

ClinGen Hereditary Hemorrhagic Telangiectasia Variant 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?	If yes, please list the genes:	Do you have any other existing or planned independent efforts that will potentially overlap with the scope of your ClinGen work? If so, please describe and also send an email describing the project(s) to the co-chairs and coordinator:	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		Open-Ended Response	Open-Ended Response
2020-09-23 18:54:29	Shoji Ichikawa	Yes	No		No	None
2019-05-17 12:43:31	Luca Jovine	No	Yes	ENG	I am co-author on a manuscript, soon to be submitted, that describes a mutational and phenotypic characterisation of HHT mutations.	None.
2019-01-10 15:40:51	Raj S. Kasthuri	No	No		No	None
2019-01-10 12:04:31	GIRAUD Sophie	Yes	Yes	ENG, ACVRL1	No	
2019-01-10 05:30:13	JK Ploos van Amstel	Yes	Yes	See PubMed, many		
2019-01-09 03:21:50	DUPUIS-GIROD	No	No		NO	NO
2019-01-08 13:24:20	Reed E. Pyeritz	No	Yes	ENG ACVRL1 SMAD4	no	None
2019-01-08 07:43:27	Carla Olivieri	No	Yes	ACVRL1 ENG SMAD4	I wrote the ACVRL1 card for the Atlas of Genetics and Cytogenetics in Oncology and Haematology ( <a href="http://AtlasGeneticsOncology.org">http://AtlasGeneticsOncology.org</a> )	No other COI
2019-01-08 05:22:10	Helen Arthur	No	Yes	ENG, ACVRL1	No	None
2019-01-07 21:47:12	Arupa Ganguly	Yes	Yes		We maintain our internal database of the mutations found in our lab.	None
2019-01-07 20:07:04	Hilary Racher	Yes	Yes	ACVRL1 and ENG	No	None
2019-01-07 16:31:56	Claire Shovlin	No	Yes	ENG, ACVRL1, SMAD4	Yes- Existing: HHT gene initiatives in the UK, either families seen in my service, or more broadly UK HHT families through the 100,000 Genomes Project- I chair the HHT and PAVM subdomains of the Genomics England Clinical Interpretation Partnership (GeCIP)	None
2019-01-07 16:12:29	Pinar Bayrak-Toydemir	Yes	Yes	ENG, ACVRL1, SMAD4		
2019-01-07 15:36:22	Murray Brilliant	No	No		NO	none
2019-01-07 14:08:38	Jamie McDonald	Yes	Yes	ENG, ACVRL1, SMAD4	No	None
2019-01-07 11:12:41	Jaime Jessen	Yes	Yes	ENG, ACVRL1, SMAD4	No	None
2019-01-02 07:03:55	Carmelo Bernabeu	No	Yes	Endoglin (ENG) ALK1 (ACVRL1)	No	None
2018-12-27 13:34:57	Desi DeMille	Yes	No		No	
2018-12-21 09:24:37	reed pyeritz	No	No		no	
2018-12-21 05:30:16	Pernille Topping	Yes	Yes	ENG, ACVRL1, SMAD4		