

#253290 MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE; LMPS

OMIM Gene ID	HGNC	UniProtAC
100690	CHRNA1	P02708
100720	CHRND	Q07001
100730	CHRNA1	P07510

Table 1: OMIM - UniProtAC mapping

Legend

- N1: #input proteins associated to the significant GO term
- N2: #proteins associated to the significant GO term
- P-value: Bonferroni-corrected p-value of Fisher's exact test
- *red*: go terms not related to the input proteins
- *blue*: go terms related to the input proteins (enriched uniquely by network-based method)
- *green*: go terms ancestors of terms enriched with the standard method (enriched uniquely by network-based method)

1 Standard enrichment

GO Term	N1	N2	P-value	Description
GO:0048630	2	3	6.82333e-07	skeletal muscle tissue growth
GO:0006936	3	261	1.76536e-05	muscle contraction
GO:0003012	3	320	3.26052e-05	muscle system process
GO:0050879	2	20	4.32015e-05	multicellular organismal movement
GO:0050881	2	20	4.32015e-05	musculoskeletal movement
GO:0042391	3	353	4.3807e-05	regulation of membrane potential
GO:0007268	3	530	0.000148691	synaptic transmission
GO:0007267	3	859	0.000634424	cell-cell signaling
GO:0023052	3	913	0.000761908	signaling
GO:0044700	3	913	0.000761908	single organism signaling
GO:0098655	3	1072	0.00123392	cation transmembrane transport
GO:0007154	3	1103	0.0013442	cell communication
GO:0050905	2	125	0.0017589	neuromuscular process
GO:0034220	3	1538	0.00364704	ion transmembrane transport
GO:0006812	3	1582	0.0039693	cation transport
GO:0003008	3	1588	0.00401467	system process
GO:0048589	2	296	0.00987887	developmental growth
GO:0055085	3	2352	0.013052	transmembrane transport
GO:0006811	3	2423	0.0142706	ion transport
GO:0070050	1	5	0.0214587	neuron cellular homeostasis
GO:0040007	2	569	0.0363867	growth

Table 2: Overrepresented GO terms with the standard enrichment

2 Network-based enrichment

No novel enriched terms