ClinGen Mitochondrial Diseases Gene Curation Expert Panel						
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
2019-01-10 12:25:43	John Shoffner, MD	No	Yes	MTTK (PMID 29449072)	No	None
2019-01-09 15:44:50	Isabelle Thiffault	No	Yes	I have published on several Mitochondrial genes: AIMP1, AARS2, MARS2, IARS2, YARS2, TRIT1, TRNT1, EPRS, CLPB, PNPLA8, NDUFS1, ARSACS	No	none

2018-05-22 3:06:13	Annette Feigenbaum	No	No	I have co-published case reports but should not influence this activity: re gene curation: SLC19A3; DARS2; IDH2;PDP1 re: variant curation: NDUFV1, PDH E2; eIF2B; mtDNA dGUOK; PDHE3, PC, etc.	no	none
2018-05-17 11:02:08	Elizabeth McCormick	No	No		No	N/A
2018-05-17 8:21:47	Marni J Falk, MD	No	No	I've discovered several novel mitochondrial disease genes (including FBXL4 that we plan to curate in later years), but none related to the genes currently under curation for Leigh syndrome.	I am a member of the ACMG mtDNA curation review committee, whose activities we are attempting to align with the ClinGen/MSeqDR mtDNA expert panel	

2018-05-17 6:59:51	Douglas C. Wallace	No	Yes	I discovered the first maternally inherited disease and have spent my entire career trying to determine the role of mtDNA mutations in disease.	No	
2018-05-16 9:35:45	Richard Rodenburg	Yes	Yes	I have published around 150 publications on the topic of mitochondrial genetics and disease, including SLC19A3 and many more.	No	There are no conflicts of interest to disclose
2018-05-13 9:43:58	Yasushi Okazaki	No	No			
2018-05-11 21:48:18	Amy Goldstein	No	No		No	
2018-05-11 20:06:27	Christine Stanley	No	No			
2018-05-11 3:03:33	Belén Pérez Dueñas	No	Yes	SLC19A3 PANK2 PLA2G6	NO	

2018-05-10 17:51:49	Russell P. Saneto, DO, PhD	No	No	Yes, for POLG (but maybe not as substantial as my ego would like to think). But none for Leigh syndrome.	No	
2018-05-10 17:54:12	Xiaowu Gai	Yes	Yes	FBXL4	No	
2018-05-10 17:30:16	procaccio vincent	Yes	Yes	Mitochondrial genome Nuclear-encoded mitochondrial genes	curation of mitomap mitochondrial database	none
2018-05-10 16:44:19	Matthew Dulik	No	No		No	Nothing to disclose.
2018-04-24 10:55:15	Marie Lott	No	Yes	MT-ND4: LHON (m.11778G>A); MT-TK:MERRF (m.8344A>G); MT-TL1:PEM (m.3271T>del); multi-locus: KSS/CPEO/aging (mtDNA 5kb deletion)	I am the curator of Mitomap	no

2018-04-23 5:02:21	Marcella Attimonelli	No	No		Yes I am curator of HmtDB database and hence I support with data available through HmtDB to the in silico assessment of pathogenicity of any mitochondrial DNA variant	no conflicts of interest
2018-04-19 15:51:08	Kierstin Keller	No	No		No	N/A
2018-04-18 12:12:22	Renkui Bai	Yes	No		Regular working on reporting mtDNA variants identified in a clinical molecular diagnostic laboratory	NA
2018-04-16 12:06:47	Austin Larson	No	No		No	None.
2018-04-11 17:47:06	Antonio Velazquez-Arellano	No	No			
2018-04-11 16:08:58	Marc Yudkoff	No	No		No	
2018-04-11 11:58:08	Mary Kay Koenig	No	No		No	
2018-04-09 21:47:44	Amel Karaa	No	No		mitochondrial DNA expert panel for ClinGen. This is for general variant interpretation.	
2018-04-09 9:20:17	George A. Diaz	No	No		No	

2018-04-09 7:25:54	Peter W. Stacpoole	No	Yes	Collated in review: Molecular Genetics and Metabolism 106 (2012) 385–394	Recruitment of patients to a clinical trial that may identify new pathological mutations.	

2018-04-09 2:48:12	David Thorburn	No	Yes	I have published 9 peer-reviewed research papers related to POLG mutations.	Only in the context that I have contributed to diagnosis of about 50 POLG patients (mostly from single gene sequencing in a research context) and have overseen the application of ACMGG criteria to such results. I also take part in multidisciplinary team meetings reporting exome sequencing results that will occasionally include POLG mutations.	No relevant conflicts of interest. I am associated with a not-for-profit genetic testing lab (Victorian Clinical Genetics Services) owned by my employer (Murdoch Children's Research Institute). My association includes being an Honorary VCGS Fellow (which allows my lab to offer diagnostic respiratory chain enzyme testing) and being on the VCGS Executive committee to provide research input. VCGS offers single-gene sequencing for POLG and whole exome sequencing. I receive no salary or other income from VCGS and think that the curation work will benefit VCGS in the same way that it will benefit any genetic pathology provider, whether for-profit or not-for-profit. Hence I don't regard it as a relevant CoI.

2018-04-07 14:07:39	William Copeland	No	Yes	We study the consequences of POLG mutations to help understand the pathogenesis from POLG mutations and study POLG enzymology and biology, in general.	No	
2018-04-06 23:52:43	Ann Saada (Reisch)	No	No		No	
2018-04-06 21:33:57	John Christodoulou	Yes	Yes	multiple (>20)genes associated with mitochondrial disorders or overlapping phenotypes	yes - Rett/Angelman	I am the approved pathology practitioner for the Victorian Clinical Genetics Service, Melbourne, Australia, which provides WES and mtDNA sequencing.
2018-04-06 21:28:44	Andrea Gropman	No	No		no	none
2018-04-06 19:30:42	Sumit Parikh	No	No		No	
2018-04-06 16:33:10	Richard Haas MD	No	No		No	
2018-04-06 13:28:51	Colleen Muraresku	No	No		No	N/A
2018-04-06 13:26:51	LISHUANG SHEN	No	No		No	No

2018-04-06 13:25:16	Zarazuela Zolkipli Cunningham	No	No			
2018-04-06 13:23:26	Greg Enns	No	Yes	mitochondrial tRNA Ile (m.4296G>A); ATP5F1D; HTRA2; HSD17B10	I'm on the ClinGen Metabolism and Fatty Acid Oxidation working groups	NA
2018-04-06 13:21:37	Rebecca Ganetzky	No	No		Natural history and curation of MT-ATP6 variants for my research program (NIH funded)	
2018-04-06 13:22:17	Shamima Rahman	No	Yes	I have published extensively on the genetics of Leigh syndrome, and was the first to report mutations in FOXRED1 and COQ9, both of which have subsequently been linked to Leigh syndrome	We have made a virtual diagnostic resource of disease genes causing Leigh syndrome, that is available as a free resource called the Leigh Map: vmh.uni.lu/#leighmap Currently this resource does not contain variant level information.	I do not own any patents or IP in this or any other area. I have had paid one-off consultancies asking about clinical trials for mitochondrial disease, but not about any disease genes.