

#1

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Wednesday, February 06, 2019 11:59:36 AM
Last Modified: Wednesday, February 06, 2019 12:03:04 PM
Time Spent: 00:03:28
IP Address: 109.190.253.11

Page 2

Q1 Your Name

pierre Morange

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **No**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel?

Yes,

If yes, please list the genes::

SERPINC1, PROS1,PROC,PROCR, F8, Willebrand, F11, F5,F2, SLC44A2, TSPAN15,HIVEP1, PROCR, ABO, FGG, KNG1

Q4 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:

NA

Q5 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):

Respondent skipped this question

#2

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Wednesday, February 06, 2019 12:07:14 PM
Last Modified: Wednesday, February 06, 2019 12:09:16 PM
Time Spent: 00:02:02
IP Address: 152.23.177.214

Page 2

Q1 Your Name

Shruthi Mohan

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **No**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel? **No**

Q4 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:

No

Q5 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):

None to disclose

#3

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Wednesday, February 06, 2019 12:29:14 PM
Last Modified: Wednesday, February 06, 2019 12:36:17 PM
Time Spent: 00:07:03
IP Address: 205.239.98.30

Page 2

Q1 Your Name

Kate Downes

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **Yes**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel?

Yes,

If yes, please list the genes::

Multiple genes-disease-mode of inheritance identified by NIHR BRIDGE-BPD project. DIAPH1, TPM4, GNE, GF11B, GP1BB, RASGRP2, KDSR.

Q4 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:

ISTH Scientific and Standardization Committee Genomics in Thrombosis and Hemostasis.
Curator of PanelApp (Genomics England).

Q5 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):

Respondent skipped this question

#4

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Thursday, February 07, 2019 4:06:39 AM
Last Modified: Thursday, February 07, 2019 4:07:57 AM
Time Spent: 00:01:18
IP Address: 213.205.242.80

Page 2

Q1 Your Name

Neil Morgan

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **No**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel?

Yes,

If yes, please list the genes::

Slfn14 and many other genes related to inherited platelet disorders

Q4 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:

No

Q5 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):

None

#5

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Thursday, February 07, 2019 4:46:47 AM
Last Modified: Thursday, February 07, 2019 4:58:53 AM
Time Spent: 00:12:06
IP Address: 131.111.92.238

Page 2

Q1 Your Name

Karyn Megy

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **No**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel? **No**

Q4 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:

ThromboGenomics gene curation efforts: generation of a curated list of genes involved in Thrombosis & Hemostasis. Publication to come.

Q5 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): **Respondent skipped this question**

#6

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Thursday, February 07, 2019 6:00:09 AM
Last Modified: Thursday, February 07, 2019 6:03:30 AM
Time Spent: 00:03:21
IP Address: 159.149.67.10

Page 2

Q1 Your Name

Marco Cattaneo

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **No**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel? **Yes,**
If yes, please list the genes::
P2Y12

Q4 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:

Collaboration with Thrombogenomics

Q5 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):

none

#7

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Friday, February 08, 2019 10:52:37 AM
Last Modified: Friday, February 08, 2019 10:52:57 AM
Time Spent: 00:00:20
IP Address: 152.23.173.125

Page 2

Q1 Your Name

Justyne Ross

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **No**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel? **No**

Q4 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: **Respondent skipped this question**

Q5 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): **Respondent skipped this question**

#8

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Friday, February 08, 2019 11:06:43 AM
Last Modified: Friday, February 08, 2019 11:10:10 AM
Time Spent: 00:03:27
IP Address: 132.187.191.4

Page 2

Q1 Your Name

Harald Schulze

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **No**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel? **Yes,**
If yes, please list the genes::
THPOR, RBM8A, FERMT3, WASP, TRPM7, GFI1B, HPS6

Q4 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:

No

Q5 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):

n/a

#9

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Monday, February 11, 2019 5:35:20 AM
Last Modified: Monday, February 11, 2019 5:38:42 AM
Time Spent: 00:03:22
IP Address: 134.58.253.56

Page 2

Q1 Your Name

Freson Kathleen

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **Yes**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel?

Yes,
 If yes, please list the genes::
 FGB FGG GGCX KNG1 LMAN1 MCFD2 SERPINE1
 SERPINF2 VKORC1 VWF ADAMTS13 HRG PIGA PLG
 PROC PROS1 SERPINC1 SERPIND1 THBD ABCG5
 ABCG8 ACTB ACTN1 ANKRD26 ANO6 AP3B1 AP3D1
 ARPC1B BLOC1S3 BLOC1S6 CDC42 CYCS DIAPH1
 DTNBP1 ETV6 FERMT3 FLI1 FLNA FYB1 GATA1 GF11B
 GNE GP1BA GP1BB GP6 GP9 HOXA11 HPS1 HPS3 HPS4
 HPS5 HPS6 ITGA2B ITGB3 KDSR LYST MECOM MPIG6B
 MPL MYH9 NBEA NBEAL2 P2RY12 PLA2G4A PLAU
 RASGRP2 RBM8A RNU4ATAC RUNX1 SLFN14 SRC
 STIM1 STXBP2 TBXA2R TBXAS1 THPO TUBB1 VIPAS39
 VPS33B WAS

Q4 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:

I have curated these genes in collaboration with other experts for the ThromboGenomics and the Subcommittee for Genomics in Thrombosis and Hemostasis of the ISTH

Q5 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):

None

#10

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Tuesday, February 19, 2019 2:09:11 PM
Last Modified: Tuesday, February 19, 2019 2:31:40 PM
Time Spent: 00:22:29
IP Address: 192.188.254.2

Page 2

Q1 Your Name

Mla Sullivan

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **Yes**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel? **No**

Q4 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:

n/a

Q5 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):

n/a

#11

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Wednesday, February 20, 2019 12:56:17 PM
Last Modified: Wednesday, February 20, 2019 12:57:12 PM
Time Spent: 00:00:55
IP Address: 192.188.254.2

Page 2

Q1 Your Name

Juliana Perez Botero

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **Yes**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel? **Yes,**
If yes, please list the genes::
ANKRD26

Q4 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:

No

Q5 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):

None

#12

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Thursday, February 21, 2019 3:43:24 AM
Last Modified: Thursday, February 21, 2019 3:44:18 AM
Time Spent: 00:00:54
IP Address: 5.169.146.60

Page 2

Q1 Your Name

Loredana Bury

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **No**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel? **Yes,**
If yes, please list the genes::
ITGB3,
MYH9

Q4 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:

No

Q5 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): **Respondent skipped this question**

#13

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Wednesday, February 20, 2019 9:55:43 AM
Last Modified: Sunday, February 24, 2019 11:03:39 AM
Time Spent: Over a day
IP Address: 5.170.120.189

Page 2

Q1 Your Name

Paolo Gresele

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **No**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel? **Yes,**
If yes, please list the genes::
ITGB3,
MYH9

Q4 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: **Respondent skipped this question**

Q5 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): **Respondent skipped this question**

#14

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Thursday, February 28, 2019 2:59:14 PM
Last Modified: Thursday, February 28, 2019 3:13:46 PM
Time Spent: 00:14:32
IP Address: 152.19.33.8

Page 2

Q1 Your Name

Kristy Lee

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **No**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel? **No**

Q4 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:

no

Q5 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):

n/a

#15

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Monday, March 04, 2019 8:53:35 AM
Last Modified: Monday, March 04, 2019 8:54:13 AM
Time Spent: 00:00:38
IP Address: 152.19.85.236

Page 2

Q1 Your Name

Leighton Telling

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **No**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel? **No**

Q4 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:

No

Q5 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):

NA

#16

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Thursday, March 07, 2019 8:15:34 AM
Last Modified: Thursday, March 07, 2019 8:16:28 AM
Time Spent: 00:00:54
IP Address: 24.218.4.161

Page 2

Q1 Your Name

Alfred Lee

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **No**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel? **No**

Q4 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:

None

Q5 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):

None

#17

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Wednesday, March 27, 2019 6:17:44 AM
Last Modified: Wednesday, March 27, 2019 6:21:14 AM
Time Spent: 00:03:30
IP Address: 137.191.238.100

Page 2

Q1 Your Name

Catriona Keenan

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **No**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel?

Yes,
If yes, please list the genes::
I have made some contributions but not 'substantial'.
However here are the related genes: F8, F9, VWF,

Q4 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:

No

Q5 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):

None

#18

COMPLETE

Collector: Hemostasis/Thrombosis GCEP (Web Link)
Started: Wednesday, March 27, 2019 10:22:10 PM
Last Modified: Wednesday, March 27, 2019 10:29:57 PM
Time Spent: 00:07:47
IP Address: 100.11.141.169

Page 2

Q1 Your Name

Michele Puskarczyk Lambert

Q2 Do you work for a laboratory that offers fee-for-service testing related to the work of your Expert Panel? **No**

Q3 Have you made substantial contributions to the literature implicating a gene:disease relationship that relates to the work of your Expert Panel?

Yes,
If yes, please list the genes::
DIAPH1, TPM4, RASPRG2, MYH9, MPL, RUNX1, TRPM7, SRC, CARD11

Q4 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:

Thrombogenomics

Q5 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):

N/a
