

ClinGen Familial Hypercholesterolemia Variant 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
2019-04-15 2:51:35	Lukas Tichy	No	Yes	LDLR, APOB, PCSK9, LDLRAP1, ABCG5/8, ABCA1, APOE	No	Nothing to disclose.
2019-04-14 15:50:27	Tomas Freiburger	No	Yes	LDLR, APOB, PCSK9	No.	Nothing to disclose.
2019-04-14 16:02:39	Pedro Mata	No	No			
2019-04-12 6:36:44	ronen durst	No	Yes	LDLr	no	none
2019-04-11 19:21:03	Mariko Harada-Shiba	No	Yes	familial hypercholesterolemia	no	no
2019-04-11 17:27:26	Raul Santos	Yes	Yes	Familial hypercholesterolemia and cardiovascular disease	No	I have consulted for pharmaceutical companies regarding medical treatment for familial hypercholesterolemia: Amgen, Sanofi/Regeneron
2019-02-25 9:18:48	Mafalda Bourbon	No	No		No	
2019-01-30 6:58:52	Joana Rita Chora	No	No			
2019-01-30 4:49:33	RABES Jean-Pierre	Yes	Yes	APOB (R3531C variant) PCSK9 (GoF mutations)	No	Paid consultancies related to molecular strategies for diagnosis of Familial Hypercholesterolemia in France
2019-01-30 3:10:29	Joep C. Defesche	Yes	Yes	LDLR, APOB, PCSK9	continuously ongoing curation for routine DNA analysis for patient care	nothing to disclose
2019-01-29 19:29:39	Eric. J.G. Sijbrands	No	Yes	genotype-phenotype studies of variants in LDLR, APOB, and multiple other genes; identification of multiple other genes involved in metabolic traits and cardiovascular disease.		none
2019-01-29 16:10:02	Benjamin Helm	No	No			I am a FH Foundation Patient Advocate, and I have also done paid consulting work for the FH Foundation (from 2017-present).
2019-01-29 15:33:27	Marina Cuchel	No	No		No	No
2019-01-29 14:56:03	Amit Khera	No	Yes	LDLR, APOB, PCSK9	No	Paid consultant for Color Genomics
2018-09-04 10:17:49	Hannah Wand	No	No		No	None. I volunteer with the FH Foundation, but I don't believe this is a conflict of interest.
2018-08-29 15:48:37	Alain Carrié	No	No			Collaboration with Agilent/Multiplicom to develop kit for FH diagnosis
2018-08-29 11:33:03	Sarah Leigh	No	Yes	LDLR	I work as a scientific curator at Genomics England and may at some point be involved in curating variants for pathogenicity	none
2018-08-29 11:32:40	Michael Iacocca	No	No		No	I have no conflicts of interest to disclose
2018-03-15 15:05:43	Margaret Chen	Yes	No		GeneDx presently submits variant classifications/interpretations to ClinVar	N/A
2018-03-07 2:40:57	Amanda J Hooper	Yes	No		Previously curator for APOB LOVD https://grenada.lumc.nl/LOVD2/mendelian_genes/home.php?select_db=APOB	

2018-03-06 13:40:14	Rob Hegele	No	Yes	LDLR, APOB, PCSK9, ARH, APOE, LIPA, ABCG5/G8	No.	None at present.
2018-03-06 11:41:54	Marianne Stef	Yes	Yes	LDLR, PCKS9	No	
2018-03-06 11:29:16	Steve Humphries	Yes	Yes	LDLR, APOB, PCSK9, APOE, LDLRAP1,	I lead the FH GeCIP for the UK 100,000 Genomes project and have been advising on Genome England on approaches to curate FH-causing mutations for incidental findings. We will also analyse the no-mutation FH families included in the WGS to try to identify novel FH-causing genes	I am a consultant for Color Genomics, a US-based company that offers DNA testing for FH. I am medical Director and minor share holder in StoreGene, a UK company that offers Genetic testing for CHD including FH
2018-03-06 10:50:34	josh knowles	No	Yes	ldlr, pcsk9, apob	no	I am the volunteer (unpaid) Chief Medical Advisor for the FH Foubdation, a patient led research and advocacy group. The FHF receives support from pharma, lab and genetic testing companies.