

ClinGen FBN1 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
2019-04-12 11:06:51	Hal Dietz	No	Yes	FBN1 - Marfan syndrome FBN1 Stiff Skin Syndrome FBN1 - MASS phenotype TGFBR1, TGFBR2, SMAD3, TGFB2, TGFB3, SMAD2 - all in Loeys-Dietz syndrome ROBO4 - bicuspid valve with aneurysm	No	We have both patents and IP related to methods of treating vascular connective tissue disorders.
2018-11-27 22:07:19	Takayuki Morisaki	No	No		no	nothing
2019-01-30 8:49:03	Dianna Milewicz	No	No		N/A	N/A
2019-01-30 5:00:31	Laura Muiño Mosquera	Yes	Yes	FBN1, TGFBR1, TGFBR2, SMAD3	No	Paper on FBN1 variant classification: Tailoring the American College of Medical Genetics and Genomics and the Association for Molecular Pathology Guidelines for the Interpretation of Sequenced Variants in the FBN1 Gene for Marfan Syndrome: Proposal for a Disease- and Gene-Specific Guideline. Circ Genom Precis Med. 2018 Jun;11(6):e002039. doi: 10.1161/CIRCGEN.117.002039.
2019-01-30 4:18:06	Katie Francis	No	No		No	
2019-01-29 16:11:34	Maral Ouzounian	No	No		No	
2019-01-29 15:22:18	Julie De Backer	No	Yes	FBN1, TGFBR1/2, TGFB2, ACTA2, SMAD3	No	
2019-01-29 14:36:54	Linnea Baudhuin	Yes	Yes	FBN1	yes; FBN1 paper under revision. Already cleared with Julie and Birgit as not conflicting	
2019-01-29 13:56:30	Loeys	Yes	Yes	FBN1	NO	
2019-01-23 15:00:56	Olga Jarinova	Yes	No		N/A	N/A
2019-01-15 7:51:08	Bert Callewaert	Yes	Yes	FBN1	no, I have not.	
2019-01-10 16:14:45	Dongchuan Guo	No	Yes	ACTA2, LOX, MYLK, PRKG1, SMAD3, TGFB2, and TGFBR2	Curation of rare variants in our own NIH founded research project and research project in the Department of Internal Medicine, University of Michigan.	No
2019-01-08 9:12:05	Katrina Kotzer	Yes	No		no	none
2019-01-07 10:11:57	Michelle Kluge	Yes	No		No	
2018-12-04 16:41:27	Lucas Bronicki	Yes	No	Our laboratory has been testing for FBN1-related conditions for the past three years, generating significant experience. We are currently working on publishing some of our experiences with this work.	Over the past ~1/2 year we have been developing an internal protocol to standardize the use the ACMG/AMP PP4 criteria for Marfan testing. We aim at publishing our findings.	
2018-12-12 16:58:41	Julie Richer	No	No		No	Nil
2018-12-12 16:01:50	Leema Robert	No	No		NA	NA

2018-12-07 2:58:53	Hiroko Morisaki	No	Yes	FBN1, TGFBR1, TGFBR2, ACTA2, COL3A1,		
2018-11-28 3:43:02	Lut Van Laer	Yes	Yes	TGFB2, TGFB3, SKI, SMAD2, SMAD3, BGN, FBN1, SLC2A10, SMAD6	no	no conflicts of interest
2018-11-28 3:32:59	Marjolijn Renard	Yes	Yes	FBN1 (PMID: 29875124)	No	None to declare
2018-11-27 13:05:04	Olga Jarinova	No	No			
2018-11-27 11:10:10	Mark Lindsay	No	Yes	TGFB2	NO	I have no COI that would relate to any variant or gene associated with the work of this expert panel