

ClinGen Rett and Angelman-like Disorders 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?		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
2019-04-30 10:16:46	Kelly Toner	No	No		No	N/A
2018-04-18 9:22:54	Mike Friez	Yes	Yes	MECP2	No	Nothing to disclose
2018-04-16 15:20:15	Bienvenu	Yes	Yes	Mecp2, CDKL5, FOXG1, MEF2C	No	no conflict of interest
2018-04-16 14:27:37	Jeffrey Neul	No	Yes	MECP2, genotype phenotype relationships	No	

2018-04-16 13:43:49	Katelyn Beattie	Yes	No		I am an genetic counselor currently working at GeneDx, Inc., a wholly-owned subsidiary of OPKO Health, Inc. We complete genetic testing that includes genes associated with Rett/Angelman syndrome. As such, I am involved in variant classification for these genes.	I am an employee of GeneDx, Inc., a wholly-owned subsidiary of OPKO Health, Inc
2018-04-11 20:28:03	Huda zoghbi	No	Yes	discovered Rett gene	no	have patent on Rett diagnostic test. it will expire end of 2019
2018-03-28 9:12:10	Carrie-Anne Barry	No	No		N/A	N/A
2018-03-28 9:01:56	Sarah Hemphill	No	No		No	
2018-03-27 18:32:58	rahul raj krishna raj	No	No		RettBASE curation Curating MECP2,CDKL5,FOXG1	N/A
2018-03-27 15:11:40	Lora Bean	Yes	No		No	
2018-03-26 11:35:59	Amy Knight Johnson	Yes	No			
2018-03-23 11:54:27	Soma Das	Yes	Yes	CDKL5, FOXG1	No	
2018-03-23 5:06:11	Kathleen Murphy	No	No			

2018-03-22 5:08:53	Simon Ramsden	Yes	No	I am the lead author on the current AS/PWS best practice guidelines (PMID: 20459762) developed for European labs.		
2018-03-21 19:28:22	Ping Fang	Yes	Yes	MECP2, UBE3A	No	No
2018-03-21 10:46:41	Izabela Karbassi	Yes	No		Not currently	Publication on a Variant Classification scheme and an associated pending patent. Also, a member of the ClinGen Sequence Variant Interpretation group.
2018-03-20 18:28:32	John Christodoulou	Yes	Yes	MECP2 CDKL5 TCF4 FOXP1	I am part of the MSeqDR group involved in the curation of several mitochondrial disease genes	none that I can think of

						Consultant with AveXis, Anavex, Parexel Participate in clinic trials in Rett syndrome with Neuron Pharmaceuticals and Newron Pharmaceuticals
2018-03-20 15:53:31	Alan Percy	No	Yes	MECP2, CDKL5, FOXP1	No	
2018-03-20 15:52:02	Erin Riggs	No	No		N/A	N/A
2018-03-20 13:18:47	Dianalee McKnight	Yes	No		No	NA
2018-03-20 12:38:43	Hanyin Cheng	Yes	No		No	No
2018-03-20 12:20:49	Lindsey Mighion	Yes	No		No	
2018-03-20 12:26:07	Linyan Meng	Yes	No		Yes, curation service for Baylor Genetics	U.S. Patent 9,617,539, "Modulation of UBE3A-ATS expression"