

ClinGen FBN1 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 |
| 2018-11-27 19:07:19 | Takayuki Morisaki | No | No | | no | nothing |
| 2019-01-30 5:49:03 | Dianna Milewicz | No | No | | N/A | N/A |
| 2019-01-30 2:00:31 | 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 1:18:06 | Katie Francis | No | No | | No | |
| 2019-01-29 13:11:34 | Maral Ouzounian | No | No | | No | |
| 2019-01-29 12:22:18 | Julie De Backer | No | Yes | FBN1, TGFBR1/2, TGFB2, ACTA2, SMAD3 | No | |
| 2019-01-29 11:36:54 | Linnea Baudhuin | Yes | Yes | FBN1 | yes; FBN1 paper under revision. Already cleared with Julie and Birgit as not conflicting | |
| 2019-01-29 10:56:30 | Loeys | Yes | Yes | FBN1 | NO | |
| 2019-01-23 12:00:56 | Olga Jarinova | Yes | No | | N/A | N/A |
| 2019-01-15 4:51:08 | Bert Callewaert | Yes | Yes | FBN1 | no, I have not. | |
| 2019-01-10 13:14:45 | 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 6:12:05 | Katrina Kotzer | Yes | No | | no | none |
| 2019-01-07 7:11:57 | Michelle Kluge | Yes | No | | No | |
| 2018-12-04 13:41:27 | 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 13:58:41 | Julie Richer | No | No | | No | Nil |
| 2018-12-12 13:01:50 | Leema Robert | No | No | | NA | NA |
| 2018-12-06 23:58:53 | Hiroko Morisaki | No | Yes | FBN1, TGFBR1, TGFBR2, ACTA2, COL3A1, | | |
| 2018-11-28 0:43:02 | Lut Van Laer | Yes | Yes | TGFB2, TGFB3, SKI, SMAD2, SMAD3, BGN, FBN1, SLC2A10, SMAD6 | no | no conflicts of interest |
| 2018-11-28 0:32:59 | Marjolijn Renard | Yes | Yes | FBN1 (PMID: 29875124) | No | None to declare |
| 2018-11-27 10:05:04 | Olga Jarinova | No | No | | | |

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| 2018-11-27 8:10:10 | 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 |
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