Test Cases

Internal ID	Clinical Indication and Phenotype (HPO)	Diagnosed Disorder	Orphanet ID (Diagnosed Disorder)	Gene	HGNC Gene ID	(NM) or Locus and Genomic Variant (zygosity)	Inheritance Pattern
Stavropoulos et al							
TCS001	HP:0010301, HP:0000252, HP:0001263	Rubinstein-Taybi Syndrome 2	ORPHA353284 (Rubinstein-Taybi syndrome due to EP300 haploinsufficiency)	EP300	3373	NM_001429.3:c.5723dupC (p.Thr1909Asnfs*164) (het)	AD
TCS003	HP:0001263, HP:0002650, HP:0001382, HP:0002079, HP:0001999	Coffin-Siris syndrome	ORPHA1465 (Coffin-Siris syndrome)	SMARCB1	11103	NM_003073.3:c.364del (p.Glu122Asnfs*21) (het)	AD
TCS004	HP:0001263, HP:0004322, HP:0100255	Alazami Syndrome	ORPHA319671 (Microcephalic primordial dwarfism, Alazami type)	LARP7	24912	NM_016648.2:c.756_757del (p.Arg253Ile*6) (hom)	AR
TCS005	HP:0007105	KAT6B-Related Disorder	ORPHA85201 (Genitopatellar syndrome), ORPHA3047 (Blepharophimosis-intellectual disability syndrome, SBBYS type) or some other KAT6B-related disorder	KAT6B	17582	NM_012330.3:c.3021+1G>C (p?) (het)	AD
TCS006	HP:0009372, HP:0001193, HP:0007598, HP:0001762	Type C Brachydactyly	ORPHA93384 (Brachydactyly type C)	GDF5	4220	NM_000557.2:c.847G>A (p.Val283Met) (het)	AD
TCS007	HP:0001263	Neurodegeneration with brain iron accumulation-1 (NBIA1).	ORPHA216866 (Classic pantothenate kinase-associated neurodegeneration) or ORPHA157850 (Pantothenate kinase-associated neurodegeneration)	PANK2	15894	NM_153638.2:c.824_825del (p.Cys276Trpfs*15) (hom)	AR
TCS008	HP:0001263, HP:0002540, HP:0001344, HP:0003693, HP:0001266, HP:0001250, HP:0000563	Congenital disorder of deglycosylation	ORPHA137 (Congenital disorder of deglycosylation	NGLY1	17646	NM_018297.3:c.1201A>T (p.Arg401*) (hom)	AR
TCS009	HP:0000256, HP:0002126, HP:0410009, HP:0012210, HP:0003302, HP:0011342	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome-1 (MPPH)	ORPHA83473 (Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome)	PIK3R2	8980	NM_181523.2:c.1117G>A (p.Gly373Arg) (het)	AD
TCS010	HP:0000252, HP:0001252, HP:0001250	Early Infantile Epileptic Encephalopathy 5.	ORPHA1934 (Early infantile epileptic encephalopathy)	SPTAN1	11273	NM_001130438.2:c.6947A>C (p.Gln2316Pro) (het)	AD
TCS012	HP:0001266, HP:0001263, HP:0001321, HP:0011968	Pontocerebellar Hypoplasia Type 2A.	ORPHA2524 (Pontocerebellar hypoplasia type 2)	TSEN54	27561	NM_207346.2:c.919G>T (p.Ala307Ser) (hom)	AR
TCS013	HP:0001519, HP:0011342, HP:0002194	Sotos Syndrome	ORPHA821 (Sotos syndrome)	NSD1	14234	NM_022455.4:c.3922-1G>C (p.?) (het)	AD
TCS014	HP:0012209, HP:0001642	Noonan Syndrome-like disorder with or without juvenile myelomonocytic leukemia	ORPHA363972 (Noonan syndrome-like disorder with juvenile myelomonocytic leukemia)	CBL	1541	NM_005188.3:c.1096-11_1109del (p.?) (het)	AD

TCS015	HP:0001508, HP:0001250, HP:0000256, HP:0001334, HP:0002921, HP:0002020, HP:0000545, HP:0001252, HP:0011675, HP:0000483, HP:0001647, HP:0001875	Autosomal Dominant Mental Retardation 17.	ORPHA329224 (Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome)	PACS1	30032	NM_018026.3:c.607C>T (p.Arg203Trp) (het)	AD
TCS016	HP:0001252, HP:0001263, HP:0000924	Mental retardation autosomal dominant 23	ORPHA404440 (Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency)	SETD5	25566	NM_001080517:c.1576_1580del (p.Glu526Lysfs*15) (het)	AD
TCS017	HP:0000558, HP:0000668, HP:0001999	SHORT syndrome	ORPHA3163 (SHORT syndrome)	PIK3R1	8979	NM_181523.2:c.1993G>A (p.Gly665Ser) (het)	AD
TCS018	HP:0000407, HP:0000717, HP:0001638	Autosomal Recessive hearing loss caused by GJB2 variant.	ORPHA90636 (Autosomal recessive non-syndromic sensorineural deafness type DFNB)	GJB2	4284	NM_004004.5:c.35delG (p.Gly12Valfs*2) (hom)	AR
TCS021	HP:0001107, HP:0001249, HP:0001513, HP:0007018	Oculocutaneous albinism type 1 and MC4R-linked obesity?	ORPHA352731 (Oculocutaneous albinism type 1)	TYR and MC4R	12442, 6932	NM_000372.4:c.1118C>A (p.Thr373Lys) (het)/c.1205G>A (p.Arg402Gln) (het) and NM_005912.2:c.307G>A (p.Val103Ile)	AR
TCS022	HP:0000519, HP:0001263	COL4A1-Related Disorder	ORPHA75326 (Retinal arterial tortuosity), ORPHA73229 (Autosomal dominant familial hematuria-retinal arteriolar tortuosity-contractures syndrome), ORPHA36383 (Familial vascular leukoencephalopathy), ORPHA2940 (Porencephaly) or ORPHA99810 (Familial porencephaly)	COL4A1	2202	NM_001845.4:c.2317G>A (p.Gly773Arg) (het)	AD
TCS024	HP:0002510, HP:0001263, HP:0001250, HP:0001508, HP:0000505, HP:0001537	Congenital disorder of glycosylation, type Iv and Congenital disorder of glycosylation, type IIi.	ORPHA263487 (COG5-CDG)	NGLY1 and COG5	17646, 14857	NM_001145294.1:c.517A>G p.Arg173Gly (hom) and NM_006348.3:c.1205C>T (p.Ser402Leu) (hom)	AR
TCS025	HP:0006852, HP:0002376, HP:0001945	CAPOS Syndrome and global developmental delay/Autism Spectrum disorder	ORPHA1171 (Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome)	ATP1A3 and NRXN1	801, 8008	arr 2p16.3 (51,021,507-51,358,841)x1 and NM_152296.4:c.2485G>A (p.Glu818Lys) (het)	AD
TCS026	HP:0005484, HP:0001263, HP:0002376, HP:0002123	Pontocerebellar hypoplasia, type 2E	ORPHA247198 (Progressive cerebello-cerebral atrophy)	VPS53	25608	NM_018289:c.1429C>T (p.Arg477*)/c.1716T>G (p.Ser572Arg)	AR

TCS027	HP:0009885, HP:0001087, HP:0001508, HP:0000252, HP:0030048, HP:0003072, HP:0000280, HP:0001252	Nicolaides-Baraitser syndrome	ORPHA3051 (Intellectual disability-sparse hair-brachydactyly syndrome)	SMARCA2	11098	NM_003070:c.2639C>T (p.Thr880Ile)	AD
TCS028	HP:0001250, HP:0001508, HP:0001999, HP:0000260, HP:0001382, HP:0000592, HP:0001263, HP:0001631, HP:0001943, HP:0002757, HP:0000938, HP:0001385, HP:0001903	Cutis laxa, type IIIA	ORPHA35664 (ALDH18A1-related De Barsy syndrome) or ORPHA2962 (De Barsy syndrome) or ORPHA90348 (Autosomal dominant cutis laxa)	ALDH18A1	9722	NM_002860:c.1321C>T (p.Arg441*)/c.191G>A (p.Arg64His)	AR
Bone et al							
TCS031	HP:0000256, HP:0000708, HP:0000718, HP:0001065, HP:0001251, HP:0001263, HP:0001290, HP:0001513, HP:0001770, HP:0001985, HP:0001999, HP:0002020, HP:0002028, HP:0002342, HP:0002360, HP:0002650, HP:0008542, HP:0011412, HP:0100512	Smith-Magenis	ORPHA819 (Smith-Magenis syndrome)	RAI1	9834	NM_030665.3:c.[2273G>A];[=]	
TCS036	HP:0000708, HP:0001250, HP:0001257, HP:0002342, HP:0007319	Combined oxidative phosphorylation deficiency 12	ORPHA314051 (Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome)	AARS2	21022	c.[1949G>A];	
TCS039	HP:0000518, HP:0001257, HP:0001288, HP:0002500, HP:0002518, HP:0003487, HP:0006858, HP:0006886, HP:0006937, HP:0010829, HP:0011448, HP:0011449, HP:0012534, HP:0100561	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	ORPHA363412 (Hypomyelination with brain stem and spinal cord involvement and leg spasticity)	DARS	2678	NM_001349.2:c.[839A>T]; [1099G>C]	
Stelzer et al							
TCS041	HP:0030005	Capillary leak syndrome	ORPHA188 (Systemic capillary leak syndrome)	TLN1	11845		
TCS043	HP:0002804, HP:0002803	Arthrogryposis multiplex congenita	ORPHA1037 (Arthrogryposis multiplex congenita)	MYBPC	7550		
Lee et al							
TCS044	HP:0002376, HP:0001250	ceroid-lipofuscinosis, neuronal 8 [MIM: 600143]	OR:168491, OR:228354, OR:79264	CLN8		p.Arg204Cys	

TCS045	HP:0001251, HP:0012444, HP:0000518, HP:0001272, HP:0002503, HP:0001263, HP:0001332, HP:0001252, HP:0002418, HP:0001270, HP:0012179, HP:0001257	autosomal recessive Joubert syndrome 3 [MIM: 608629]	OR:220493	AHI1		p.Glu1086Gly	
TCS046	HP:0007703, HP:0000662, HP:0000662, HP:0000510, HP:0000572	autosomal recessive Retinitis Pigmentosa type 39 [MIM: 613809]	OR:791	USH2A		p.Cys759Phe	
TCS047	HP:0001251, HP:0001272, HP:0001251	autosomal recessive spinocerebellar ataxia type 8 (SCAR8) [MIM: 610743]	OR:88644	SYNE1		Splice Defect	
TCS048	HP:0001274, HP:0002019, HP:0001999, HP:0000365, HP:0001609, HP:0000601, HP:0001302, HP:0000750	autosomal recessive primary microcephaly type 3 [MIM: 604804]	OR:2512	CDK5RAP2		p.Arg1481*	
TCS049	HP:0007366, HP:0001272, HP:0001347, HP:0001251, HP:0000365, HP:0001347	autosomal recessive Spastic Paraplegia type 7 [MIM: 607259]	OR:99013	SPG7		p.Gly577Ser	
TCS050	HP:0007875, HP:0000622, HP:0000551, HP:0000639, HP:0007702, HP:0007980, HP:0000510, HP:0000510	autosomal recessive cone-rod dystrophy 13 [MIM: 608194]	OR:1872	RPGRIP1		p.Asp966Ilefs	
TCS051	HP:0002804, HP:0001533, HP:0000518, HP:0001999, HP:0001328, HP:0001324, HP:0003560, HP:0002650, HP:0004322	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14 [MIM: 615350]; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14 [MIM: 615351]; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14 [MIM: 615352]	OR:363623, OR:370959, OR:370968, OR:588	GMPPB		p.Arg185Cys	
TCS052	HP:0000823, HP:0001263, HP:0001288, HP:0001347, HP:0040075, HP:0030353, HP:0004322, HP:0012506, HP:0008734	autosomal recessive Woodhouse-Sakati Syndrome [MIM: 241080]	OR:3464	DCAF17		p.Ile97Asnfs*22	
TCS053	HP:0001251, HP:0002415, HP:0009830, HP:0002500	autosomal recessive adult polyglucosan body disease [MIM: 263570]; glycogen storage disease type IV [MIM: 232500]	OR:206583, OR:308621, OR:308638, OR:308655, OR:308670, OR:308684, OR:308698, OR:308712, OR:367, OR:367	GBE1		p.Tyr329Ser	
TCS054	HP:0000717, HP:0001638, HP:0003560, HP:0003201	Duchenne muscular dystrophy (DMD) [MIM: 310200]	OR:98896	DMD		p.Gly1334*	

TCS055	HP:0002910, HP:0010526, HP:0002415, HP:0100513, HP:0001328, HP:0001257, HP:0006827, HP:0002493, HP:0002500	X-linked chronic non-spherocytic hemolytic anemia due to G6PD deficiency [MIM: 305900]		G6PD		p.Ser188Phe	
TCS056	HP:0005110, HP:0001638, HP:0001633, HP:0001634	Cardiomyopathy, dilated, 1G [MIM: 604145]; Cardiomyopathy, familial hypertrophic, 9 [MIM: 613765]	OR:154	TTN		p.Asp17434Gly, p.Cys12844*	
TCS057	HP:0001251	SYNE1-related autosomal recessive cerebellar ataxia (also known as autosomal recessive cerebellar ataxia type 1 or ARCA1) [MIM: 610743]	OR:88644	SYNE1		Splice defect, p.Lys3216*	
TCS058	HP:0001251, HP:0001272, HP:0012103	SYNE1-related autosomal recessive cerebellar ataxia (also known as autosomal recessive cerebellar ataxia type 1 or ARCA1) [MIM: 610743]	OR:88644	SYNE1		p.Arg6684*, p.Trp2646*	
TCS059	HP:0000978, HP:0001873	autosomal recessive Glanzmann thrombasthenia [MIM: 273800]	OR:849	ITGA2B		p.Ile596Thr, p.Leu147Val	
TCS060	HP:0011854, HP:0001733, HP:0012330, HP:0001977	autosomal recessive Wilson Disease [MIM:277900]	OR:905	ATP7B		p.Asn1270Ser, p.Pro984Ala	
TCS061	HP:0007754, HP:0000608, HP:0007754	autosomal recessive Stargardt Disease [MIM: 248200]	OR:827	ABCA4		p.Gly1961Glu, p.Gly863Ala	
TCS062	HP:0000023, HP:0100790	autosomal recessive 17-beta hydroxysteroid dehydrogenase III deficiency (a.k.a. male pseudohermaphroditism with gynecomastia) [MIM: 264300]	OR:46, OR:752	HSD17B3		p.Arg80Gln, p.Ser65Leu	
TCS063	HP:0004430, HP:0002721, HP:0001876	Severe combined immunodeficiency due to ADA deficiency [MIM: 102700]	OR:277	ADA		Splice defect, p.Ala329Val	
TCS064	HP:0001347, HP:0002415, HP:0009830, HP:0001257, HP:0002500	autosomal recessive spastic paraplegia type 7 [MIM: 607259]	OR:99013	SPG7		p.Ala510Val, p.Ala708Lysfs	
TCS065	HP:0003326, HP:0002099, HP:0003687, HP:0100749, HP:0012378, HP:0007185, HP:0001324, HP:0003560, HP:0003474, HP:0012531	autosomal recessive Miyoshi muscular dystrophy-1 [MIM: 254130]; autosomal recessive limb-girdle muscular dystrophy type 2B [MIM: 253601]; distal myopathy with anterior tibial onset [MIM: 606768]	OR:178400, OR:268, OR:45448	DYSF		p.Arg1693Gln, p.Ser1173*	

TCS066	HP:0003326, HP:0003326, HP:0003198	autosomal recessive limb-girdle muscular dystrophy type 2L [MIM: 611397]		ANO5		p.Asn52Ser, p.Asn64Lys*15	
TCS067	HP:0003789, HP:0001324, HP:0003560, HP:0003198	autosomal recessive congenital neuromuscular disease; central core disease [MIM: 117000]	OR:178145, OR:597, OR:598	RYR1		p.Arg1999Cys, p.Arg2241*, p.Asp708Asn	
TCS068	HP:0003202, HP:0001324, HP:0003202, HP:0003560	limb-girdle muscular dystrophy type 2B [MIM: 253601]	OR:268	DYSF		Splice defect, p.Lys1526Thr	
TCS069	HP:0002194, HP:0001252, HP:0001324, HP:0003560, HP:0003198, HP:0000508	autosomal recessive fetal akinesia deformation sequence [MIM: 208150]; familial limb-girdle myasthenia [MIM: 254300]	OR:590, OR:98913, OR:994	DOK7		p.Ser422Hisfs*34, p.Ser422Leufs*97	
TCS070	HP:0001260, HP:0002094, HP:0002875, HP:0001324, HP:0003560, HP:0003473, HP:0003198, HP:0012764	autosomal recessive familial limb-girdle myasthenia [MIM: 254300]	OR:590, OR:98913	DOK7		Splice defect, p.Ala378Serfs*30	
TCS071	HP:0001324, HP:0003560	autosomal recessive limb-girdle muscular dystrophy (LGMD) type 2A [MIM: 253600]	OR:267	CAPN3		p.Arg440Gln, p.Glu107*	
TCS072	HP:0001263, HP:0001324, HP:0003560, HP:0004322	autosomal recessive congenital muscular dystrophy (megaconial type) [MIM: 602541]	OR:280671	CHKB		p.Gln51*, p.Glu283Lys, p.Thr301Ile	
TCS073	HP:0011096, HP:0009830, HP:0003474, HP:0003401	Mitochondrial DNA depletion syndrome 4A (Alpers type) [MIM: 203700]; Mitochondrial DNA depletion syndrome 4B (MNGIE type) [MIM: 613662]; Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE) [MIM: 607459]; Progressive external ophthalmoplegia, autosomal dominant [MIM: 157640]; Progressive external ophthalmoplegia, autosomal recessive [MIM: 258450]	OR:254881, OR:254886, OR:254892, OR:298, OR:70595, OR:726	POLG		p.Arg852Cys, p.Gly11Asp	
TCS074	HP:0009049, HP:0001251, HP:0002196, HP:0009830, HP:0000639, HP:0001257	autosomal recessive spastic ataxia Charlevoix-Saguenay type (ARSACS) [MIM: 270550]	OR:98	SACS		p.Arg2426*, p.Arg3875His	
TCS075	HP:0001251, HP:0001263, HP:0001250, HP:0001257, HP:0001337	autosomal recessive citrullinemia [MIM: 215700]	OR:247525	ASS1		p.Arg307Cys, see eTable 3	
TCS076	HP:0000551, HP:0000510, HP:0007754, HP:0000662, HP:0000510	late onset retinal degeneration (LORD) [MIM:605670]	OR:67042	C1QTNF5		p.Ser163Arg	

TCS077	HP:0000169, HP:0001067, HP:0001631, HP:0001647, HP:0002202, HP:0001638, HP:0001627, HP:0001635, HP:0030148, HP:0100026, HP:0011571, HP:0002202, HP:0001642, HP:0001629	Aortic valve disease [MIM: 109730]	OR:402075	NOTCH1		p.Glu794SerfsX8	
TCS078	HP:0011675, HP:0001695, HP:0001638, HP:0001699	dilated cardiomyopathy (DCM) [MIM: 115200]; hypertrophic cardiomyopathy (HCM) [MIM: 115197]	OR:300751	MYBPC3		Splice Defect	
TCS079	HP:0004749, HP:0000836, HP:0001638, HP:0012622, HP:0001635, HP:0003077, HP:0000822, HP:0002092, HP:0001645, HP:0004756	Atrial fibrillation, familial, 10 [MIM: 614022]; Brugada syndrome 1 [MIM: 601144]; Cardiomyopathy, dilated, 1E [MIM: 601154]; Heart block, nonprogressive [MIM: 113900]; Heart block, progressive, type IA [MIM: 113900]; Long QT syndrome-3 [MIM: 603830]; Sick sinus syndrome 1 [MIM: 608567]; Ventricular fibrillation, familial, 1 [MIM: 603829]	OR:101016, OR:130, OR:154, OR:166282, OR:228140, OR:334, OR:768, OR:871, OR:871	SCN5A		p.Arg523Cys	
TCS080	HP:0005110, HP:0001638, HP:0001252, HP:0011106, HP:0002202	Cardiomyopathy, dilated, 1S [MIM: 613426]; Cardiomyopathy, familial hypertrophic, 1 [MIM: 192600]; Laing distal myopathy [MIM: 160500]; Left ventricular noncompaction 5 [MIM: 613426]; Myopathy, myosin storage [MIM: 608358]; Scapuloperoneal syndrome, myopathic type [MIM: 181430]	OR:154, OR:154, OR:437572, OR:53698, OR:54260, OR:54260, OR:59135	MYH7		p.Met515Thr	
TCS081	HP:0001631, HP:0000518, HP:0000175, HP:0000568, HP:0001643	oculofaciocardiodental (OFCD) syndrome [MIM: 300166]	OR:2712, OR:568	BCOR		p.Gln1337*	
TCS082	HP:0002617, HP:0012727, HP:0012727	autosomal dominant familial thoracic aortic aneurysm type 7 [MIM: 613780]	OR:91387	MYLK		p.Asp717Tyr	
TCS083	HP:0001382, HP:0011729, HP:0003011 and HP:0000924, HP:0012531, HP:0010647	autosomal dominant Ehlers-Danlos syndrome, hypermobility type [MIM: 130020]	OR:285	TNXB		p.Asp2025Val	

TCS084	HP:0009049, HP:0002380, HP:0009027, HP:0001288, HP:0002380, HP:0001324, HP:0009830	Charcot-Marie-Tooth disease, dominant intermediate D [MIM: 607791]; Charcot-Marie-Tooth disease, type 1B [MIM: 118200]; Charcot-Marie-Tooth disease, type 2I [MIM: 607677]; Charcot-Marie-Tooth disease, type 2J [MIM: 607736]; Dejerine-Sottas disease [MIM: 145900]; Neuropathy, congenital hypomyelinating [MIM: 605253]; Roussy-Levy syndrome [MIM:180800]	OR:100046, OR:101082, OR:3115, OR:64748, OR:99942, OR:99943, OR:99951	MPZ		p.Pro151Thr	
TCS085	HP:0003234, HP:0003394, HP:0012378, HP:0001945, HP:0001427, HP:0003326, HP:0001954, HP:0002094	autosomal dominant familial periodic fever [MIM: 142680]	OR:32960	TNFRSF1A		p.Arg121Gln	
TCS086	HP:0002067, HP:0002483, HP:0001268, HP:0002307, HP:0001332, HP:0000712, HP:0000298, HP:0003470, HP:0002371, HP:0002015, HP:0000511	autosomal dominant Parkinson Disease type 11 [MIM: 607688]	OR:411602	GIGYF2		p.Ala793Val	
TCS087	HP:0010463, HP:0002617, HP:0001369, HP:0008724, HP:0010511/HP:0100807, HP:0002758, HP:0000939, HP:0000541, HP:0002650	autosomal dominant Loeys-Dietz syndrome [MIM: 610168, 610380]	OR:284973, OR:558, OR:60030, OR:91387	TGFBR2		p.Tyr470Asp	
TCS088	HP:0001430, HP:0009049, HP:0009027, HP:0003394, HP:0002487, HP:0009830	2-aminoadipic 2-oxoadipic aciduria [MIM: 204750]; Charcot-Marie-Tooth disease, axonal, type 2Q [MIM: 615025]	OR:329258, OR:79154	DHTKD1		p.Arg834*	
TCS089	HP:0000048, HP:0000047, HP:0100600, HP:0000028	46XY sex reversal type 6 [MIM: 613762]	OR:242, OR:251510	MAP3K1		p.Pro257Leu	
TCS090	HP:0002863, HP:0001903, HP:0001876	Emberger syndrome [MIM: 614038]; Immunodeficiency 21 [MIM: 614172]	OR:228423, OR:3226	GATA2		p.Asn371Lys	
TCS091	HP:0003560, HP:0003198, HP:0002380, HP:0002515, HP:0003324	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia [MIM: 613954]; Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1 [MIM: 167320]	OR:275872, OR:52430, OR:803	VCP		p.Arg155Gly	

TCS092	HP:0007269, HP:0003324	autosomal dominant spastic paraplegia type 4 [MIM: 182601]	OR:100985	SPAST		p.Asp542Gly	
TCS093	HP:0001251, HP:0001272, HP:0000365, HP:0000726, HP:0000365, HP:0009830, HP:0001257	autosomal dominant hereditary sensory neuropathy type E1 [MIM: 614116]	OR:456318	DNMT1		p.Tyr511Cys	
TCS094	HP:0011675, HP:0001279, HP:0012668	autosomal dominant Brugada syndrome-1 [MIM: 601144]	OR:130	SCN5A		p.Ser1710Leu	
TCS095	HP:0001999, HP:0001562, HP:0000938, HP:0002652	autosomal dominant osteogenesis imperfecta (OI), type II [MIM:166210]; OI, type III [MIM: 259420]; OI, type IV [MIM: 166220]; autosomal dominant Ehlers-Danlos syndrome type VIIB [MIM: 130060]	OR:1899, OR:216804, OR:216812, OR:216820, OR:666, OR:666, OR:666, OR:99875, OR:99876	COL1A2		p.Gly1012Ser	
TCS096	HP:0001288, HP:0100021, HP:0001263, HP:0002141, HP:0002136	autosomal dominant Angelman syndrome [MIM: 105830]	OR:72	UBE3A		p.Leu825Phefs*15	
TCS097	HP:0002888, HP:0002888	autosomal dominant Li-Fraumeni Syndrome [MIM: 151623]	OR:524	TP53		p.Arg158His	
TCS098	HP:0002607, HP:0100568, HP:0012378, HP:0000131, HP:0002895, HP:0002668, HP:0001337	autosomal dominant hereditary paraganglioma type 1 [MIM: 168000]	OR:29072	SDHD		p.Pro81Leu	
TCS099	HP:0100568, HP:0002668	autosomal dominant paragangliomas [MIM: 115310]	OR:29072	SDHB		p.Arg90*	
TCS100	HP:0000589, HP:0001263, HP:0000639, HP:0001642, HP:0004322, HP:0004381, HP:0001642	autosomal dominant Noonan Syndrome 1 [MIM: 163950]	OR:648	PTPN11		p.Asn308Asp	
TCS101	HP:0001724, HP:0001647, HP:0000164, HP:0001679, HP:0001627, HP:0000975, HP:0008404	autosomal dominant pachyonychia congenita [MIM: 167200]	OR:2309	KRT16		p.Gln122Arg	

TCS102	HP:0005321, HP:0002059, HP:0000453, HP:0000028, HP:0001263, HP:0001999, HP:0002032, HP:0002937, HP:0000023, HP:0040064, HP:0000272, HP:0000308, HP:0002878	autosomal dominant mandibulofacial dysostosis, Guion-Almeida type [MIM: 610536]	OR:79113	EFTUD2		Splice Defect	
TCS103	HP:0001251, HP:0001268, HP:0000726, HP:0002415, HP:0009830, HP:0000572, HP:0002500	autosomal dominant cerebral arteriopathy with subcortical infarcts; leukoencephalopathy [MIM: 125310]	OR:136	NOTCH3		p.Arg1231Cys	
TCS104	HP:0012244, HP:0008723	46XY sex reversal 3 [MIM: 612965]; Adrenocortical insufficiency; Premature ovarian failure 7 [MIM: 612964]; Spermatogenic failure 8 [MIM: 613957]	OR:242, OR:251510, OR:399805	NR5A1		p.Tyr404Asp	
TCS105	HP:0005227, HP:0002097, HP:0002097, HP:0010566	autosomal dominant Birt-Hogg-Dube syndrome [MIM: 135150]; autosomal dominant primary spontaneous pneumothorax [MIM: 173600]	OR:122, OR:2903	FLCN		p.His429Thrfs*39	
TCS106	HP:0002313, HP:0001257	autosomal dominant spastic paraplegia 4 [MIM: 182601]	OR:100985	SPAST		p.Arg562Gln	
TCS107	HP:0007018, HP:0003761, HP:0000646, HP:0012803, HP:0001396, HP:0001263, HP:0000565, HP:0010313, HP:0001399, HP:0000722, HP:0001250, HP:0007902	autosomal dominant hyperphosphatemic familial tumoral calcinosis [MIM: 211900]	OR:306661, OR:53715	FGF23		p.Gly87Asp	
TCS108	HP:0011157, HP:0001510, HP:0001328, HP:0001250	X-linked female-restricted early infantile epileptic encephalopathy type 9 (EIEE9) or Juberg-Hellman Syndrome [MIM: 300088]	OR:101039, OR:2076	PCDH19		p.Ala153Ile	
TCS109	HP:0000365, HP:0000365, HP:0001875, HP:0002895	autosomal dominant Emberger syndrome [MIM: 614038]	OR:3226	GATA2		p.Tyr376Profs*9	
TCS110	HP:0001251, HP:0000819, HP:0000639	autosomal dominant spinocerebellar ataxia-6 [MIM:183086]; episodic ataxia type 2 [MIM:108500]; familial hemiplegic migraine-1 with progressive cerebellar ataxia [MIM:141500]	OR:569, OR:97, OR:98758	CACNA1A		p.Asp302Asn	
TCS111	HP:0009049, HP:0001288, HP:0009830	autosomal recessive Charcot-Marie-Tooth disease [MIM: 607831, 607706, 608340,; 21440]	OR:101097, OR:101097, OR:217055, OR:440, OR:90797, OR:99944	GDAP1		p.Asp21Alafs*23, p.Met116Thr, p.Pro392Ser	

TCS112	HP:0100021, HP:0002019, HP:0001263, HP:0002151, HP:0030895, HP:0001276, HP:0001252	Pitt-Hopkins Syndrome [MIM: 610954]	OR:2896	TCF4		Splice defect	
TCS113	HP:0009049, HP:0003391, HP:0003011, HP:0003202, HP:0001324, HP:0009830, HP:0007269, HP:0040083	Bethlem myopathy [MIM: 158810]; Myosclerosis, congenital [MIM: 255600]; Ullrich congenital muscular dystrophy [MIM: 254090]	OR:289380, OR:610, OR:75840	COL6A2		p.Gly699Asp	
TCS114	HP:0000787, HP:0012444, HP:0001263, HP:0000826, HP:0011157, HP:0001276, HP:0001257	Mowat-Wilson syndrome [MIM: 235730]	OR:2152	ZEB2		p.Asp978Tyr	
TCS115	HP:0001263, HP:0011157, HP:0001263, HP:0001250, HP:0003643	malignant migrating partial seizures of infancy (MMPSI)		KCNT1		p.Arg474His	
TCS116	HP:0001999, HP:0002553, HP:0000218, HP:0000750, HP:0000400, HP:0000527, HP:0000637, HP:0000750, HP:0000682	autosomal dominant Cornelia de Lange syndrome 4 [MIM: 614701]	OR:199	RAD21		p.Leu603Pro	
TCS117	HP:0011927, HP:0100249, HP:0002363, HP:0004484, HP:0000365, HP:0001999, HP:0000968, HP:0000238, HP:0011729, HP:0001270, HP:0000407, HP:0000924, HP:0000407	fibrodysplasia ossificans progressive (FOP) [MIM: 135100]	OR:337	ACVR1		p.Arg258Gly	
TCS118	HP:0001263, HP:0000388, HP:0001250, HP:0002013	early infantile epileptic encephalopathy type 11 [MIM: 613721] or benign familial infantile seizures type 3 [MIM: 607745]	OR:140927, OR:1934, OR:306	SCN2A		p.Ser987Ile	
TCS119	HP:0000365, HP:0001263, HP:0000365, HP:0012469, HP:0001250	Cognitive impairment with or without cerebellar ataxia [MIM: 614306]; Epileptic encephalopathy, early infantile, 13 [MIM: 614558]	OR:442835	SCN8A		p.Arg850Gln	
TCS120	HP:0000483, HP:0001263, HP:0001999, HP:0000316, HP:0001252, HP:0000639, HP:0001642, HP:0000026	autosomal dominant Noonan Syndrome type 3 [MIM: 609942]	OR:648	KRAS		p.Asp153Val	

TCS121	HP:0011448, HP:0012443, HP:0001272, HP:0001321, HP:0002169, HP:0005280, HP:0001263, HP:0001347, HP:0002509, HP:0008936	Dystonia 4, torsion, autosomal dominant [MIM: 128101]; Leukodystrophy, hypomyelinating, 6 [MIM: 612438]	OR:139441, OR:98805	TUBB4A		p.Val255Ile	
TCS122	HP:0030148, HP:0000238, HP:0004415, HP:0001263, HP:0001999, HP:0001250	autosomal dominant Cardiofaciocutaneous Syndrome (CFCS) [MIM: 115150]	OR:1340	MAP2K2		p.Lys61Glu	
TCS123	HP:0001263, HP:0001999, HP:0001891, HP:0001508, HP:0004322, HP:0000486, HP:0004324	autosomal dominant mental retardation type 7 [MIM:614104]	OR:178469	DYRK1A		p.Tyr104 *	
TCS124	HP:0008689, HP:0002019, HP:0001263, HP:0012572, HP:0002039, HP:0000238, HP:0001252, HP:0000023, HP:0001601, HP:0100559, HP:0100259	megalencephaly-capillary malformation-polymicrogyria syndrome (MCAP) [MIM: 602501]	OR:60040	PIK3CA		p.Gly914Arg	
TCS125	HP:0001288, HP:0007018, HP:0000717, HP:0001999, HP:0001621, HP:0001427, HP:0003560, HP:0000722	intellectual disability with language impairment; autistic features [MIM: 613670]	OR:391372	FOXP1		p.Val423His fs*37	
TCS126	HP:0003487, HP:0002415, HP:0001427, HP:0003808, HP:0002385, HP:0001257, HP:0003324	autsomal dominant Aicardi-Goutieres syndrome type 6 [MIM: 615010]; dyschromatosis symmetrica hereditaria [MIM: 127400]	OR:41, OR:51	ADAR		p.Gly1007Arg	
TCS127	HP:0001263, HP:0001999, HP:0001510, HP:0001252, HP:0000347, HP:0000568, HP:0000588, HP:0001144, HP:0000767, HP:0001510, HP:0000541, HP:0002967, HP:0004492	Wiedemann-Steiner syndrome [MIM: 605130]	OR:319182	KMT2A(MLL)		Splice defect	
TCS128	HP:0007875, HP:0007401, HP:0000639, HP:0001105, HP:0030532	Cone-rod retinal dystrophy-2 [MIM: 120970]; Leber congenital amaurosis 7 [MIM: 613829]	OR:1872, OR:65	CRX		p.Glu42Lys	
TCS129	HP:0001263, HP:0001999, HP:0011157, HP:0001385, HP:0001382, HP:0001388, HP:0001250	Turner type intellectual disability syndrome [MIM: 300706]	OR:85328	HUWE1		p.Ala1338Val	
TCS130	HP:0001263, HP:0011157, HP:0011968, HP:0002020, HP:0004305, HP:0000787, HP:0000787, HP:0001583, HP:0001250, HP:0002179, HP:0002013	Epileptic encephalopathy, early infantile, 11 [MIM: 613721]; seizures, benign familial infantile, 3 [MIM: 607745]	OR:140927, OR:1934, OR:306	SCN2A		p.Arg853Gln	

TCS131	HP:0000717, HP:0003688, HP:0001263, HP:0011157, HP:0001276, HP:0001427, HP:0001250, HP:0002179, HP:0000637	Epileptic encephalopathy, early infantile, 11 [MIM: 613721]; seizures, benign familial infantile, 3 [MIM: 607745]	OR:140927, OR:1934, OR:306	SCN2A		p.Glu1211Lys	
TCS132	HP:0000324, HP:0001263, HP:0004325, HP:0004322	X-linked dominant Cornelia de Lange Syndrome [MIM: 30882]		HDAC8		Splice defect	
TCS133	HP:0001643, HP:0012443, HP:0004484, HP:0001263, HP:0001999, HP:0001252, HP:0002637, HP:0000347, HP:0001643	autosomal dominant Beckwith-Wiedemann syndrome [MIM: 130650]; autosomal dominant Sotos syndrome 1 [MIM: 117550]	OR:116, OR:821	NSD1		p.Gln784*	
TCS134	HP:0000987, HP:0006337, HP:0010566, HP:0000365, HP:0002808, HP:0000132, HP:0002076, HP:0000826, HP:0100699, HP:0002650, HP:0000360	autosomal dominant PTEN hamartoma tumor syndrome including Bannayan-Riley-Ruvalcaba syndrome [MIM: 153480]; Cowden syndrome 1 [MIM: 158350]; macrocephaly/autism syndrome [MIM: 605309]	OR:109, OR:201, OR:210548, OR:2969, OR:65285	PTEN		p.Ser287*	
TCS135	HP:0001288, HP:0003560, HP:0003198, HP:0009830, HP:0002086, HP:0003324	autosomal dominant Charcot-Marie-Tooth disease type 2 [MIM: 601472]; distal hereditary motor neuronopathy type V [MIM: 600794]	OR:139536, OR:99938	GARS		p.Glu333Val	
TCS136	HP:0003473, HP:0000954, HP:0000598, HP:0030148, HP:0000126, HP:0001252, HP:0000579	autosomal dominant Say-Barber-Biesecker-Young-Simpson Syndrome (SBBYSS) [MIM: 603736]	OR:3047	KAT6B		p.Gln1321Argfs*20	
TCS137	HP:0000717, HP:0001999, HP:0000286, HP:0000218, HP:0001328, HP:0001166, HP:0000275, HP:0008050, HP:0002650, HP:0000098, HP:0001238, HP:0000446, HP:0000820, HP:0001780	autism type 18 [MIM: 615032]		CHD8		p.Arg773*	
TCS138	HP:0001263, HP:0001999, HP:0001276, HP:0000347, HP:0001257, HP:0001182	intellectual disability type 7 [MIM: 614104]	OR:178469	DYRK1A		p.Lys188Ile	
TCS139	HP:0001251, HP:0012443, HP:0001321, HP:0002019, HP:0001263, HP:0001310, HP:0000486, HP:0001320	Spinocerebellar ataxia 15 [MIM: 606658]; Spinocerebellar ataxia 29, congenital nonprogressive [MIM: 117360]	OR:208513, OR:98769	ITPR1		p.Ser277Ile	
TCS140	HP:0001347, HP:0001268, HP:0001276, HP:0001347, HP:0001276, HP:0001257, HP:0000572	autosomal dominant early onset dementia with or without spastic paraparesis [MIM: 607822]	OR:1020	PSEN1		p.Leu381Val	

TCS141	HP:0004322, HP:0011328, HP:0001999, HP:0000964, HP:0001627, HP:0001252, HP:0001270, HP:0002652, HP:0000750	Noonan syndrome-like disorder with loose anagen hair [MIM: 607721]	OR:2701	SHOC2		p.Ser2Gly	
TCS142	HP:0002019, HP:0100704, HP:0001263, HP:0001999, HP:0001510, HP:0001627, HP:0000047, HP:0001252, HP:0001649, HP:0000505	Mowat-Wilson syndrome [MIM: 235730]	OR:2152	ZEB2		p.Leu727Tyrfs*7	
TCS143	HP:0000717, HP:0001263, HP:0001508, HP:0008551, HP:0001250, HP:0000407, HP:0002179	Bosch-Boonstra-Schaaf optic atrophy syndrome [MIM: 615722]	OR:401777	NR2F1		p.Cys128Arg	
TCS144	HP:0100021, HP:0001263, HP:0001252, HP:0002650, HP:0001250	generalized epilepsy; paroxysmal dyskinesia [MIM: 609446]	OR:79137	KCNMA1		p.Thr352Ala	
TCS145	HP:0002020, HP:0000717, HP:0001263, HP:0004789	autosomal dominant intellectual disability type 5 [MIM: 612621]	OR:178469	SYNGAP1		p.Arg299Profs*48	
TCS146	HP:0007018, HP:0001263, HP:0001252, HP:0000750, HP:0001328, HP:0002650	autosomal dominant intellectual disability type 6 [MIM: 613970]	OR:178469	GRIN2B		p.Arg682Cys	
TCS147	HP:0002019, HP:0001263, HP:0000824, HP:0000126, HP:0001250, HP:0004322, HP:0000074	Cortical dysplasia, complex, with other brain malformations 5 [MIM: 615763]		TUBB2A		p.Gln291Pro	
TCS148	HP:0100021, HP:0100704, HP:0001263, HP:0001250	autosomal dominant early infantile epileptic encephalopathy [MIM: 61459]; autosomal dominant nocturnal frontal lobe epilepsy 5 [MIM: 615005]	OR:98784	KCNT1		p.Arg464His	
TCS149	HP:0009049, HP:0009830	Charcot-Marie-Tooth disease, dominant intermediate D [MIM: 607791]; Charcot-Marie-Tooth disease, type 1B [MIM: 118200]; Charcot-Marie-Tooth disease, type 2I [MIM: 607677]; Charcot-Marie-Tooth disease, type 2J [MIM: 607736]; Dejerine-Sottas disease [MIM: 145900]; Neuropathy, congenital hypomyelinating [MIM: 605253]; Roussy-Levy syndrome [MIM: 180800]	OR:100046, OR:101082, OR:3115, OR:64748, OR:99942, OR:99943, OR:99951	MPZ		p.Gly167Arg	

TCS150	HP:0003269, HP:0000483, HP:0001251, HP:0003429, HP:0006789, HP:0000545, HP:0000639, HP:0002352	autosomal dominant spastic paraplegia 13 [MIM: 605280]; autosomal recessive hypomyelinating leukodystrophy 4 [MIM: 612233]	OR:100994, OR:280270, OR:280288	HSPD1		p.Ala536Val	
TCS151	HP:0003234, HP:0100814, HP:0001999, HP:0001298, HP:0001252, HP:0001480, HP:0000767, HP:0001250, HP:0002079	autosomal dominant early infantile epileptic encephalopathy-7 [MIM: 613720]	OR:439218	KCNQ2		p.Gly281Trp	
TCS152	HP:0007018, HP:0000717, HP:0001263, HP:0000709	autosomal dominant nonprogressive cerebellar ataxia with intellectual disability [MIM: 614756]	OR:314647	CAMTA1		p.Tyr1077Cys	
TCS153	HP:0001263, HP:0011157, HP:0012469, HP:0012650, HP:0001250	Cortical dysplasia, complex, with other brain malformations 5 [MIM: 615763]		TUBB2A		p.Ile345Phe	
TCS154	HP:0010296, HP:0001251, HP:0001272, HP:0001263, HP:0001999, HP:0000577, HP:0000618, HP:0001250, HP:0001257, HP:0000486	autosomal dominant dystonia 12 [MIM: 128235]; autosomal dominant alternating hemiplegia of childhood 2 [MIM: 614820]	OR:2131, OR:71517	ATP1A3		p.Gly325Asp	
TCS155	HP:0000717, HP:0001263, HP:0000819, HP:0001427, HP:0003560	autosomal dominant susceptibility to autism type 18 [MIM: 615032]		CHD8		p.Arg582*	
TCS156	HP:0001007, HP:0001263, HP:0001999, HP:0002020, HP:0011471, HP:0000998, HP:0001252, HP:0000473	autosomal dominant Wiedemann-Steiner syndrome [MIM: 605130]	OR:319182	KMT2A		p.Lys1218Glufs*4	
TCS157	HP:0001263, HP:0001999, HP:0000508, HP:0004322, HP:0000750	autosomal dominant Floating-Harbor syndrome [MIM: 136140]	OR:2044	SRCAP		p.Arg2435*	
TCS158	HP:0001640, HP:0001627, HP:0002019, HP:0001263, HP:0001999, HP:0000545, HP:0000767, HP:0000508, HP:0001250, HP:0004322, HP:0002138	autosomal dominant Noonan syndrome 1 [MIM: 163950]	OR:648	PTPN11		p.Asn308Asp	
TCS159	HP:0001263, HP:0001999, HP:0002415, HP:0001250, HP:0000026, HP:0002500	autosomal dominant Nicolaides-Baraitser syndrome [MIM: 601358]	OR:3051	SMARCA2		p.His1161Arg	
TCS160	HP:0001627, HP:0001680, HP:0001263, HP:0012572, HP:0002020, HP:0001627, HP:0001252, HP:0000388, HP:0004755	autosomal dominant Kabuki syndrome [MIM: 147920]	OR:2322	KMT2D		p.Gln3759*	

TCS161	HP:0000483, HP:0001263, HP:0000565, HP:0001276, HP:0001252, HP:0002415, HP:0000545, HP:0000648, HP:0001290, HP:0002500	autosomal dominant Pitt-Hopkins syndrome [MIM: 610954]	OR:2896	TCF4		p.Gly656Argfs*55	
TCS162	HP:0001263, HP:0001999, HP:0000347, HP:0000639	autosomal dominant Nicolaides-Baraitser Syndrome [MIM: 601358]	OR:3051	SMARCA2		p.Asp534Tyr	
TCS163	HP:0001263, HP:0001999, HP:0001252, HP:0000023, HP:0002058, HP:0002928, HP:0011069, HP:0000028	autosomal dominant Glass Syndrome [MIM: 612313]	OR:251019	SATB2		p.Thr390Ile	
TCS164	HP:0001251, HP:0003560, HP:0003198	autosomal dominant limb-girdle muscular dystrophy [MIM: 159001], autosomal dominant congenital muscular dystrophy [MIM: 613205]; autosomal dominant Emery-Dreifuss muscular dystrophy 2 [MIM: 181350]	OR:157973, OR:261, OR:264, OR:98853	LMNA		Splice defect	
TCS165	HP:0100021, HP:0001263, HP:0001999, HP:0001288, HP:0002650	autosomal dominant intellectual disability 19 [MIM: 615075]	OR:404473	CTNNB1		p.Leu424Arg	
TCS166	HP:0000483, HP:0011995, HP:0001631, HP:0001647, HP:0002019, HP:0004484, HP:0000365, HP:0001263, HP:0001999, HP:0002020, HP:0000365, HP:0001601, HP:0001655	autosomal dominant CHARGE syndrome[MIM: 214800]		CHD7		p.His1734Serfs*3	
TCS167	HP:0002126, HP:0000365, HP:0001263, HP:0011157, HP:0000365, HP:0001252, HP:0000639, HP:0006879, HP:0001250	X-linked dominant intellectual disability; microcephaly with pontine; cerebellar hypoplasia (MICPCH) [MIM: 300749]	OR:163937	CASK		Splice defect	
TCS168	HP:0000175, HP:0004484, HP:0001263, HP:0001999, HP:0000842, HP:0000316, HP:0001943, HP:0001252, HP:0001511, HP:0000347, HP:0000278	autosomal dominant Birk-Barel intellectual disability dysmorphism syndrome [MIM: 612292]	OR:166108	KCNK9		p.Gly236Arg	

TCS169	HP:0001500, HP:0004484, HP:0001263, HP:0001999, HP:0000286, HP:0000286, HP:0011157, HP:0000218, HP:0003186, HP:0001388, HP:0002808, HP:0002003, HP:0000276 and HP:0000275, HP:0000368, HP:0000272, HP:0000767, HP:0000275, HP:0000767, HP:0002650, HP:0001250, HP:0001781	Sotos syndrome [MIM:117550]	OR:821	NSD1		p.Glu1184*	
TCS170	HP:0012443, HP:0001263, HP:0001347, HP:0001257, HP:0002136	autosomal dominant lissencephaly 3 [MIM: 611603]	OR:171680	TUBA1A		p.Val118Met	
TCS171	HP:0001263, HP:0001252, HP:0000508, HP:0004322	autosomal dominant Wiedemann-Steiner syndrome [MIM: 605130]	OR:319182	KMT2A		p.Ser3446Phefs*29	
TCS172	HP:0001508, HP:0001252, HP:0003128	Leigh syndrome, due to COX deficiency [MIM: 256000]	OR:255210, OR:255241, OR:506, OR:70474	SURF1		Splice Defect	
TCS173	HP:0001397, HP:0004420, HP:0003128, HP:0001942, HP:0012103, HP:0002928, HP:0002878, HP:0001714, HP:0002500	Leigh syndrome [MIM:256000]	OR:255210, OR:255241, OR:506, OR:70474	NDUFS8		p.Lys115Glu	
TCS174	HP:0007401, HP:0000551, HP:0000510, HP:0000510, HP:0007875, HP:0007401, HP:0000662, HP:0000639, HP:0000613	Leber congenital amaurosis [MIM: 614186]	OR:65	KCNJ13		p.Thr153Ile	
TCS175	HP:0012444, HP:0003688, HP:0001263, HP:0012572, HP:0001508, HP:0011968, HP:0011968, HP:0001510, HP:0040083	leukocyte adhesion deficiency II (LAD2) [MIM: 266265]	OR:2968, OR:99843	SLC35C1		p.Thr291Ile	
TCS176	HP:0000717, HP:0100543, HP:0001263, HP:0001263, HP:0001252, HP:0004305, HP:0001250, HP:0100716, HP:0002360	autosomal recessive cutis laxa, type IIIA (ARCL3A) [MIM: 219150]	OR:2962, OR:35664	ALDH18A1		p.Arg765Gln	
TCS177	HP:0001999, HP:0001508, HP:0001252, HP:0000347, HP:0011220, HP:0002878, HP:0001250	autosomal recessive non-ketotic hyperglycinemia [MIM: 605899]	OR:407	GLDC		p.Pro581Arg	
TCS178	HP:0000365, HP:0000365	autosomal recessive Usher Syndrome type 2A [MIM: 276901]	OR:231178, OR:886	USH2A		p.Ser3276*	
TCS179	HP:0001263, HP:0000238, HP:0001250	pyridoxine-dependent epilepsy [MIM: 266100]	OR:3006	ALDH7A1		p.Glu427Gln	

TCS180	HP:0001324, HP:0003560, HP:0003198	autosomal recessive Limb-girdle muscular dystrophy type 2G [MIM: 601954]	OR:34514	TCAP		p.Ser11*	
TCS181	HP:0012179, HP:0030038, HP:0000365, HP:0001250	Bart-Pumphrey syndrome [MIM: 149200]; Deafness, autosomal dominant 3A [MIM: 601544]; Deafness, autosomal recessive 1A [MIM: 220290]; Hystrix-like ichthyosis with deafness [MIM: 602540]; Keratitis-ichthyosis-deafness syndrome [MIM: 148210]; Keratoderma, palmoplantar, with deafness [MIM: 148350]; Vohwinkel syndrome [MIM: 124500]	OR:2202, OR:2698, OR:477, OR:477, OR:494, OR:90635, OR:90636	GJB2		p.Val37Ile	
TCS182	HP:0001263, HP:0001999, HP:0011968, HP:0000126, HP:0001276, HP:0000347	autosomal recessive Crisponi syndrome (cold-induced sweating syndrome type 1) [MIM: 272430]	OR:157820	CRLF1		p.Pro239Alafs*91	
TCS183	HP:0000483, HP:0001251, HP:0000009, HP:0012444, HP:0002363, HP:0100543, HP:0000551, HP:0002283, HP:0011096, HP:0001263, HP:0003469, HP:0000648, HP:0007256, HP:0002273, HP:0001257	autosomal recessive adrenoleukodystrophy due to peroxisomal acyl-CoA oxidase deficiency [MIM: 264470]	OR:2971	ACOX1		p.Arg59Pro	
TCS184	HP:0009049, HP:0005109, HP:0001284, HP:0100021, HP:0001371, HP:0011096, HP:0002380, HP:0001324, HP:0009830, HP:0002650, HP:0002179	autosomal recessive Charcot-Marie-Tooth Disease (CMT) type 4C [MIM: 601596]	OR:99949	SH3TC2		p.Arg954*	
TCS185	HP:0012178, HP:0001945, HP:0002721, HP:0001744	autosomal recessive familial hemophagocytic lymphohistiocytosis type 2 [MIM: 603553]	OR:540	PRF1		p.Gln446Pro	
TCS186	HP:0001288, HP:0001347, HP:0007443, HP:0001427, HP:0001324, HP:0001257	autosomal recessive spastic paraplegia type 35 (SPG35) [MIM: 612319]	OR:171629	FA2H		p.Pro173Ser	
TCS187	HP:0001263, HP:0003128, HP:0001252, HP:0000816, HP:0012103, HP:0011968, HP:0002878, HP:0000488	autosomal recessive mitochondrial DNA depletion syndrome types 8A; 8B [MIM: 612075]	OR:254803, OR:255235, OR:298	RRM2B		p.Gly212_Leu213insSer	

TCS188	HP:0002586, HP:0001903, HP:0004348, HP:0100546, HP:0001263, HP:0001999, HP:0003774, HP:0001508, HP:0001627, HP:0000822, HP:0001642, HP:0001250, HP:0001297, HP:0000076	autosomal recessive combined oxidative phosphorylation deficiency type 15 [MIM: 611766]		MTFMT		p.Pro373Glnfs*19	
TCS189	HP:0001903, HP:0002908, HP:0002904, HP:0003128, HP:0001399, HP:0012103, HP:0001873, HP:0002910	autosomal recessive transient infantile liver failure [MIM: 613070]	OR:217371	TRMU		p.Leu253Pro	
TCS190	HP:0001251, HP:0000717, HP:0100021, HP:0001263, HP:0002376, HP:0001298, HP:0001252, HP:0001257	autosomal recessive infantile neuroaxonal dystrophy 1 (neurodegeneration with brain iron accumulation types 2A; 2B) [MIM: 256600] [MIM: 610217]	OR:35069, OR:35069	PLA2G6		p.Gly373Arg	
TCS191	HP:0000646, HP:0000483, HP:0001363, HP:0001263, HP:0001999, HP:0000621, HP:0011157, HP:0001252, HP:0000545, HP:0000639, HP:0000508, HP:0001250, HP:0000026	Alpha-thalassemia myelodysplasia syndrome, somatic [MIM: 300448]; Alpha-thalassemia/mental retardation syndrome [MIM: 301040]; Mental retardation-hypotonic facies syndrome, X-linked [MIM: 309580]	OR:231401, OR:73220, OR:847, OR:93970, OR:93971, OR:93972, OR:93973, OR:93974, OR:93975	ATRX		p.Pro190Ala	
TCS192		X-linked cone-rod dystrophy [MIM: 304020]	OR:1872	RPGR		p.Glu1060ArgfsX18	
TCS193	HP:0002027, HP:0000463, HP:0001263, HP:0001999, HP:0001508, HP:0001945, HP:0001276, HP:0009058, HP:0000527, HP:0030434, HP:0000520, HP:0001250, HP:0003196	Kabuki Syndrome [MIM: 300867]	OR:2322	KDM6A		p.Arg1279*	
TCS194	HP:0000717, HP:0004484, HP:0001999, HP:0000286, HP:0002463, HP:0011800	X-Linked Intellectual Disability Syndromes: Lujan-Fryns Syndrome [MIM:309520]; Opitz-Kaveggia Syndrome [MIM: 305450]	OR:776	MED12		p.Thr617Ala	
TCS195	HP:0004430, HP:0002955, HP:0002721, HP:0000999, HP:0000988, HP:0200043	X-linked recessive severe combined immunodeficiency [MIM: 300400]	OR:276	IL2RG		p.Val152Ala	
TCS196	HP:0003113, HP:0002900, HP:0002902, HP:0001254	autosomal recessive Bartter Syndrome type 3 [MIM: 607364]	OR:112, OR:93605	CLCNKB		p.Leu633*	

TCS197	HP:0001671, HP:0100021, HP:0001263, HP:0001999, HP:0001508, HP:0001627, HP:0001252, HP:0001684	X-linked recessive intellectual disability with hypotonic facies syndrome [MIM: 309580]	OR:73220, OR:93970, OR:93971, OR:93972, OR:93973, OR:93974, OR:93975	ATRX		p.Arg2131Gln	
TCS198	HP:0000717, HP:0001263, HP:0001999, HP:0001252	X-linked intellectual disability type 93 [MIM: 300659]		BRWD3		p.Leu1419Val	
TCS199	HP:0007663, HP:0007994, HP:0000510, HP:0000510, HP:0000572	glucose-6-phosphate deficiency in males [MIM: 305900]		G6PD		p.Ser188Phe	
TCS200	HP:0001284, HP:0001371, HP:0005684, HP:0001252, HP:0001511, HP:0001511, HP:0003198, HP:0200136, HP:0002098	autosomal recessive congenital myasthenic syndrome associated with facial dysmorphism; acetylcholine receptor deficiency [MIM: 608931]; autosomal recessive fetal akinesia deformation sequence [MIM: 208150]	OR:590, OR:98913, OR:994	RAPSN		p.Gln175Arg, p.Thr277Ala	
TCS201	HP:0009806, HP:0001510, HP:0003072, HP:0000843, HP:0002900, HP:0002902, HP:0012408, HP:0000103, HP:0004322	autosomal recessive Bartter syndrome, antenatal, type 1 [MIM: 601678]	OR:112, OR:93604	SLC12A1		p.Ala508Thr, p.Tyr245*	
TCS202	HP:0000717, HP:0000803, HP:0001263, HP:0001252, HP:0003560, HP:0002126	autosomal recessive congenital muscular dystrophy-dystroglycanopathy with brain; eye anomalies type A, 11 [MIM: 615181]	OR:588, OR:899	B3GALNT 2		p.Asp327Asn, p.Glu480_Trp485 dup	
TCS203	HP:0003150, HP:0001263, HP:0011157, HP:0002079, HP:0001250, HP:0001257	familial or infantile myoclonic epilepsy [MIM: 613577]		TBC1D24		p.Cys424Arg, p.Trp406Cys	
TCS204	HP:0007018, HP:0007018, HP:0000708, HP:0000510, HP:0000752, HP:0007018, HP:0000510, HP:0001250, HP:0000572	neuronal ceroid lipofuscinosis (NCL) [MIM:600143]	OR:168491, OR:228354, OR:79264	CLN8		p.Ala71Val, p.Leu188Valfs	
TCS205	HP:0011504, HP:0001133, HP:0000510	autosomal recessive juvenile onset macular dystrophy (Stargardt Disease) [MIM: 248200]	OR:827	CNGB3		p.Arg403Gln, p.Tyr398Cys	
TCS206	HP:0001251, HP:0000365, HP:0006292, HP:0000968, HP:0000365, HP:0000376, HP:0000695, HP:0004322, HP:0006288, HP:0004324	autosomal recessive hypomyelinating leukodystrophy type 8 [MIM:614381]	OR:88637	POLR3B		p.Thr663Ile, p.Val523Glu	

TCS207	HP:0011504, HP:0000551, HP:0030514, HP:0000662, HP:0001133, HP:0000510	autosomal recessive retinitis pigmentosa type 26 (RP26) [MIM: 608380]	OR:791	CERKL		p.Arg257*, p.Asp225Val	
TCS208	HP:0002650, HP:0002910, HP:0000961, HP:0001263, HP:0000363, HP:0002213, HP:0001252, HP:0012429, HP:0003560, HP:0000028, HP:0001583, HP:0002079	autosomal recessive progressive myoclonic epilepsy, type 6 [MIM: 614018]	OR:280620	GOSR2		Splice defect, p.Gly144Trp	
TCS209	HP:0002870, HP:0001385, HP:0001272, HP:0001321, HP:0002472, HP:0001263, HP:0000565, HP:0001385, HP:0001252, HP:0001427, HP:0003198, HP:0001321, HP:0025267, HP:0002515, HP:0002500	GM1-Gangliosidosis type 1 [MIM:230500]	OR:354, OR:79255	GLB1		p.Gly123Arg, p.Thr82Met	
TCS210	HP:0000565, HP:0012072, HP:0002321, HP:0001508, HP:0002315, HP:0010908, HP:0002076, HP:0002018, HP:0000246, HP:0001643, HP:0030148, HP:0004365	autosomal recessive 2-aminoadipic 2-oxoadipic aciduria [MIM:204750]	OR:79154	DHTKD1		p.Arg715Cys, p.Gly729Arg	
TCS211	HP:0001297, HP:0002381, HP:0001298, HP:0000738, HP:0001427, HP:0002180, HP:0001337, HP:0003324	autosomal recessive familial Mediterranean fever [MIM: 249100]	OR:342	MEFV		p.Ala744Ser, p.Lys695Arg	
TCS212	HP:0012115, HP:0001399	autosomal recessive Niemann-Pick disease type C1 (NPC1) [MIM: 257220]	OR:646	NPC1		p.Pro1080IlefsX18, p.Ser738*	
TCS213	HP:0001251, HP:0009830	Amyotrophic lateral sclerosis 4, juvenile [MIM: 602433]; Ataxia-ocular apraxia-2 [MIM: 606002]	OR:357043, OR:64753	SETX		Splice defect, p.Gly2036Arg	
TCS214	HP:0001709, HP:0001320, HP:0001263, HP:0012722, HP:0000545, HP:0001107, HP:0001010, HP:0001539, HP:0000609, HP:0011683	oculocutaneous albinism type IV [MIM: 606574]	OR:79435	SLC45A2, SLC45A2		p.Gly89Aspfs*24, p.Leu485Pro	
TCS215	HP:0001263, HP:0002013, HP:0001298, HP:0003128, HP:0001942	autosomal recessive acyl-CoA dehydrogenase-9 deficiency [MIM: 611126]	OR:99901	ACAD9, ACAD9		p.Phe120Serfs*9, p.Pro616Ser	
TCS216	HP:0006918, HP:0006789, HP:0001251, HP:0003128, HP:0001250, HP:0000988, HP:0001297, HP:0002401	autosomal recessive recurrent infections with encephalopathy, hepatic dysfunction; cardiovascular malformations [MIM: 613759]	OR:306550, OR:306550	FADD		p.Cys105Arg, p.Ser18*	

TCS217	HP:0012378, HP:0001324, HP:0003560, HP:0003198, HP:0012531, HP:0003201	autosomal recessive early-onset myopathy [MIM: 611705]; limb-girdle muscular dystrophy type 2J [MIM: 608807]	OR:140922, OR:289377	TTN		p.Arg32684Thrfs*47, p.Arg33084*	
TCS218	HP:0003128, HP:0001942, HP:0001427, HP:0002098	autosomal recessive lipoylation defect of the 2-ketoacid dehydrogenase complexes		LIPT1		p.Leu327*, p.Trp269*	
TCS219	HP:0002308, HP:0002020, HP:0000670, HP:0001263, HP:0001276, HP:0001250, HP:0001257	autosomal recessive Joubertâ€[?]s syndrome type 3 [MIM: 608629]	OR:220493	AHI1		p.Cys854Phe, p.Ser1123Phe	
TCS220	HP:0000083, HP:0001394, HP:0001394, HP:0001508, HP:0001399, HP:0012120	autosomal recessive methylmalonic aciduria [MIM: 251000]	OR:27, OR:289916, OR:79312	MUT		p.Ala631Glnfs*17, p.Arg108Cys	
TCS221	HP:0001638	autosomal recessive combined oxidative phosphorylation deficiency type 3 (COPD3) [MIM: 61050]		TSFM		p.Arg333Trp, p.Val119Leu	
TCS222	HP:0001263, HP:0001999, HP:0001252, HP:0003202, HP:0003560	autosomal recessive merosin-deficient muscular dystrophy [MIM: 607855]	OR:258	LAMA2		p.Arg683Serfs*21, p.Glu175*	
TCS223	HP:0002910, HP:0011157, HP:0001943, HP:0003128, HP:0001399, HP:0001324, HP:0003201, HP:0001250	autosomal recessive glutaric acidemia IIC [MIM: 231680]	OR:26791	ETFDH		p.Leu496Pro, p.Pro456Leu	
TCS224	HP:0000726, HP:0002415, HP:0001300, HP:0001337	autosomal recessive hypomyelinating leukodystrophy-7 [MIM: 607694]	OR:137639, OR:447893, OR:447896, OR:77295, OR:88637	POLR3A		p.Lys123del, p.Met852Val	
TCS225	HP:0009049, HP:0001251, HP:0009830	autosomal dominant myofibrillar myopathy type 4 [MIM: 609452]; autosomal dominant dilated cardiomyopathy type 1C with or without noncompaction of the left ventricular myocardium [MIM: 601493]	OR:154, OR:54260, OR:98912	LDB3		p.Arg268Cys	
TCS226	HP:0002647	autosomal dominant Ehlers-Danlos syndrome type IV [MIM: 130050]; autosomal dominant Ehlers-Danlos syndrome type III [MIM: 130020]	OR:285, OR:286	COL3A1		p.Gln1366*	

TCS227	HP:0011516, HP:0000551, HP:0000510, HP:0000551, HP:0030532	autosomal dominant optic atrophy type 1 (OPA1) [MIM: 165500]	OR:98673	OPA1		p.Arg711*	
TCS228	HP:0001640, HP:0001638, HP:0002566	dilated cardiomyopathy type 1FF [MIM: 613286]; type 2A [MIM: 611880]; familial hypertrophic cardiomyopathy type 7 [MIM: 613690]; familial restrictive cardiomyopathy [MIM: 115210]	OR:154, OR:154, OR:75249	TNNI3		p.Arg170Gly	
TCS229	HP:0002748, HP:0002757, HP:0000938, HP:0002652, HP:0100512	autosomal dominant osteogenesis imperfecta type II [MIM:166210], type III [MIM: 259420]; type IV [MIM:166220]	OR:216804, OR:216812, OR:216820, OR:666, OR:666, OR:666	COL1A2		p.Pro986Leu	
TCS230	HP:0002750, HP:0001510, HP:0001878, HP:0001971, HP:0002721, HP:0001744	chronic non-spherocytic hemolytic anemia (CNSHA) [MIM: 305900]		G6PD, STAT1		p.Arg274Trp, p.Ser188Phe	
TCS231	HP:0001263, HP:0001999, HP:0011157, HP:0000365, HP:0001302, HP:0001250	autosomal recessive auditory neuropathy; deafness [MIM:601071]	OR:178469, OR:284232, OR:90636, OR:98878	DYNC1H1, OTOF		p.Arg1567Leu, p.Glu1057Lys, p.Glu1700Gln, p.Lys1310dup	
TCS232	HP:0004313, HP:0002721, HP:0001891	common variable immunodeficiency type 2 [MIM: 240500]	OR:1572, OR:98879	TNFRSF13B		p.Ala279Thr, p.Cys104Arg	
TCS233	HP:0011516, HP:0000551, HP:0000510, HP:0000639, HP:0001133, HP:0000613	achromatopsia; cone dystrophy type 4 (COD4) [MIM: 613093]	OR:1871, OR:49382	PDE6C		p.Leu653Pro	
TCS234	HP:0003131, HP:0000097, HP:0000093, HP:0000556, HP:0003131, HP:0000110	autosomal recessive Bothnia retinal dystrophy [MIM: 607475]; autosomal recessive Retinitis punctata albescens [MIM: 136880]	OR:227796, OR:52427, OR:85128	RLBP1		p.Phe96_Phe99del	
TCS235	HP:0000510, HP:0000488	autosomal recessive retinitis pigmentosa type 38 [MIM: 613862]	OR:791	MERTK		p.Leu731Ser	
TCS236	HP:0002721, HP:0005352	autosomal recessive dyskeratosis congenita type 5 [MIM:615190]	OR:1775, OR:3322	RTEL1		p.Arg1010*, p.Ile449Thr	
TCS237	HP:0005110, HP:0002253, HP:0001997, HP:0000822, HP:0003233, HP:0000135, HP:0000821, HP:0001513, HP:0000510, HP:0010535, HP:0100512	autosomal recessive Retinitis Pigmentosa 49 (RP49) [MIM: 613756]	OR:791	CNGA1		p.Arg218*, p.Ser320Phe	
TCS238	HP:0001067	autosomal dominant neurofibromatosis type 1 (NF1) [MIM: 162200]	OR:363700, OR:636	NF1		p.Arg2616*	

TCS239	HP:0001999, HP:0000316, HP:0002885, HP:0008551	autosomal dominant desmoplastic medulloblastoma [MIM: 155255]; susceptibility to familial meningioma [MIM: 607174]	OR:251858, OR:251863, OR:251867, OR:263662, OR:616	SUFU		p.Tyr38Thrfs*58	
TCS240	HP:0005616, HP:0000646, HP:0001263, HP:0001999, HP:0011157, HP:0001250, HP:0004322, HP:0010049, HP:0003298, HP:0000076	autosomal dominant pseudohypoparathyroidism types Ia (PHP Ia) [MIM: 103580]; Ic (PHP Ic) [MIM: 612462]	OR:79443, OR:79444	GNAS		p.Gln294*	
TCS241	HP:0001263, HP:0001999, HP:0001510, HP:0001382, HP:0002079	autosomal dominant SHORT syndrome [MIM: 269880]	OR:3163	PIK3R1		p.Arg649Trp	
TCS242	HP:0000062, HP:0001328	autosomal dominant sex-limited 46,XY sex reversal 3 [MIM:612965]	OR:242, OR:251510	NR5A1		p.Glu51*	
TCS243	HP:0000365, HP:0000365, HP:0007703, HP:0000662, HP:0007703, HP:0000510, HP:0007843	autosomal dominant retinitis pigmentosa (type 37) [MIM: 611131]; enhanced S-cone syndrome [MIM: 268100]	OR:53540, OR:791	NR2E3		p.Gly56Arg	
TCS244	HP:0001631, HP:0000717, HP:0001263, HP:0001999, HP:0000577, HP:0000126, HP:0002904, HP:0000388, HP:0000488, HP:0001537	autosomal dominant Sotos Syndrome [MIM: 117550]	OR:821	NSD1		Splice defect	

Abbreviations used in this Supplementary File

HPO	Human Phenotype Ontology
HGNC	HUGO Gene Nomenclature Committee
NM	*RefSeq* Accession
AD	Autosomal Dominant
AR	Autosomal Recessive