

## Supplementary Table 8. 320 Genetic Disorders of 237 hMitChip3-Genes

Genetic Disorder (ID)		
Gene ID	Symbol	Name
<u>Mental retardation, X-linked nonspecific, 63 (MIM: 300387)</u>		
2182	ACSL4	acyl-CoA synthetase long-chain family member 4
<u>Retinitis pigmentosa-26 (MIM: 608380)</u>		
375298	CERKL	ceramide kinase-like
<u>Charcot-Marie-Tooth disease, type 2A2 (MIM: 609260)</u>		
9927	MFN2	mitofusin 2
<u>Charcot-Marie-Tooth disease, type 2A1 (MIM: 118210)</u>		
23095	KIF1B	kinesin family member 1B
<u>Retinitis pigmentosa-19 (MIM: 601718)</u>		
24	ABCA4	ATP-binding cassette, sub-family A (ABC1), member 4
<u>Spastic paraplegia-13 (MIM: 605280)</u>		
3329	HSPD1	heat shock 60kDa protein 1 (chaperonin)
<u>Dystonia-12 (MIM: 128235)</u>		
478	ATP1A3	ATPase, Na <sup>+</sup> /K <sup>+</sup> transporting, alpha 3 polypeptide
<u>Retinitis pigmentosa-10 (MIM: 180105)</u>		
3614	IMPDH1	IMP (inosine monophosphate) dehydrogenase 1
<u>Spastic paraplegia-7 (MIM: 607259)</u>		
6687	SPG7	spastic paraplegia 7, paraplegin (pure and complicated autosomal recessive)
<u>Cone-rod dystrophy 3 (MIM: 604116)</u>		
24	ABCA4	ATP-binding cassette, sub-family A (ABC1), member 4
<u>Ceroid-lipofuscinosis, neuronal-3, juvenile (MIM: 204200)</u>		
1201	CLN3	ceroid-lipofuscinosis, neuronal 3, juvenile (Batten, Spielmeyer-Vogt disease)
<u>HMG-CoA synthase-2 deficiency (MIM: 605911)</u>		
3158	HMGCS2	3-hydroxy-3-methylglutaryl-Coenzyme A synthase 2 (mitochondrial)
<u>Charcot-Marie-Tooth disease, type 2D (MIM: 601472)</u>		
2617	GARS	glycyl-tRNA synthetase
<u>Charcot-Marie-Tooth disease, type 2B (MIM: 600882)</u>		
7879	RAB7	RAB7, member RAS oncogene family
<u>Hypomagnesemia-2, renal (MIM: 154020)</u>		
486	FXVD2	FXVD domain containing ion transport regulator 2

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<u>Macular degeneration, age-related, 2 (MIM: 153800)</u>		
24	ABCA4	ATP-binding cassette, sub-family A (ABC1), member 4
<u>Leukemia/lymphoma, B-cell, 2 (MIM: 151430)</u>		
596	BCL2	B-cell CLL/lymphoma 2
<u>Deafness, X-linked 1, progressive (MIM: 300356)</u>		
1678	TIMM8A	translocase of inner mitochondrial membrane 8 homolog A (yeast)
<u>Alpha-1-antichymotrypsin deficiency (MIM: 107280)</u>		
12	SERPINA3	serpin peptidase inhibitor, clade A (alpha-1 antiproteinase, antitrypsin), member 3
<u>Alzheimer disease-1, APP-related (MIM: 104760)</u>		
351	APP	amyloid beta (A4) precursor protein (peptidase nexin-II, Alzheimer disease)
<u>Myopathy, myofibrillar, ZASP-related (MIM: 609452)</u>		
11155	LDB3	LIM domain binding 3
<u>Epilepsy, neonatal myoclonic, with suppression-burst pattern (MIM: 609304)</u>		
79751	SLC25A22	solute carrier family 25 (mitochondrial carrier: glutamate), member 22
<u>Progressive external ophthalmoplegia with mitochondrial DNA deletions (MIM: 609286)</u>		
56652	PEO1	progressive external ophthalmoplegia 1
<u>Kanzaki disease (MIM: 609242)</u>		
4668	NAGA	N-acetylgalactosaminidase, alpha-
<u>Schindler disease, type I (MIM: 609241)</u>		
4668	NAGA	N-acetylgalactosaminidase, alpha-
<u>Combined oxidative phosphorylation deficiency (MIM: 609060)</u>		
85476	GFM1	G elongation factor, mitochondrial 1
<u>Myopathy, cardioskeletal, desmin-related, with cataract (MIM: 608810)</u>		
1410	CRYAB	crystallin, alpha B
<u>Hemangioblastoma, cerebellar, somatic (MIM: 608537)</u>		
7428	VHL	von Hippel-Lindau tumor suppressor
<u>Adenomas, multiple colorectal (MIM: 608456)</u>		
4595	MUTYH	mutY homolog (E. coli)
<u>Weill-Marchesani syndrome, dominant (MIM: 608328)</u>		
2200	FBN1	fibrillin 1 (Marfan syndrome)

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<u>Sensory ataxia neuropathy, dysarthria, and ophthalmoparesis (MIM: 607459)</u>		
56652	PEO1	progressive external ophthalmoplegia 1
<u>Autoimmune lymphoproliferative syndrome, type IIB (MIM: 607271)</u>		
841	CASP8	caspace 8, apoptosis-related cysteine peptidase
<u>Microcephaly, Amish type (MIM: 607196)</u>		
60386	SLC25A19	solute carrier family 25 (mitochondrial deoxynucleotide carrier), member 19
<u>Fumarase deficiency (MIM: 606812)</u>		
2271	FH	fumarate hydratase
<u>Goiter, congenital (MIM: 606765)</u>		
7173	TPO	thyroid peroxidase
<u>Hyperinsulinism-hyperammonemia syndrome (MIM: 606762)</u>		
2746	GLUD1	glutamate dehydrogenase 1
<u>Glaucoma, normal tension, susceptibility to (MIM: 606657)</u>		
4976	OPA1	optic atrophy 1 (autosomal dominant)
<u>Propionicacidemia (MIM: 606054)</u>		
5095	PCCA	propionyl Coenzyme A carboxylase, alpha polypeptide
5096	PCCB	propionyl Coenzyme A carboxylase, beta polypeptide
<u>Parkinson disease, early onset (MIM: 605909)</u>		
65018	PINK1	PTEN induced putative kinase 1
<u>Cardiomyopathy, dilated (MIM: 605906)</u>		
11155	LDB3	LIM domain binding 3
<u>Glycine encephalopathy (MIM: 605899)</u>		
2653	GCSH	glycine cleavage system protein H (aminomethyl carrier)
2731	GLDC	glycine dehydrogenase (decarboxylating; glycine decarboxylase, glycine cleavage system
<u>Dimethylglycine dehydrogenase deficiency (MIM: 605850)</u>		
29958	DMGDH	dimethylglycine dehydrogenase
<u>Leiomyomatosis and renal cell cancer (MIM: 605839)</u>		
2271	FH	fumarate hydratase
<u>Amyloidosis, cerebroarterial, Iowa type (MIM: 605714)</u>		
351	APP	amyloid beta (A4) precursor protein (peptidase nexin-II, Alzheimer disease)

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<u>Non-Hodgkin lymphoma, somatic (MIM: 605027)</u>		
843	CASP10	caspase 10, apoptosis-related cysteine peptidase
<u>Cortisone reductase deficiency (MIM: 604931)</u>		
3290	HSD11B1	hydroxysteroid (11-beta) dehydrogenase 1
<u>Emery-Dreifuss muscular dystrophy, AR (MIM: 604929)</u>		
4000	LMNA	lamin A/C
<u>Ventricular tachycardia, stress-induced polymorphic (MIM: 604772)</u>		
845	CASQ2	calsequestrin 2 (cardiac muscle)
<u>Lipodystrophy, familial partial, with decreased subcutaneous fat of face and neck (MIM: 604517)</u>		
10891	PPARGC1A	peroxisome proliferative activated receptor, gamma, coactivator 1, alpha
<u>Alpha-methylacyl-CoA racemase deficiency (MIM: 604489)</u>		
23600	AMACR	alpha-methylacyl-CoA racemase
<u>Myelodysplastic syndrome (MIM: 604443)</u>		
23305	ACSL6	acyl-CoA synthetase long-chain family member 6
<u>Diabetes mellitus, insulin-resistant, with acanthosis nigricans and hypertension (MIM: 604367)</u>		
5468	PPARG	peroxisome proliferative activated receptor, gamma
<u>Non-small cell lung cancer (MIM: 604331)</u>		
26512	DDX26	DEAD/H (Asp-Glu-Ala-Asp/His) box polypeptide 26
<u>MASS syndrome (MIM: 604308)</u>		
2200	FBN1	fibrillin 1 (Marfan syndrome)
<u>Renal tubular acidosis, proximal, with ocular abnormalities (MIM: 604278)</u>		
8671	SLC4A4	solute carrier family 4, sodium bicarbonate cotransporter, member 4
<u>Complex V, mitochondrial respiratory chain, deficiency of (MIM: 604273)</u>		
91647	ATPAF2	ATP synthase mitochondrial F1 complex assembly factor 2
<u>Peters anomaly (MIM: 604229)</u>		
1545	CYP1B1	cytochrome P450, family 1, subfamily B, polypeptide 1
<u>Leukemia, acute myeloid, therapy-related (MIM: 604061)</u>		
10801	SEPT9	septin 9
<u>Autoimmune lymphoproliferative syndrome, type II (MIM: 603909)</u>		
843	CASP10	caspase 10, apoptosis-related cysteine peptidase

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<u>Thalassemia-beta, dominant inclusion-body (MIM: 603902)</u>		
3043	HBB	hemoglobin, beta
<u>Leukoencephalopathy with vanishing white matter (MIM: 603896)</u>		
8893	EIF2B5	eukaryotic translation initiation factor 2B, subunit 5 epsilon, 82kDa
<u>Tyrosinemia, type Ib (MIM: 603758)</u>		
2954	GSTZ1	glutathione transferase zeta 1 (maleylacetoacetate isomerase)
<u>Cardiomyopathy, hypertrophic, early-onset fatal (MIM: 603646)</u>		
1355	COX15	COX15 homolog, cytochrome c oxidase assembly protein (yeast)
<u>Hepatic failure, early onset, and neurologic disorder (MIM: 603644)</u>		
6341	SCO1	SCO cytochrome oxidase deficient homolog 1 (yeast)
<u>GRACILE syndrome (MIM: 603358)</u>		
617	BCS1L	BCS1-like (yeast)
<u>Pseudohypoparathyroidism, type Ib (MIM: 603233)</u>		
2778	GNAS	GNAS complex locus
<u>Methylmalonate semialdehyde dehydrogenase deficiency (MIM: 603178)</u>		
4329	ALDH6A1	aldehyde dehydrogenase 6 family, member A1
<u>Mitochondrial neurogastrointestinal encephalomyopathy syndrome (MIM: 603041)</u>		
1890	ECGF1	endothelial cell growth factor 1 (platelet-derived)
<u>Merkel cell carcinoma, somatic (MIM: 602690)</u>		
6392	SDHD	succinate dehydrogenase complex, subunit D, integral membrane protein
<u>Carbohydrate-deficient glycoprotein syndrome, type Ib (MIM: 602579)</u>		
4351	MPI	mannose phosphate isomerase
<u>Ossification of the posterior longitudinal spinal ligaments (MIM: 602475)</u>		
1291	COL6A1	collagen, type VI, alpha 1
<u>Ethylmalonic encephalopathy (MIM: 602473)</u>		
23474	ETHE1	ethylmalonic encephalopathy 1
<u>AGAT deficiency (MIM: 602360)</u>		
2628	GATM	glycine amidinotransferase (L-arginine:glycine amidinotransferase)
<u>Encephalopathy, progressive mitochondrial, with proximal renal tubulopathy due to cytochrome c oxidase deficiency (MIM: 602125)</u>		
1352	COX10	COX10 homolog, cytochrome c oxidase assembly protein, heme A: farnesyltransferase (yeast)

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<u>Autoimmune lymphoproliferative syndrome (MIM: 601859)</u>		
355	FAS	Fas (TNF receptor superfamily, member 6)
<u>Glaucoma, early-onset, digenic (MIM: 601771)</u>		
1545	CYP1B1	cytochrome P450, family 1, subfamily B, polypeptide 1
<u>Obesity, severe (MIM: 601665)</u>		
5468	PPARG	peroxisome proliferative activated receptor, gamma
7350	UCP1	uncoupling protein 1 (mitochondrial, proton carrier)
7351	UCP2	uncoupling protein 2 (mitochondrial, proton carrier)
7352	UCP3	uncoupling protein 3 (mitochondrial, proton carrier)
<u>Neural tube defects, maternal risk of (MIM: 601634)</u>		
4522	MTHFD1	methylenetetrahydrofolate dehydrogenase (NADP+ dependent) 1, methenyltetrahydrofolate
<u>Blood group, Froese (MIM: 601551)</u>		
6521	SLC4A1	solute carrier family 4, anion exchanger, member 1 (erythrocyte membrane protein band 3, Diego)
<u>Blood group, Swann (MIM: 601550)</u>		
6521	SLC4A1	solute carrier family 4, anion exchanger, member 1 (erythrocyte membrane protein band 3, Diego)
<u>Obesity, resistance to (MIM: 601487)</u>		
5468	PPARG	peroxisome proliferative activated receptor, gamma
<u>Myopathy, desmin-related, cardioskeletal (MIM: 601419)</u>		
1674	DES	desmin
<u>Brody myopathy (MIM: 601003)</u>		
487	ATP2A1	ATPase, Ca <sup>++</sup> transporting, cardiac muscle, fast twitch 1
<u>Fatty liver, acute, of pregnancy (MIM: 600890)</u>		
3030	HADHA	hydroxyacyl-Coenzyme A dehydrogenase/3-ketoacyl-Coenzyme A thiolase/enoyl-Coenzyme
<u>Spinal muscular atrophy, distal, type V (MIM: 600794)</u>		
2617	GARS	glycyl-tRNA synthetase
<u>Epilepsy, idiopathic generalized, susceptibility to (MIM: 600669)</u>		
4200	ME2	malic enzyme 2, NAD(+)-dependent, mitochondrial
<u>Mitochondrial myopathy and sideroblastic anemia (MIM: 600462)</u>		
80324	PUS1	pseudouridylate synthase 1
<u>Dementia, frontotemporal (MIM: 600274)</u>		
5663	PSEN1	presenilin 1 (Alzheimer disease 3)

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<u>Colorectal cancer (MIM: 600040)</u>		
581	BAX	BCL2-associated X protein
<u>pyruvate decarboxylase deficiency (MIM: 312170)</u>		
5160	PDHA1	pyruvate dehydrogenase (lipoamide) alpha 1
<u>Ornithine transcarbamylase deficiency (MIM: 311250)</u>		
5009	OTC	ornithine carbamoyltransferase
<u>Jensen syndrome (MIM: 311150)</u>		
1678	TIMM8A	translocase of inner mitochondrial membrane 8 homolog A (yeast)
<u>Brunner syndrome (MIM: 309850)</u>		
4128	MAOA	monoamine oxidase A
<u>Keratosis follicularis spinulosa decalvans (MIM: 308800)</u>		
6303	SAT	spermidine/spermine N1-acetyltransferase
<u>Glycerol kinase deficiency (MIM: 307030)</u>		
2710	GK	glycerol kinase
<u>Chronic granulomatous disease, X-linked (MIM: 306400)</u>		
1536	CYBB	cytochrome b-245, beta polypeptide (chronic granulomatous disease)
<u>Mohr-Tranebjaerg syndrome (MIM: 304700)</u>		
1678	TIMM8A	translocase of inner mitochondrial membrane 8 homolog A (yeast)
<u>Anemia, sideroblastic, with ataxia (MIM: 301310)</u>		
22	ABCB7	ATP-binding cassette, sub-family B (MDR/TAP), member 7
<u>Anemia, sideroblastic/hypochromic (MIM: 301300)</u>		
212	ALAS2	aminolevulinate, delta-, synthase 2 (sideroblastic/hypochromic anemia)
<u>Adrenoleukodystrophy (MIM: 300100)</u>		
215	ABCD1	ATP-binding cassette, sub-family D (ALD), member 1
<u>Wilson disease (MIM: 277900)</u>		
540	ATP7B	ATPase, Cu <sup>++</sup> transporting, beta polypeptide
<u>Thyroid hormone organification defect IIA (MIM: 274500)</u>		
7173	TPO	thyroid peroxidase
<u>Sulfite oxidase deficiency (MIM: 272300)</u>		
6821	SUOX	sulfite oxidase

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<u>Succinic semialdehyde dehydrogenase deficiency (MIM: 271980)</u>		
7915	ALDH5A1	aldehyde dehydrogenase 5 family, member A1 (succinate-semialdehyde dehydrogenase)
<u>Sarcosinemia (MIM: 268900)</u>		
1757	SARDH	sarcosine dehydrogenase
<u>Saccharopinuria (MIM: 268700)</u>		
10157	AASS	aminoadipate-semialdehyde synthase
<u>Pyruvate carboxylase deficiency (MIM: 266150)</u>		
5091	PC	pyruvate carboxylase
<u>Glutathione synthetase deficiency (MIM: 266130)</u>		
2937	GSS	glutathione synthetase
<u>vitamin D-dependent rickets type I (MIM: 264700)</u>		
1594	CYP27B1	cytochrome P450, family 27, subfamily B, polypeptide 1
<u>Porphyria, congenital erythropoietic (MIM: 263700)</u>		
7390	UROS	uroporphyrinogen III synthase (congenital erythropoietic porphyria)
<u>Polycythemia, benign familial (MIM: 263400)</u>		
7428	VHL	von Hippel-Lindau tumor suppressor
<u>Myopathy due to phosphoglycerate mutase deficiency (MIM: 261670)</u>		
5224	PGAM2	phosphoglycerate mutase 2 (muscle)
<u>Pseudo-Zellweger syndrome (MIM: 261515)</u>		
30	ACAA1	acetyl-Coenzyme A acyltransferase 1 (peroxisomal 3-oxoacyl-Coenzyme A thiolase)
<u>Pancreatic cancer (MIM: 260350)</u>		
7157	TP53	tumor protein p53 (Li-Fraumeni syndrome)
<u>Hyperoxaluria, primary, type II (MIM: 260000)</u>		
9380	GRHPR	glyoxylate reductase/hydroxypyruvate reductase
<u>Leigh syndrome (MIM: 256000)</u>		
617	BCS1L	BCS1-like (yeast)
1355	COX15	COX15 homolog, cytochrome c oxidase assembly protein (yeast)
4724	NDUFS4	NADH dehydrogenase (ubiquinone) Fe-S protein 4, 18kDa (NADH-coenzyme Q reductase)
374291	NDUFS7	NADH dehydrogenase (ubiquinone) Fe-S protein 7, 20kDa (NADH-coenzyme Q reductase)
4728	NDUFS8	NADH dehydrogenase (ubiquinone) Fe-S protein 8, 23kDa (NADH-coenzyme Q reductase)
4723	NDUFV1	NADH dehydrogenase (ubiquinone) flavoprotein 1, 51kDa

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6389	SDHA	succinate dehydrogenase complex, subunit A, flavoprotein (Fp)
6834	SURF1	surfeit 1
<u>CPT deficiency, hepatic, type IA (MIM: 255120)</u>		
1374	CPT1A	carnitine palmitoyltransferase 1A (liver)
<u>Myeloperoxidase deficiency (MIM: 254600)</u>		
4353	MPO	myeloperoxidase
<u>Ullrich congenital muscular dystrophy (MIM: 254090)</u>		
1291	COL6A1	collagen, type VI, alpha 1
<u>Mitochondrial respiratory chain complex II deficiency (MIM: 252011)</u>		
6389	SDHA	succinate dehydrogenase complex, subunit A, flavoprotein (Fp)
<u>Mitochondrial complex I deficiency (MIM: 252010)</u>		
4719	NDUFS1	NADH dehydrogenase (ubiquinone) Fe-S protein 1, 75kDa (NADH-coenzyme Q reductase)
4720	NDUFS2	NADH dehydrogenase (ubiquinone) Fe-S protein 2, 49kDa (NADH-coenzyme Q reductase)
4724	NDUFS4	NADH dehydrogenase (ubiquinone) Fe-S protein 4, 18kDa (NADH-coenzyme Q reductase)
4726	NDUFS6	NADH dehydrogenase (ubiquinone) Fe-S protein 6, 13kDa (NADH-coenzyme Q reductase)
4723	NDUFV1	NADH dehydrogenase (ubiquinone) flavoprotein 1, 51kDa
<u>Mitochondrial DNA-depletion syndrome, hepatocerebral form (MIM: 251880)</u>		
1716	DGUOK	deoxyguanosine kinase
8803	SUCLA2	succinate-CoA ligase, ADP-forming, beta subunit
7084	TK2	thymidine kinase 2, mitochondrial
<u>Methylmalonic aciduria, muttype (MIM: 251000)</u>		
4594	MUT	methylmalonyl Coenzyme A mutase
<u>Methemoglobinemia, type I (MIM: 250800)</u>		
1727	CYB5R3	cytochrome b5 reductase 3
<u>Maple syrup urine disease, type Ib (MIM: 248611)</u>		
594	BCKDHB	branched chain keto acid dehydrogenase E1, beta polypeptide (maple syrup urine disease)
<u>Maple syrup urine disease, type II (MIM: 248610)</u>		
1629	DBT	dihydrolipoamide branched chain transacylase E2
<u>Maple syrup urine disease, type Ia (MIM: 248600)</u>		
593	BCKDHA	branched chain keto acid dehydrogenase E1, alpha polypeptide

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<u>Malonyl-CoA decarboxylase deficiency (MIM: 248360)</u>		
23417	MLYCD	malonyl-CoA decarboxylase
<u>Malaria, resistance to (MIM: 248310)</u>		
4843	NOS2A	nitric oxide synthase 2A (inducible, hepatocytes)
<u>Fundus flavimaculatus (MIM: 248200)</u>		
24	ABCA4	ATP-binding cassette, sub-family A (ABC1), member 4
<u>Miller-Dieker lissencephaly (MIM: 247200)</u>		
7531	YWHAE	tyrosine 3-monooxygenase/tryptophan 5-monooxygenase activation protein, epsilon
<u>HMG-CoA lyase deficiency (MIM: 246450)</u>		
3155	HMGCL	3-hydroxymethyl-3-methylglutaryl-Coenzyme A lyase (hydroxymethylglutaricaciduria)
<u>Ketoacidosis due to SCOT deficiency (MIM: 245050)</u>		
5019	OXCT1	3-oxoacid CoA transferase 1
<u>Hyperprolinemia, type II (MIM: 239510)</u>		
8659	ALDH4A1	aldehyde dehydrogenase 4 family, member A1
<u>Hyperprolinemia, type I (MIM: 239500)</u>		
5625	PRODH	proline dehydrogenase (oxidase) 1
<u>Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (MIM: 238970)</u>		
10166	SLC25A15	solute carrier family 25 (mitochondrial carrier; ornithine transporter) member 15
<u>Hyperlysinemia (MIM: 238700)</u>		
10157	AASS	aminoadipate-semialdehyde synthase
<u>N-acetylglutamate synthase deficiency (MIM: 237310)</u>		
162417	NAGS	N-acetylglutamate synthase
<u>Carbamoylphosphate synthetase I deficiency (MIM: 237300)</u>		
1373	CPS1	carbamoyl-phosphate synthetase 1, mitochondrial
<u>Homocystinuria-megaloblastic anemia, cbl E type (MIM: 236270)</u>		
4552	MTRR	5-methyltetrahydrofolate-homocysteine methyltransferase reductase
<u>Glycogen storage disease VII (MIM: 232800)</u>		
5213	PFKM	phosphofructokinase, muscle
<u>Hemolytic anemia due to glutathione synthetase deficiency (MIM: 231900)</u>		
2937	GSS	glutathione synthetase

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<u>Glutaricaciduria, type IIA (MIM: 231680)</u>		
2108	ETF A	electron-transfer-flavoprotein, alpha polypeptide (glutaric aciduria II)
2109	ETFB	electron-transfer-flavoprotein, beta polypeptide
2110	ETFDH	electron-transferring-flavoprotein dehydrogenase
<u>Glutaricaciduria, type I (MIM: 231670)</u>		
2639	GCDH	glutaryl-Coenzyme A dehydrogenase
<u>Gaucher disease with cardiovascular calcification (MIM: 231005)</u>		
2629	GBA	glucosidase, beta; acid (includes glucosylceramidase)
<u>Gaucher disease (MIM: 230800)</u>		
2629	GBA	glucosidase, beta; acid (includes glucosylceramidase)
<u>Friedreich ataxia (MIM: 229300)</u>		
2395	FXN	frataxin
<u>Epidermolysis bullosa, Herlitz junctional type (MIM: 226700)</u>		
3914	LAMB3	laminin, beta 3
<u>Epidermolysis bullosa, generalized atrophic benign (MIM: 226650)</u>		
3914	LAMB3	laminin, beta 3
<u>Hemolytic anemia due to bisphosphoglycerate mutase deficiency (MIM: 222800)</u>		
669	BPGM	2,3-bisphosphoglycerate mutase
<u>Leigh syndrome, French-Canadian type (MIM: 220111)</u>		
10128	LRPPRC	leucine-rich PPR-motif containing
<u>Cerebrotendinous xanthomatosis (MIM: 213700)</u>		
1593	CYP27A1	cytochrome P450, family 27, subfamily A, polypeptide 1
<u>Carnitine-acylcarnitine translocase deficiency (MIM: 212138)</u>		
788	SLC25A20	solute carrier family 25 (carnitine/acylcarnitine translocase), member 20
<u>Adenocarcinoma of lung, somatic (MIM: 211980)</u>		
673	BRAF	v-raf murine sarcoma viral oncogene homolog B1
5519	PPP2R1B	protein phosphatase 2 (formerly 2A), regulatory subunit A (PR 65), beta isoform
<u>Aspartylglucosaminuria (MIM: 208400)</u>		
175	AGA	aspartylglucosaminidase
<u>Alpha-methylacetoacetic aciduria (MIM: 203750)</u>		
38	ACAT1	acetyl-Coenzyme A acetyltransferase 1 (acetoacetyl Coenzyme A thiolase)

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<u>Alexander disease (MIM: 203450)</u>		
4723	NDUFV1	NADH dehydrogenase (ubiquinone) flavoprotein 1, 51kDa
<u>Adrenal cortical carcinoma (MIM: 202300)</u>		
7157	TP53	tumor protein p53 (Li-Fraumeni syndrome)
<u>Lipoid adrenal hyperplasia (MIM: 201710)</u>		
6770	STAR	steroidogenic acute regulator
<u>VLCAD deficiency (MIM: 201475)</u>		
37	ACADVL	acyl-Coenzyme A dehydrogenase, very long chain
<u>Acyl-CoA dehydrogenase, medium chain, deficiency of (MIM: 201450)</u>		
34	ACADM	acyl-Coenzyme A dehydrogenase, C-4 to C-12 straight chain
<u>von Hippel-Lindau syndrome (MIM: 193300)</u>		
7428	VHL	von Hippel-Lindau tumor suppressor
<u>Cardiomyopathy, familial hypertrophic (MIM: 192600)</u>		
4624	MYH6	myosin, heavy polypeptide 6, cardiac muscle, alpha (cardiomyopathy, hypertrophic 1)
<u>Histiocytoma (MIM: 191170)</u>		
7157	TP53	tumor protein p53 (Li-Fraumeni syndrome)
<u>Hemolytic anemia due to triosephosphate isomerase deficiency (MIM: 190450)</u>		
7167	TPI1	triosephosphate isomerase 1
<u>Colon cancer, advanced (MIM: 190090)</u>		
6714	SRC	v-src sarcoma (Schmidt-Ruppin A-2) viral oncogene homolog (avian)
<u>Hypertension, pregnancy-induced (MIM: 189800)</u>		
4846	NOS3	nitric oxide synthase 3 (endothelial cell)
<u>Spinal muscular atrophy, late-onset, Finkel type (MIM: 182980)</u>		
9217	VAPB	VAMP (vesicle-associated membrane protein)-associated protein B and C
<u>Shprintzen-Goldberg syndrome (MIM: 182212)</u>		
2200	FBN1	fibrillin 1 (Marfan syndrome)
<u>Sepiapterin reductase deficiency (MIM: 182125)</u>		
6697	SPR	sepiapterin reductase (7,8-dihydrobiopterin:NADP+ oxidoreductase)
<u>Schizophrenia, susceptibility to (MIM: 181500)</u>		
1312	COMT	catechol-O-methyltransferase

**Supplementary Table 8. 320 Genetic Disorders of 237 hMitChip3-Genes**

<b>Genetic Disorder (ID)</b>		
<b>Gene ID</b>	<b>Symbol</b>	<b>Name</b>
<u>Emery-Dreifuss muscular dystrophy, AD (MIM: 181350)</u>		
4000	LMNA	lamin A/C
<u>Pyloric stenosis, infantile hypertrophic, susceptibility to (MIM: 179010)</u>		
4842	NOS1	nitric oxide synthase 1 (neuronal)
<u>Protoporphyrinemia, erythropoietic (MIM: 177000)</u>		
2235	FECH	ferrochelatase (protoporphyrinemia)
<u>Diabetes mellitus, rare form (MIM: 176730)</u>		
3630	INS	insulin
<u>Breast cancer, sporadic (MIM: 176705)</u>		
5245	PHB	prohibitin
<u>Porphyria variegata (MIM: 176200)</u>		
5498	PPOX	protoporphyrinogen oxidase
<u>McCune-Albright syndrome (MIM: 174800)</u>		
2778	GNAS	GNAS complex locus
<u>Pick disease (MIM: 172700)</u>		
5663	PSEN1	presenilin 1 (Alzheimer disease 3)
<u>Hemolytic anemia due to glucosephosphate isomerase deficiency (MIM: 172400)</u>		
2821	GPI	glucose phosphate isomerase
<u>Hemolytic anemia due to phosphofructokinase deficiency (MIM: 171860)</u>		
5211	PFKL	phosphofructokinase, liver
<u>Pheochromocytoma (MIM: 171300)</u>		
6390	SDHB	succinate dehydrogenase complex, subunit B, iron sulfur (lp)
6392	SDHD	succinate dehydrogenase complex, subunit D, integral membrane protein
7428	VHL	von Hippel-Lindau tumor suppressor
<u>Hyperapobetalipoproteinemia, susceptibility to (MIM: 170998)</u>		
5465	PPARA	peroxisome proliferative activated receptor, alpha
<u>Parkinson disease, susceptibility to (MIM: 168600)</u>		
4729	NDUFV2	NADH dehydrogenase (ubiquinone) flavoprotein 2, 24kDa
<u>Panic disorder, susceptibility to (MIM: 167870)</u>		
1312	COMT	catechol-O-methyltransferase

**Supplementary Table 8. 320 Genetic Disorders of 237 hMitChip3-Genes**

<b>Genetic Disorder (ID)</b>		
<b>Gene ID</b>	<b>Symbol</b>	<b>Name</b>
<u>Buschke-Ollendorff syndrome (MIM: 166700)</u>		
23592	LEMD3	LEM domain containing 3
<u>Osseous heteroplasia, progressive (MIM: 166350)</u>		
2778	GNAS	GNAS complex locus
<u>Optic atrophy and cataract (MIM: 165300)</u>		
80207	OPA3	optic atrophy 3 (autosomal recessive, with chorea and spastic paraplegia)
<u>Colorectal cancer, somatic (MIM: 164757)</u>		
673	BRAF	v-raf murine sarcoma viral oncogene homolog B1
<u>Viral infection, susceptibility to (MIM: 164350)</u>		
4938	OAS1	2',5'-oligoadenylate synthetase 1, 40/46kDa
<u>Nucleoside phosphorylase deficiency, immunodeficiency due to (MIM: 164050)</u>		
4860	NP	nucleoside phosphorylase
<u>Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency (MIM: 164008)</u>		
4792	NFKBIA	nuclear factor of kappa light polypeptide gene enhancer in B-cells inhibitor, alpha
<u>Coronary spasms, susceptibility to (MIM: 163729)</u>		
4846	NOS3	nitric oxide synthase 3 (endothelial cell)
<u>Nasopharyngeal carcinoma (MIM: 161550)</u>		
7157	TP53	tumor protein p53 (Li-Fraumeni syndrome)
<u>Bethlem myopathy (MIM: 158810)</u>		
1291	COL6A1	collagen, type VI, alpha 1
<u>Progressive external ophthalmoplegia with mitochondrial DNA deletions (MIM: 157640)</u>		
291	SLC25A4	solute carrier family 25 (mitochondrial carrier; adenine nucleotide translocator), member 4
<u>Migraine, susceptibility to (MIM: 157300)</u>		
2099	ESR1	estrogen receptor 1
<u>Melorheostosis with osteopoikilosis (MIM: 155950)</u>		
23592	LEMD3	LEM domain containing 3
<u>Marfan syndrome (MIM: 154700)</u>		
2200	FBN1	fibrillin 1 (Marfan syndrome)
<u>Longevity, reduced (MIM: 152430)</u>		
11216	AKAP10	A kinase (PRKA) anchor protein 10

**Supplementary Table 8. 320 Genetic Disorders of 237 hMitChip3-Genes**

<b>Genetic Disorder (ID)</b>		
<b>Gene ID</b>	<b>Symbol</b>	<b>Name</b>
<u>Lipodystrophy, familial partial (MIM: 151660)</u>		
4000	LMNA	lamin A/C
5468	PPARG	peroxisome proliferative activated receptor, gamma
<u>Li-Fraumeni syndrome (MIM: 151623)</u>		
7157	TP53	tumor protein p53 (Li-Fraumeni syndrome)
<u>Multiple cutaneous and uterine leiomyomata (MIM: 150800)</u>		
2271	FH	fumarate hydratase
<u>Charcot-Marie-Tooth disease, (MIM: 150330)</u>		
4000	LMNA	lamin A/C
<u>Lactate dehydrogenase-B deficiency (MIM: 150100)</u>		
3945	LDHB	lactate dehydrogenase B
<u>Exertional myoglobinuria due to deficiency of LDH-A (MIM: 150000)</u>		
3939	LDHA	lactate dehydrogenase A
<u>Hypertension, susceptibility to (MIM: 145500)</u>		
4843	NOS2A	nitric oxide synthase 2A (inducible, hepatocytes)
4846	NOS3	nitric oxide synthase 3 (endothelial cell)
<u>Renal cell carcinoma, clear cell, somatic (MIM: 144700)</u>		
4968	OGG1	8-oxoguanine DNA glycosylase
<u>Trifunctional protein deficiency, type II (MIM: 143450)</u>		
3032	HADHB	hydroxyacyl-Coenzyme A dehydrogenase/3-ketoacyl-Coenzyme A thiolase/enoyl-Coenzyme
<u>Hemolytic anemia due to hexokinase deficiency (MIM: 142600)</u>		
3098	HK1	hexokinase 1
<u>Thrombophilia due to heparin cofactor II deficiency (MIM: 142360)</u>		
3053	SERPIND1	serpin peptidase inhibitor, clade D (heparin cofactor), member 1
<u>HPFH, nondeletion type G (MIM: 142250)</u>		
3048	HBG2	hemoglobin, gamma G
<u>Erythremias, beta- (MIM: 141900)</u>		
3043	HBB	hemoglobin, beta
<u>Pituitary ACTH secreting adenoma (MIM: 139320)</u>		
2778	GNAS	GNAS complex locus

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Genetic Disorder (ID)		
Gene ID	Symbol	Name
<u>Glyoxalase II deficiency (MIM: 138760)</u>		
3029	HAGH	hydroxyacylglutathione hydrolase
<u>Apolipoprotein H deficiency (MIM: 138700)</u>		
350	APOH	apolipoprotein H (beta-2-glycoprotein I)
<u>Hemolytic anemia due to glutathione peroxidase deficiency (MIM: 138320)</u>		
2876	GPX1	glutathione peroxidase 1
<u>Hemolytic anemia due to glutathione reductase deficiency (MIM: 138300)</u>		
2936	GSR	glutathione reductase
<u>Hyperammonemia with hypooornithinemia, hypocitrullinemia, hypoargininemia, and hypoprolinemia (MIM: 138250)</u>		
5832	ALDH18A1	aldehyde dehydrogenase 18 family, member A1
<u>Cortisol resistance (MIM: 138040)</u>		
2908	NR3C1	nuclear receptor subfamily 3, group C, member 1 (glucocorticoid receptor)
<u>Glioblastoma, susceptibility to (MIM: 137800)</u>		
5468	PPARG	peroxisome proliferative activated receptor, gamma
<u>Glaucoma, primary open angle, adult-onset (MIM: 137760)</u>		
1545	CYP1B1	cytochrome P450, family 1, subfamily B, polypeptide 1
<u>Glaucoma, primary open angle, juvenile-onset (MIM: 137750)</u>		
1545	CYP1B1	cytochrome P450, family 1, subfamily B, polypeptide 1
<u>Gastric cancer, somatic (MIM: 137215)</u>		
843	CASP10	caspase 10, apoptosis-related cysteine peptidase
4595	MUTYH	mutY homolog (E. coli)
<u>GABA-transaminase deficiency (MIM: 137150)</u>		
18	ABAT	4-aminobutyrate aminotransferase
<u>Aortic aneurysm, ascending, and dissection (MIM: 134797)</u>		
2200	FBN1	fibrillin 1 (Marfan syndrome)
<u>Squamous cell carcinoma, burn scar-related, somatic (MIM: 134637)</u>		
355	FAS	Fas (TNF receptor superfamily, member 6)
<u>Trichothiodystrophy (MIM: 133510)</u>		
2071	ERCC3	excision repair cross-complementing rodent repair deficiency, complementation group 3

**Supplementary Table 8. 320 Genetic Disorders of 237 hMitChip3-Genes**

<b>Genetic Disorder (ID)</b>		
<b>Gene ID</b>	<b>Symbol</b>	<b>Name</b>
<u>Breast cancer (MIM: 133430)</u>		
2099	ESR1	estrogen receptor 1
<u>Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas (MIM: 132600)</u>		
4595	MUTYH	mutY homolog (E. coli)
<u>Ectopia lentis, familial (MIM: 129600)</u>		
2200	FBN1	fibrillin 1 (Marfan syndrome)
<u>DNA topoisomerase I, camptothecin-resistant (MIM: 126420)</u>		
7150	TOP1	topoisomerase (DNA) I
<u>Diabetes mellitus, noninsulin-dependent (MIM: 125853)</u>		
2820	GPD2	glycerol-3-phosphate dehydrogenase 2 (mitochondrial)
<u>MODY, one form (MIM: 125850)</u>		
3630	INS	insulin
<u>Darier disease (MIM: 124200)</u>		
488	ATP2A2	ATPase, Ca <sup>++</sup> transporting, cardiac muscle, slow twitch 2
<u>Mephenytoin poor metabolizer (MIM: 124020)</u>		
1557	CYP2C19	cytochrome P450, family 2, subfamily C, polypeptide 19
<u>Mitochondrial complex III deficiency (MIM: 124000)</u>		
617	BCS1L	BCS1-like (yeast)
7381	UQCRB	ubiquinol-cytochrome c reductase binding protein
<u>Coproporphyrinuria (MIM: 121300)</u>		
1371	CPOX	coproporphyrinogen oxidase
<u>Paroxysmal nonkinesigenic dyskinesia (MIM: 118800)</u>		
25953	PNKD	paroxysmal nonkinesigenic dyskinesia
<u>Acatlasemia (MIM: 115500)</u>		
847	CAT	catalase
<u>Cardiomyopathy, dilated (MIM: 115200)</u>		
4607	MYBPC3	myosin binding protein C, cardiac
<u>Carcinoid tumors, intestinal (MIM: 114900)</u>		
6392	SDHD	succinate dehydrogenase complex, subunit D, integral membrane protein

**Supplementary Table 8. 320 Genetic Disorders of 237 hMitChip3-Genes**

<b>Genetic Disorder (ID)</b>		
<b>Gene ID</b>	<b>Symbol</b>	<b>Name</b>
<u>Hepatocellular carcinoma, somatic (MIM: 114550)</u>		
841	CASP8	caspase 8, apoptosis-related cysteine peptidase
7157	TP53	tumor protein p53 (Li-Fraumeni syndrome)
<u>Colonic adenoma recurrence, reduced risk of (MIM: 114500)</u>		
4953	ODC1	ornithine decarboxylase 1
5320	PLA2G2A	phospholipase A2, group IIA (platelets, synovial fluid)
7157	TP53	tumor protein p53 (Li-Fraumeni syndrome)
<u>Breast cancer (MIM: 114480)</u>		
7157	TP53	tumor protein p53 (Li-Fraumeni syndrome)
<u>Blood group, Wright (MIM: 112050)</u>		
6521	SLC4A1	solute carrier family 4, anion exchanger, member 1 (erythrocyte membrane protein band 3, Diego)
<u>Blood group, Waldner (MIM: 112010)</u>		
6521	SLC4A1	solute carrier family 4, anion exchanger, member 1 (erythrocyte membrane protein band 3, Diego)
<u>Blood group, OK (MIM: 111380)</u>		
682	BSG	basigin (Ok blood group)
<u>Blood group, MN (MIM: 111300)</u>		
2993	GYPA	glycophorin A (MNS blood group)
<u>Blood group, Diego (MIM: 110500)</u>		
6521	SLC4A1	solute carrier family 4, anion exchanger, member 1 (erythrocyte membrane protein band 3, Diego)
<u>Acanthocytosis, one form (MIM: 109270)</u>		
6521	SLC4A1	solute carrier family 4, anion exchanger, member 1 (erythrocyte membrane protein band 3, Diego)
<u>Amyloidosis, renal (MIM: 105200)</u>		
4069	LYZ	lysozyme (renal amyloidosis)
<u>Amyloidosis, Finnish type (MIM: 105120)</u>		
2934	GSN	gelsolin (amyloidosis, Finnish type)
<u>Amyloidosis, secondary, susceptibility to (MIM: 104770)</u>		
325	APCS	amyloid P component, serum
<u>Alzheimer disease, susceptibility to (MIM: 104300)</u>		
4353	MPO	myeloperoxidase
4846	NOS3	nitric oxide synthase 3 (endothelial cell)

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<b>Genetic Disorder (ID)</b>		
<b>Gene ID</b>	<b>Symbol</b>	<b>Name</b>
<u>Aldosteronism, glucocorticoid-remediable (MIM: 103900)</u>		
1584	CYP11B1	cytochrome P450, family 11, subfamily B, polypeptide 1
<u>Aldolase A deficiency (MIM: 103850)</u>		
226	ALDOA	aldolase A, fructose-bisphosphate
<u>Pseudohypoparathyroidism, type Ia (MIM: 103580)</u>		
2778	GNAS	GNAS complex locus
<u>Hemolytic anemia due to adenylate kinase deficiency (MIM: 103000)</u>		
203	AK1	adenylate kinase 1
<u>Acromegaly (MIM: 102200)</u>		
2778	GNAS	GNAS complex locus
<u>Acrokeratosis verruciformis (MIM: 101900)</u>		
488	ATP2A2	ATPase, Ca <sup>++</sup> transporting, cardiac muscle, slow twitch 2
<u>Fetal alcohol syndrome (MIM: 100650)</u>		
217	ALDH2	aldehyde dehydrogenase 2 family (mitochondrial)
<u>Amyotrophic lateral sclerosis, due to SOD1 deficiency (MIM: 105400)</u>		
6647	SOD1	superoxide dismutase 1, soluble (amyotrophic lateral sclerosis 1 (adult))
<u>Cardiomyopathy, dilated, 1A (MIM: 115200)</u>		
4000	LMNA	lamin A/C
<u>Optic atrophy 1 (MIM: 165500)</u>		
4976	OPA1	optic atrophy 1 (autosomal dominant)
<u>Paragangliomas, familial nonchromaffin, 1, with or without deafness (MIM: 168000)</u>		
6392	SDHD	succinate dehydrogenase complex, subunit D, integral membrane protein
<u>Pyruvate dehydrogenase E1-beta deficiency (MIM: 179060)</u>		
5162	PDHB	pyruvate dehydrogenase (lipoamide) beta
<u>Lacticacidemia due to PDX1 deficiency (MIM: 245349)</u>		
8050	PDHX	pyruvate dehydrogenase complex, component X
<u>Hyperoxaluria, primary, type 1 (MIM: 259900)</u>		
189	AGXT	alanine-glyoxylate aminotransferase (oxalosis I; hyperoxaluria I; glycolicaciduria; serine-pyruvate
<u>Cardiomyopathy, dilated, 1D (MIM: 601494)</u>		
7139	TNNT2	troponin T type 2 (cardiac)

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<b>Genetic Disorder (ID)</b>		
<b>Gene ID</b>	<b>Symbol</b>	<b>Name</b>
<u>Cardiomyopathy, dilated, 1I (MIM: 604765)</u>		
1674	DES	desmin
<u>Cardiomyopathy, dilated, 1P (MIM: 609909)</u>		
5350	PLN	phospholamban
<u>Aminoacylase 1 deficiency (MIM: 609924)</u>		
95	ACY1	aminoacylase 1
<u>Cardiomyopathy, familial hypertrophic, 2 (MIM: 115195)</u>		
7139	TNNT2	troponin T type 2 (cardiac)
<u>Cataract, posterior polar 2 (MIM: 123590)</u>		
1410	CRYAB	crystallin, alpha B
<u>Fanconi anemia, complementation group D2 (MIM: 227646)</u>		
2177	FANCD2	Fanconi anemia, complementation group D2
<u>2-methylbutyrylglucosuria (MIM: 600301)</u>		
36	ACADSB	acyl-Coenzyme A dehydrogenase, short/branched chain
<u>Hypothyroidism, congenital, due to DUOX2 deficiency (MIM: 607200)</u>		
50506	DUOX2	dual oxidase 2
<u>progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2 (MIM: 609283)</u>		
291	SLC25A4	solute carrier family 25 (mitochondrial carrier; adenine nucleotide translocator), member 4
<u>Amyloidosis, 3 or more types (MIM: 107680)</u>		
335	APOA1	apolipoprotein A-I
<u>Atrial septal defect 3 (MIM: 160710)</u>		
4624	MYH6	myosin, heavy polypeptide 6, cardiac muscle, alpha (cardiomyopathy, hypertrophic 1)
<u>3-beta-hydroxysteroid dehydrogenase, type II, deficiency (MIM: 201810)</u>		
3284	HSD3B2	hydroxy-delta-5-steroid dehydrogenase, 3 beta- and steroid delta-isomerase 2
<u>Glaucoma 3A, primary congenital (MIM: 231300)</u>		
1545	CYP1B1	cytochrome P450, family 1, subfamily B, polypeptide 1
<u>3-hydroxyacyl-CoA dehydrogenase deficiency (MIM: 231530)</u>		
3033	HADHSC	L-3-hydroxyacyl-Coenzyme A dehydrogenase, short chain
<u>3-methylglutaconic aciduria, type I (MIM: 250950)</u>		
549	AUH	AU RNA binding protein/enoyl-Coenzyme A hydratase

## Supplementary Table 8. 320 Genetic Disorders of 237 hMitChip3-Genes

Genetic Disorder (ID)		
Gene ID	Symbol	Name
<u>3-methylglutaconicaciduria, type III (MIM: 258501)</u>		
80207	OPA3	optic atrophy 3 (autosomal recessive, with chorea and spastic paraplegia)
<u>Paragangliomas, familial nonchromaffin, 3 (MIM: 605373)</u>		
6391	SDHC	succinate dehydrogenase complex, subunit C, integral membrane protein, 15kDa
<u>Alzheimer disease, type 3 (MIM: 607822)</u>		
5663	PSEN1	presenilin 1 (Alzheimer disease 3)
<u>Cardiomyopathy, familial hypertrophic, 4 (MIM: 115197)</u>		
4607	MYBPC3	myosin binding protein C, cardiac
<u>Paraganglioma, familial chromaffin, 4 (MIM: 115310)</u>		
6390	SDHB	succinate dehydrogenase complex, subunit B, iron sulfur (lp)
<u>Hemochromatosis, type 4 (MIM: 606069)</u>		
30061	SLC40A1	solute carrier family 40 (iron-regulated transporter), member 1
<u>Immunodeficiency with hyper IgM, type 4 (MIM: 608106)</u>		
7374	UNG	uracil-DNA glycosylase
<u>Methemoglobinemia due to cytochrome b5 deficiency (MIM: 250790)</u>		
1528	CYB5A	cytochrome b5 type A (microsomal)
<u>Gyrate atrophy of choroid and retina with ornithinemia, B6 responsive or unresponsive (MIM: 258870)</u>		
4942	OAT	ornithine aminotransferase (gyrate atrophy)
<u>Episodic ataxia, type 6 (MIM: 600111)</u>		
6507	SLC1A3	solute carrier family 1 (glial high affinity glutamate transporter), member 3
<u>Amyotrophic lateral sclerosis 8 (MIM: 608627)</u>		
9217	VAPB	VAMP (vesicle-associated membrane protein)-associated protein B and C
<u>Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency (MIM: 202010)</u>		
1584	CYP11B1	cytochrome P450, family 11, subfamily B, polypeptide 1
<u>Methylmalonic aciduria, vitamin B12-responsive (MIM: 251100)</u>		
166785	MMAA	methylmalonic aciduria (cobalamin deficiency) cbIA type
<u>Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cbIB complementation type (MIM: 251100)</u>		
326625	MMAB	methylmalonic aciduria (cobalamin deficiency) cbIB type
<u>2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency (MIM: 300438)</u>		
3028	HADH2	hydroxyacyl-Coenzyme A dehydrogenase, type II

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<b>Genetic Disorder (ID)</b>		
<b>Gene ID</b>	<b>Symbol</b>	<b>Name</b>
<u>3-Methylcrotonyl-CoA carboxylase 2 deficiency (MIM: 210210)</u>		
64087	MCCC2	methylcrotonoyl-Coenzyme A carboxylase 2 (beta)
<u>CD59 deficiency (MIM: 107271)</u>		
966	CD59	CD59 antigen, complement regulatory protein
<u>Renal tubular acidosis, distal, 179800 (MIM: 602722)</u>		
6521	SLC4A1	solute carrier family 4, anion exchanger, member 1 (erythrocyte membrane protein band 3, Diego
<u>Spina bifida, risk of}, 601634 (MIM: 182940)</u>		
4552	MTRR	5-methyltetrahydrofolate-homocysteine methyltransferase reductase
<u>Prostate cancer 1, 176807 (MIM: 601518)</u>		
6041	RNASEL	ribonuclease L (2',5'-oligoadenylate synthetase-dependent)
<u>Schizophrenia, susceptibility to, 4} 600850 (MIM: 606810)</u>		
5625	PRODH	proline dehydrogenase (oxidase) 1