

ClinGen Limb Girdle Muscular Dystrophy Gene Curation Expert Panel

Start Date	Your Name	Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel?	Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel?		Do you have any other existing or planned independent curation efforts that will potentially overlap with the scope of your ClinGen work? If so, please describe:	Please disclose any other relevant conflicts of interest (e.g. patents, intellectual property ownership, or paid consultancies related to any variants or genes associated with the work of your Expert Panel):
	Open-Ended Response	Response	Response	If yes, please list the genes:	Open-Ended Response	Open-Ended Response
2019-12-05 08:38:11	Elizabeth Kearns	No	No		No	
2019-11-26 14:05:16	SAMYA CHAKRAVORTY	No	No		No	None
2019-11-18 10:59:00	Shannon McNulty	No	No		No	Nothing to disclose
2019-11-05 13:25:08	Shruthi Mohan	No	No		No	None to disclose
2019-11-04 13:57:30	May Flowers	No	No		N/A	N/A
2019-10-15 09:41:00	Madhuri Hegde	Yes	Yes	LGMD gene	No	None
2019-10-14 04:03:23	Volker Straub	No	No			
2019-10-10 12:42:52	Anthony Amato	No	Yes	Led AAN practice guideline on LGMD/distal myopathies in regard to clinical phenotype, genotype and diagnosis Co-author on paper reporting mutations in dysferlin as cause of Miyoshi	no	none
2019-10-10 11:54:19	Hanns Lochmuller	No	Yes	ANOS, GMPPB, DOK7, GFPT1, INPP5K, ALG2, ALG14	I was the coordinator of RD-Connect, a European project with a Genome Phenome Analysis Platform. I am the co-editor in chief for the Journal of Neuromuscular Diseases	none

2019-10-08 14:08:17	Conrad Wehl	No	Yes	Myopathy gene discovery	no	none
2019-10-08 14:07:20	Monkol Lek	No	No		No	None
2019-10-07 07:31:41	Vincenzo Nigro	No	Yes	delta sarcoglycan, calpain 3, dysferlin, titin, TNPO3		no conflict
2019-10-04 19:14:45	Laura Rufibach	No	Yes	DYSF	yes, I have been working with researchers at Emory University for a number of years to do analysis to help with the clarification of pathogenicity of DYSF variants. A paper of the data for this project is currently under review and further analysis is ongoing. The Jain Foundation is also funding another researcher at MIT to perform assays to evaluate the pathogenicity of missense mutations. This work is not yet published.	none
2019-10-04 14:54:32	Lucas Bronicki	Yes	No		none	none
2019-10-04 14:39:59	Courtney Thaxton	No	Yes	I am a current ClinGen biocurator and have curated and published curation for myopathies previously, as part of ClinGen expert panels.	no	no
2019-10-03 13:55:59	Chandler Douglas	No	No		No	No
2019-09-25 08:50:11	Katherine Mathews	No	No		none	

2019-09-24 18:40:36	Peter B. Kang	No	Yes	PYROXD1, have published several articles covering a range of LGMD genes.	I hold a CDC grant for MD Starnet, I'm not sure if this will overlap or not.	I have served on an advisory board for Sarepta Therapeutics, which is developing gene therapies for some LGMDs. My other relationships are not directly relevant to LGMD.
2019-09-24 17:00:45	Matthew P. Wicklund, MD	No	No		No	Advisory board for Sarepta Therapeutics
2019-09-24 16:51:56	Marco Savarese	No	Yes	ANOS, TNPO3, TTN, GAA	A submitted manuscript on the interpretation of titin variants	None
2019-09-24 16:53:13	Melissa Spencer	No	Yes	I don't know if they are substantial but, I have published on a few CAPN3 and TRIM32 pathogenic mutations	no	none
2019-09-24 16:17:52	Sander Pajusalu	Yes	No		No	Nothing to disclose
2019-09-10 14:06:06	Giorgio Tasca	No	Yes	DNAJB6, ISPD, POPDC1	No	None
2019-09-10 14:05:24	Vijay Ganesh	No	No		No.	None.