

Appendix F. Functional enrichment analysis of the 221 genes containing rare variants in ACEi-A samples

#Term ID	Term description	Observed gene count	Background gene count	False discovery rate	Genes	No. terms
Biological Process						
GO:0007010	cytoskeleton organization	32	953	7.01E-05	AKAP2,AKAP9,ANK3,CCDC40,CNTN2,CUL7,DAAM2,DNAH1,DNAH8,EPPK1,FAT1,FRY,HYDIN,LAMA5,LLGL1,MYH11,MYH3,MYOM2,NDE1,OBSCN,PCLO,PCNT,PDE4DIP,PPL,RANBP2,RLTPR,SYNE1,SYNE2,TRIOBP,TTN,TUBGCP6,XIRP1	45
GO:0007155	cell adhesion	29	843	0.0001	CELSR1,CNTN2,COL6A3,DCHS2,DPP4,FAT1,GPR56,GPR98,LAMA5,LAMC3,LRRC4C,MADCAM1,MUC16,MUC4,NRXN1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PKD1,PKD1L1,SNED1,SVEP1,TM9SF4,TNC,VWF	
GO:0006928	movement of cell or subcellular component	37	1355	0.00017	CCDC40,CELSR1,CNTN2,CSPG4,DAB2IP,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,DPP4,FAT1,FCAMR,GPR56,HYDIN,LAMA5,MADCAM1,MEGF8,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,NDE1,NEB,NRXN1,PIK3C2B,PLXNA2,PLXNA3,PLXNA4,RLTPR,RYR2,SYNE2,TNK2,TNS1,TTN	
GO:0048856	anatomical structure development	90	5085	0.00017	AACS,ACE,ADNP2,AKAP13,ANK3,ANKLE2,ARHGAP11B,BIRC6,C12orf57,CACNA1H,CCDC40,CELSR1,CFTR,CNTN2,COL24A1,COL6A3,CSDM3,CSPG4,CUL7,CYFIP1,DAAM2,DAB2IP,DCLRE1C,DDX39B,DNAH1,DNAH11,FAT1,FBN3,FLG,FLG2,FOXO1,FRY,GPR56,GPR98,GTTF2IRD1,HIVEP3,HRNR,HSPG2,HUS1,HYDIN,IGF2R,KMT2D,LAMA5,LAMC3,LLGL1,LRP2,LRRC4C,MACF1,MAML1,MEGF8,MSH6,MYH11,MYH3,MYO15A,MYOM2,NDE1,NEB,NRXN1,OBSCN,OSBP2,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCLO,PKD1,PKD1L1,PLXNA2,PLXNA3,PLXNA4,PPL,RECQL4,RIMS2,RLTPR,RNF213,RYR2,SECTM1,SOX13,SPEG,SREBF1,SYNE1,SYNE2,TH,TNC,TRIOBP,TTN,XIRP1	
GO:0007018	microtubule-based movement	15	276	0.00034	CCDC40,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,HYDIN,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,NDE1,SYNE2	

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GO:0048731	system development	76	4144	0.00042	AACS,ACE,ADNP2,AKAP13,ANK3,ANKLE2,ARHGAP11B,BIRC6,C12orf57,CACNA1H,CCDC40,CELSR1,CNTN2,COL24A1,COL6A3,CSMD3,CSPG4,CUL7,CYFIP1,DAAM2,DAB2IP,DCLRE1C,DDX39B,DNAH11,FAT1,FLG,FLG2,FOXO1,FRY,GPR56,GPR98,HIVEP3,HRNR,HSPG2,HYDIN,IGF2R,LAMA5,LAMC3,LLGL1,LRP2,LRRC4C,MACF1,MAML1,MEGF8,MSH6,MYH11,MYH3,MYO15A,MYOM2,NDE1,NEB,NRXN1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCLO,PKD1,PLXNA2,PLXNA3,PLXNA4,PPL,RIMS2,RNF213,RYR2,SPEG,SREBF1,SYNE2,TH,TNC,TRIOBP,TTN,XIRP1	
GO:0007017	microtubule-based process	22	605	0.00047	AKAP9,CCDC40,CNTN2,CUL7,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,HYDIN,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,NDE1,PCNT,PDE4DIP,RANBP2,SYNE2,TUBGCP6	
GO:0032502	developmental process	91	5401	0.00061	AACS,ACE,ADNP2,AKAP13,ANK3,ANKLE2,ARHGAP11B,BIRC6,C12orf57,CACNA1H,CCDC40,CELSR1,CFTR,CNTN2,COL24A1,COL6A3,CSMD3,CSPG4,CUL7,CYFIP1,DAAM2,DAB2IP,DCLRE1C,DDX39B,DNAH1,DNAH11,FAT1,FBN3,FLG,FLG2,FOXO1,FRY,GPR56,GPR98,GTTF2IRD1,HIVEP3,HRNR,HSPG2,HUS1,HYDIN,IGF2R,KMT2D,LAMA5,LAMC3,LLGL1,LRP2,LRRC4C,MACF1,MAML1,MEGF8,MSH6,MYH11,MYH3,MYO15A,MYOM2,NDE1,NEB,NRXN1,OBSCN,OSBP2,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCLO,PKD1,PKD1L1,PLXNA2,PLXNA3,PLXNA4,PPL,RECQL4,RIMS2,RLTPR,RNF213,RYR2,SECTM1,SOX13,SPEG,SREBF1,SYNE1,SYNE2,TH,TNC,TNK2,TRIOBP,TTN,XIRP1	
GO:0007156	homophilic cell adhesion via plasma membrane adhesion molecules	11	158	0.00066	CELSR1,DCHS2,FAT1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PKD1	

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GO:0032501	multicellular organismal process	104	6507	0.00066	AACS,ACE,ADNP2,AKAP13,AKAP9,ANK3,ANKLE2,ARHGAP11B,BEST2,BIRC6,C12orf57,CACNA1H,CACNA2D4,CCDC40,CELSR1,CFTR,CGB,CGB2,CNTN2,COL24A1,COL6A3,CSMD3,CSPG4,CUBN,CUL7,CYFIP1,DAAM2,DAB2IP,DCLRE1C,DDX39B,DNAH1,DNAH11,DOCK9,DPP4,FAT1,FBXO11,FLG,FLG2,FOXO1,FRY,GPR56,GPR98,GTF2IRD1,HIVEP3,HMCN1,HMCN2,HRNR,HSPG2,HUS1,HYDIN,IGF2R,KMT2D,LAMA5,LAMC3,LLGL1,LRP2,LRRC4C,MACF1,MAML1,MEGF8,MSH6,MUC4,MYH1,MYH11,MYH2,MYH3,MYO15A,MYOM2,NDE1,NEB,NRXN1,OBSCN,OR10C1,OR11A1,OR5V1,OSBP2,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCLO,PKD1,PKD1L1,PLXNA2,PLXNA3,PLXNA4,PPL,RECQL4,RIMS2,RNF213,RYR2,SPEG,SREBF1,SYNE1,SYNE2,TH,TNC,TRIOBP,TTN,VWF,XIRP1	
GO:0098742	cell-cell adhesion via plasma-membrane adhesion molecules	13	230	0.00066	CELSR1,DCHS2,FAT1,LRRC4C,NRXN1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PKD1	
GO:0007275	multicellular organism development	81	4726	0.0013	AACS,ACE,ADNP2,AKAP13,ANK3,ANKLE2,ARHGAP11B,BIRC6,C12orf57,CACNA1H,CCDC40,CELSR1,CNTN2,COL24A1,COL6A3,CSMD3,CSPG4,CUL7,CYFIP1,DAAM2,DAB2IP,DCLRE1C,DDX39B,DNAH11,FAT1,FLG,FLG2,FOXO1,FRY,GPR56,GPR98,GTF2IRD1,HIVEP3,HRNR,HSPG2,HUS1,HYDIN,IGF2R,LAMA5,LAMC3,LLGL1,LRP2,LRRC4C,MACF1,MAML1,MEGF8,MSH6,MYH11,MYH3,MYO15A,MYOM2,NDE1,NEB,NRXN1,OBSCN,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCLO,PKD1,PKD1L1,PLXNA2,PLXNA3,PLXNA4,PPL,RECQL4,RIMS2,RNF213,RYR2,SPEG,SREBF1,SYNE2,TH,TNC,TRIOBP,TTN,XIRP1	
GO:0051642	centrosome localization	5	25	0.0035	AKAP9,NDE1,PLXNA2,RANBP2,SYNE2	
GO:0098609	cell-cell adhesion	16	416	0.0038	CELSR1,DCHS2,FAT1,GPR98,LRRC4C,MADCAM1,NRXN1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PKD1,PKD1L1	

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GO:0048513	animal organ development	55	2926	0.0052	AACS,ACE,AKAP13,ARHGAP11B,BIRC6,C12orf57,CACNA1H,CCDC40,CELSR1,CNTN2,COL24A1,COL6A3,CUL7,DAAM2,DAB2IP,DCLRE1C,DDX39B,DNAH11,FAT1,FLG,FLG2,FOXO1,GPR56,GPR98,HIVEP3,HRNR,HSPG2,HYDIN,IGF2R,LAMA5,LAMC3,LRP2,MAML1,MEGF8,MYH11,MYH3,MYO15A,MYOM2,NDE1,NEB,NRXN1,PKD1,PLXNA2,PLXNA3,PLXNA4,PPL,RYR2,SPEG,SREBF1,SYNE2,TH,TNC,TRIOBP,TTN,XIRP1	
GO:0097435	supramolecular fiber organization	15	383	0.0052	AKAP2,AKAP9,EPPK1,FAT1,FRY,MYH11,MYH3,MYOM2,NDE1,OBSCN,PDE4DIP,RLTPR,TRIOBP,TTN,TUBGCP6	
GO:0030029	actin filament-based process	17	493	0.0065	AKAP2,DAAM2,FAT1,FRY,LLGL1,MYH11,MYH2,MYH3,MYOM2,NEB,OBSCN,RLTPR,RYR2,SYNE2,TRIOBP,TTN,XIRP1	
GO:0007399	nervous system development	44	2206	0.0083	ADNP2,ANK3,ANKLE2,ARHGAP11B,C12orf57,CELSR1,CNTN2,CSMD3,CSPG4,CUL7,CYFIP1,DAAM2,DAB2IP,DNAH11,FRY,GPR56,GPR98,HSPG2,HYDIN,LAMC3,LLGL1,LRP2,LRR4C,MACF1,MEGF8,NDE1,NRXN1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCLO,PKD1,PLXNA2,PLXNA3,PLXNA4,RIMS2,SYNE2,TH,TNC,TRIOBP	
GO:0016266	O-glycan processing	6	57	0.0083	MUC12,MUC16,MUC17,MUC4,MUC5B,ST6GAL1	
GO:0032989	cellular component morphogenesis	21	720	0.0083	ANK3,C12orf57,CNTN2,CYFIP1,DAB2IP,FAT1,FRY,LAMA5,LAMC3,LLGL1,LRP2,MYH11,MYH3,MYOM2,NRXN1,OBSCN,PLXNA2,PLXNA3,PLXNA4,TRIOBP,TTN	
GO:0048841	regulation of axon extension involved in axon guidance	4	16	0.0083	MEGF8,PLXNA2,PLXNA3,PLXNA4	
GO:0061061	muscle structure development	16	457	0.0083	AKAP13,CACNA1H,COL6A3,HIVEP3,LAMA5,LRP2,MAML1,MYH11,MYH3,MYOM2,NEB,OBSCN,RYR2,SPEG,SYNE1,TTN	

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GO:0009653	anatomical structure morphogenesis	40	1992	0.015	ANK3,C12orf57,CACNA1H,CCDC40,CELSR1,CNTN2,CSPG4,CUL7,CYFIP1,DAB2IP,DNAH11,FAT1,FBN3,FOXO1,FRY,GPR56,HSPG2,LAMA5,LAMC3,LLGL1,LRP2,MEGF8,MYH11,MYH3,MYO15A,MYOM2,NRXN1,OBSCN,PKD1,PLXNA2,PLXNA3,PLXNA4,RLTPR,RNF213,RYR2,SOX13,TH,TNC,TRIOBP,TTN	
GO:0021785	branchiomotor neuron axon guidance	3	7	0.0165	PLXNA2,PLXNA3,PLXNA4	
GO:0048870	cell motility	23	914	0.0215	CCDC40,CELSR1,CNTN2,CSPG4,DAB2IP,DNAH1,DNAH11,DNAH6,DNAH8,DPP4,FAT1,FCAMR,GPR56,LAMA5,MADCAM1,MEGF8,NDE1,PIK3C2B,PLXNA2,RLTPR,SYNE2,TNK2,TNS1	
GO:0120036	plasma membrane bounded cell projection organization	25	1034	0.0215	AKAP9,ANK3,C12orf57,CCDC40,CNTN2,CYFIP1,DAB2IP,DNAH1,DNAH14,DNAH6,DNAH8,FRY,HYDIN,LAMA5,LLGL1,LRP2,MEGF8,NDE1,NRXN1,PCNT,PLXNA2,PLXNA3,PLXNA4,TNC,TRIOBP	
GO:0001539	cilium or flagellum-dependent cell motility	4	25	0.0249	DNAH1,DNAH11,DNAH6,DNAH8	
GO:0072359	circulatory system development	21	807	0.0249	AKAP13,CCDC40,CSPG4,CUL7,DAB2IP,DNAH11,FOXO1,GPR56,HSPG2,LAMA5,LRP2,MAML1,MEGF8,MYH11,NRXN1,PKD1,RNF213,RYR2,TH,TTN,XIRP1	
GO:0042692	muscle cell differentiation	10	230	0.0267	AKAP13,CACNA1H,MAML1,MYH11,MYH3,MYOM2,OBSCN,SPEG,SYNE1,TTN	
GO:0007267	cell-cell signaling	25	1073	0.0294	AKAP9,CELSR1,CGB,CGB2,CPZ,DAAM2,FAT1,GPR56,MACF1,NRXN1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCLO,RIMS2,RYR2,SLC1A6,SLC22A2,SNAP47,TH,TNC	
GO:0021537	telencephalon development	10	237	0.0307	ARHGAP11B,C12orf57,CNTN2,DAB2IP,GPR56,NDE1,PLXNA3,PLXNA4,SYNE2,TH	
GO:0030154	cell differentiation	58	3457	0.0307	AACS,ACE,ADNP2,AKAP13,ANK3,C12orf57,CACNA1H,CELSR1,CFTR,CNTN2,COL24A1,CSMD3,CSPG4,CUL7,CYFIP1,DAAM2,DAB2IP,DCLRE1C,DNAH1,FAT1,FLG,FOXO1,FRY,GPR56,HIVEP3,HRNR,HSPG2,HYDIN,KMT2D,LAMA5,LAMC3,LLGL1,LRP2,LRRC4C,MACF1,MAML1,MEGF8,MYH11,MYH3,MYOM2,NDE1,NRXN1,OBSCN,OSBP2,PLXNA2,PLXNA3,PLXNA4,PPL,RIMS2,SPEG,SREBF1,SYNE1,SYNE2,TH,TNC,TNK2,TRIOBP,TTN	

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GO:0030516	regulation of axon extension	6	82	0.0307	CYFIP1,MACF1,MEGF8,PLXNA2,PLXNA3,PLXNA4	
GO:0040011	locomotion	26	1144	0.0307	CCDC40,CELSR1,CNTN2,CSPG4,DAB2IP,DNAH1,DNAH11,DNAH6,DNAH8,DPP4,FAT1,FCAMR,GPR56,LAMA5,MADCAM1,MEGF8,NDE1,NRXN1,PIK3C2B,PLXNA2,PLXNA3,PLXNA4,RLTPR,SYNE2,TNK2,TNS1	
GO:0007517	muscle organ development	11	287	0.0311	CACNA1H,COL6A3,HIVEP3,LAMA5,LRP2,MYH3,MYOM2,NEB,RYR2,SPEG,TTN	
GO:0006936	muscle contraction	10	244	0.0336	CACNA1H,HMCN2,MYH1,MYH11,MYH2,MYH3,MYOM2,NEB,RYR2,TTN	
GO:0070925	organelle assembly	18	666	0.0336	AKAP9,CCDC40,DNAH1,DNAH14,DNAH6,DNAH8,HYDIN,LAMA5,MYH11,MYH3,MYOM2,NDE1,NRXN1,OBSCN,PACS2,PCNT,TTN,TUBGCP6	
GO:0000226	microtubule cytoskeleton organization	13	393	0.0344	AKAP9,CCDC40,CNTN2,CUL7,DNAH1,DNAH8,HYDIN,NDE1,PCNT,PDE4DIP,RANBP2,SYNE2,TUBGCP6	
GO:0050770	regulation of axonogenesis	8	162	0.0365	CNTN2,CYFIP1,LRRC4C,MACF1,MEGF8,PLXNA2,PLXNA3,PLXNA4	
GO:0030239	myofibril assembly	5	59	0.0424	MYH11,MYH3,MYOM2,OBSCN,TTN	
GO:0071688	striated muscle myosin thick filament assembly	3	13	0.0424	MYH11,MYOM2,TTN	
GO:0003341	cilium movement	5	61	0.0463	CCDC40,DNAH1,DNAH11,DNAH8,HYDIN	
GO:0002223	stimulatory C-type lectin receptor signaling pathway	5	62	0.0469	MUC12,MUC16,MUC17,MUC4,MUC5B	
GO:0010769	regulation of cell morphogenesis involved in differentiation	10	263	0.0469	CNTN2,CUL7,CYFIP1,LRRC4C,MACF1,MEGF8,PLXNA2,PLXNA3,PLXNA4,TRIOBP	
GO:0030036	actin cytoskeleton organization	13	418	0.048	AKAP2,DAAM2,FAT1,FRY,LLGL1,MYH11,MYH3,MYOM2,OBSCN,RLTPR,TRIOBP,TTN,XIRP1	
Molecular Function						
GO:0003777	microtubule motor activity	12	110	3.05E-06	DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,KLC4,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A	30
GO:0005509	calcium ion binding	28	700	3.05E-06	CELSR1,CUBN,DCHS2,FAT1,FBN3,FLG,FLG2,GPR98,HMCN1,HMCN2,HRNR,HSPG2,LRP2,MACF1,MEGF8,NRXN1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCLO,RYR2,SNED1,SVEP1,TTN	
GO:0003779	actin binding	19	413	3.41E-05	ACE,CYFIP1,DAAM2,MACF1,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,MYOM2,NEB,PLEC,SYNE1,SYNE2,TNS1,TRIOBP,TTN,XIRP1	

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GO:0051015	actin filament binding	12	158	3.71E-05	CYFIP1,MACF1,MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,SYNE1,SYNE2,TRIOBP,TTN	
GO:0008569	ATP-dependent microtubule motor activity, minus-end-directed	5	15	0.00018	DNAH1,DNAH11,DNAH14,DNAH6,DNAH8	
GO:0005198	structural molecule activity	22	679	0.0006	AKAP13,AKAP9,ANK3,COL24A1,FBN3,FLG,FLG2,LAMC3,LLGL1,MUC17,MUC4,MYH11,MYOM2,NEB,OBSCN,PCNT,PDE4DIP,PLEC,PPL,RPS9,TTN,TUBGCP6	
GO:0008307	structural constituent of muscle	6	45	0.001	MYH11,MYOM2,NEB,OBSCN,PLEC,TTN	
GO:0016462	pyrophosphatase activity	24	819	0.001	ABCA13,ATP13A5,ATP2C2,CFTR,DDX39B,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,HBS1L,KLC4,MACF1,MSH6,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,PRUNE2,RABL6,RECQL4,RNF213	
GO:0017111	nucleoside-triphosphatase activity	23	778	0.001	ABCA13,ATP13A5,ATP2C2,CFTR,DDX39B,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,HBS1L,KLC4,MACF1,MSH6,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,RABL6,RECQL4,RNF213	
GO:0045503	dynein light chain binding	5	25	0.001	DNAH1,DNAH11,DNAH14,DNAH6,DNAH8	
GO:0045505	dynein intermediate chain binding	5	28	0.001	DNAH1,DNAH11,DNAH14,DNAH6,DNAH8	
GO:0051959	dynein light intermediate chain binding	5	29	0.0011	DNAH1,DNAH11,DNAH14,DNAH6,DNAH8	
GO:0016887	ATPase activity	15	392	0.0013	ABCA13,ATP13A5,ATP2C2,CFTR,DDX39B,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,MACF1,MSH6,MYH3,RECQL4,RNF213	
GO:0008092	cytoskeletal protein binding	24	882	0.0015	ACE,ANK3,CYFIP1,DAAM2,EPPK1,MACF1,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,MYOM2,NDE1,NEB,OBSCN,PLEC,SYNE1,SYNE2,TNS1,TRIOBP,TTN,TUBGCP6,XIRP1	
GO:0042623	ATPase activity, coupled	13	320	0.0021	ABCA13,ATP13A5,ATP2C2,CFTR,DDX39B,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,MSH6,MYH3,RECQL4	
GO:0005516	calmodulin binding	10	194	0.0022	MYH1,MYH11,MYH2,MYH3,MYO15A,OBSCN,PCNT,RYR2,TTN,UBR4	
GO:0051018	protein kinase A binding	5	50	0.0083	AKAP13,AKAP2,AKAP8,AKAP9,RYR2	

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GO:0043167	ion binding	90	6066	0.0096	AACS,ABCA13,ACE,ADAM20,ADNP2,AKAP13,AKAP8,AMZ1,ATP13A5,ATP2C2,BRF1,CACNA1H,CACNA2D4,CELSR1,CFTR,CPZ,CUBN,DAB2IP,DCHS2,DDX39B,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,DNPEP,DROSHA,FAT1,FBN3,FBXO11,FLG,FLG2,GPR56,GPR98,HBS1L,HIVEP3,HMCN1,HMCN2,HRNR,HSPG2,IGF2R,KMT2D,LRP2,MACF1,MEGF8,MKI67,MSH6,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,NRXN1,OBSCN,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCLO,PIK3C2B,PRUNE2,RABL6,RANBP2,RECQL4,RIMS2,RLTPR,RNF213,RSF1,RYR2,SLC19A1,SLC1A6,SNED1,SPEG,SVEP1,TH,TNK2,TTN,UBR4,VARS2,ZFYVE28,ZNF17,ZNF417,ZNF749,ZNF772,ZSCAN5A,ZSCAN5DP	
GO:0017154	semaphorin receptor activity	3	11	0.0101	PLXNA2,PLXNA3,PLXNA4	
GO:0044877	protein-containing complex binding	23	968	0.0106	COG2,CYFIP1,DAB2IP,DDX39B,EPPK1,GPR56,HSPG2,LAMA5,MACF1,MACAM1,MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,RLTPR,SREBF1,SYNE1,SYNE2,TRIOBP,TTN,VWF	
GO:0015085	calcium ion transmembrane transporter activity	7	135	0.02	ATP13A5,ATP2C2,CACNA1H,CACNA2D4,PKD1,PKD1L1,RYR2	
GO:0008240	tripeptidyl-peptidase activity	2	3	0.0246	ACE,TPP2	

#Term ID	Term description	Observed gene count	Background gene count	False discovery rate	Genes	No. terms
GO:0005488	binding	151	11878	0.0275	AACS,ABCA13,ACE,ACOT11,ADAM20,ADNP2,AKAP13,AKAP2,AKAP8,AKAP9,ALS2CL,AMZ1,ANK3,ANKLE2,ANKRD30A,ATP13A5,ATP2C2,BRF1,CACNA1H,CACNA2D4,CELSR1,CFTR,CGB,CGB2,CNTN2,COG2,CPZ,CSPG4,CUBN,CUL7,CYFIP1,DAAM2,DAB2IP,DCHS2,DCLRE1C,DDX39B,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,DNPEP,DOCK9,DPP4,DROSHA,EPPK1,ESX1,FAT1,FBN3,FBXO11,FLG,FLG2,FOXO1,GPR56,GPR98,GTF2IRD1,HBS1L,HIVEP3,HMCN1,HMCN2,HRNR,HSPG2,IGF2R,KMT2D,LAMA5,LLGL4,LLGL1,LRP2,LRRC4C,MACF1,MADCAM1,MAML1,MDC1,MEGF8,MGA,MKI67,MSH6,MUC17,MUC4,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,MYOM2,NABP1,NDE1,NEB,NOL12,NRXN1,OBSCN,OSBP2,PACS2,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCLO,PCNT,PDE4DIP,PDIA2,PIK3C2B,PKD1,PLEC,PLXNA2,PRUNE2,RABL6,RANBP2,RECQL4,RIMS2,RLTPR,RNF213,RPS9,RSF1,RYR2,SCAF1,SECTM1,SLC19A1,SLC1A6,SNAP47,SNED1,SOX13,SPEG,SREBF1,SVEP1,SYNE1,SYNE2,TBC1D30,TH,TNC,TNK2,TNS1,TPP2,TRIOBP,TTN,TUBGCP6,UBR4,VARS2,VWF,XIRP1,ZFYVE28,ZNF17,ZNF417,ZNF749,ZNF772,ZSCAN5A,ZSCAN5DP	
GO:0030197	extracellular matrix constituent, lubricant activity	2	4	0.0329	MUC17,MUC4	
GO:0034237	protein kinase A regulatory subunit binding	3	19	0.0329	AKAP8,AKAP9,RYR2	
GO:0047961	glycine N-acyltransferase activity	2	4	0.0329	GLYATL1,GLYATL2	
GO:0008144	drug binding	32	1710	0.0362	AACS,ABCA13,ACE,ATP13A5,ATP2C2,CFTR,CUBN,DDX39B,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,LRP2,MKI67,MSH6,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,OBSCN,PIK3C2B,RECQL4,RYR2,SLC19A1,SPEG,TH,TNK2,TTN,VARS2	
GO:0008017	microtubule binding	9	253	0.0389	MACF1,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,NDE1,TUBGCP6	
GO:0044325	ion channel binding	6	120	0.042	AKAP9,ANK3,PACS2,PKD1,RIMS2,RYR2	

#Term ID	Term description	Observed gene count	Background gene count	False discovery rate	Genes	No. terms
GO:0046872	metal ion binding	62	4087	0.0494	ACE,ADAM20,ADNP2,AKAP13,AKAP8,AMZ1,ATP13A5,ATP2C2,BRF1,CACNA1H,CACNA2D4,CELSR1,CPZ,CUBN,DCHS2,DNPEP,DROSHA,FAT1,FBN3,FBXO11,FLG,FLG2,GPR98,HIVEP3,HMCN1,HMCN2,HRNR,HSPG2,KMT2D,LRP2,MACF1,MEGF8,NRXN1,OBSCN,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCL0,PRUNE2,RANBP2,RIMS2,RNF213,RSF1,RYR2,SLC1A6,SNED1,SVEP1,TH,TNK2,TTN,UBR4,ZFYVE28,ZNF17,ZNF417,ZNF749,ZNF772,ZSCAN5A,ZSCAN5DP	
Cellular Component						
GO:0043292	contractile fiber	17	228	6.65E-07	AHNAK2,ANK3,CMYA5,MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,NEB,OBSCN,PDE4DIP,PLEC,RYR2,SYNE1,SYNE2,TTN	47
GO:0030016	myofibril	16	216	1.12E-06	AHNAK2,ANK3,CMYA5,MYH1,MYH2,MYH3,MYH7B,MYOM2,NEB,OBSCN,PDE4DIP,PLEC,RYR2,SYNE1,SYNE2,TTN	
GO:0099512	supramolecular fiber	31	873	1.77E-06	AHNAK2,AKAP13,ANK3,CMYA5,COL6A3,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,EPPK1,FLG,KLC4,MACF1,MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,NDE1,NEB,OBSCN,PCNT,PDE4DIP,PLEC,RYR2,SYNE1,SYNE2,TTN,TUBGCP6	
GO:0044449	contractile fiber part	15	212	2.04E-06	AHNAK2,ANK3,CMYA5,MYH1,MYH11,MYH2,MYH3,MYOM2,NEB,OBSCN,PLEC,RYR2,SYNE1,SYNE2,TTN	
GO:0030017	sarcomere	13	195	2.89E-05	AHNAK2,ANK3,CMYA5,MYH1,MYH2,MYH3,MYOM2,NEB,OBSCN,RYR2,SYNE1,SYNE2,TTN	
GO:0032982	myosin filament	6	22	2.89E-05	MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2	

#Term ID	Term description	Observed gene count	Background gene count	False discovery rate	Genes	No. terms
GO:0071944	cell periphery	90	5254	5.31E-05	ABCA13,ACE,ADAM20,AHNAK2,AKAP13,AKAP9,ANK3,ATP13A5,BEST2,CACNA1H,CACNA2D4,CELSR1,CFTR,CNTN2,COL6A3,CRIPAK,CSMD3,CSPG4,CUBN,DAB2IP,DCHS2,DPP4,EPPK1,FAT1,FCAMR,FLG,FRY,GPR56,GPR78,GPR98,HMCN1,HMCN2,HRNR,HSPG2,IGF2R,LILRA4,LILRB3,LLGL1,LRP2,LRRRC4C,MACF1,MADCAM1,MUC12,MUC16,MUC17,MUC4,MUC5B,MYH2,NDE1,NRXN1,OBSCN,OR10C1,OR11A1,OR5V1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCLO,PIGN,PIK3C2B,PKD1,PKD1L1,PLEC,PLXNA2,PLXNA3,PLXNA4,PPL,RFTN2,RIMS2,RLTPR,RYR2,SECTM1,SLC19A1,SLC1A6,SLC22A10,SLC22A2,SLC22A25,SNAP47,SYNE1,SYNE2,TBC1D30,TH,TJP3,TNK2,TRIOBP,UBR4	
GO:0005859	muscle myosin complex	6	27	5.95E-05	MYH1,MYH11,MYH2,MYH3,MYOM2,TTN	
GO:0016459	myosin complex	8	69	7.55E-05	MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,MYOM2,TTN	
GO:0005856	cytoskeleton	44	2068	0.00047	AKAP13,AKAP9,ANK3,BIRC6,CCDC40,CUL7,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,EPPK1,FLG,FRY,HYDIN,KLC4,LLGL1,MACF1,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,MYOM2,NDE1,NEB,PCLO,PCNT,PDE4DIP,PLEC,PPL,RABL6,RANBP2,RLTPR,SLC1A6,SYNE1,SYNE2,TBC1D30,TNS1,TRIOBP,TTN,TUBGCP6,UBR4	

#Term ID	Term description	Observed gene count	Background gene count	False discovery rate	Genes	No. terms
GO:0005886	plasma membrane	85	5159	0.00047	ABCA13,ACE,ADAM20,AHNAK2,AKAP9,ANK3,ATP13A5,BEST2,CACNA1H,CACNA2D4,CELSR1,CFTR,CNTN2,COL6A3,CRIPAK,CSMD3,CSPG4,CUBN,DAB2IP,DCHS2,DPP4,EPPK1,FAT1,FCAMR,FLG,GPR56,GPR78,GPR98,HMCN1,HMCN2,HRNR,HSPG2,IGF2R,LILRA4,LILRB3,LLGL1,LRP2,LRRC4C,MACF1,MADCAM1,MUC12,MUC16,MUC17,MUC4,MUC5B,NDE1,NRXN1,OBSCN,OR10C1,OR11A1,OR5V1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PIGN,PIK3C2B,PKD1,PKD1L1,PLEC,PLXNA2,PLXNA3,PLXNA4,PPL,RFTN2,RIMS2,RLTPR,RYR2,SECTM1,SLC19A1,SLC1A6,SLC22A10,SLC22A2,SLC22A25,SNAP47,SYNE1,SYNE2,TBC1D30,TH,TJP3,TNK2,UBR4	
GO:0031672	A band	6	45	0.00052	CMYA5,MYH1,MYH2,MYOM2,OBSCN,TTN	
GO:0044459	plasma membrane part	52	2651	0.00052	ACE,AHNAK2,AKAP9,ANK3,ATP13A5,CACNA1H,CACNA2D4,CELSR1,CFTR,CNTN2,CSPG4,CUBN,DAB2IP,DPP4,EPPK1,FAT1,GPR56,HMCN1,HMCN2,HSPG2,IGF2R,LILRA4,LILRB3,LRP2,LRRC4C,MACF1,MUC12,MUC17,MUC4,NDE1,NRXN1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PKD1,PKD1L1,PLXNA2,RIMS2,RLTPR,SLC19A1,SLC1A6,SLC22A10,SLC22A2,SLC22A25,SYNE1,SYNE2,TH,TNK2	
GO:0044447	axoneme part	5	31	0.0011	DNAH1,DNAH11,DNAH6,DNAH8,HYDIN	
GO:0030018	Z disc	8	122	0.0021	AHNAK2,ANK3,MYOM2,NEB,OBSCN,RYR2,SYNE2,TTN	
GO:0044430	cytoskeletal part	34	1547	0.0021	AKAP13,AKAP9,BIRC6,CCDC40,CUL7,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,EPPK1,FLG,FRY,HYDIN,KLC4,LLGL1,MACF1,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,MYOM2,NDE1,PCNT,PDE4DIP,RABL6,RANBP2,TBC1D30,TRIOBP,TTN,TUBGCP6,UBR4	
GO:0002116	semaphorin receptor complex	3	7	0.0033	PLXNA2,PLXNA3,PLXNA4	
GO:0005796	Golgi lumen	7	101	0.0038	CSPG4,HSPG2,MUC12,MUC16,MUC17,MUC4,MUC5B	
GO:0099568	cytoplasmic region	14	402	0.0038	AKAP13,CCDC40,DNAH1,DNAH11,DNAH6,DNAH8,FRY,HMCN1,HMCN2,HYDIN,LLGL1,MYH2,PCLO,TRIOBP	
GO:0030286	dynein complex	5	46	0.0043	DNAH1,DNAH11,DNAH14,DNAH6,DNAH8	

#Term ID	Term description	Observed gene count	Background gene count	False discovery rate	Genes	No. terms
GO:0005794	Golgi apparatus	31	1474	0.0069	AKAP8,AKAP9,ANK3,ATP2C2,BIRC6,COG2,CSPG4,CUBN,CUL7,DCLRE1C,GOLGA6L2,HSPG2,IGF2R,LLGL1,LRP2,MACF1,MSH6,MUC12,MUC16,MUC17,MUC4,MUC5B,MYH2,PDE4DIP,PKD1,SECTM1,SLC1A6,SREBF1,ST6GAL1,SYNE1,TM9SF4	
GO:0031430	M band	4	31	0.0095	CMYA5,MYOM2,OBSCN,TTN	
GO:0005604	basement membrane	6	91	0.0111	HMCN1,HMCN2,HSPG2,LAMA5,LAMC3,TNC	
GO:0043232	intracellular non-membrane-bounded organelle	64	4005	0.0111	AHNAK2,AKAP13,AKAP8,AKAP9,ANK3,BIRC6,CCDC40,CMYA5,CUL7,DCLRE1C,DDX39B,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,DROSHA,EPPK1,FBXO11,FLG,FRY,HUS1,HYDIN,KLC4,LLGL1,MACF1,MDC1,MKI67,MSH6,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,MYOM2,NABP1,NDE1,NEB,NOL12,NRXN1,OBSCN,PCLO,PCNT,PDE4DIP,PLEC,PPL,RABL6,RANBP2,RECQL4,RLTPR,RNF213,RPS9,RSF1,RYR2,SLC1A6,SYNE1,SYNE2,TBC1D30,TNS1,TRIOBP,TTN,TUBGCP6,UBR4	
GO:0032155	cell division site part	5	61	0.0114	HMCN1,HMCN2,MYH2,NDE1,TUBGCP6	
GO:0015629	actin cytoskeleton	13	432	0.0165	AKAP13,LLGL1,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,MYOM2,NEB,RLTPR,TRIOBP,TTN	
GO:0036457	keratohyalin granule	2	3	0.0165	FLG,HRNR	
GO:0005858	axonemal dynein complex	3	17	0.017	DNAH1,DNAH6,DNAH8	
GO:0005875	microtubule associated complex	7	144	0.0171	DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,KLC4,NDE1	
GO:0031012	extracellular matrix	10	283	0.0175	COL6A3,FBN3,HMCN1,HMCN2,HSPG2,LAMA5,LAMC3,MUC4,TNC,VWF	
GO:0005930	axoneme	6	107	0.018	CCDC40,DNAH1,DNAH11,DNAH6,DNAH8,HYDIN	
GO:0043034	costamere	3	19	0.0197	AHNAK2,ANK3,PLEC	
GO:0031258	lamellipodium membrane	3	22	0.028	CSPG4,DPP4,SYNE2	

#Term ID	Term description	Observed gene count	Background gene count	False discovery rate	Genes	No. terms
GO:0012505	endomembrane system	65	4347	0.037	ABCA13,ACE,AKAP8,AKAP9,ANK3,ANKLE2,ATP2C2,BIRC6,CFTR,COG2,COL24A1,COL6A3,CRIPAK,CSPG4,CUBN,CUL7,CYB5R2,CYFIP1,DCLRE1C,DOCK9,FLG2,GLYATL2,GOLGA6L2,HRNR,HSPG2,IGF2R,LILRB3,LLGL1,LRP2,MACF1,MSH6,MUC12,MUC16,MUC17,MUC4,MUC5B,MYH2,NRXN1,PACS2,PCDHA1,PCDHA4,PCDHA7,PDE4DIP,PDIA2,PIGN,PIK3C2B,PKD1,RANBP2,RLTPR,RYR2,SECTM1,SLC1A6,SNAP47,SREBF1,ST6GAL1,SYNE1,SYNE2,TBC1D30,TH,TM9SF4,TNC,TNK2,UBR4,VWF,ZFYVE28	
GO:0098590	plasma membrane region	22	1061	0.037	AKAP9,ANK3,CFTR,CSPG4,CUBN,DPP4,EPPK1,FAT1,HMCN1,HMCN2,LRP2,LRR4C,MACF1,MUC17,NDE1,NRXN1,PKD1,PKD1L1,RIMS2,SLC19A1,SYNE1,SYNE2	
GO:0005887	integral component of plasma membrane	29	1564	0.0405	AKAP9,ATP13A5,CACNA1H,CELSR1,CFTR,CNTN2,CSPG4,FAT1,GPR56,IGF2R,LILRA4,LILRB3,MUC12,MUC4,NRXN1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PKD1,PLXNA2,SLC19A1,SLC1A6,SLC22A10,SLC22A2,SLC22A25	
GO:0030054	cell junction	21	1006	0.0405	ANK3,DPP4,EPPK1,FAT1,HMCN1,HMCN2,LRR4C,MDC1,MYH1,MYH2,NRXN1,PCLO,PLEC,PLXNA3,PPL,RIMS2,SYNE2,TJP3,TNK2,TNS1,XIRP1	
GO:0030056	hemidesmosome	2	7	0.0405	EPPK1,PLEC	
GO:0031226	intrinsic component of plasma membrane	30	1641	0.0405	AKAP9,ATP13A5,CACNA1H,CELSR1,CFTR,CNTN2,CSPG4,DAB2IP,FAT1,GPR56,IGF2R,LILRA4,LILRB3,MUC12,MUC4,NRXN1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PKD1,PLXNA2,SLC19A1,SLC1A6,SLC22A10,SLC22A2,SLC22A25	
GO:0005938	cell cortex	8	230	0.0438	AKAP13,FRY,HMCN1,HMCN2,LLGL1,MYH2,PCLO,TRIOBP	
GO:0034704	calcium channel complex	4	60	0.0481	CACNA1H,CACNA2D4,PKD1L1,RYR2	
GO:0034993	meiotic nuclear membrane microtubule tethering complex	2	8	0.0481	SYNE1,SYNE2	

#Term ID	Term description	Observed gene count	Background gene count	False discovery rate	Genes	No. terms
GO:0042995	cell projection	34	1969	0.0481	AKAP9,ANK3,BEST2,CCDC40,CNTN2,CSPG4,CUBN,CYFIP1,DAB2IP,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,DPP4,EPPK1,FAT1,GPR56,GPR98,HYDIN,LLGL1,LRP2,MACF1,MYO15A,NRXN1,OSBP2,PKD1,PKD1L1,RLTPR,SNAP47,SYNE2,TBC1D30,TH,TRIOBP	
GO:0098862	cluster of actin-based cell projections	6	143	0.0481	CUBN,GPR98,LRP2,MYO15A,PLEC,TRIOBP	
GO:0120025	plasma membrane bounded cell projection	33	1900	0.0481	AKAP9,ANK3,BEST2,CCDC40,CNTN2,CSPG4,CUBN,CYFIP1,DAB2IP,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,DPP4,FAT1,GPR56,GPR98,HYDIN,LLGL1,LRP2,MACF1,MYO15A,NRXN1,OSBP2,PKD1,PKD1L1,RLTPR,SNAP47,SYNE2,TBC1D30,TH,TRIOBP	
GO:0120038	plasma membrane bounded cell projection part	25	1316	0.0481	AKAP9,ANK3,CCDC40,CNTN2,CSPG4,CUBN,CYFIP1,DAB2IP,DNAH1,DNAH11,DNAH6,DNAH8,DPP4,HYDIN,LRP2,MACF1,NRXN1,OSBP2,PKD1,PKD1L1,SNAP47,SYNE2,TBC1D30,TH,TRIOBP	
GO:0045202	synapse	18	849	0.0483	AKAP9,ANK3,CNTN2,CYFIP1,DROSHA,GPR98,LAMA5,LRRC4C,NDE1,NRXN1,PCDHA4,PCDHA7,PCLO,RIMS2,SLC22A2,SNAP47,SYNE1,TH	
rence Publications						
PMID:27484992	(2016) Parallel or convergent evolution in human population genomic data revealed by genotype networks.	24	49	3.06E-24	AHNAK2,DCHS2,DNAH11,FBN3,FCGBP,FLG,GPR98,HRNR,LAMA5,LILRA6,LILRB3,MKI67,MUC12,MUC16,MUC17,MUC4,MUC5B,NEB,OBSCN,PCL O,PKD1L1,PRUNE2,SYNE2,TTN	100
PMID:23206951	(2012) Identification and characterization of alternative exon usage linked glioblastoma multiforme survival.	11	36	2.81E-07	GPR98,HMCN1,MACF1,NEB,OBSCN,PDE4DIP,RYR2,SYNE1,SYNE2,TTN,UBR4	
PMID:29255378	(2017) The human, F-actin-based cytoskeleton as a mutagen sensor.	10	35	3.38E-06	DNAH11,DNAH8,MUC16,MUC17,MUC4,PCLO,PLEC,SYNE1,SYNE2,TTN	
PMID:25555989	(2015) Titin and obscurin: giants holding hands and discovery of a new Ig domain subset.	9	30	1.71E-05	CUL7,MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,OBSCN,SPEG	
PMID:23770567	(2013) Mutational heterogeneity in cancer and the search for new cancer-associated genes.	9	32	2.22E-05	CSMD3,DNAH11,KMT2D,MUC16,MUC4,NRXN1,PCLO,RYR2,TTN	
PMID:26179023	(2015) Fibre typing of intrafusal fibres.	8	24	5.56E-05	MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,OBSCN,TTN	

#Term ID	Term description	Observed gene count	Background gene count	False discovery rate	Genes	No. terms
PMID:28694483	(2017) Somatic mutations in ZFHX4 gene are associated with poor overall survival of Chinese esophageal squamous cell carcinoma patients.	9	37	5.56E-05	CSMD3,FAT1,KMT2D,LRP2,MUC16,NRXN1,PCLO,SYNE1,TTN	
PMID:15327201	(2004) Sarcomeric cytoskeletal proteins and myosin phenotype in stretched soleus of hindlimb-suspended rats	7	16	9.44E-05	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,TTN	
PMID:15526475	(2004) Human soleus fibers contractile characteristics and sarcomeric cytoskeletal proteins after gravitational unloading. Contribution of support stimulus	7	16	9.44E-05	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,TTN	
PMID:23331320	(2013) Unfolding dynamics of the mucin SEA domain probed by force spectroscopy suggest that it acts as a cell-protective device.	8	27	9.44E-05	CFTR,MUC12,MUC16,MUC17,MUC4,MUC5B,TTN,VWF	
PMID:2474551	(1989) Myogenesis in the mouse embryo: differential onset of expression of myogenic proteins and the involvement of titin in myofibril assembly.	7	16	9.44E-05	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,TTN	
PMID:25761688	(2015) Seasonal changes in isoform composition of giant proteins of thick and thin filaments and titin (connectin) phosphorylation level in striated muscles of bears (Ursidae, Mammalia).	7	16	9.44E-05	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,TTN	
PMID:25977021	(2015) Evidence of peptide oxidation from major myofibrillar proteins in dry-cured ham.	7	16	9.44E-05	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,TTN	
PMID:29203134	(2018) Proteomic profiling of large myofibrillar proteins from dried and long-term stored polyacrylamide gels.	7	16	9.44E-05	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,TTN	
PMID:8818801	(1996) Effect of electrical stimulation on postmortem titin, nebulin, desmin, and troponin-T degradation and ultrastructural changes in bovine longissimus muscle.	7	16	9.44E-05	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,TTN	
PMID:28396857	(2017) Proteome Profiles of Digested Products of Commercial Meat Sources.	7	18	0.0001	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,TTN	

#Term ID	Term description	Observed gene count	Background gene count	False discovery rate	Genes	No. terms
PMID:22400118	(2012) At the Start of the Sarcomere: A Previously Unrecognized Role for Myosin Chaperones and Associated Proteins during Early Myofibrillogenesis.	8	32	0.00014	HSPG2,MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,NEB	
PMID:20350540	(2010) Nonmuscle myosin IIB, a sarcomeric component in the extraocular muscles.	8	33	0.00016	MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,NEB,TTN	
PMID:24988079	(2014) Integrated analysis of mutation data from various sources identifies key genes and signaling pathways in hepatocellular carcinoma.	11	90	0.00016	ABCA13,CSMD3,CUBN,DNAH8,GPR98,HMCN1,MUC16,PCLO,RYR2,SYNE1,TTN	
PMID:25541946	(2014) Two desmin gene mutations associated with myofibrillar myopathies in Polish families.	8	34	0.00018	CMYA5,MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,TTN	
PMID:26759717	(2016) Genomic analyses reveal FAM84B and the NOTCH pathway are associated with the progression of esophageal squamous cell carcinoma.	8	34	0.00018	CSMD3,FAT1,KMT2D,MAML1,MUC16,PCLO,SYNE1,TTN	
PMID:24360430	(2014) LC MSMS identification of large structural proteins from bull muscle and their degradation products during post mortem storage.	6	11	0.00019	MYH1,MYH2,MYOM2,NEB,RYR2,TTN	
PMID:20359464	(2010) Carbonylation of myosin heavy chains in rat heart during diabetes.	7	22	0.00022	MYH1,MYH11,MYH2,MYH3,MYH7B,RYR2,TTN	
PMID:28044126	(2016) Mechanotransduction and Metabolism in Cardiomyocyte Microdomains.	8	37	0.00026	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,RYR2,TTN	
PMID:25125171	(2014) Localization of sarcomeric proteins during myofibril assembly in cultured mouse primary skeletal myotubes.	8	39	0.00036	MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,NEB,TTN	
PMID:18218129	(2008) Diaphragm adaptations in patients with COPD.	7	25	0.00041	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,TTN	
PMID:27009202	(2016) The SH3 domain of UNC-89 (obscurin) interacts with paramyosin, a coiled-coil protein, in Caenorhabditis elegans muscle.	8	40	0.00041	AACS,MYH1,MYH11,MYH2,MYH3,MYH7B,OBSCN,TTN	
PMID:28435450	(2017) Exome Sequencing of Oral Squamous Cell Carcinoma Reveals Molecular Subgroups and Novel Therapeutic Opportunities.	8	40	0.00041	CSMD3,FAT1,KMT2D,MUC16,MUC4,MUC5B,PCLO,TTN	

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PMID:23776483	(2013) Mucins as diagnostic and prognostic biomarkers in a fish-parasite model: transcriptional and functional analysis.	6	14	0.00045	MUC12,MUC16,MUC17,MUC4,MUC5B,VWF	
PMID:24119284	(2013) Determining the role of sarcomeric proteins in facioscapulohumeral muscular dystrophy: a study protocol.	7	26	0.00045	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,TTN	
PMID:24804796	(2014) Molecular remodeling of left and right ventricular myocardium in chronic anthracycline cardiotoxicity and post-treatment follow up.	7	26	0.00045	MYH1,MYH11,MYH2,MYH3,MYH7B,RYR2,TTN	
PMID:24578791	(2013) Assessment of Developmental Toxicants using Human Embryonic Stem Cells.	7	27	0.00052	MYH1,MYH11,MYH2,MYH3,MYH7B,RYR2,XIRP1	
PMID:10661318	(1999) Genetic variability of foetal bovine myoblasts in primary culture.	6	15	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:15100236	(2004) Fiber types in rat laryngeal muscles and their transformations after denervation and reinnervation.	6	15	0.00054	CNTN2,MYH1,MYH11,MYH2,MYH3,MYH7B	
PMID:15494719	(2004) Multiple roles of mucins in pancreatic cancer, a lethal and challenging malignancy.	7	29	0.00054	MAGEC1,MUC12,MUC16,MUC17,MUC4,MUC5B,VWF	
PMID:15612538	(2004) The behavior of titin and the proteins of its family from skeletal muscles of ground squirrel (Citellus undulatus) during hibernation and rats under conditions of simulated microgravity	6	15	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:1571325	(1992) Fetal development of the urethral sphincter.	6	15	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:1704877	(1990) Myofibrillar M-band proteins in rat skeletal muscles during development.	6	15	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2	
PMID:2144849	(1990) Expression of myosin heavy chain isoforms in developing rat muscle spindles.	6	15	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2	
PMID:22054492	(1981) Proteolytic degradation of connectin, a high molecular weight myofibrillar protein, during heating of meat.	6	16	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	

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PMID:22752243	(2012) The velocity of cardiac sarcomere shortening: mechanisms and implications.	6	16	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:22790982	(2012) Splicing of internal large exons is defined by novel cis-acting sequence elements.	6	15	0.00054	MUC12,MUC16,MUC17,MUC4,MUC5B,TTN	
PMID:23272578	(2012) Seasonal changes in the isoform composition of the myosin heavy chains in skeletal muscles of hibernating ground squirrels <i>Spermophilus undulatus</i>	6	15	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:23277479	(2013) Effect of muscle length on cross-bridge kinetics in intact cardiac trabeculae at body temperature.	6	15	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:25081385	(2014) Cardiac tissue structure, properties, and performance: a materials science perspective.	6	15	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:25399805	(2014) The pepsin digestibility of thermal gel products made from white croaker (<i>Pennahia argentata</i>) muscle in associating with myosin polymerization levels.	6	16	0.00054	ACE,MYH1,MYH11,MYH2,MYH3,MYH7B	
PMID:25664316	(2015) Isoform composition and gene expression of thick and thin filament proteins in striated muscles of mice after 30-day space flight.	8	45	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,PLEC,TTN	
PMID:28115019	(2017) Correlation between change in muscle excursion and collagen content after tendon rupture and delayed repair.	6	15	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:29137374	(2017) Hypoxia favors myosin heavy chain beta gene expression in an Hif-1alpha-dependent manner.	7	28	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,TTN	
PMID:3536962	(1986) Titin and myosin, but not desmin, are linked during myofibrillogenesis in postmitotic mononucleated myoblasts.	6	15	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:7174202	(1982) Studies on the structure of connectin in muscle.	6	16	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:8025340	(1993) Development of the diaphragmatic muscle	6	15	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	

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PMID:9314534	(1997) Desmin is essential for the tensile strength and integrity of myofibrils but not for myogenic commitment, differentiation, and fusion of skeletal muscle.	7	28	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,TTN	
PMID:9713857	(1998) The role of immunocytochemistry in congenital myopathies.	6	15	0.00054	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB	
PMID:24037260	(2013) Localisation of AMPK Gamma subunits in cardiac and skeletal muscles.	7	30	0.00056	MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,PLEC	
PMID:10205233	(1999) Angiotensin II type 1 receptor antagonist downregulates nonmuscle myosin heavy chains in spontaneously hypertensive rat aorta.	6	17	0.00058	ACE,MYH1,MYH11,MYH2,MYH3,MYH7B	
PMID:20703484	(2011) Muscle plasticity in hibernating ground squirrels (<i>Spermophilus lateralis</i>) is induced by seasonal, but not low-temperature, mechanisms.	6	17	0.00058	FOXO1,MYH1,MYH11,MYH2,MYH3,MYH7B	
PMID:25466818	(2014) FLAGS, frequently mutated genes in public exomes.	6	17	0.00058	EPPK1,HRNR,KMT2D,MUC16,TTN,VPS13B	
PMID:29575536	(2018) Mutation analysis of adenomas and carcinomas of the colon: Early and late drivers.	6	17	0.00058	BIRC6,CSMD3,MUC17,OBSCN,RYR2,TTN	
PMID:19130271	(2008) Calcium-dependent signaling mechanisms and soleus fiber remodeling under gravitational unloading.	7	31	0.00063	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,TTN	
PMID:20414365	(2010) Molecular structure of sarcomere-to-membrane attachment at M-Lines in <i>C. elegans</i> muscle.	7	32	0.00071	HSPG2,MYH1,MYH11,MYH2,MYH3,MYH7B,OBSCN	
PMID:20824529	(2010) Short-range mechanical properties of skeletal and cardiac muscles.	6	18	0.00071	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:23204842	(2012) Skeletal muscle molecular alterations precede whole-muscle dysfunction in NYHA Class II heart failure patients.	6	18	0.00071	ACE,MYH1,MYH11,MYH2,MYH3,MYH7B	
PMID:25400587	(2014) O-GlcNAcylation, contractile protein modifications and calcium affinity in skeletal muscle.	7	32	0.00071	MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,TTN	
PMID:27824934	(2016) Controlled Heat Stress Promotes Myofibrillogenesis during Myogenesis.	8	49	0.00071	FOXO1,MYH1,MYH11,MYH2,MYH3,MYH7B,NEB,TTN	

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PMID:28042345	(2016) Biomimetic microstructure morphology in electrospun fiber mats is critical for maintaining healthy cardiomyocyte phenotype.	6	18	0.00071	MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2	
PMID:29491150	(2018) ELKS active zone proteins as multitasking scaffolds for secretion.	7	32	0.00071	MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,PLEC	
PMID:29344379	(2018) Changes in contractile protein expression are linked to ventricular stiffness in infants with pulmonary hypertension or right ventricular hypertrophy due to congenital heart disease.	7	33	0.0008	CMYA5,MYOM2,OBSCN,RYR2,SPEG,TTN,XIRP1	
PMID:18769205	(2008) Functions of ocular surface mucins in health and disease.	6	19	0.00082	MUC12,MUC16,MUC17,MUC4,MUC5B,VWF	
PMID:20563650	(2011) Impact of polymorphism of the regulatory subunit of the my-calpain (CAPN1S) on the proteolysis process and meat tenderness of young cattle.	6	19	0.00082	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:22919607	(2012) Defense and adaptation: the complex inter-relationship between Campylobacter jejuni and mucus.	6	19	0.00082	MUC12,MUC16,MUC17,MUC4,MUC5B,VWF	
PMID:23478383	(2013) The gastrointestinal mucus system in health and disease.	6	19	0.00082	CFTR,MUC12,MUC17,MUC4,MUC5B,VWF	
PMID:16887038	(2006) An inventory of mucin genes in the chicken genome shows that the mucin domain of Muc13 is encoded by multiple exons and that ovomucin is part of a locus of related gel-forming mucins.	6	20	0.001	MUC12,MUC16,MUC17,MUC4,MUC5B,VWF	
PMID:20647489	(2010) Making the case for skeletal myopathy as the major limitation of exercise capacity in heart failure.	6	20	0.001	ACE,MYH1,MYH11,MYH2,MYH3,MYH7B	
PMID:21297871	(2011) Molecular mechanisms of sarcomere dysfunction in dilated and hypertrophic cardiomyopathy.	6	20	0.001	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:23523554	(2013) Tracking unfolding and refolding reactions of single proteins using atomic force microscopy methods.	6	20	0.001	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:27034009	(2016) The degree of intratumor mutational heterogeneity varies by primary tumor sub-site.	13	188	0.001	CRIPAK,CSMD3,HMCN1,HYDIN,KMT2D,LRP2,MSH6,PCLO,RIMS2,RNF213,RYR2,SYNE1,SYNE2	

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PMID:17319961	(2007) A musculoskeletal model of low grade connective tissue inflammation in patients with thyroid associated ophthalmopathy (TAO): the WOMED concept of lateral tension and its general implications in disease.	8	54	0.0011	MYH1,MYH11,MYH2,MYH3,MYH7B,OBSCN,RYR2,TTN	
PMID:29271938	(2017) Chaperones and the Proteasome System: Regulating the Construction and Demolition of Striated Muscle.	8	54	0.0011	MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,NEB,OBSCN	
PMID:29339694	(2018) Aortic Alfa-smooth muscle actin expressions in aortic disorders and coronary artery disease: An immunohistochemical study.	6	21	0.0012	ACE,MYH1,MYH11,MYH2,MYH3,MYH7B	
PMID:21146919	(2011) Role of mucins in the skin during benign and malignant conditions.	7	37	0.0013	MUC12,MUC16,MUC17,MUC4,MUC5B,TNS1,VWF	
PMID:19598116	(2009) S. macrurus myogenic regulatory factors (MRFs) induce mammalian skeletal muscle differentiation; evidence for functional conservation of MRFs.	6	22	0.0014	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:23936073	(2013) Cardiac myosin binding protein-C plays no regulatory role in skeletal muscle structure and function.	6	22	0.0014	MYH1,MYH11,MYH2,MYH3,MYH7B,TTN	
PMID:25822412	(2015) GAlfaq protein carboxyl terminus imitation polypeptide GCIP-27 improves cardiac function in chronic heart failure rats.	6	22	0.0014	ACE,MYH1,MYH11,MYH2,MYH3,MYH7B	
PMID:25634251	(2015) Genetically engineered mucin mouse models for inflammation and cancer.	7	38	0.0015	CFTR,MADCAM1,MUC16,MUC17,MUC4,MUC5B,VWF	
PMID:21385362	(2011) Genome wide analysis of the bovine mucin genes and their gastrointestinal transcription profile.	6	23	0.0017	MUC12,MUC16,MUC17,MUC4,MUC5B,VWF	
PMID:21798101	(2011) Novel mutations in NEB cause abnormal nebulin expression and markedly impaired muscle force generation in severe nemaline myopathy.	6	23	0.0017	MYH1,MYH11,MYH2,MYH3,MYH7B,NEB	
PMID:23138638	(2013) Muscle development, regeneration and laminopathies: how lamins or lamina-associated proteins can contribute to muscle development, regeneration and disease.	10	107	0.0017	ANKLE2,MYH1,MYH11,MYH2,MYH3,MYH7B,SREBF1,SYNE1,SYNE2,TTN	

[illegible]

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HSA-977068	Termination of O-glycan biosynthesis	6	22	0.00021	MUC12,MUC16,MUC17,MUC4,MUC5B,ST6GAL1	10
HSA-5083625	Defective GALNT3 causes familial hyperphosphatemic tumoral calcinosis (HFTC)	5	16	0.00056	MUC12,MUC16,MUC17,MUC4,MUC5B	
HSA-5083632	Defective C1GALT1C1 causes Tn polyagglutination syndrome (TNPS)	5	16	0.00056	MUC12,MUC16,MUC17,MUC4,MUC5B	
HSA-5083636	Defective GALNT12 causes colorectal cancer 1 (CRCS1)	5	16	0.00056	MUC12,MUC16,MUC17,MUC4,MUC5B	
HSA-5621480	Dectin-2 family	5	26	0.0017	MUC12,MUC16,MUC17,MUC4,MUC5B	
HSA-3000171	Non-integrin membrane-ECM interactions	5	58	0.0384	HSPG2,LAMA5,LAMC3,NRXN1,TNC	
HSA-399955	SEMA3A-Plexin repulsion signaling by inhibiting Integrin adhesion	3	14	0.043	PLXNA2,PLXNA3,PLXNA4	
HSA-3781865	Diseases of glycosylation	7	136	0.0471	CSPG4,HSPG2,MUC12,MUC16,MUC17,MUC4,MUC5B	
HSA-399954	Sema3A PAK dependent Axon repulsion	3	16	0.0471	PLXNA2,PLXNA3,PLXNA4	
HSA-399956	CRMPs in Sema3A signaling	3	16	0.0471	PLXNA2,PLXNA3,PLXNA4	
UniProt Keywords						
KW-0677	Repeat	108	4751	3.54E-14	ABCA13,ACE,ACOT11,ADNP2,AKAP8,ALS2CL,ANK3,ANKRD30A,BRF1,CACNA1H,CELSR1,CFTR,CMYA5,CNTN2,COL24A1,COL6A3,CRIPAK,CSMD3,CSPG4,CUBN,DCHS2,DNAH11,DNAH6,DNAH8,DOCK9,DROSHA,EPPK1,ESX1,FAT1,FBN3,FBXO11,FLG,FLG2,GPR98,GTF2IRD1,HIVEP3,HMCN1,HMCN2,HRNR,HSPG2,IGF2R,KLC4,KMT2D,LAMA5,LAMC3,LILRA4,LILRA6,LILRB3,LLGL1,LRP2,LRRC4C,LRRIQ1,LRTM2,MACF1,MADCAM1,MAGEC1,MDC1,MEGF8,MKI67,MUC12,MUC16,MUC17,MUC4,MUC5B,MYO15A,MYOM2,NRXN1,NSMAF,OBSCN,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCL0,PDIA2,PKD1,PKD1L1,PLEC,PLXNA2,PLXNA3,PLXNA4,PPL,RANBP2,RIMS2,RLTPR,RYR2,SCAF1,SNAP47,SNED1,SPEG,SVEP1,SYNE1,SYNE2,TJP3,TNC,TTN,TUBGCP6,VWF,XIRP1,ZNF17,ZNF417,ZNF749,ZNF772,ZSCAN5A,ZSCAN5DP	31

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KW-0106	Calcium	33	866	8.61E-08	ATP2C2,BEST2,CACNA1H,CACNA2D4,CELSR1,CUBN,DCHS2,FAT1,FBN3,FLG,FLG2,GPR98,HMCN1,HMCN2,HRNR,HSPG2,LRP2,MACF1,MEGF8,NRXN1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCLO,PKD1L1,RYR2,SNED1,SVEP1,TTN	
KW-0621	Polymorphism	168	11707	2.89E-07	AACS,ABCA13,ACE,ACOT11,AHNAK2,AKAP13,AKAP8,AKAP9,ALS2CL,AMZ1,ANK3,ANKLE2,ANKRD30A,ATP13A5,ATP2C2,BRF1,C16orf59,C20orf96,C9orf84,CACNA1H,CACNA2D4,CCDC40,CELSR1,CFTR,CMYA5,CNTN2,COG2,COL24A1,COL6A3,CPZ,CRIPAK,CSMD3,CSPG4,CUBN,CUL7,CYB5R2,CYFIP1,DAAM2,DAB2IP,DCHS2,DCLRE1C,DNAH1,DNAH11,DNAH6,DNAH8,DOCK9,DROSHA,ESX1,ETFB,FAM151A,FAM90A1,FAT1,FBN3,FBXO11,FLG,FLG2,FRY,GLYATL2,GOLGA6L2,GPR56,GPR78,GPR98,GSE1,GTTF2IRD1,HBS1L,HIVEP3,HMCN1,HRNR,HSPG2,HUS1,HYDIN,IGF2R,KLC4,KMT2D,LAMA5,LAMC3,LILRA4,LILRA6,LILRB3,LLGL1,LRP2,LRRIQ1,LRTM2,MACF1,MADCAM1,MAGEC1,MAML1,MDC1,MGA,MKI67,MSH6,MUC12,MUC16,MUC17,MUC4,MUC5B,MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2,NABP1,NRXN1,NSMAF,OR10C1,OR11A1,OR5V1,OSBP2,PACS2,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCLO,PCNT,PDE4DIP,PDIA2,PIGN,PKD1,PKD1L1,PLEC,PLXNA2,PLXNA3,PPL,PRUNE2,RABL6,RANBP2,RECQL4,RPS9,RSF1,RYR2,SCAF1,SLC19A1,SLC22A2,SLC22A25,SNAP47,SNED1,SOX13,SPEG,SREBF1,SVEP1,SYNE1,SYNE2,TBC1D30,TH,TJP3,TNC,TNK2,TNS1,TRIOBP,TTN,TUBGCP6,UBR4,USP41,VAR2,VPS13B,VWF,XIRP1,ZFYVE28,ZNF17,ZNF417,ZNF749,ZNF772,ZSCAN5A	
KW-0245	EGF-like domain	16	229	6.88E-07	ADAM20,CELSR1,CUBN,FAT1,FBN3,HMCN1,HMCN2,HSPG2,LRP2,MEGF8,MUC12,MUC4,NRXN1,SNED1,SVEP1,TNC	

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KW-0175	Coiled coil	52	2158	1.90E-06	AKAP13,AKAP9,ANKRD30A,C20orf96,CCDC40,CMYA5,DAAM2,DAB2IP,DNAH1,DNAH11,DNAH14,DNAH6,DNAH8,DOCK9,EPPK1,FLG,GOLGA6L2,GSE1,HIVEP3,HYDIN,KLC4,KMT2D,LAMA5,LAMC3,MGA,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,NDE1,NEB,NOL12,OBSCN,PCNT,PDE4DIP,PKD1,PLEC,PLXNA2,PPL,RNF213,RSF1,RYR2,SNAP47,SYNE1,SYNE2,TCP10L2,TNC,TRIOBP,TTN,XIRP1	
KW-0130	Cell adhesion	21	476	4.71E-06	CNTN2,COL6A3,DCHS2,DPP4,FAT1,GPR56,LAMA5,LAMC3,MADCAM1,MUC4,NRXN1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,SVEP1,TNC,VWF	
KW-0787	Thick filament	6	17	4.77E-06	MYH1,MYH11,MYH2,MYH3,MYH7B,MYOM2	
KW-0505	Motor protein	11	131	1.34E-05	DNAH1,DNAH11,DNAH6,DNAH8,KLC4,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A	
KW-0009	Actin-binding	14	266	6.57E-05	CYFIP1,MACF1,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,PLEC,SYNE1,SYNE2,TNS1,TRIOBP,XIRP1	
KW-0518	Myosin	6	50	0.0008	MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A	
KW-0424	Laminin EGF-like domain	5	30	0.00083	CELSR1,HSPG2,LAMA5,LAMC3,MEGF8	
KW-0112	Calmodulin-binding	9	152	0.0014	MYH1,MYH11,MYH2,MYH3,MYO15A,PCNT,RYR2,TTN,UBR4	
KW-1015	Disulfide bond	57	3276	0.0032	ACE,ADAM20,CACNA2D4,CELSR1,CGB2,CNTN2,COL6A3,CPZ,CSMD3,CSPG4,CUBN,DPP4,FAT1,FBN3,FCAMR,GPR78,HMCN1,HMCN2,HSPG2,IGF2R,LAMA5,LAMC3,LILRA4,LILRA6,LILRB3,LRP2,LRR4C,MADCAM1,MEGF8,MUC12,MUC16,MUC17,MUC4,MUC5B,NRXN1,OBSCN,OR10C1,OR11A1,OR5V1,PCDHA4,PCDHA7,PDIA2,PKD1,PLXNA2,PLXNA3,PLXNA4,RANBP2,SECTM1,SNED1,SPEG,ST6GAL1,SVEP1,SYNE1,SYNE2,TNC,TTN,VWF	
KW-0272	Extracellular matrix	11	263	0.0035	COL24A1,COL6A3,CPZ,FBN3,HMCN1,HMCN2,HSPG2,LAMA5,LAMC3,TNC,VWF	
KW-1012	Kartagener syndrome	4	27	0.0054	CCDC40,DNAH1,DNAH11,HYDIN	

#Term ID	Term description	Observed gene count	Background gene count	False discovery rate	Genes	No. terms
KW-0025	Alternative splicing	136	10223	0.0069	AACS,ABCA13,ACE,ACOT11,AHNAK2,AKAP13,AKAP9,ALS2CL,AMZ1,ANK3,ANKLE2,ATP2C2,BRF1,C16orf59,C9orf84,CACNA1H,CACNA2D4,CCDC40,CELSR1,CFTR,COG2,COL24A1,COL6A3,CPZ,CSMD3,CUL7,CYB5R2,CYFIP1,DAAM2,DAB2IP,DCHS2,DCLRE1C,DDX39B,DNAH1,DNAH6,DNAH8,DOCK9,DROSHA,ETFB,FAM151A,FBXO11,FCAMR,GLYATL1,GOLGA6L2,GPR56,GPR98,GSE1,GTF2IRD1,HBS1L,HIVEP3,HMCN1,HMCN2,HUS1,HYDIN,KLC4,KMT2D,LAMA5,LILRA4,LILRA6,LILRB3,LRRIQ1,MACF1,MADCAM1,MAGEC1,MDC1,MEGF8,MGA,MKI67,MSH6,MUC12,MUC17,MUC4,MYH11,MYH2,MYH7B,MYO15A,NABP1,NDE1,NRXN1,NSMAF,OSBP2,PACS2,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PCLO,PCNT,PDE4DIP,PDIA2,PKD1,PKD1L1,PLEC,PLXNA2,PLXNA4,PRUNE2,RABL6,RIMS2,RLTPR,RNF213,RSF1,RYR2,SLC19A1,SLC1A6,SLC22A10,SLC22A2,SNAP47,SNED1,SPEG,SREBF1,ST6GAL1,SVEP1,SYNE1,SYNE2,TBC1D30,TH,TJP3,TNC,TNK2,TNS1,TRIOBP,TTN,TUBGCP6,UBR4,VARS2,VPS13B,VWF,XIRP1,ZFYVE28,ZNF17,ZNF417,ZNF772,ZSCAN5A	
KW-0325	Glycoprotein	69	4353	0.0069	ACE,ADAM20,ATP13A5,CACNA1H,CACNA2D4,CELSR1,CFTR,CGB2,CNTN2,COL24A1,COL6A3,CPZ,CSMD3,CSPG4,CUBN,DCHS2,DPP4,FAT1,FBN3,FCAMR,GPR56,HMCN1,HMCN2,HSPG2,IGF2R,LAMA5,LAMC3,LILRA4,LILRA6,LILRB3,LRP2,LRTM2,MADCAM1,MEGF8,MUC12,MUC16,MUC17,MUC4,MUC5B,NRXN1,OR10C1,OR11A1,OR5V1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PDIA2,PIGN,PKD1,PKD1L1,PLXNA2,PLXNA3,PLXNA4,RABL6,SECTM1,SLC19A1,SLC1A6,SLC22A10,SLC22A2,SLC22A25,SNED1,ST6GAL1,SVEP1,TNC,VWF	
KW-0393	Immunoglobulin domain	14	448	0.0071	CNTN2,FCAMR,HMCN1,HMCN2,HSPG2,LILRA4,LILRA6,LILRB3,LRRC4C,MADCAM1,MYOM2,OBSCN,SPEG,TTN	
KW-0243	Dynein	4	32	0.0078	DNAH1,DNAH11,DNAH6,DNAH8	

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KW-0206	Cytoskeleton	26	1198	0.0091	AKAP9,ANK3,BIRC6,CUL7,DNAH1,DNAH11,DNAH6,DNAH8,EPPK1,FRY,KLC4,LLGL1,MACF1,MYO15A,NDE1,PCNT,PDE4DIP,PLEC,PPL,RLTPR,SYNE1,SYNE2,TNS1,TRIOBP,TUBGCP6,UBR4	
KW-9995	Disease	61	3799	0.0098	AKAP13,AKAP9,ANK3,ANKLE2,BRF1,C12orf57,CACNA1H,CCDC40,CELSR1,CFTR,CNTN2,COG2,COL6A3,CUBN,CUL7,DAB2IP,DCLRE1C,DNAH1,DNAH11,ETFB,FLG,FLG2,FOXO1,GPR56,GPR98,GTF2IRD1,HMCN1,HSPG2,HYDIN,KMT2D,LAMC3,LRP2,MEGF8,MSH6,MYH11,MYH2,MYH3,MYO15A,NDE1,NRXN1,PACS2,PCLO,PCNT,PIGN,PKD1,PKD1L1,PLEC,RECQL4,RLTPR,RNF213,RYR2,SPEG,SYNE1,SYNE2,TH,TNC,TRIOBP,TTN,VARS2,VPS13B,VWF	
KW-1003	Cell membrane	53	3208	0.0117	ACE,ANK3,BEST2,CACNA1H,CELSR1,CFTR,CNTN2,CSMD3,CSPG4,CUBN,DAB2IP,DPP4,EPPK1,FAT1,FCAMR,GPR56,GPR78,GPR98,LILRA4,LILRB3,LRP2,LRRC4C,MACF1,MUC16,MUC17,MUC4,NRXN1,OR10C1,OR11A1,OR5V1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PIK3C2B,PKD1,PKD1L1,PLXNA2,PLXNA3,PLXNA4,PPL,RFTN2,RIMS2,RLTPR,SECTM1,SLC1A6,SYNE2,TBC1D30,TJP3,TNK2	
KW-0084	Basement membrane	4	40	0.0134	HMCN1,HSPG2,LAMA5,LAMC3	
KW-0732	Signal	53	3237	0.0134	ACE,ADAM20,CACNA2D4,CELSR1,CGB2,CNTN2,COL24A1,COL6A3,CPZ,CSPG4,CUBN,FAT1,FBN3,FCAMR,GPR56,GPR98,HMCN1,HMCN2,HSPG2,IGF2R,LAMA5,LAMC3,LILRA4,LILRA6,LILRB3,LRP2,LRRC4C,LRTM2,MADCAM1,MEGF8,MUC12,MUC17,MUC4,MUC5B,NRXN1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7,PDIA2,PKD1,PLXNA2,PLXNA3,PLXNA4,SECTM1,SNED1,SVEP1,TM9SF4,TNC,VWF	
KW-0488	Methylation	21	959	0.021	AKAP13,AKAP8,ETFB,FOXO1,GSE1,HMCN2,HRNR,IGF2R,KMT2D,MDC1,MGA,MYH1,MYH11,MYH2,MYH3,PLEC,RANBP2,RLTPR,SPEG,TNK2,TRIOBP	

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KW-0966	Cell projection	20	910	0.0247	ANK3,CCDC40,CSPG4,CYFIP1,DAB2IP,DNAH1,DNAH11,DNAH6,DNAH8,DPP4,EPPK1,GPR98,HYDIN,LLGL1,LRP2,MACF1,MYO15A,PKD1,PKD1L1,RLTPR	
KW-1067	Emery-Dreifuss muscular dystrophy	2	6	0.0283	SYNE1,SYNE2	
KW-0728	SH3 domain	8	221	0.0306	MACF1,MYO15A,NEB,OBSCN,PLEC,PPL,TJP3,TNK2	
KW-0493	Microtubule	9	275	0.0322	DNAH1,DNAH11,DNAH6,DNAH8,KLC4,MACF1,NDE1,PCNT,TUBGCP6	
KW-0031	Aminopeptidase	3	27	0.0334	DNPEP,DPP4,TPP2	
KW-0109	Calcium transport	5	100	0.0449	ATP2C2,CACNA1H,CACNA2D4,PKD1L1,RYR2	
PFAM Protien Domains						
PF00028	Cadherin domain	10	113	0.00013	CELSR1,DCHS2,FAT1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7	54
PF02210	Laminin G domain	7	39	0.00013	CELSR1,COL24A1,CSPG4,FAT1,HSPG2,LAMA5,NRXN1	
PF02736	Myosin N-terminal SH3-like domain	5	14	0.00013	MYH1,MYH11,MYH2,MYH3,MYH7B	
PF15974	Cadherin C-terminal cytoplasmic tail, catenin-binding region	7	37	0.00013	PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7	
PF00054	Laminin G domain	6	34	0.00026	CELSR1,CSPG4,FAT1,HSPG2,LAMA5,NRXN1	
PF00063	Myosin head (motor domain)	6	38	0.00026	MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A	
PF01576	Myosin tail	5	18	0.00026	MYH1,MYH11,MYH2,MYH3,MYH7B	
PF07645	Calcium-binding EGF domain	8	78	0.00026	CUBN,FBN3,HMCN1,HMCN2,LRP2,MEGF8,SNED1,SVEP1	
PF13895	Immunoglobulin domain	12	212	0.00026	CNTN2,HMCN1,HMCN2,HSPG2,LILRA4,LILRA6,LILRB3,LRR4C4,MYOM2,OBSCN,SPEG,TTN	
PF08266	Cadherin-like	7	63	0.00033	PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7	
PF00047	Immunoglobulin domain	12	243	0.00065	CNTN2,HMCN1,HMCN2,HSPG2,LILRA4,LILRA6,LILRB3,LRR4C4,MYOM2,OBSCN,SPEG,TTN	
PF12780	P-loop containing dynein motor region D4	4	13	0.00075	DNAH1,DNAH11,DNAH6,DNAH8	
PF12781	ATP-binding dynein motor region D5	4	13	0.00075	DNAH1,DNAH11,DNAH6,DNAH8	
PF00008	EGF-like domain	7	81	0.00082	CELSR1,CUBN,FAT1,HSPG2,NRXN1,SNED1,SVEP1	
PF00053	Laminin EGF domain	5	31	0.00082	CELSR1,HSPG2,LAMA5,LAMC3,MEGF8	
PF00094	von Willebrand factor type D domain	4	14	0.00082	FCGBP,MUC4,MUC5B,VWF	
PF03028	Dynein heavy chain and region D6 of dynein motor	4	14	0.00082	DNAH1,DNAH11,DNAH6,DNAH8	
PF08393	Dynein heavy chain, N-terminal region 2	4	14	0.00082	DNAH1,DNAH11,DNAH6,DNAH8	

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PF12774	Hydrolytic ATP binding site of dynein motor region D1	4	14	0.00082	DNAH1,DNAH11,DNAH6,DNAH8	
PF12775	P-loop containing dynein motor region D3	4	14	0.00082	DNAH1,DNAH11,DNAH6,DNAH8	
PF12777	Microtubule-binding stalk of dynein motor	4	14	0.00082	DNAH1,DNAH11,DNAH6,DNAH8	
PF00052	Laminin B (Domain IV)	3	8	0.0028	HSPG2,LAMA5,LAMC3	
PF08337	Plexin cytoplasmic RasGAP domain	3	9	0.0036	PLXNA2,PLXNA3,PLXNA4	
PF01437	Plexin repeat	4	26	0.0039	MEGF8,PLXNA2,PLXNA3,PLXNA4	
PF01826	Trypsin Inhibitor like cysteine rich domain	3	10	0.0042	FCGBP,MUC5B,VWF	
PF08742	C8 domain	3	10	0.0042	FCGBP,MUC5B,VWF	
PF07679	Immunoglobulin I-set domain	9	202	0.0058	CNTN2,HMCN1,HMCN2,HSPG2,LRRC4C,MYOM2,OBSCN,SPEG,TTN	
PF10495	Pericentrin-AKAP-450 domain of centrosomal targeting protein	2	2	0.0079	AKAP9,PCNT	
PF07728	AAA domain (dynein-related subfamily)	4	34	0.0081	DNAH1,DNAH11,DNAH6,DNAH8	
PF00041	Fibronectin type III domain	7	133	0.0088	CMYA5,CNTN2,MYOM2,OBSCN,SNED1,TNC,TTN	
PF13927	Immunoglobulin domain	9	221	0.0094	CNTN2,HMCN1,HMCN2,HSPG2,LRRC4C,MYOM2,OBSCN,SPEG,TTN	
PF12662	Complement C1r-like EGF-like	4	40	0.0128	FBN3,HMCN1,HMCN2,LRP2	
PF12947	EGF domain	3	18	0.0139	CUBN,FBN3,MEGF8	
PF00681	Plectin repeat	2	4	0.016	EPPK1,PLEC	
PF01390	SEA domain	3	20	0.016	MUC12,MUC16,MUC17	
PF06021	Aralkyl acyl-CoA:amino acid N-acyltransferase	2	4	0.016	GLYATL1,GLYATL2	
PF07474	G2F domain	2	4	0.016	HMCN1,HMCN2	
PF08444	Aralkyl acyl-CoA:amino acid N-acyltransferase, C-terminal region	2	4	0.016	GLYATL1,GLYATL2	
PF10541	Nuclear envelope localisation domain	2	4	0.016	SYNE1,SYNE2	
PF12661	Human growth factor-like EGF	4	46	0.0166	CUBN,FBN3,SNED1,SVEP1	
PF00007	Cystine-knot domain	3	22	0.0185	CGB,CGB2,ENSG00000267335	
PF00435	Spectrin repeat	3	22	0.0185	MACF1,SYNE1,SYNE2	
PF06119	Nidogen-like	2	5	0.0185	MUC4,SNED1	
PF01833	IPT/TIG domain	3	24	0.0219	PLXNA2,PLXNA3,PLXNA4	
PF02010	REJ domain	2	6	0.0223	PKD1,PKD1L1	
PF00801	PKD domain	2	7	0.0278	PKD1,PKD1L1	
PF02207	Putative zinc finger in N-recognin (UBR box)	2	7	0.0278	FBXO11,UBR4	
PF00092	von Willebrand factor type A domain	4	58	0.0299	CACNA2D4,COL6A3,SVEP1,VWF	

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PF01023	S-100/ICaBP type calcium binding domain	3	28	0.0299	FLG,FLG2,HRNR	
PF13519	von Willebrand factor type A domain	4	58	0.0299	CACNA2D4,COL6A3,SVEP1,VWF	
PF01403	Sema domain	3	30	0.0336	PLXNA2,PLXNA3,PLXNA4	
PF08385	Dynein heavy chain, N-terminal region 1	2	9	0.0371	DNAH11,DNAH8	
PF00307	Calponin homology (CH) domain	4	67	0.0443	MACF1,PLEC,SYNE1,SYNE2	
PF08016	Polycystin cation channel	3	34	0.0443	CACNA1H,PKD1,PKD1L1	
INTERPRO Protein Domains and Features						
IPR000742	EGF-like domain	18	225	8.64E-08	CELSR1,CUBN,FAT1,FBN3,FCGBP,HMCN1,HMCN2,HSPG2,LAMA5,LAMC3,LRP2,MEGF8,MUC17,MUC4,NRXN1,SNED1,SVEP1,TNC	86
IPR000152	EGF-type aspartate/asparagine hydroxylation site	11	100	8.86E-06	CELSR1,CUBN,FAT1,FBN3,HMCN1,HMCN2,LRP2,MEGF8,NRXN1,SNED1,SVEP1	
IPR013032	EGF-like, conserved site	14	193	1.05E-05	CELSR1,CUBN,FAT1,FBN3,HMCN1,HMCN2,HSPG2,LRP2,MEGF8,MUC12,MUC17,SNED1,SVEP1,TNC	
IPR001881	EGF-like calcium-binding domain	11	121	2.66E-05	CELSR1,CUBN,FAT1,FBN3,HMCN1,HMCN2,HSPG2,LRP2,MEGF8,SNED1,SVEP1	
IPR013783	Immunoglobulin-like fold	25	708	4.11E-05	CMYA5,CNTN2,COL6A3,FCAMR,HMCN1,HMCN2,HSPG2,HYDIN,LILRA4,LILRA6,LILRB3,LRRC4C,MADCAM1,MYOM2,OBSCN,PKD1,PKD1L1,PLXNA2,PLXNA3,PLXNA4,SECTM1,SNED1,SPEG,TNC,TTN	
IPR031904	Cadherin, C-terminal catenin-binding domain	7	37	4.11E-05	PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7	
IPR020894	Cadherin conserved site	10	108	4.73E-05	CELSR1,DCHS2,FAT1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7	
IPR002126	Cadherin-like	10	113	6.08E-05	CELSR1,DCHS2,FAT1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7	
IPR015919	Cadherin-like superfamily	10	115	6.27E-05	CELSR1,DCHS2,FAT1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7	
IPR018097	EGF-like calcium-binding, conserved site	9	96	0.00011	CUBN,FAT1,FBN3,HMCN1,HMCN2,LRP2,MEGF8,SNED1,SVEP1	
IPR026983	Dynein heavy chain	5	16	0.00012	DNAH1,DNAH11,DNAH14,DNAH6,DNAH8	
IPR027401	Myosin IQ motif-containing domain superfamily	5	16	0.00012	MYH1,MYH11,MYH2,MYH3,MYH7B	
IPR000048	IQ motif, EF-hand binding site	8	81	0.00017	LRRIQ1,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,OBSCN	
IPR001101	Plectin repeat	4	7	0.00017	EPPK1,MACF1,PLEC,PPL	
IPR002928	Myosin tail	5	18	0.00017	MYH1,MYH11,MYH2,MYH3,MYH7B	

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IPR004009	Myosin, N-terminal, SH3-like	5	19	0.00017	MYH1,MYH11,MYH2,MYH3,MYH7B	
IPR035915	Plakin repeat superfamily	4	7	0.00017	EPPK1,MACF1,PLEC,PPL	
IPR001791	Laminin G domain	7	58	0.00019	CELSR1,COL24A1,CSPG4,FAT1,HSPG2,LAMA5,NRXN1	
IPR008989	Myosin S1 fragment, N-terminal	5	20	0.00019	MYH1,MYH11,MYH2,MYH3,MYH7B	
IPR001609	Myosin head, motor domain	6	38	0.0002	MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A	
IPR013164	Cadherin, N-terminal	7	63	0.00027	PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7	
IPR003598	Immunoglobulin subtype 2	12	237	0.00043	CNTN2,HMCN1,HMCN2,HSPG2,LILRA4,LILRA6,LILRB3,LRRC4C,MYOM2,OBSCN,SPEG,TTN	
IPR013098	Immunoglobulin I-set	9	133	0.00057	CNTN2,HMCN1,HMCN2,HSPG2,LRRC4C,MYOM2,OBSCN,SPEG,TTN	
IPR011704	ATPase, dynein-related, AAA domain	4	13	0.00064	DNAH1,DNAH11,DNAH6,DNAH8	
IPR018159	Spectrin/alpha-actinin	5	29	0.00064	MACF1,PLEC,PPL,SYNE1,SYNE2	
IPR024317	Dynein heavy chain, AAA module D4	4	13	0.00064	DNAH1,DNAH11,DNAH6,DNAH8	
IPR035706	Dynein heavy chain, ATP-binding dynein motor region	4	13	0.00064	DNAH1,DNAH11,DNAH6,DNAH8	
IPR001846	von Willebrand factor, type D domain	4	14	0.0007	FCGBP,MUC4,MUC5B,VWF	
IPR004273	Dynein heavy chain region D6 P-loop domain	4	14	0.0007	DNAH1,DNAH11,DNAH6,DNAH8	
IPR013602	Dynein heavy chain, domain-2	4	14	0.0007	DNAH1,DNAH11,DNAH6,DNAH8	
IPR024743	Dynein heavy chain, coiled coil stalk	4	14	0.0007	DNAH1,DNAH11,DNAH6,DNAH8	
IPR035699	Dynein heavy chain, hydrolytic ATP-binding dynein motor region	4	14	0.0007	DNAH1,DNAH11,DNAH6,DNAH8	
IPR013320	Concanavalin A-like lectin/glucanase domain superfamily	11	220	0.00072	CELSR1,CMYA5,COL24A1,CSPG4,FAT1,GPR98,HSPG2,LAMA5,NRXN1,R YR2,SVEP1	
IPR002049	Laminin EGF domain	5	36	0.0012	CELSR1,HSPG2,LAMA5,LAMC3,MEGF8	
IPR000082	SEA domain	4	22	0.0026	HSPG2,MUC12,MUC16,MUC17	
IPR003599	Immunoglobulin subtype	14	401	0.0026	CNTN2,FCAMR,HMCN1,HMCN2,HSPG2,LILRA4,LILRA6,LILRB3,LRRC4C ,MADCAM1,MYOM2,OBSCN,SPEG,TTN	
IPR000034	Laminin IV	3	8	0.0028	HSPG2,LAMA5,LAMC3	
IPR001589	Actinin-type actin-binding domain, conserved site	4	23	0.0028	MACF1,PLEC,SYNE1,SYNE2	
IPR013548	Plexin, cytoplasmic RasGAP domain	3	9	0.0036	PLXNA2,PLXNA3,PLXNA4	
IPR031148	Plexin family	3	9	0.0036	PLXNA2,PLXNA3,PLXNA4	
IPR002165	Plexin repeat	4	26	0.0039	MEGF8,PLXNA2,PLXNA3,PLXNA4	
IPR036961	Kinesin motor domain superfamily	6	79	0.0041	MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A	
IPR002919	Trypsin Inhibitor-like, cysteine rich domain	3	10	0.0042	FCGBP,MUC5B,VWF	
IPR026823	Complement C1r-like EGF domain	4	27	0.0042	FBN3,HMCN1,HMCN2,LRP2	

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IPR014853	Uncharacterised domain, cysteine-rich	3	11	0.005	FCGBP,MUC5B,VWF	
IPR036084	Serine protease inhibitor-like superfamily	3	11	0.005	FCGBP,MUC5B,VWF	
IPR003961	Fibronectin type III	9	203	0.0059	CMYA5,CNTN2,COL6A3,MYOM2,OBSCN,SNED1,SPEG,TNC,TTN	
IPR036116	Fibronectin type III superfamily	9	204	0.006	CMYA5,CNTN2,COL6A3,MYOM2,OBSCN,SNED1,SPEG,TNC,TTN	
IPR009030	Growth factor receptor cysteine-rich domain superfamily	7	125	0.0065	CELSR1,CUBN,FBN3,HMCN1,HMCN2,LRP2,SVEP1	
IPR003303	Filaggrin	2	2	0.0075	FLG,FLG2	
IPR019528	Pericentrin/AKAP-450 centrosomal targeting domain	2	2	0.0075	AKAP9,PCNT	
IPR026980	Dynein heavy chain 6, axonemal	2	2	0.0075	DNAH14,DNAH6	
IPR028745	A-kinase anchor protein 9/Pericentrin	2	2	0.0075	AKAP9,PCNT	
IPR029926	Dystonin-like	2	2	0.0075	EPPK1,PLEC	
IPR030269	Plectin	2	2	0.0075	EPPK1,PLEC	
IPR000203	GPS motif	4	35	0.0079	CELSR1,GPR56,GPR98,PKD1	
IPR007110	Immunoglobulin-like domain	14	470	0.0079	CNTN2,FCAMR,HMCN1,HMCN2,HSPG2,LILRA4,LILRA6,LILRB3,LRRC4C,MADCAM1,MYOM2,OBSCN,SPEG,TTN	
IPR036465	von Willebrand factor A-like domain superfamily	6	97	0.0083	CACNA2D4,COL6A3,HMCN1,HMCN2,SVEP1,VWF	
IPR036179	Immunoglobulin-like domain superfamily	14	478	0.0087	CNTN2,FCAMR,HMCN1,HMCN2,HSPG2,LILRA4,LILRA6,LILRB3,LRRC4C,MADCAM1,MYOM2,OBSCN,SPEG,TTN	
IPR027417	P-loop containing nucleoside triphosphate hydrolase	20	849	0.0108	ABCA13,CFTR,DDX39B,DNAH1,DNAH11,DNAH6,DNAH8,HBS1L,LRRIQ1,MSH6,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,RABL6,RECQL4,RNF213,TJP3	
IPR034325	S-100	3	17	0.0111	FLG,FLG2,HRNR	
IPR016201	PSI domain	4	42	0.0133	MEGF8,PLXNA2,PLXNA3,PLXNA4	
IPR036364	SEA domain superfamily	3	19	0.0142	MUC12,MUC16,MUC17	
IPR006605	G2 nidogen/fibulin G2F	2	4	0.0145	HMCN1,HMCN2	
IPR009017	Green fluorescent protein	2	4	0.0145	HMCN1,HMCN2	
IPR010313	Glycine N-acyltransferase	2	4	0.0145	GLYATL1,GLYATL2	
IPR012315	KASH domain	2	4	0.0145	SYNE1,SYNE2	
IPR013652	Glycine N-acyltransferase, C-terminal	2	4	0.0145	GLYATL1,GLYATL2	
IPR015938	Glycine N-acyltransferase, N-terminal	2	4	0.0145	GLYATL1,GLYATL2	
IPR002017	Spectrin repeat	3	22	0.0184	MACF1,SYNE1,SYNE2	
IPR003886	NIDO domain	2	5	0.0184	MUC4,SNED1	
IPR014010	REJ domain	2	5	0.0184	PKD1,PKD1L1	

#Term ID	Term description	Observed gene count	Background gene count	False discovery rate	Genes	No. terms
IPR016332	Alpha-1B-glycoprotein/leukocyte immunoglobulin-like receptor	2	5	0.0184	LILRA4,LILRA6	
IPR002859	PKD/REJ-like domain	2	6	0.023	PKD1,PKD1L1	
IPR001751	S100/Calbindin-D9k, conserved site	3	25	0.0243	FLG,FLG2,HRNR	
IPR001452	SH3 domain	8	222	0.0257	MACF1,MYO15A,NEB,OBSCN,PLEC,PPL,TJP3,TNK2	
IPR008936	Rho GTPase activation protein	5	90	0.0261	ARHGAP11B,DAB2IP,PLXNA2,PLXNA3,PLXNA4	
IPR013106	Immunoglobulin V-set domain	7	177	0.0272	FCAMR,HMCN1,HMCN2,HSPG2,LRRC4C,OBSCN,TTN	
IPR003593	AAA+ ATPase domain	6	134	0.0287	ABCA13,CFTR,DNAH11,DNAH6,DNAH8,RNF213	
IPR003126	Zinc finger, UBR-type	2	8	0.0334	FBXO11,UBR4	
IPR013787	S100/CaBP-9k-type, calcium binding, subdomain	3	29	0.0334	FLG,FLG2,HRNR	
IPR002909	IPT domain	3	30	0.0355	PLXNA2,PLXNA3,PLXNA4	
IPR001627	Sema domain	3	31	0.0382	PLXNA2,PLXNA3,PLXNA4	
IPR011993	PH-like domain superfamily	11	408	0.0382	AKAP13,DAB2IP,DOCK9,MYO15A,OBSCN,OSBP2,RANBP2,RLTPR,SNAP47,TNS1,TRIOBP	
IPR013594	Dynein heavy chain, domain-1	2	9	0.0382	DNAH11,DNAH8	33
IPR036352	Sema domain superfamily	3	31	0.0382	PLXNA2,PLXNA3,PLXNA4	
SMART Protein Domains						
SM00001	EGF domain, unclassified subfamily	11	76	3.44E-07	FAT1,FCGBP,HSPG2,LAMA5,LAMC3,LRP2,MEGF8,MUC12,MUC17,MUC4,TNC	
SM00181	Epidermal growth factor-like domain.	14	208	9.86E-06	CELSR1,CUBN,FAT1,FBN3,HMCN1,HSPG2,LRP2,MEGF8,MUC17,MUC4,NRXN1,SNED1,SVEP1,TNC	
SM00112	Cadherin repeats.	10	115	4.92E-05	CELSR1,DCHS2,FAT1,PCDHA1,PCDHA2,PCDHA3,PCDHA4,PCDHA5,PCDHA6,PCDHA7	
SM00179	Calcium-binding EGF-like domain	9	98	8.53E-05	CUBN,FAT1,FBN3,HMCN1,HMCN2,LRP2,MEGF8,SNED1,SVEP1	
SM00015	Short calmodulin-binding motif containing conserved Ile and Gln residues.	8	83	0.00018	LRRIQ1,MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A,OBSCN	
SM00242	Myosin. Large ATPases.	6	38	0.00018	MYH1,MYH11,MYH2,MYH3,MYH7B,MYO15A	
SM00282	Laminin G domain	6	44	0.00032	CELSR1,CSPG4,FAT1,HSPG2,LAMA5,NRXN1	
SM00200	Domain found in sea urchin sperm protein, enterokinase, agrin	4	14	0.00064	HSPG2,MUC12,MUC16,MUC17	
SM00216	von Willebrand factor (vWF) type D domain	4	15	0.00071	FCGBP,MUC4,MUC5B,VWF	

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SM00150	Spectrin repeats	5	33	0.00075	MACF1,PLEC,PPL,SYNE1,SYNE2	
SM00180	Laminin-type epidermal growth factor-like domai	5	33	0.00075	CELSR1,HSPG2,LAMA5,LAMC3,MEGF8	
SM00250	Plectin repeat	3	5	0.0008	EPPK1,PLEC,PPL	
SM00060	Fibronectin type 3 domain	9	189	0.0034	CMYA5,CNTN2,COL6A3,MYOM2,OBSCN,SNED1,SPEG,TNC,TTN	
SM00409	Immunoglobulin	12	328	0.0034	CNTN2,FCAMR,HMCN1,HMCN2,LILRA4,LILRA6,LILRB3,MADCAM1,MYOM2,OBSCN,SPEG,TTN	
SM00068	Glycoprotein hormone beta chain homologues.	3	11	0.0039	CGB,CGB2,ENSG00000267335	
SM00327	von Willebrand factor (vWF) type A domain	6	85	0.0039	CACNA2D4,COL6A3,HMCN1,HMCN2,SVEP1,VWF	
SM00832	This domain contains 8 conserved cysteine residues, but this family only contains 7 of them to overlaps with other domains. It is found in disease-related proteins including von Willebrand factor, Alpha tectorin, Zonadhesin and Mucin.	3	11	0.0039	FCGBP,MUC5B,VWF	
SM00410	Immunoglobulin like	6	88	0.0043	LILRA4,LILRA6,LILRB3,MYOM2,OBSCN,TTN	
SM00303	G-protein-coupled receptor proteolytic site domain	4	33	0.005	CELSR1,GPR56,GPR98,PKD1	
SM00408	Immunoglobulin C-2 Type	9	211	0.005	CNTN2,HMCN1,HMCN2,HSPG2,LRR4C,MYOM2,OBSCN,SPEG,TTN	
SM01249	Nuclear envelope localisation domain	2	4	0.0115	SYNE1,SYNE2	
SM00423	domain found in Plexins, Semaphorins and Integrins	4	45	0.0124	MEGF8,PLXNA2,PLXNA3,PLXNA4	
SM00539	Extracellular domain of unknown function in nidogen (entactin) and hypothetical proteins.	2	5	0.0146	MUC4,SNED1	
SM00221	Protein kinase	2	6	0.0186	OBSCN,SPEG	
SM00281	Laminin B domain	2	7	0.0228	HSPG2,LAMA5	
SM00429	ig-like, plexins, transcription factors	3	27	0.0228	PLXNA2,PLXNA3,PLXNA4	
SM00011	VWC_def	2	8	0.0262	FCGBP,MUC5B	
SM00630	semaphorin domain	3	30	0.0272	PLXNA2,PLXNA3,PLXNA4	
SM00089	Repeats in polycystic kidney disease 1 (PKD1) and other proteins	2	9	0.0286	PKD1,PKD1L1	
SM00382	ATPases associated with a variety of cellular activities	6	144	0.0286	ABCA13,CFTR,DNAH11,DNAH6,DNAH8,RNF213	
SM00033	Calponin homology domain	4	64	0.0289	MACF1,PLEC,SYNE1,SYNE2	

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SM00214	von Willebrand factor (vWF) type C domain	3	35	0.0354	FCGBP,MUC5B,VWF	
SM00274	Follistatin-N-terminal domain-like	2	11	0.0363	FCGBP,SNED1	