-hand/foot_malformation (1430)

8629	JRK	Neurological	1	16	1	Epilepsy (495)
8643	PTCH2	Cancer	2	4	1	Basal_cell_carcinoma (188), Medulloblastoma (970)
8647	ABCB11	Gastrointestinal	1	3	1	Cholestasis (310)
8659	ALDH4A1	Metabolic	1	1	1	Hyperprolinemia (749)
8660	IRS2	Endocrine	1	26	1	Diabetes_mellitus (427)
8671	SLC4A4	Renal	1	4	1	Renal_tubular_acidosis (7308)
8701	DNAH11	Grey	2	3	2	Kartagener_syndrome (840), Situs_ambiguus (1399)
8706	B3GALT3	Hematological	1	22	1	Blood_group (212)
8787	RGS9	Ophthamological	1	1	1	Bradyopsia (225)
8792	TNFRSF11A	Bone	2	3	1	Osteolysis (1157), Paget_disease (1173)
8795	TNFRSF10B	Cancer	1	2	1	Squamous_cell_carcinoma (1437)
8803	SUCLA2	multiple	1	1	1	Mitochondrial_DNA_depletion_syndrome (5016)
8805	TRIM24	Cancer	1	10	1	Thyroid_carcinoma (1503)
8813	DPM1	Metabolic	1	12	1	Congenital_disorder_of_glycosylation (354)
8820	HESX1	Grey	2	3	2	Pituitary_hormone_deficiency (5232), Septooptic_dysplasia (1380)
8833	GMPS	Cancer	1	36	1	Leukemia (886)
8838	WISP3	Grey	2	4	2	Arthropathy (147), Spondyloepiphyseal_dysplasia (1435)
8842	PROM1	Ophthamological	1	3	1	Retinal_cone_dsytrophy (1315)
8864	PER2	Neurological	1	0	1	Advanced_sleep_phase_syndrome (60)
8869	ST3GAL5	Neurological	1	0	1	Amish_infantile_epilepsy_syndrome (100)
8878	SQSTM1	Bone	1	2	1	Paget_disease (1173)
8890	EIF2B4	Neurological	2	4	1	Leukoencephalopathy_with_vanishing_white_matter (891), Ovarioleukodystrophy (1171)
8891	EIF2B3	Neurological	1	4	1	Leukoencephalopathy_with_vanishing_white_matter (891)
8892	EIF2B2	Neurological	2	4	1	Leukoencephalopathy_with_vanishing_white_matter (891), Ovarioleukodystrophy (1171)
8893	EIF2B5	Neurological	2	4	1	Leukoencephalopathy_with_vanishing_white_matter (891), Ovarioleukodystrophy (1171)
8898	MTMR2	Neurological	1	17	1	Charcot-Marie-Tooth_disease (301)
8910	SGCE	Neurological	1	5	1	Dystonia (462)
8915	BCL10	Cancer	5	40	1	Colon_cancer (346), Germ_cell_tumor (580), Lymphoma (925), Mesothelioma (995), Sezary_syndrome (1385)
8929	PHOX2B	Grey	3	8	3	Central_hypoventilation_syndrome (287), Hirschsprung_disease (681), Neuroblastoma (1091)
9051	PSTPIP1	Dermatological	1	0	1	Pyogenic_sterile_arthritis,_pyoderma_gangrenosum,_and_acne (1295)
9056	SLC7A7	Metabolic	1	0	1	Lysinuric_protein_intolerance (931)
9060	PAPSS2	Connective_tissue_disorder	1	0	1	SEMD,_Pakistani_type (1374)
9076	CLDN1	multiple	1	0	1	Neonatal_ichthyosis-sclerosing_cholangitis_syndrome (3079)
9095	TBX19	Endocrine	1	0	1	Adrenocorticotropic_hormone_deficiency (55)
9129	PRPF3	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
9132	KCNQ4	Ear,Nose,Throat	1	40	1	Deafness (406)
9150	CTDP1	Ophthamological	1	0	1	Congenital_cataracts (4354)
9210	BMP15	Endocrine	1	1	1	Ovarian_dysgenesis (3171)
9211	LGI1	Grey	2	20	2	Epilepsy (495), Glioblastoma (590)
9215	LARGE	Muscular	1	17	1	Muscular_dystrophy (1040)
9217	VAPB	Grey	2	11	2	Amyotrophic_lateral_sclerosis (104), Spinal_muscular_atrophy (1426)
9227	LRAT	Ophthamological	1	3	1	Retinal_cone_dsytrophy (1315)
9231	DLG5	Gastrointestinal	1	1	1	Crohn_disease (390)
9241	NOG	Grey	4	0	2	Stapes_ankylosis_syndrome_without_symphalangism (1438), Symphalangism,_proximal (1467), Synostoses_syndrome (1469), Tarsal-carpal_coalition_syndrome (1475)
9244	CRLF1	multiple	1	0	1	Cold-induced_sweating_syndrome (2344)
9247	GCM2	Endocrine	1	1	1	Hypoparathyroidism (779)
9289	GPR56	Neurological	1	0	1	Polymicrogyria (1244)
9342	SNAP29	Neurological	1	0	1	Cerebral_dysgenesis,_neuropathy,_ichthyosis,_and_palmoplantar_keratoderma_syndrome (6291)

9365	KL	Cardiovascular	1	5	1	Coronary_artery_disease (365)
9370	ADIPOQ	Endocrine	1	0	1	Adiponectin_deficiency (52)
9380	GRHPR	Metabolic	1	1	1	Hyperoxaluria (742)
9381	OTOF	Ear,Nose,Throat	1	40	1	Deafness (406)
9401	RECQL4	multiple	2	0	1	RAPADILINO_syndrome (1303), Rothmund-Thomson_syndrome (1338)
9409	PEX16	multiple	1	10	1	Zellweger_syndrome (1615)
9414	TJP2	Gastrointestinal	1	2	1	Hypercholanemia (723)
9420	CYP7B1	Gastrointestinal	1	0	1	Giant_cell_hepatitis (2584)
9445	ITM2B	Neurological	1	5	1	Dementia (412)
9451	EIF2AK3	Bone	1	0	1	Wolcott-Rallison_syndrome (1600)
9459	ARHGEF6	Neurological	1	23	1	Mental_retardation (990)
9469	CHST3	Skeletal	1	4	1	Spondyloepiphyseal_dysplasia (1435)
9479	MAPK8IP1	Endocrine	1	26	1	Diabetes_mellitus (427)
9496	TBX4	Skeletal	1	0	1	Small_patella_syndrome (1406)
9499	MYOT	Muscular	2	17	1	Muscular_dystrophy (1040), Myotilinopathy (1061)
9509	ADAMTS2	Connective_tissue_disorder	1	8	1	Ehlers-Danlos_syndrome (470)
9516	LITAF	Neurological	1	17	1	Charcot-Marie-Tooth_disease (301)
9526	MPDU1	Metabolic	1	12	1	Congenital_disorder_of_glycosylation (354)
9562	MINPP1	Cancer	1	10	1	Thyroid_carcinoma (1503)
9563	H6PD	Metabolic	1	1	1	Cortisone_reductase_deficiency (368)
9568	GPR51	Psychiatric	1	2	1	Nicotine_addiction (1106)
9607	CART	Nutritional	1	20	1	Obesity (1126)
9622	KLK4	Bone	1	3	1	Amelogenesis_imperfecta (99)
9627	SNCAIP	Neurological	1	10	1	Parkinson_disease (1192)
9639	ARHGEF10	Neurological	1	0	1	Slowed_nerve_conduction_velocity,_AD (1404)
9657	IQCB1	Renal	1	2	1	Senior-Loken_syndrome (1375)
9685	ENTH	Psychiatric	1	8	1	Schizophrenia (1359)
9820	CUL7	multiple	1	0	1	3-M_syndrome (10)
9821	RB1CC1	Cancer	1	18	1	Breast_cancer (228)
9839	ZFHX1B	Developmental	1	0	1	Mowat-Wilson_syndrome (1028)
9856	KIAA0319	Psychiatric	1	1	1	Dyslexia (459)
9927	MFN2	Neurological	1	17	1	Charcot-Marie-Tooth_disease (301)
9950	GOLGA5	Cancer	1	10	1	Thyroid_carcinoma (1503)
9990	SLC12A6	Neurological	1	0	1	Agenesis_of_the_corpus_callosum_with_peripheral_neuropathy (65)
9992	KCNE2	Cardiovascular	2	9	1	Atrial_fibrillation (166), Long_QT_syndrome (912)
9997	SCO2	Cardiovascular	1	0	1	Cardioencephalomyopathy,_fatal_infantile,_due_to_cytochrome_c_oxidase_deficiency (267)
10002	NR2E3	Ophthamological	2	29	1	Enhanced_S-cone_syndrome (483), Retinitis_pigmentosa (1316)
10008	KCNE3	Renal	1	2	1	Hypokalemic_periodic_paralysis (776)
10020	GNE	Grey	3	2	2	Inclusion_body_myopathy (805), Nonaka_myopathy (1112), Sialuria (1392)
10060	ABCC9	Cardiovascular	1	24	1	Cardiomyopathy (268)
10083	USH1C	Grey	2	44	2	Deafness (406), Usher_syndrome (1551)
10084	PQBP1	Neurological	1	0	1	Renpenning_syndrome (1309)
10117	ENAM	Grey	2	3	2	Amelogenesis_imperfecta (99), Hypoplastic_enamel_pitting,_localized (785)
10128	LRPPRC	Neurological	1	11	1	Leigh_syndrome (877)
10133	OPTN	Ophthamological	1	3	1	Glaucoma (589)
10157	AASS	Metabolic	1	0	1	Hyperlysinemia (738)
10165	SLC25A13	Metabolic	1	1	1	Citrullinemia (335)
10166	SLC25A15	Metabolic	1	0	1	Hyperornithinemia-hyperammonemia-homocitrullinemia_syndrome (740)
10195	ALG3	Metabolic	1	12	1	Congenital_disorder_of_glycosylation (354)

10216	PRG4	Skeletal	1	0	1	Camptodactyly-arthropathy-coxa_vara-pericarditis_syndrome (256)
10243	GPHN	Metabolic	1	2	1	Molybdenum_cofactor_deficiency (1023)
10269	ZMPSTE24	Grey	2	0	2	Mandibuloacral_dysplasia_with_type_B_lipodystrophy (945), Restrictive_dermopathy,_lethal (1314)
10312	TCIRG1	Bone	1	3	1	Osteopetrosis (1161)
10320	ZNFN1A1	Cancer	1	36	1	Leukemia (886)
10395	DLC1	Cancer	1	33	1	Colon_cancer (346)
10397	NDRG1	Neurological	1	17	1	Charcot-Marie-Tooth_disease (301)
10461	MERTK	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
10516	FBLN5	Grey	2	6	2	Cutis_laxa (395), Macular_degeneration (937)
10555	AGPAT2	Metabolic	1	4	1	Lipodystrophy (902)
10558	SPTLC1	Neurological	1	7	1	Neuropathy (1099)
10560	SLC19A2	Hematological	1	0	1	Thiamine-responsive_megaloblastic_anemia_syndrome (1491)
10564	ARFGEF2	Neurological	1	0	1	Periventricular_heterotopia_with_microcephaly (1209)
10577	NPC2	Metabolic	1	2	1	Niemann-Pick_disease (1107)
10585	POMT1	Grey	2	18	2	Muscular_dystrophy (1040), Walker-Warburg_syndrome (1578)
10594	PRPF8	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
10682	EBP	Connective_tissue_disorder	1	2	1	Chondrodysplasia_punctata (315)
10683	DLL3	Skeletal	1	1	1	Spondylocostal_dysostosis (1433)
10686	CLDN16	Renal	1	2	1	Hypomagnesemia (778)
10743	RAI1	multiple	1	0	1	Smith-Magenis_syndrome (1411)
10747	MASP2	Immunological	1	0	1	MASP2_deficiency (956)
10804	GJB6	Grey	2	47	2	Deafness (406), Ectodermal_dysplasia (466)
10841	FTCD	Metabolic	1	0	1	Glutamate_formiminotransferase_deficiency (602)
10842	C7orf16	Metabolic	1	7	1	Hypercholesterolemia (724)
10891	PPARGC1A	Metabolic	1	4	1	Lipodystrophy (902)
10892	MALT1	Cancer	1	0	1	MALT_lymphoma (944)
10913	EDAR	Dermatological	1	7	1	Ectodermal_dysplasia (466)
10962	MLLT11	Cancer	1	36	1	Leukemia (886)
10984	KCNQ1OT1	multiple	1	3	1	Beckwith-Wiedemann_syndrome (194)
11005	SPINK5	Grey	2	4	2	Atopy (164), Netherton_syndrome (1088)
11081	KERA	Ophthamological	1	6	1	Corneal_dystrophy (362)
11093	ADAMTS13	Hematological	1	8	1	Thrombophilia (1497)
11132	CAPN10	Endocrine	1	26	1	Diabetes_mellitus (427)
11136	SLC7A9	Renal	1	1	1	Cystinuria (401)
11141	IL1RAPL1	Neurological	1	23	1	Mental_retardation (990)
11146	GLMN	Cardiovascular	1	0	1	Glomuvenous_malformations (597)
11178	LZTS1	Cancer	1	3	1	Esophageal_cancer (508)
11200	CHEK2	Cancer	4	27	1	Breast_cancer (228), Li-Fraumeni_syndrome (898), Osteosarcoma (1166), Prostate_cancer (1272)
11216	AKAP10	Developmental	1	1	1	Longevity (911)
11231	SEC63	Gastrointestinal	1	1	1	Polycystic_liver_disease (3241)
11234	HPS5	multiple	1	6	1	Hermansky-Pudlak_syndrome (670)
11235	PDCD10	Neurological	1	2	1	Cerebral_cavernous_malformations (4291)
11236	RNF139	Cancer	1	6	1	Renal_cell_carcinoma (1308)
11254	SLC6A14	Nutritional	1	20	1	Obesity (1126)
11281	POU6F2	Cancer	1	3	1	Wilms_tumor (1595)
11285	B4GALT7	Connective_tissue_disorder	1	8	1	Ehlers-Danlos_syndrome (470)
11315	PARK7	Neurological	1	10	1	Parkinson_disease (1192)
11322	EVER1	Dermatological	1	1	1	Epidermodysplasia_verruciformis (492)

22800	RRAS2	Cancer	1	7	1	Ovarian_cancer (1170)
22891	ZNF365	Renal	1	1	1	Nephrolithiasis (1081)
22930	RAB3GAP1	multiple	1	2	1	Walker-Warburg_syndrome (1578)
22934	RPIA	Metabolic	1	0	1	Ribose_5-phosphate_isomerase_deficiency (1329)
22954	TRIM32	Muscular	1	17	1	Muscular_dystrophy (1040)
22976	PAXIP1	Neurological	1	11	1	Alzheimer_disease (98)
23064	ALS4	Neurological	1	4	1	Ataxia (154)
23092	ARHGAP26	Cancer	1	36	1	Leukemia (886)
23095	KIF1B	Neurological	1	17	1	Charcot-Marie-Tooth_disease (301)
23111	SPG20	Neurological	1	0	1	Troyer_syndrome (1530)
23209	MLC1	Neurological	1	0	1	Megalencephalic_leukoencephalopathy_with_subcortical_cysts (974)
23230	VPS13A	Neurological	1	0	1	Choreoacanthocytosis (319)
23305	ACSL6	Grey	2	4	2	Myelodysplastic_syndrome (1045), Myelogenous_leukemia (1047)
23365	ARHGEF12	Cancer	1	36	1	Leukemia (886)
23373	MECT1	Cancer	1	1	1	Mucoepidermoid_salivary_gland_carcinoma (1031)
23389	THRAP2	Developmental	1	1	1	Transposition_of_great_arteries,_dextro-looped (1519)
23414	ZFPM2	Cardiovascular	1	2	1	Tetralogy_of_Fallot (1483)
23417	MLYCD	Metabolic	1	0	1	Malonyl-CoA_decarboxylase_deficiency (943)
23418	CRB1	Ophthamological	3	32	1	Leber_congenital_amaurosis (873), Pigmented_paravenous_chorioretinal_atrophy (1229), Retinitis_pigmentosa (1316)
23474	ETHE1	Metabolic	1	0	1	Ethylmalonic_encephalopathy (510)
23495	TNFRSF13B	Immunological	1	0	1	Immunoglobulin_A_deficiency (804)
23562	CLDN14	Ear,Nose,Throat	1	40	1	Deafness (406)
23569	PADI4	Connective_tissue_disorder	1	7	1	Rheumatoid_arthritis (1324)
23592	LEMD3	Grey	3	0	3	Buschke-Ollendorff_syndrome (238), Melorheostosis_with_osteopoikilosis (982), Osteopoikilosis (1162)
23600	AMACR	Metabolic	1	0	1	Alpha-methylacyl-CoA_racemase_deficiency (91)
23607	CD2AP	Renal	1	2	1	Glomerulosclerosis (596)
23746	AIPL1	Ophthamological	3	33	1	Cone_dystrophy (352), Leber_congenital_amaurosis (873), Retinitis_pigmentosa (1316)
24140	FTSJ1	Neurological	1	23	1	Mental_retardation (990)
25788	RAD54B	Cancer	2	39	1	Colon_cancer (346), Lymphoma (925)
25794	FSCN2	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
25814	ATXN10	Neurological	1	12	1	Spinocereballar_ataxia (1428)
25836	NIPBL	Developmental	1	0	1	Cornelia_de_Lange_syndrome (364)
25861	DFNB31	Ear,Nose,Throat	1	40	1	Deafness (406)
25894	PLEKHG4	Neurological	1	12	1	Spinocereballar_ataxia (1428)
25953	MR-1	Neurological	1	0	1	Paroxysmal_kinesigenic_choreoathetosis (1195)
26012	NELF	Endocrine	1	3	1	Hypogonadotropic_hypogonadism (774)
26119	LDLRAP1	Metabolic	1	7	1	Hypercholesterolemia (724)
26121	PRPF31	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
26128	KIAA1279	multiple	1	0	1	Goldberg-Shprintzen_megacolon_syndrome (618)
26154	ABCA12	Dermatological	1	6	1	Ichthyosis (794)
26191	PTPN22	Grey	3	36	3	Diabetes_mellitus (427), Rheumatoid_arthritis (1324), Systemic_lupus_erythematosus (1471)
26227	PHGDH	Metabolic	1	0	1	Phosphoglycerate_dehydrogenase_deficiency (1220)
26276	VPS33B	Gastrointestinal	1	0	1	ARC_syndrome (139)
26278	SACS	Neurological	1	8	1	Spastic_ataxia/paraplegia (1418)
26353	HSPB8	Neurological	1	7	1	Neuropathy (1099)
26503	SLC17A5	Metabolic	2	1	1	Salla_disease (1346), Sialidosis (1391)
26511	CHIC2	Cancer	1	36	1	Leukemia (886)
26580	BSCL2	Grey	3	10	3	Lipodystrophy (902), Silver_spastic_paraplegia_syndrome (1396), Spinal_muscular_atrophy (1426)

26762	HAVCR1	Immunological	1	4	1	Atopy (164)
27019	DNAI1	Grey	2	2	2	Ciliary_dyskinesia (331), Kartagener_syndrome (840)
27030	MLH3	Cancer	1	33	1	Colon_cancer (346)
27031	NPHP3	Renal	1	3	1	Nephronophthisis (1082)
27032	ATP2C1	Dermatological	1	0	1	Hailey-Hailey_disease (633)
27130	INVS	Renal	1	3	1	Nephronophthisis (1082)
27185	DISC1	Psychiatric	1	8	1	Schizophrenia (1359)
27232	GNMT	Metabolic	1	3	1	Glycine_encephalopathy (608)
28962	OSTM1	Bone	1	3	1	Osteopetrosis (1161)
29760	BLNK	Immunological	1	0	1	Hypoglobulinemia_and_absent_B_cells (771)
29851	ICOS	Immunological	1	0	1	ICOS_deficiency (795)
29881	NPC1L1	Unclassified	1	0	0	Ezetimibe,_nonresponse_to (517)
29929	ALG6	Metabolic	1	12	1	Congenital_disorder_of_glycosylation (354)
29958	DMGDH	Metabolic	1	0	1	Dimethylglycine_dehydrogenase_deficiency (435)
30061	SLC40A1	Metabolic	1	4	1	Hemochromatosis (657)
30813	VSX1	Grey	3	6	2	Corneal_dystrophy (362), Craniofacial_anomalies,_empty_sella_turcica,_corneal_endothelial_changes,_and_abnormal_retinal_and_auditory_bipolar_cells (377), Keratoconus (844)
30835	CD209	Immunological	1	0	1	Dengue_fever,_protection_against (413)
50485	SMARCAL1	Connective_tissue_disorder	1	0	1	Schimke_immunoosseous_dysplasia (1354)
50506	DUOX2	Endocrine	1	6	1	Hypothyroidism (787)
50511	SYCP3	Endocrine	1	1	1	Azoospermia (178)
50617	ATP6V0A4	Renal	1	4	1	Renal_tubular_acidosis (7308)
50814	NSDHL	Developmental	1	0	1	CHILD_syndrome (306)
50846	DHH	Endocrine	1	1	1	Gonadal_dysgenesis (619)
50943	FOXP3	Grey	2	26	2	Diabetes_mellitus (427), Immunodysregulation,_polyendocrinopathy,_and_enteropathy,_X-linked (803)
50945	TBX22	Developmental	1	2	1	Cleft_palate (336)
50964	SOST	Skeletal	1	0	1	Sclerosteosis (1363)
51062	SPG3A	Neurological	1	8	1	Spastic_ataxia/paraplegia (1418)
51099	ABHD5	Metabolic	1	0	1	Chanarin-Dorfman_syndrome (300)
51119	SBDS	multiple	1	0	1	Shwachman-Diamond_syndrome (1389)
51128	SAR1B	Gastrointestinal	2	0	1	Anderson_disease (108), Chylomicron_retention_disease (330)
51131	PHF11	Grey	2	12	2	Asthma (153), IgE_levels_QTL (797)
51135	IRAK4	Immunological	1	0	1	IRAK4_deficiency (821)
51151	SLC45A2	Grey	2	2	2	Ocular_albinism (1130), Pigmentation_of_hair,_skin,_and_eyes,_variation_in (1227)
51156	SERPINA10	Cardiovascular	1	1	1	Venous_thrombosis (1557)
51168	MYO15A	Ear,Nose,Throat	1	40	1	Deafness (406)
51185	CRBN	Neurological	1	23	1	Mental_retardation (990)
51251	NT5C3	Hematological	1	8	1	Anemia (110)
51324	SPG21	Neurological	1	0	1	Mast_syndrome (960)
51422	PRKAG2	Cardiovascular	2	24	1	Cardiomyopathy (268), Wolff-Parkinson-White_syndrome (1601)
51592	TRIM33	Cancer	1	10	1	Thyroid_carcinoma (1503)
51684	SUFU	Cancer	1	1	1	Medulloblastoma (970)
51733	UPB1	Metabolic	1	0	1	Beta-ureidopropionase_deficiency (199)
51738	GHRL	Nutritional	1	20	1	Obesity (1126)
51741	WWOX	Cancer	1	3	1	Esophageal_cancer (508)
53904	MYO3A	Ear,Nose,Throat	1	40	1	Deafness (406)
53947	A4GALT	Hematological	1	22	1	Blood_group (212)
54209	TREM2	Bone	1	1	1	Nasu-Hakola_disease (1075)

54332	GDAP1	Neurological	1	17	1	Charcot-Marie-Tooth_disease (301)
54345	SOX18	Dermatological	1	3	1	Hypotrichosis (788)
54361	WNT4	Developmental	1	0	1	Rokitansky-Kuster-Hauser_syndrome (1337)
54413	NLGN3	Psychiatric	2	3	1	Asperger_syndrome (151), Autism (173)
54658	UGT1A1	Grey	3	0	3	Crigler-Najjar_syndrome (389), Gilbert_syndrome (586), Hyperbilirubinemia (720)
54714	CNGB3	Ophthamological	2	6	1	Achromatopsia (29), Macular_degeneration (937)
54806	AHI1	multiple	1	1	1	Joubert_syndrome (831)
54808	DYM	Grey	2	0	2	Dyggve-Melchior-Clausen_disease (452), Smith-McCort_dysplasia (1412)
54829	ASPN	Connective_tissue_disorder	1	3	1	Osteoarthritis (1154)
54840	APTX	Neurological	1	4	1	Ataxia (154)
54880	BCOR	Grey	2	2	2	Microphthalmia (1012), Oculofaciocardiodental_syndrome (1133)
54904	WHSC1L1	Cancer	1	36	1	Leukemia (886)
54982	CLN6	Neurological	1	5	1	Ceroid-lipofuscinosis (296)
55109	AGGF1	multiple	1	0	1	Klippel-Trenaunay_syndrome (853)
55120	FANCL	multiple	1	10	1	Fanconi_anemia (523)
55212	BBS7	multiple	1	7	1	Bardet-Biedl_syndrome (183)
55343	SLC35C1	Metabolic	1	12	1	Congenital_disorder_of_glycosylation (354)
55605	KIF21A	Ophthamological	1	1	1	Fibrosis (538)
55612	C20orf42	Dermatological	1	0	1	Kindler_syndrome (850)
55624	POMGNT1	multiple	1	0	1	Muscle-eye-brain_disease (1038)
55630	SLC39A4	Dermatological	1	0	1	Acrodermatitis_enteropathica (34)
55636	CHD7	multiple	1	0	1	CHARGE_syndrome (302)
55670	PEX26	Grey	3	13	2	Adrenoleukodystrophy (56), Refsum_disease (1307), Zellweger_syndrome (1615)
55755	CDK5RAP2	Neurological	1	4	1	Microcephaly (1007)
55775	TDP1	Neurological	1	12	1	Spinocereballar_ataxia (1428)
55806	HR	Dermatological	2	0	1	Alopecia_universalis (85), Atrichia_with_papular_lesions (167)
55811	SAC	Renal	1	0	1	Hypercalciuria (721)
55835	CENPJ	Neurological	1	4	1	Microcephaly (1007)
55997	CFC1	Grey	3	2	3	Double-outlet_right_ventricle (444), Heterotaxy (675), Transposition_of_great_arteries,_dextro-looped (1519)
56052	ALG1	Metabolic	1	12	1	Congenital_disorder_of_glycosylation (354)
56172	ANKH	Grey	2	0	2	Chondrocalcinosis (314), Craniometaphyseal_dysplasia (382)
56244	BTNL2	Immunological	1	2	1	Sarcoidosis (1349)
56246	MRAP	Endocrine	1	1	1	Glucocorticoid_deficiency (598)
56262	LRRC8A	Hematological	1	3	1	Agammaglobulinemia (63)
56652	PEO1	Ophthamological	1	2	1	Progressive_external_ophthalmoplegia_with_mitochondrial_DNA_deletions (1265)
56729	RETN	Grey	2	37	2	Diabetes_mellitus (427), Hypertension (752)
56922	MCCC1	Metabolic	1	1	1	3-Methylcrotonyl-CoA_carboxylase_deficiency (7)
57096	RPGRIP1	Ophthamological	2	9	1	Cone_dystrophy (352), Leber_congenital_amaurosis (873)
57152	SLURP1	Dermatological	1	0	1	Meleda_disease (979)
57165	GJA12	Neurological	1	0	1	(null) (1201)
57167	SALL4	multiple	1	0	1	Duane_syndrome (449)
57190	SEPN1	Muscular	1	17	1	Muscular_dystrophy (1040)
57192	MCOLN1	Metabolic	1	2	1	Mucolipidosis (1032)
57214	KIAA1199	Ear,Nose,Throat	1	40	1	Deafness (406)
57338	JPH3	Neurological	1	3	1	Huntington_disease (708)
57379	AICDA	Immunological	1	5	1	Immunodeficiency (802)
57502	NLGN4X	Grey	3	25	2	Asperger_syndrome (151), Autism (173), Mental_retardation (990)
57591	MKL1	Cancer	1	0	1	Megakaryoblastic_leukemia,_acute (973)
57679	ALS2	Neurological	3	13	1	Amyotrophic_lateral_sclerosis (104), Primary_lateral_sclerosis (1261), Spastic_ataxia/paraplegia (1418)

57697	FANCM	multiple	1	10	1	Fanconi_anemia (523)
57716	PRX	multiple	1	3	1	Dejerine-Sottas_disease (410)
57817	HAMP	Metabolic	1	4	1	Hemochromatosis (657)
59344	ALOXE3	Dermatological	1	2	1	Ichthyosiform_erythroderma (793)
60386	SLC25A19	Neurological	1	4	1	Microcephaly (1007)
60506	NYX	Ophthamological	1	4	1	Night_blindness (1110)
60528	ELAC2	Cancer	1	11	1	Prostate_cancer (1272)
60529	ALX4	Skeletal	1	1	1	Parietal_foramina (1190)
63869	PSORS6	Dermatological	1	1	1	Psoraisis (1288)
64072	CDH23	Grey	2	44	2	Deafness (406), Usher_syndrome (1551)
64087	MCCC2	Metabolic	1	1	1	3-Methylcrotonyl-CoA_carboxylase_deficiency (7)
64127	CARD15	Grey	4	4	4	Blau_syndrome (208), Crohn_disease (390), Psoraisis (1288), Sarcoidosis (1349)
64221	ROBO3	Neurological	1	0	1	Gaze_palsy (576)
64240	ABCG5	Metabolic	1	1	1	Sitosterolemia (1398)
64241	ABCG8	Metabolic	1	1	1	Sitosterolemia (1398)
64324	NSD1	Grey	3	3	2	Beckwith-Wiedemann_syndrome (194), Sotos_syndrome (1417), Weaver_syndrome (1582)
64327	LMBR1	Skeletal	1	0	1	Acheiropody (25)
64342	HS1BP3	Neurological	1	0	1	Tremor,_familial_essential (1521)
64421	DCLRE1C	Immunological	2	7	1	Omenn_syndrome (1141), Severe_combined_immunodeficiency (1383)
64699	TMPRSS3	Ear,Nose,Throat	1	40	1	Deafness (406)
64805	P2RY12	Hematological	1	3	1	Platelet_defect/deficiency (1237)
65018	PINK1	Neurological	1	10	1	Parkinson_disease (1192)
65078	RTN4R	Psychiatric	1	8	1	Schizophrenia (1359)
65125	WNK1	Endocrine	1	5	1	Pseudohypoaldosteronism (1281)
65217	PCDH15	Grey	2	44	2	Deafness (406), Usher_syndrome (1551)
65266	WNK4	Endocrine	1	5	1	Pseudohypoaldosteronism (1281)
78987	CRELD1	Cardiovascular	1	2	1	Atrioventricular_block (168)
79001	VKORC1	Hematological	2	3	1	Vitamin_K-dependent_coagulation_defect (1565), Warfarin_resistance/sensitivity (1580)
79053	ALG8	Metabolic	1	12	1	Congenital_disorder_of_glycosylation (354)
79058	ASPSCR1	Cancer	1	0	1	Alveolar_soft-part_sarcoma (97)
79083	MLPH	Dermatological	1	2	1	Griscelli_syndrome (627)
79087	ALG12	Metabolic	1	12	1	Congenital_disorder_of_glycosylation (354)
79147	FKRP	Muscular	1	17	1	Muscular_dystrophy (1040)
79158	GNPTAB	Metabolic	1	2	1	Mucolipidosis (1032)
79577	CDC73	Grey	2	2	2	Hyperparathyroidism (743), Parathyroid_adenoma (1189)
79628	SH3TC2	Neurological	1	17	1	Charcot-Marie-Tooth_disease (301)
79648	MCPH1	Neurological	2	4	1	Microcephaly (1007), Premature_chromosome_condensation_with_microcephaly_and_mental_retardation (3260)
79751	SLC25A22	Neurological	1	16	1	Epilepsy (495)
79784	MYH14	Ear,Nose,Throat	1	40	1	Deafness (406)
79796	ALG9	Metabolic	1	12	1	Congenital_disorder_of_glycosylation (354)
79803	HPS6	multiple	1	6	1	Hermansky-Pudlak_syndrome (670)
79944	L2HGDH	Metabolic	1	0	1	L-2-hydroxyglutaric_aciduria (857)
79977	GRHL2	Ear,Nose,Throat	1	40	1	Deafness (406)
80025	PANK2	Grey	2	0	2	HARP_syndrome (640), Neurodegeneration (1092)
80144	FRAS1	multiple	1	1	1	Fraser_syndrome (551)
80207	OPA3	Grey	2	1	2	3-methylglutaconicaciduria (9), Optic_atrophy (1144)
80270	HSD3B7	Gastrointestinal	1	3	1	Cholestasis (310)
80324	PUS1	multiple	1	0	1	Mitochondrial_myopathy_and_sideroblastic_anemia (7016)
80704	SLC19A3	Neurological	1	1	1	Basal_ganglia_disease (4188)
80781	COL18A1	multiple	1	0	1	Knobloch_syndrome (855)

81693	AMN	Hematological	1	1	1	Megaloblastic_anemia (975)
81794	ADAMTS10	Connective_tissue_disorder	1	1	1	Weill-Marchesani_syndrome (1585)
81846	SBF2	Neurological	1	17	1	Charcot-Marie-Tooth_disease (301)
83605	CCM2	Neurological	1	2	1	Cerebral_cavernous_malformations (4291)
83715	ESPN	Ear,Nose,Throat	1	40	1	Deafness (406)
83844	USP26	Renal	1	0	1	Sertoli-cell-only_syndrome (1381)
83872	HMCN1	Ophthamological	1	4	1	Macular_degeneration (937)
83990	BRIP1	Grey	2	27	2	Breast_cancer (228), Fanconi_anemia (523)
84059	MASS1	Grey	2	7	2	Convulsions (359), Usher_syndrome (1551)
84062	DTNBP1	multiple	1	6	1	Hermansky-Pudlak_syndrome (670)
84100	ARL6	multiple	1	7	1	Bardet-Biedl_syndrome (183)
84295	PHF6	multiple	1	0	1	Borjeson-Forssman-Lehmann_syndrome (218)
84343	HPS3	multiple	1	6	1	Hermansky-Pudlak_syndrome (670)
84441	MAML2	Cancer	1	1	1	Mucoepidermoid_salivary_gland_carcinoma (1031)
84572	GNPTG	Metabolic	1	2	1	Mucolipidosis (1032)
84634	GPR54	Endocrine	1	3	1	Hypogonadotropic_hypogonadism (774)
84916	CIRH1A	Gastrointestinal	1	2	1	Cirrhosis (334)
84930	MASTL	Hematological	1	3	1	Thrombocytopenia (1494)
85300	ATCAY	Neurological	1	4	1	Ataxia (154)
85358	SHANK3	multiple	1	0	1	Chromosome_22q13.3_deletion_syndrome (326)
85365	ALG2	Metabolic	1	12	1	Congenital_disorder_of_glycosylation (354)
85366	MYLK2	Cardiovascular	1	24	1	Cardiomyopathy (268)
85476	GFM1	multiple	1	0	1	Combined_oxidative_phosphorylation_deficiency (6350)
89781	HPS4	multiple	1	6	1	Hermansky-Pudlak_syndrome (670)
89884	LHX4	Skeletal	1	2	1	Short_stature (1387)
90411	MCFD2	Hematological	1	7	1	Factor_x_deficiency (520)
91647	ATPAF2	multiple	1	1	1	Complex_mitochondrial_respiratory_chain,_deficiency_of (351)
91949	COG7	Metabolic	1	12	1	Congenital_disorder_of_glycosylation (354)
93986	FOXP2	Neurological	1	0	1	Specific_language_impairment_QTL (1419)
114327	EFHC1	Neurological	1	2	1	Myoclonic_epilepsy (1055)
114548	CIAS1	Grey	3	0	2	CINCA_syndrome (332), Cold-induced_autoinflammatory_syndrome (344), Muckle-Wells_syndrome (1030)
114902	C1QTNF5	Ophthamological	1	3	1	Retinal_cone_dsytrophy (1315)
115761	ARL11	Cancer	1	36	1	Leukemia (886)
116085	SLC22A12	Renal	1	0	1	Hypouricemia (791)
116519	APOA5	Metabolic	1	3	1	Hypertriglyceridemia (757)
117156	SCGB3A2	Respiratory	1	12	1	Asthma (153)
117531	TMC1	Ear,Nose,Throat	1	40	1	Deafness (406)
118429	ANTXR2	Grey	2	1	2	Fibromatosisl (537), Hyalinosis,_infantile_systemic (710)
120227	CYP2R1	Bone	1	1	1	Rickets (1330)
120329	CASP12P1	Immunological	1	1	1	Sepsis (1378)
120892	LRRK2	Neurological	1	10	1	Parkinson_disease (1192)
121278	TPH2	Psychiatric	1	0	1	Unipolar_depression (1544)
122042	LGR8	Renal	1	1	1	Cryptorchidism (392)
123016	TTC8	multiple	1	7	1	Bardet-Biedl_syndrome (183)
123606	NIPA1	Neurological	1	8	1	Spastic_ataxia/paraplegia (1418)
124590	USH1G	multiple	1	7	1	Usher_syndrome (1551)
127534	GJB4	Dermatological	1	2	1	Erythrokeratoderma (507)
128178	EDARADD	Dermatological	1	7	1	Ectodermal_dysplasia (466)
129880	BBS5	multiple	1	7	1	Bardet-Biedl_syndrome (183)

132884	EVC2	Skeletal	1	1	1	Ellis-van_Creveld_syndrome (473)
136647	C7orf11	Dermatological	1	3	1	Trichothiodystrophy (1525)
140803	TRPM6	Renal	1	2	1	Hypomagnesemia (778)
145226	RDH12	Ophthamological	1	6	1	Leber_congenital_amaurosis (873)
145873	MESP2	Skeletal	1	1	1	Spondylocostal_dysostosis (1433)
146059	CDAN1	Hematological	1	8	1	Anemia (110)
146183	OTOA	Ear,Nose,Throat	1	40	1	Deafness (406)
147138	EVER2	Dermatological	1	1	1	Epidermodysplasia_verruciformis (492)
147409	DSG4	Dermatological	1	3	1	Hypotrichosis (788)
148738	HFE2	Metabolic	1	4	1	Hemochromatosis (657)
149998	LIPI	Metabolic	1	3	1	Hypertriglyceridemia (757)
157570	ESCO2	Developmental	1	0	1	Roberts_syndrome (1334)
157680	VPS13B	multiple	1	0	1	Cohen_syndrome (342)
161497	STRC	Ear,Nose,Throat	1	40	1	Deafness (406)
161582	DYX1C1	Psychiatric	1	1	1	Dyslexia (459)
162417	NAGS	Metabolic	1	0	1	N-acetylglutamate_synthase_deficiency (1067)
166785	MMAA	Metabolic	1	2	1	Methylmalonic_aciduria (1004)
170302	ARX	Grey	6	28	2	Infantile_spasm_syndrome (807), Lissencephaly (908), Mental_retardation (990), Myoclonic_epilepsy (1055), Partington_syndrome (1196), Proud_syndrome (1277)
200576	PIP5K3	Ophthamological	1	6	1	Corneal_dystrophy (362)
201163	FLCN	Grey	4	39	3	Birt-Hogg-Dube_syndrome (205), Colon_cancer (346), Pneumothorax,_primary_spontaneous (1239), Renal_cell_carcinoma (1308)
201294	UNC13D	Hematological	1	1	1	Hemophagocytic_lymphohistiocytosis (662)
203859	TMEM16E	Bone	1	0	1	Gnthodiaphyseal_dysplasia (615)
219736	STOX1	Cardiovascular	1	2	1	Preeclampsia (1257)
255738	PCSK9	Metabolic	1	7	1	Hypercholesterolemia (724)
256297	PTF1A	Endocrine	1	26	1	Diabetes_mellitus (427)
257054	MGC25181	Metabolic	1	0	1	D-2-hydroxyglutaric_aciduria (402)
259236	TMIE	Ear,Nose,Throat	1	40	1	Deafness (406)
259266	ASPM	Neurological	1	4	1	Microcephaly (1007)
261734	NPHP4	Renal	2	4	1	Nephronophthisis (1082), Senior-Loken_syndrome (1375)
283120	H19	multiple	1	3	1	Beckwith-Wiedemann_syndrome (194)
285362	SUMF1	Metabolic	1	0	1	Multiple_sulfatase_deficiency (9039)
285440	CYP4V2	Ophthamological	1	0	1	Bietti_crystalline_corneoretinal_dystrophy (201)
319100	TAAR6	Psychiatric	1	8	1	Schizophrenia (1359)
326625	MMAB	Metabolic	1	2	1	Methylmalonic_aciduria (1004)
338435	SCA25	Neurological	1	12	1	Spinocereballar_ataxia (1428)
338917	CHX10	Ophthamological	1	2	1	Microphthalmia (1012)
340024	SLC6A19	Metabolic	1	0	1	Hartnup_disorder (641)
341640	FREM2	multiple	1	1	1	Fraser_syndrome (551)
342880	R9AP	Ophthamological	1	1	1	Bradyopsia (225)
347344	ZNF81	Neurological	1	23	1	Mental_retardation (990)
348938	ICHTHYIN	Dermatological	1	6	1	Ichthyosis (794)
374291	NDUFS7	Neurological	1	11	1	Leigh_syndrome (877)
375298	CERKL	Ophthamological	1	29	1	Retinitis_pigmentosa (1316)
375611	SLC26A5	Ear,Nose,Throat	1	40	1	Deafness (406)
378465	HSN2	Neurological	1	7	1	Neuropathy (1099)
378884	NHLRC1	Neurological	1	16	1	Epilepsy (495)
387082	SUMO4	Endocrine	1	26	1	Diabetes_mellitus (427)
387129	GPR154	Respiratory	1	12	1	Asthma (153)

387281	LCRB	Hematological	1	4	1	Thalassemias (1486)
404672	GTF2H5	Dermatological	1	3	1	Trichothiodystrophy (1525)
493818	MYMY3	Neurological	1	0	1	Moyamoya_disease (1029)