

Supplementary table 3. Genes containing a rare predicted loss of function variant in cases and controls

Gene Symbol	Gene Name	Pathway	Chr	AMD Locus	AMD Locus Name	Total rare variants in cases	Total rare variants in controls
<i>ABCA1</i>	ATP-binding cassette, sub-family A (ABC1), member 1	Lipids	9	16	<i>ABCA1</i>	12	8
<i>ABCA7</i>	ATP-binding cassette, sub-family A (ABC1), member 7	Lipids	19	29	<i>CNN2</i>	6	8
<i>ACAD10</i>	acyl-CoA dehydrogenase family, member 10	Lipids	12	20	<i>ACAD10</i>	5	6
<i>ADAMTS9</i>	ADAM metalloproteinase with thrombospondin type 1 motif, 9	Extracellular Matrix	3	3	<i>ADAMTS9-AS2</i>	5	3
<i>AGER</i>	advanced glycosylation end product-specific receptor	Immune System	6	8	<i>C2/CFB/SKIV2L</i>	2	1
<i>ALDH2</i>	aldehyde dehydrogenase 2 family (mitochondrial)	Unknown/other	12	20	<i>ACAD10</i>	2	2
<i>ALG2</i>	ALG2, alpha-1,3/1,6-mannosyltransferase	Unknown/other	9	15	<i>TGFBR1</i>	0	2
<i>ARHGAP21</i>	Rho GTPase activating protein 21	Unknown/other	10	17	<i>ARHGAP21</i>	7	3
<i>ARL16</i>	ADP-ribosylation factor-like 16	Unknown/other	17	27	<i>NPLOC4/TSPAN10</i>	2	1
<i>ASPM</i>	asp (abnormal spindle) homolog, microcephaly associated (Drosophila)	Unknown/other	1	1	<i>CFH</i>	1	3
<i>ATF6B</i>	activating transcription factor 6 beta	Cell Survival	6	8	<i>C2/CFB/SKIV2L</i>	1	0
<i>B3GALTL</i>	beta 1,3-galactosyltransferase-like	Unknown/other	13	21	<i>B3GALTL</i>	3	6
<i>BAHCC1</i>	BAH domain and coiled-coil containing 1	Unknown/other	17	27	<i>NPLOC4/TSPAN10</i>	6	0
<i>BAIAP2L2</i>	BAI1-associated protein 2-like 2	Unknown/other	13	34	<i>SLC16A8</i>	1	0
<i>BCAM</i>	basal cell adhesion molecule (Lutheran blood group)	Extracellular Matrix	19	30	<i>APOE</i>	2	0
<i>BCAR1</i>	breast cancer anti-estrogen resistance 1	Unknown/other	16	25	<i>CTRB2/CTRB1</i>	4	8
<i>BLOC1S1</i>	biogenesis of lysosomal organelles complex-1, subunit 1	Metabolism	12	19	<i>RDH5/CD63</i>	1	0
<i>C2</i>	complement component 2	Complement	6	8	<i>C2/CFB/SKIV2L</i>	2	0
<i>C3</i>	complement component 3	Complement	19	28	<i>C3</i>	1	1
<i>C6orf10</i>	chromosome 6 open reading frame 10	Unknown/other	6	8	<i>C2/CFB/SKIV2L</i>	0	1
<i>C6orf48</i>	chromosome 6 open reading frame 48	Unknown/other	6	8	<i>C2/CFB/SKIV2L</i>	1	1
<i>C7orf61</i>	chromosome 7 open reading frame 61	Unknown/other	7	11	<i>PILRB/PILRA</i>	0	1
<i>C9</i>	complement component 9	Complement	5	6	<i>C9</i>	2	1
<i>CAPS</i>	calcyphosine	Metabolism	19	28	<i>C3</i>	0	1
<i>CASP6</i>	caspace 6, apoptosis-related cysteine peptidase	Cell Survival	4	5	<i>CFI</i>	3	1
<i>CATSPERD</i>	catsper channel auxiliary subunit delta	Unknown/other	19	28	<i>C3</i>	1	1
<i>CCDC109B</i>	coiled-coil domain containing 109B	Unknown/other	4	5	<i>CFI</i>	1	5
<i>CD3EAP</i>	CD3e molecule, epsilon associated protein	Unknown/other	19	30	<i>APOE</i>	2	3
<i>CETP</i>	cholesteryl ester transfer protein, plasma	Lipids	16	24	<i>CETP</i>	2	0
<i>CFB</i>	complement factor B	Complement	6	8	<i>C2/CFB/SKIV2L</i>	1	3
<i>CFH</i>	complement factor H	Complement	1	1	<i>CFH</i>	5	0
<i>CFHR3</i>	complement factor H-related 3	Complement	1	1	<i>CFH</i>	3	1
<i>CFHR4</i>	complement factor H-related 4	Complement	1	1	<i>CFH</i>	1	0
<i>CFHR5</i>	complement factor H-related 5	Complement	1	1	<i>CFH</i>	1	0
<i>CFI</i>	complement factor I	Complement	4	5	<i>CFI</i>	12	2
<i>CKM</i>	creatine kinase, muscle	Unknown/other	19	30	<i>APOE</i>	4	0
<i>CLPTM1</i>	cleft lip and palate associated transmembrane protein 1	Unknown/other	19	30	<i>APOE</i>	0	1
<i>CNN2</i>	calponin 2	Immune System	19	29	<i>CNN2</i>	1	0
<i>COL15A1</i>	collagen, type XV, alpha 1	Extracellular Matrix	9	15	<i>TGFBR1</i>	5	4
<i>COL4A3</i>	collagen, type IV, alpha 3 (Goodpasture antigen)	Extracellular Matrix	2	2	<i>COL4A3</i>	4	5
<i>COL8A1</i>	collagen, type VIII, alpha 1	Extracellular Matrix	3	4	<i>COL8A1</i>	1	1
<i>CTRB1</i>	chymotrypsinogen B1	Extracellular Matrix	16	25	<i>CTRB2/CTRB1</i>	1	0
<i>CTSA</i>	cathepsin A	Unknown/other	20	31	<i>MMP9</i>	1	2
<i>DAB2</i>	Dab, mitogen-responsive phosphoprotein, homolog 2 (Drosophila)	Unknown/other	5	6	<i>C9</i>	2	2
<i>DOM3Z</i>	decapping exoribonuclease	Unknown/other	6	8	<i>C2/CFB/SKIV2L</i>	0	1
<i>EGFL8</i>	EGF-like-domain, multiple 8	Unknown/other	6	8	<i>C2/CFB/SKIV2L</i>	1	3
<i>EHMT2</i>	euchromatic histone-lysine N-methyltransferase 2	Cell Survival	6	8	<i>C2/CFB/SKIV2L</i>	1	0

<i>ERCC2</i>	excision repair cross-complementing rodent repair deficiency, complementation group 2	Cell Survival	19	30	<i>APOE</i>	5	5
<i>F13B</i>	coagulation factor XIII, B polypeptide	Complement	1	1	<i>CFH</i>	2	4
<i>FKBP1</i>	FK506 binding protein like	Unknown/other	6	8	<i>C2/CFB/SKIV2L</i>	0	1
<i>FSCN2</i>	fascin homolog 2, actin-bundling protein, retinal (Strongylocentrotus purpuratus)	Extracellular Matrix	17	27	<i>NPLOC4/TSPAN10</i>	4	4
<i>FUT6</i>	fucosyltransferase 6 (alpha (1,3) fucosyltransferase)	Unknown/other	19	28	<i>C3</i>	3	1
<i>GAR1</i>	GAR1 ribonucleoprotein	Cell Survival	4	5	<i>CFI</i>	2	3
<i>GDF11</i>	growth differentiation factor 11	Unknown/other	12	19	<i>RDH5/CD63</i>	1	1
<i>GPR108</i>	G protein-coupled receptor 108	Unknown/other	19	28	<i>C3</i>	2	1
<i>GPSM3</i>	G-protein signaling modulator 3	Immune System	6	8	<i>C2/CFB/SKIV2L</i>	1	0
<i>GRIN3B</i>	glutamate receptor, ionotropic, N-methyl-D-aspartate 3B	Unknown/other	19	29	<i>CNN2</i>	0	1
<i>HERPUD1</i>	homocysteine-inducible, endoplasmic reticulum stress-inducible, ubiquitin-like domain member 1	Cell Survival	16	24	<i>CETP</i>	2	1
<i>HGS</i>	hepatocyte growth factor-regulated tyrosine kinase substrate	Unknown/other	17	27	<i>NPLOC4/TSPAN10</i>	6	6
<i>HSPA1L</i>	heat shock 70kDa protein 1-like	Cell Survival	6	8	<i>C2/CFB/SKIV2L</i>	1	0
<i>HSPH1</i>	heat shock 105kDa/110kDa protein 1	Cell Survival	13	21	<i>B3GALT1</i>	2	2
<i>HTRA1</i>	HtrA serine peptidase 1	Extracellular Matrix	10	18	<i>ARMS2/HTRA1</i>	2	1
<i>ITGA7</i>	integrin, alpha 7	Extracellular Matrix	12	19	<i>RDH5/CD63</i>	12	13
<i>KCNT2</i>	potassium channel, subfamily T, member 2	Unknown/other	1	1	<i>CFH</i>	0	2
<i>KIAA1217</i>	KIAA1217	Unknown/other	10	17	<i>ARHGAP21</i>	7	3
<i>KLC3</i>	kinesin light chain 3	Unknown/other	19	30	<i>APOE</i>	2	3
<i>KMT2E</i>	lysine (K)-specific methyltransferase 2E	Cell Survival	7	10	<i>KMT2E/SRPK2</i>	1	2
<i>LDHD</i>	lactate dehydrogenase D	Metabolism	16	25	<i>CTRB2/CTRB1</i>	2	1
<i>LIPC</i>	lipase, hepatic	Lipids	15	23	<i>LIPC</i>	0	2
<i>LOC654841</i>		non-coding RNA	2	2	<i>COL4A3</i>	1	0
<i>LONP1</i>	lon peptidase 1, mitochondrial	Metabolism	19	28	<i>C3</i>	3	2
<i>LOXL2</i>	lysyl oxidase-like 2	Extracellular Matrix	8	12	<i>TNFRSF10A</i>	7	4
<i>LRIT3</i>	leucine-rich repeat, immunoglobulin-like and transmembrane domains 3	Unknown/other	4	5	<i>CFI</i>	0	1
<i>METTL7B</i>	methyltransferase like 7B	Unknown/other	12	19	<i>RDH5/CD63</i>	4	1
<i>MICAL1</i>	MICAL-like 1	Unknown/other	22	34	<i>SLC16A8</i>	0	1
<i>MMP19</i>	matrix metalloproteinase 19	Extracellular Matrix	12	19	<i>RDH5/CD63</i>	0	1
<i>MMP9</i>	matrix metalloproteinase 9 (gelatinase B, 92kDa gelatinase, 92kDa type IV collagenase)	Extracellular Matrix	20	31	<i>MMP9</i>	7	10
<i>MRPL12</i>	mitochondrial ribosomal protein L12	Metabolism	17	27	<i>NPLOC4/TSPAN10</i>	2	1
<i>NCOA5</i>	nuclear receptor coactivator 5	Unknown/other	20	31	<i>MMP9</i>	2	0
<i>NDUFA11</i>	NADH dehydrogenase (ubiquinone) 1 alpha subcomplex, 11, 14.7kDa	Metabolism	19	28	<i>C3</i>	1	1
<i>NEU1</i>	sialidase 1 (lysosomal sialidase)	Unknown/other	6	8	<i>C2/CFB/SKIV2L</i>	3	2
<i>NEURL2</i>	neutralized E3 ubiquitin protein ligase 2	Unknown/other	20	31	<i>MMP9</i>	1	1
<i>NOTCH4</i>	notch 4	Unknown/other	6	8	<i>C2/CFB/SKIV2L</i>	3	3
<i>NPLOC4</i>	nuclear protein localization 4 homolog (S. cerevisiae)	Cell Survival	17	27	<i>NPLOC4/TSPAN10</i>	1	2
<i>NRTN</i>	neurturin	Immune System	19	28	<i>C3</i>	0	1
<i>NYAP1</i>	neuronal tyrosine-phosphorylated phosphoinositide-3-kinase adaptor 1	Unknown/other	7	11	<i>PILRB/PILRA</i>	4	1
<i>ORMDL2</i>	ORM1-like 2 (S. cerevisiae)	Lipids	12	19	<i>RDH5/CD63</i>	4	1
<i>PBX2</i>	pre-B-cell leukemia homeobox 2	Unknown/other	6	8	<i>C2/CFB/SKIV2L</i>	2	0
<i>PCIF1</i>	PDX1 C-terminal inhibiting factor 1	Unknown/other	20	31	<i>MMP9</i>	1	2
<i>PDE6G</i>	phosphodiesterase 6G	Lipids	17	27	<i>NPLOC4/TSPAN10</i>	0	1
<i>PICK1</i>	protein interacting with PRKCA 1	Unknown/other	22	34	<i>SLC16A8</i>	1	0
<i>PILRA</i>	paired immunoglobulin-like type 2 receptor alpha	Immune System	7	11	<i>PILRB/PILRA</i>	5	3
<i>PILRB</i>	paired immunoglobulin-like type 2 receptor beta	Immune System	7	11	<i>PILRB/PILRA</i>	1	0
<i>PLA2G12A</i>	phospholipase A2, group XIA	Lipids	4	5	<i>CFI</i>	8	3
<i>PLA2G6</i>	phospholipase A2, group VI (cytosolic, calcium-independent)	Lipids	22	34	<i>SLC16A8</i>	4	4
<i>PLEKHA1</i>	pleckstrin homology domain containing, family A (phosphoinositide binding specific) member 1	Lipids	10	18	<i>ARMS2/HTRA1</i>	4	3
<i>PLTP</i>	phospholipid transfer protein	Lipids	20	31	<i>MMP9</i>	3	2

<i>POLR2E</i>	polymerase (RNA) II (DNA directed) polypeptide E, 25kDa	Cell Survival	19	29	<i>CNN2</i>	2	0
<i>PPP1R13L</i>	protein phosphatase 1, regulatory subunit 13 like	Cell Survival	19	30	<i>APOE</i>	2	2
<i>PPP1R37</i>	protein phosphatase 1, regulatory subunit 37	Unknown/other	19	30	<i>APOE</i>	0	2
<i>PPT2</i>	palmitoyl-protein thioesterase 2	Lipids	6	8	<i>C2/CFB/SKIV2L</i>	1	1
<i>PRTFDC1</i>	phosphoribosyl transferase domain containing 1	Unknown/other	10	17	<i>ARHGAP21</i>	0	1
<i>PVRL2</i>	poliovirus receptor-related 2 (herpesvirus entry mediator B)	Unknown/other	19	30	<i>APOE</i>	1	1
<i>R3HCC1</i>	R3H domain and coiled-coil containing 1	Unknown/other	8	12	<i>TNFRSF10A</i>	3	3
<i>RAD51B</i>	RAD51 paralog B	Cell Survival	14	22	<i>RAD51B</i>	1	1
<i>RANBP3</i>	RAN binding protein 3	Unknown/other	19	28	<i>C3</i>	1	3
<i>RDH5</i>	retinol dehydrogenase 5 (11-cis/9-cis)	Unknown/other	12	19	<i>RDH5/CD63</i>	0	1
<i>RELB</i>	v-rel avian reticuloendotheliosis viral oncogene homolog B	Immune System	19	30	<i>APOE</i>	1	0
<i>RRH</i>	retinal pigment epithelium-derived rhodopsin homolog	Unknown/other	4	5	<i>CFI</i>	2	0
<i>SARM1</i>	sterile alpha and TIR motif containing 1	Immune System	17	26	<i>TMEM97/VTN</i>	1	0
<i>SBNO2</i>	strawberry notch homolog 2 (Drosophila)	Immune System	19	29	<i>CNN2</i>	2	1
<i>SH2D3A</i>	SH2 domain containing 3A	Unknown/other	19	28	<i>C3</i>	0	1
<i>SKIV2L</i>	superkiller viralicidic activity 2-like (S. cerevisiae)	Unknown/other	6	8	<i>C2/CFB/SKIV2L</i>	4	2
<i>SLC12A3</i>	solute carrier family 12 (sodium/chloride transporter), member 3	Unknown/other	16	24	<i>CETP</i>	8	6
<i>SLC12A5</i>	solute carrier family 12 (potassium/chloride transporter), member 5	Unknown/other	20	31	<i>MMP9</i>	1	1
<i>SLC13A2</i>	solute carrier family 13 (sodium-dependent dicarboxylate transporter), member 2	Unknown/other	20	26	<i>TMEM97/VTN</i>	1	2
<i>SLC16A8</i>	solute carrier family 16 (monocarboxylate transporter), member 8	Metabolism	22	34	<i>SLC16A8</i>	4	2
<i>SLC25A10</i>	solute carrier family 25 (mitochondrial carrier; dicarboxylate transporter), member 10	Metabolism	17	27	<i>NPLOC4/TSPAN10</i>	0	1
<i>SLC44A4</i>	solute carrier family 44, member 4	Unknown/other	6	8	<i>C2/CFB/SKIV2L</i>	1	0
<i>SOX10</i>	SRY (sex determining region Y)-box 10	Unknown/other	22	34	<i>SLC16A8</i>	0	1
<i>SPATA25</i>	spermatogenesis associated 25	Unknown/other	20	31	<i>MMP9</i>	1	3
<i>SRPK2</i>	SRSF protein kinase 2	Unknown/other	7	10	<i>KMT2E/SRPK2</i>	3	0
<i>STAG3L5P-PVRIG2P-PILRB</i>	STAG3L5P-PVRIG2P-PILRB readthrough	non-coding RNA	7	11	<i>PILRB/PILRA</i>	2	0
<i>STK19</i>	serine/threonine kinase 19	Unknown/other	6	8	<i>C2/CFB/SKIV2L</i>	1	0
<i>TMEM199</i>	transmembrane protein 199	Unknown/other	17	26	<i>TMEM97/VTN</i>	0	1
<i>TNFAIP1</i>	tumor necrosis factor, alpha-induced protein 1 (endothelial)	Unknown/other	17	26	<i>TMEM97/VTN</i>	4	1
<i>TNFRSF10A</i>	tumor necrosis factor receptor superfamily, member 10a	Cell Survival	8	12	<i>TNFRSF10A</i>	3	1
<i>TNXB</i>	tenascin XB	Extracellular Matrix	6	8	<i>C2/CFB/SKIV2L</i>	7	11
<i>TRIP10</i>	thyroid hormone receptor interactor 10	Unknown/other	19	28	<i>C3</i>	0	1
<i>TSC22D4</i>	TSC22 domain family, member 4	Unknown/other	7	11	<i>PILRB/PILRA</i>	4	2
<i>VARS</i>	valyl-tRNA synthetase 2, mitochondrial	Unknown/other	6	8	<i>C2/CFB/SKIV2L</i>	5	1
<i>VEGFA</i>	vascular endothelial growth factor A	Cell Survival	6	9	<i>VEGFA</i>	1	0
<i>VTN</i>	vitronectin	Complement	17	26	<i>TMEM97/VTN</i>	7	2
<i>VWA7</i>	von Willebrand factor A domain containing 7	Unknown/other	6	8	<i>C2/CFB/SKIV2L</i>	2	0
<i>WDR18</i>	WD repeat domain 18	Unknown/other	19	29	<i>CNN2</i>	3	4
<i>ZCWPW1</i>	zinc finger, CW type with PWWP domain 1	Unknown/other	7	11	<i>PILRB/PILRA</i>	5	4
<i>ZFP1</i>	ZFP1 zinc finger protein	Unknown/other	16	25	<i>CTRB2/CTRB1</i>	0	1
<i>ZNF335</i>	zinc finger protein 335	Unknown/other	20	31	<i>MMP9</i>	5	9
Total						337	273