

Researcher-identified potential diagnoses and Contact recruiting clinician forms

If you identify a potential causative variant in one of the project participants that you think should be fed back to the recruiting clinician, you can use the **Researcher-identified potential diagnosis** form to alert Genomics England. This will be reviewed internally and, if it is thought to be clinically relevant, we will contact the clinician with this information. We will let you know whether we feed this information back or not.

You can also use the **Contact recruiting clinician** form to cover more general cases where you want to contact the clinician, such as where you have found a variant of research interest.

You can find these forms in the Research Environment. Click the **Research Environment Documentation** icon on the desktop, then **GeCIP**, then **Researcher identified potential diagnoses** form or **Contact recruiting clinician** form in the side menu.



Click Research Environment Documentation to find the form


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Researcher-identified potential diagnoses form

Created by Clodagh McGuire, last modified by Freya Boardman-Pretty 25 minutes ago

If you have identified a potential causative variant, please complete this form so that it can be validated and fed back to the treating physician.

Feedback to GMCs

Your full name *

Email *

Username *


User type *

What is your expertise in this area?

If necessary, would you be willing to discuss this case with the treating clinician?

☐ Yes
 ☐ No

Fill in the Researcher identified potential diagnosis form to alert us of potential causative variants


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Contact recruiting clinician form

Created by Freya Boardman-Pretty, last modified on Mar 25, 2019

If you have identified a finding of research interest and wish to contact the recruiting clinician, please fill in the below form and we will facilitate this.

If you wish to feed back about a variant you think is **causative**, please instead fill in the Researcher-identified potential diagnoses form.

Contact clinician

Your full name *

Email *

Username *

User type *

What is your expertise in this area?

Participant ID *

Gene of interest

Variant of interest

Genome build

Disease of interest

Clinical details of interest

Why do you want to contact the clinician? Please provide supporting evidence if available. *

Please include chromosomal position and variant description using HGVS nomenclature

Please enter the clinical details that you are interested in (e.g. specific disease, HPO terms, ICD-10 code, other)

Fill in the Contact recruiting clinician form to let us know what you have found