PORTAL USER GUIDE

Portal v3.0

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# Registering your account with Microsoft

- When your account is created by the RCIGM staff, you will receive an email from Microsoft that the account has been created with instructions on how to register your account and login
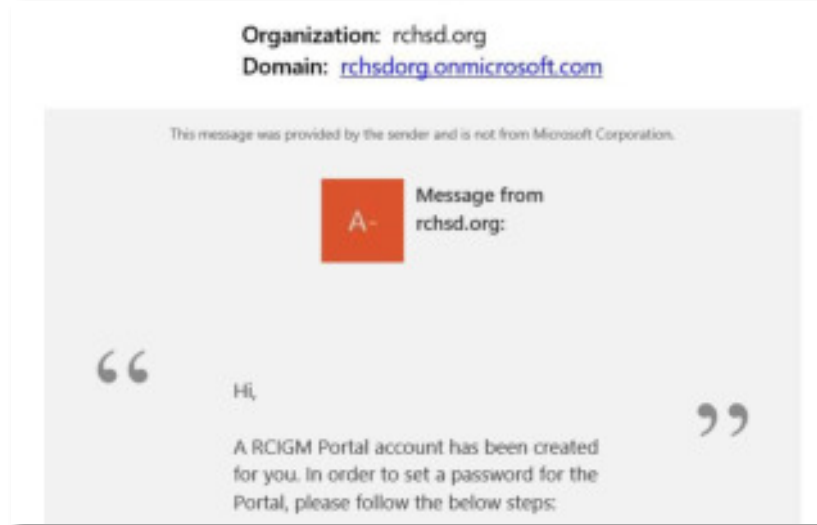

- Navigate to the link provided in the email
- You will be redirected to a Microsoft M365 login page to enter your email address
- Once you have input your email address, follow the instructions on the ensuing screens for creating a password* and receiving/entering a confirmation code
  - You will be asked to select a method for receiving a confirmation code (either by downloading the Microsoft app or opting to receive the code via SMS text message)
    *If you already have a M365 account, you can use the email and password you already have
- If you elect to use your phone instead of the Microsoft app, you can choose to receive the code via phone call or text message
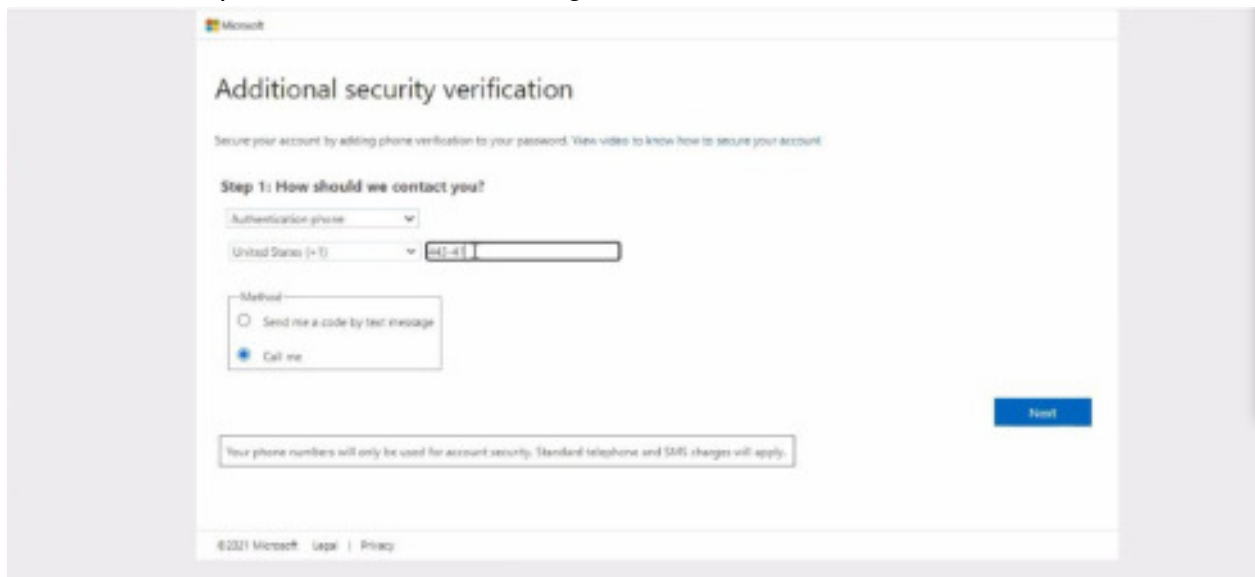
- Once you receive a code, enter it where prompted:
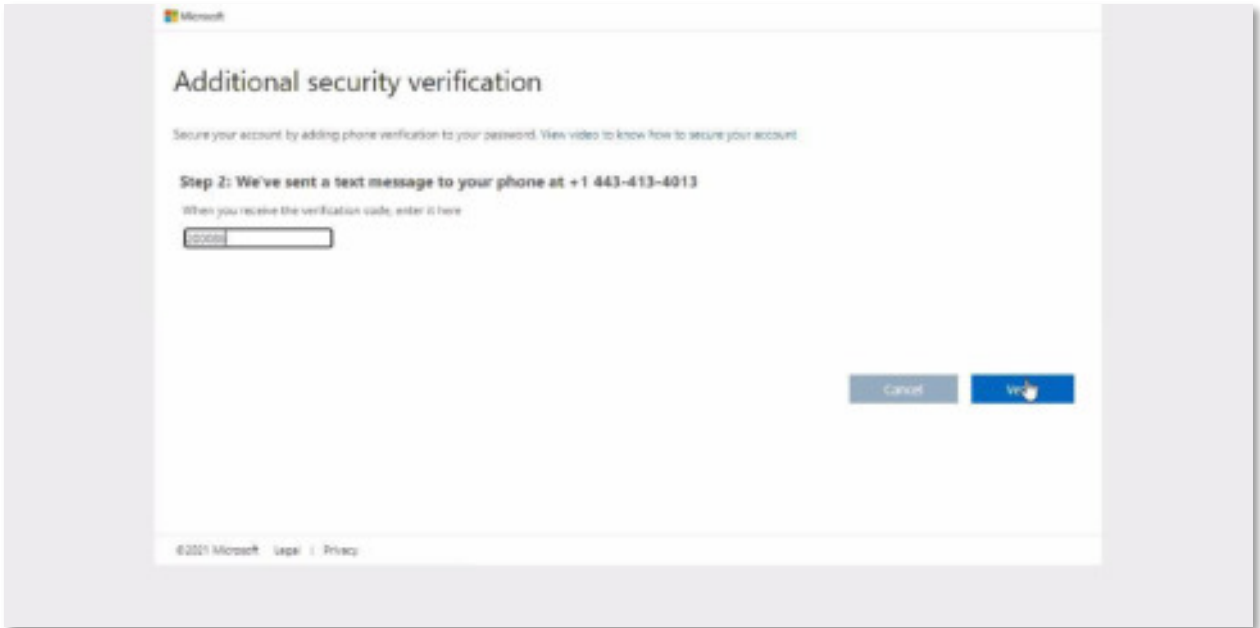
- After entering in the code, you can click the link to the portal:
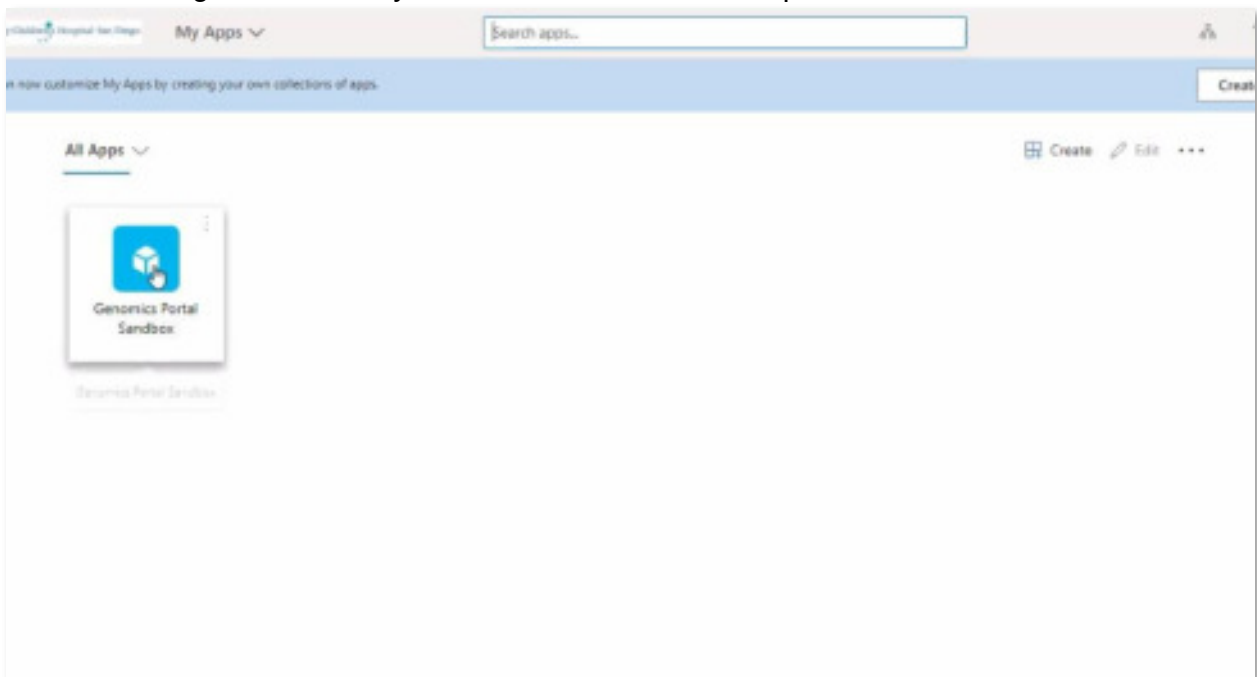
NOTE, you will be prompted for multi-factor authentication each time you log in.

Logging into portal

- Once you have entered your confirmation code and clicked the portal link as described above, you will be directed to the portal login screen
- You may use the email and password you used for the previous portal; if you have forgotten your password you can click the link shown below to reset it

- Once you follow the instructions in the email to reset your portal password, you can enter your email and new password into the login screen

- **NOTE**: the Portal is currently only compatible with Chrome, which you can download from Google if you do not already have it installed. Note, if Chrome is configured to block all cookies you will not be able to login.

Site Navigation

The following sections outline the basic navigation within the Portal.
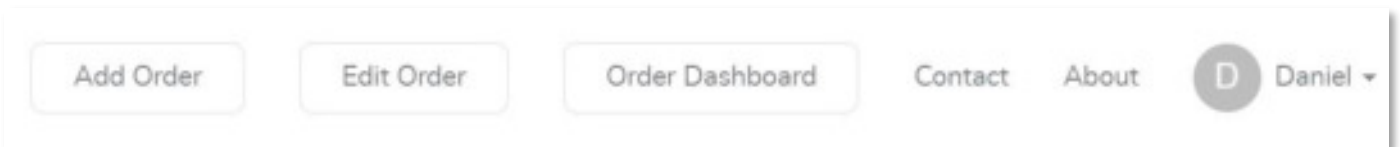
Add Order

Clicking *Add Order* kicks off the process of creating a new case, and allows you to enroll individuals into the appropriate study. The steps and enrollment process are described in a section below.

Edit Order

Clicking *Edit Order* opens a window for you to search existing cases by name or case ID. Once you select a case, you can click the Edit button to open the workflow and edit details for that particular case. More detail on editing orders are described in a section below.

Order Dashboard
Clicking *Order Dashboard* navigates to the dashboard page described in detail in a section below. The page includes the following features (this list is not all inclusive):
- A list of all families and individuals enrolled in the study/project
- Global search across studies/projects based on user access
- The ability to expand individual rows to see more detail about each person enrolled
- The ability to download results reports

Contact
Clicking *Contact* navigates to a Contact Form page. Within this page, enter details how best to contact you and any questions, concerns, or comments you have regarding the Portal or RCIGM. Someone from the Genomics Institute will reach out regarding any submission.

About
Clicking *About* navigates to the RCIGM site for more details about the Genomics Institute and team members who contribute to the Institute's work and research.

User Preferences
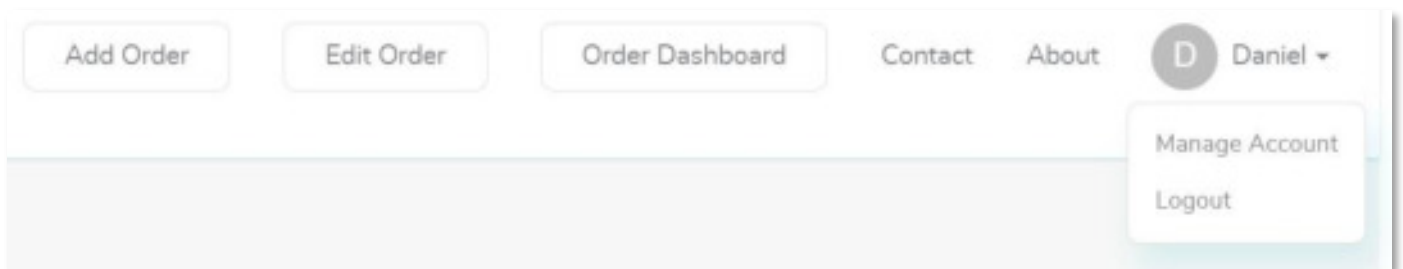
The menu item displays the first letter associated with your user profile. The following submenu options are available when the preferences menu item is clicked:
- *Manage Account*: allows editing profile information including the following details:
    - Name
    - Phone
    - Fax
    - NPI (physicians only)
    - Address
- *Log Out* logs out of the Portal and navigates to the home/login page
- Change Password: to change your Portal password, click Manage Account under user preferences, then Change Password

Page Footer

The following are available within the footer of each page:
- The *Contact* section provides a phone number and email address to reach out to RCIGM as well as the hours of operation
- *Terms of Service* navigates to a page detailing the terms of service agreed upon when initially logging into the Portal (one for clinical, one for research).
- *HIPAA Compliance Statement* navigates to a page that outlines details related to the Portal security and HIPAA compliance.
- *What is WGS?* Leads to information about genomic testing and the team at RCIGM
- *Guides & Training* will lead to a section of our Resources page that provides user guides and links to training videos
- *Licenses & Forms* will lead to a section of our Resources page that provides CAP certification, CLIA accreditation, and state license information.
- *Contact Us/Submit a Problem* leads to the issue form mentioned above

Edit Order
- To edit a previously created order or enroll a subject into an existing case, click the *Edit Order* button as mentioned above.
  - A popup box is opened to search for the existing case based on the system-generated case ID, the user-specified family ID, or last name.
  - As you type, there will be suggestions of matching cases/families that you can click.

    o Click *Edit this Order* to pick the case and close the popup

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Enrollment workflow

Clicking either of the *Add Order* buttons kicks off the process of creating a new case, where you can enroll individuals into the appropriate study
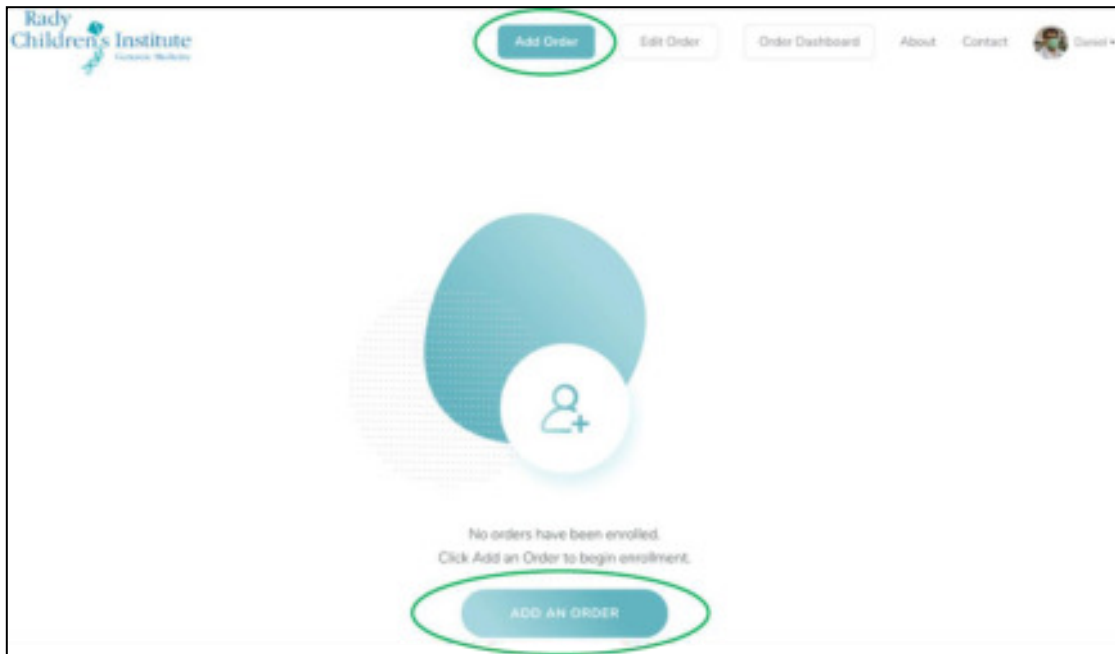
Order Information

- The first step in the research enrollment workflow is to select the study/project for the subject/family using the field '*Where should this case be created?*'. Available selections are based on user profile access. If the appropriate study/project for enrollment does not appear in the list, please contact RCIGM.
    - To select a study, click the input field where it says 'Please make a selection' and choose from the list.

- *Custom Family ID*
    - Family ID can be any identifier used within the site or study organization to reference the family being enrolled.
    - Entering a Family ID is not required if the organization does not have or use one.
- *Order Types:* Select at least one order type based on the test you want performed (e.g. rWGS Proband Only, Ultra Rapid Trio, etc.)
- *Samples Expected:* Select which family members you expect the lab to receive samples for
- Once you are done making these selections, click *Save & Continue* to move to the next step.

Register Individual

- Note that the Project/Study, Case ID, Family ID (when input), and order type(s) will display in an expandable menu at each stage of the workflow.

ORDER DETAILS

Test Project

System-Generated Case ID: **CS51AR9**
Family ID: **Test 123**

Orders: — Collapse

1. rWGS Duo (for proband and one parent)
2. Custom Variant Analysis
3. DNA Isolation and Hold

- Enter relevant details for the subject being enrolled:
    - Familial Relationship: This will be selected by default based on the prior Samples Expected field on the Order Information stage, but you can change this if needed
    - Subject Consent Information: Enter the person's name who obtained the consent of the subject and the date consent was given. These are optional fields.
    - Subject Personal Information:
        - Opt-Out for Use of De-Identified Info/Samples
            - Select this checkbox if the subject wishes to opt-out of having their de-identified test results used for additional research (this is relevant to the Clinical/Fee for Service workflow only and will not appear for research studies)
        - Opt-Out of Incidental Findings
            - Select this checkbox if the subject wishes not to receive incidental findings for family members of the proband that directly relate to a finding for the proband
        - First Name – Required
        - Middle Name – Optional
        - Last Name – Required
        - Alias – can be used if the subject's name is not available at the time of enrollment
        - Is the Patient De-Identified?
            - Select yes for this radio button if the person needs to be de-identified for any particular reason.
            - This will make all required fields optional and two new identification fields will appear and values for these fields are required to proceed with enrollment
        - Custom Subject ID (Optional) - Can be any identifier used in the study organization
          to reference the subject being enrolled
        - Date of Birth – Required if no Medical Record Number provided
        - Medical Record Number – Required if no Date of Birth provided
            - Note: either Date of Birth or Medical Record Number is required but not both
        - Sex – Required for Proband
        - Race – Required for Proband
        - Ethnicity – Not required for Proband
        - Other Notes – Enter comments regarding the subject being enrolled that is specific to that subject but not necessarily the order or sample

Adding Information Later

- If you do not have any of the required information for non-probands, you have the option of selecting the checkbox 'I'll add this information later'

    o This will allow you to continue with enrollment of at lease one, and you can return to this stage to complete the information at a later time

    o There will be an orange indicator in the left navigation panel to remind you to return and complete this information later

Deleting an Individual

- If you would like to delete an individual from the case, you can click the Delete Individual button

Registering Another Individual/Saving

- If you need to add another individual at any point, click the 'Register Another Individual' button shown below

- Once details have been entered for all individuals, click *Save & Continue* to save the subjects' information and move onto the next step.

Enter Clinical Info

- An expandable Order Details banner will appear at the top of this stage, as mentioned in the Register Individual stage
- Enter relevant clinical notes for the subject being enrolled:
  - Drag and drop a file into the area noted on the screen, or click the link that opens a file browser window to browse and upload clinical notes for the subject.
    - Recommended clinical notes include: recent Progress Notes, Consult Notes (Genetics, Neurology, Cardiology, Genetic Counseling, etc.), Pedigrees, Genetic tests, other laboratory tests and Imaging reports.
    - Details regarding file types and sizes are outlined in the drag & drop area
    - Once files have been uploaded the page will display file details
    - Files can be deleted by clicking the red delete/trashcan icon
    - Files can be downloaded by clicking the file name or the download icon next to the delete icon
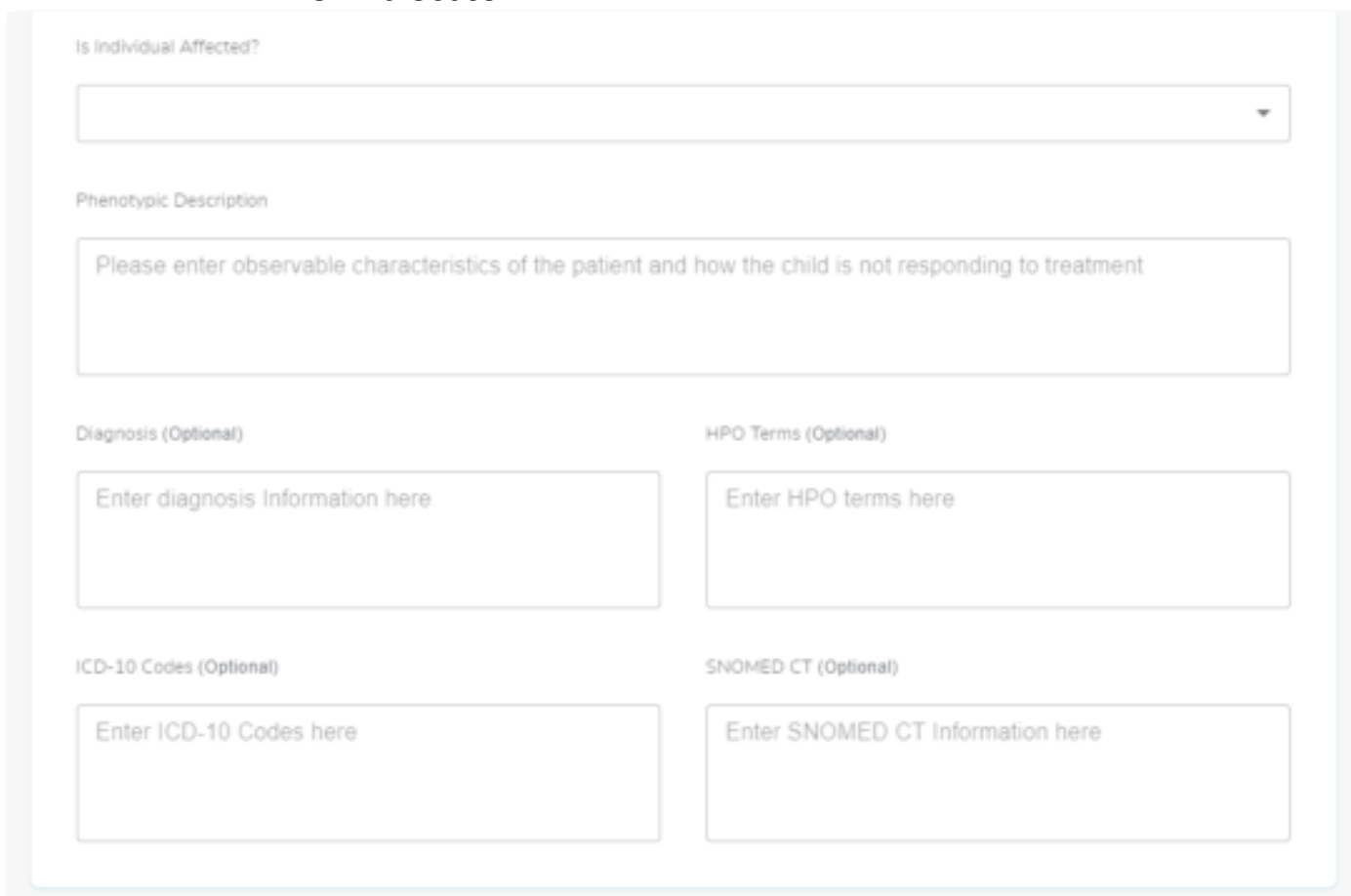
- o On the Clinical Info page, continue entering details for the subject
  - ▪ Is the subject affected? - Required
  - ▪ Phenotypic Description – Required for Proband; include a brief summary of clinical presentation.
  - ▪ Diagnosis – Can enter relevant clinical data such as differential diagnosis and/or genetic disorder suspected.
  - ▪ SNOMED CT
  - ▪ HPO Terms
  - ▪ ICD-10 Codes

Is Individual Affected?

Phenotypic Description

Please enter observable characteristics of the patient and how the child is not responding to treatment

Diagnosis (Optional)

Enter diagnosis Information here

HPO Terms (Optional)

Enter HPO terms here

ICD-10 Codes (Optional)

Enter ICD-10 Codes here

SNOMED CT (Optional)

Enter SNOMED CT Information here

- Once all details have been entered, click *Save & Continue* to save the subjects' information and move onto the next step in enrollment.
  - o **NOTE**: Clinical notes are not required to submit the order, but it is strongly recommended that you upload them if possible. If additional details regarding the subject are captured or available at a later time, the Clinical Information can be updated at that time.
- Reminder: At any point during the enrollment workflow, you may click a previously completed stage in the left navigation panel to return to that stage and edit as needed
  - o If any details are not saved before navigating away from a stage, a warning will be presented that unsaved information will be discarded.

Proband's Physician / Ordering Provider Information

- On the Sample Details stage, there will be a section to enter information for the proband/patient's physician, and another for ordering provider.
- First, enter the proband/patient's physician information (Note, this information identifies where the results of the testing are to be returned)
  - First Name (Required)
  - Last Name (Required)
  - Email (Required)
  - Phone (Required)
  - Fax (Optional)
  - NPI (Optional)
- If a physician has been linked to the study where you are enrolling the patient, you can pull the information from the dropdown which will auto-populate the required fields

- For Ordering Provider, enter the required information (Note, this information identifies the person who is ordering the test for the subject and may or may not be the same as the subject's physician):
  - Provider Type – if this is the same as the Proband's physician, you can make this selection and previously entered details will auto-complete. Or choose an alternate provider type.
  - Select from list: You can select a physician or yourself from this dropdown
  - Provider Name (Required)
  - Email (Required)
  - Phone (Required)
  - Fax (Optional)

- Once all information has been added for Proband's Physician and Ordering Provider, scroll down to complete sample details (these fields need to be entered before the sample details can saved). See more detail below

Sample Details

- Click 'Add Sample' for each individual that you are ready to enter sample information for
- RCIGM-CGC Test Description: clicking this button opens a popup window that provides detailed descriptions of tests performed, how the testing is performed, what is included in the reports and other comprehensive reference information.

- On the form that populates after the Add Sample button is clicked, enter details about the sample:
  - o Sample Type – Required (the values provided in the dropdown are specific to the study chosen when enrolling the subject)
  - o Date/Time Sample Collected (Optional)
  - o Sample Name (Optional)
  - o Sample Volume/Weight (Optional)
  - o Sample Volume Unit (Optional)

- o Sample Notes (Optional)
- Click the 'Add Sample' button for each sample you plan on sending (e.g. if you plan on sending 2 blood samples for proband, proband would have 2 sample forms to complete)
  - o Note: the new sample will have a label of Sample #2 to distinguish the fields from those previously entered.

- If the sample type is DNA (extracted), a new field *Location of DNA Isolation* will be presented with two options for a CLIA laboratory or a research laboratory.
  - o If the CLIA laboratory option is selected, a field will be presented to input the *CLIA Laboratory Name.*

- If a sample was added by mistake, click *Delete Sample* and the fields will no longer appear on the screen

- **NOTE:** Once a sample has been received by the lab (i.e. is already accessioned), details for that particular sample can no longer be edited. Contact RCIGM to update any details that were entered incorrectly.
- Once all details have been entered, click *Save & Continue* to save all information and move onto the Sign & Print stage.
- **NOTE**: If additional details regarding the sample information are captured or available at a later time, the Sample Details can be updated at that time

Sign & Print

- The Project/Study, Case ID, Family ID (when input), and order type(s) will display in the expandable Order Details menu, as with previous stages
- If you bypassed entering information for any individual (e.g. you clicked the 'Add this Information later' checkbox, or didn't add a sample for an individual) an orange message will appear, and the left navigation panel will alert you to what area needs completion
  - You can still sign and print the req form for the individuals who are ready to have their samples sent to the lab

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Electronic Signature

- There will be a certification message for the signer to review
- The ordering provider, representative thereof, or another authorized signer like a genetic counselor can electronically sign and save their signature in the box shown below (this could either be a physician or other medical professional acting as a delegate)
  - This is not a required field
  - The signer can delete and redo their signature at any time
  - If electronic signature is saved, it will appear on the requisition form


Printing Requisition & Shipping Samples

- Shipment instructions are provided to help guide on how to continue if all subjects have been entered and the samples are ready for shipment (need to be added above print req button)
- When you are ready to print the requisition form, click the button on the bottom right corner
- This will open a window where you can select which sample(s) you will be sending to the lab along with the requisition form
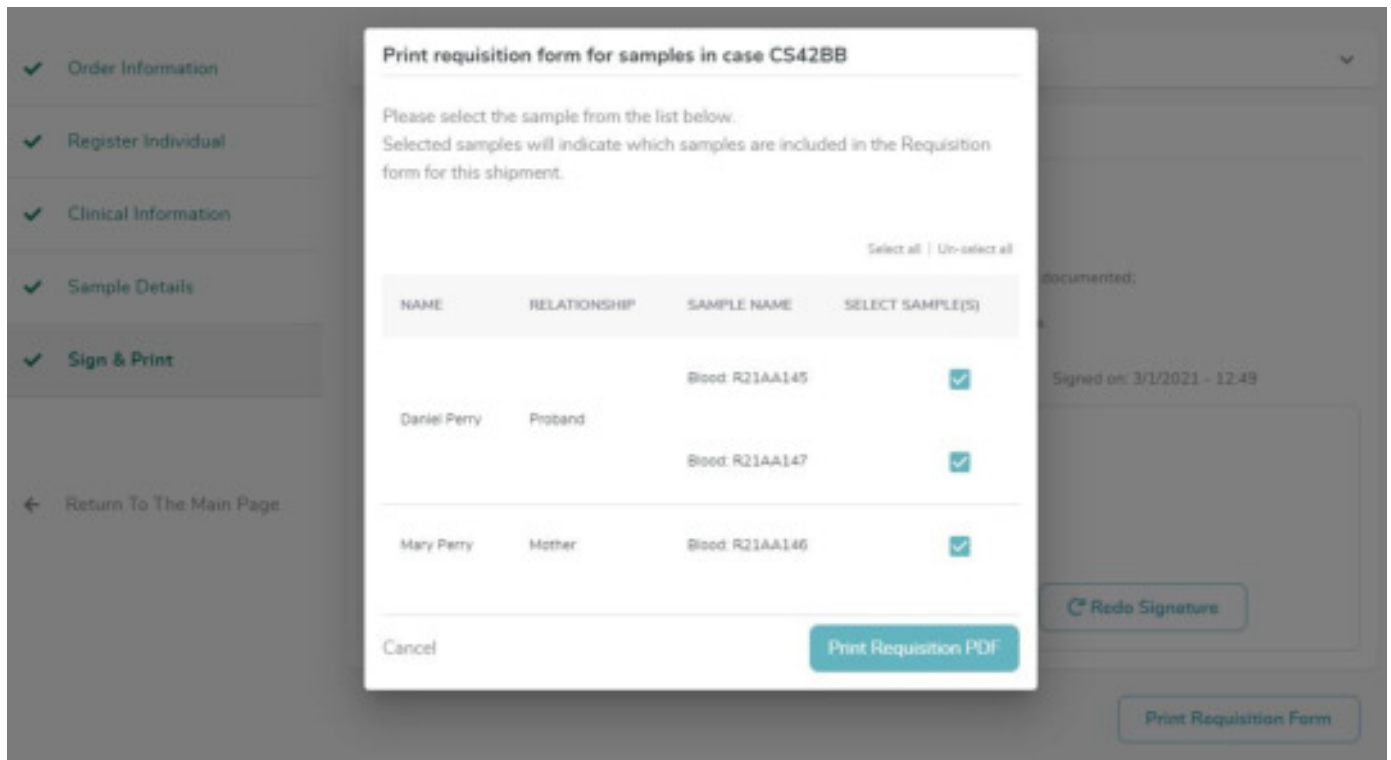
- Once all appropriate samples have been selected, click Print Requisition PDF to generate a printable version of the requisition.
  - A warning message will be displayed prior to printing the requisition as a reminder to indicate all samples that will be included in the shipment and ensure that it is downloaded to an appropriate file location to protect PHI.
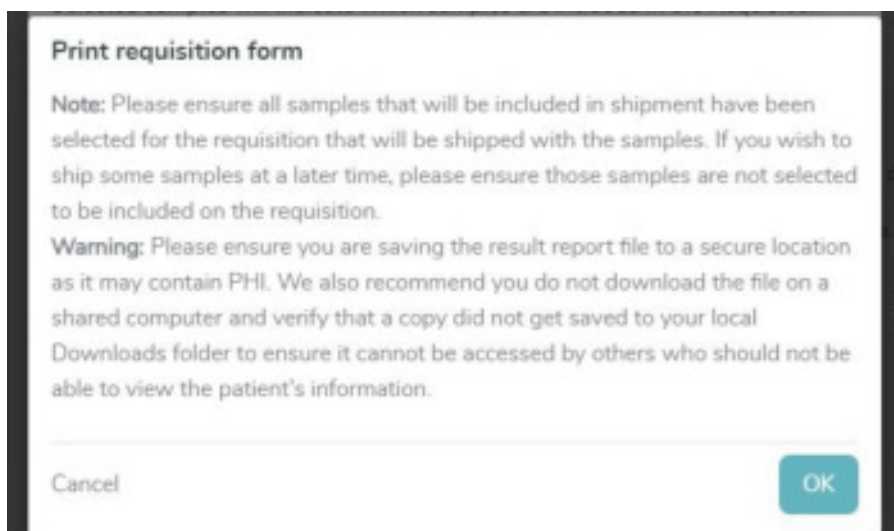
- After clicking *OK* to confirm the warning message was read, the requisition is generated and will automatically download the file to the main browser as well as your computer's local downloads folder

Order Dashboard

Within the main top menu bar, clicking *Order Dashboard* navigates to a page that includes the following features:

- A list of all families and individuals enrolled in the study/project, grouped by family/case/order
- One row will be displayed for every sample added to an individual
- Search across studies/projects based on user access
- Ability to expand individual rows for additional individual, clinical, and sample information, with ability to link back to workflow to make edits to an order
- Ability to download results reports, clinical notes, and raw genomic data

Search

Users may choose to search the Order Dashboard by specific areas (e.g. name, study, etc.) using the 'Search By' box. **NOTE:** only one search term can be applied at a time

Search includes the following searchable areas:
- o Project/Study Name
- o Project/Study Code
- o Institute Abbreviation
- o Case ID
- o Custom Family ID
- o Order Type
- o Case Created On
- o Portal Individual ID
- o First Name
- o Middle Name
- o Last Name
- o Alias Name
- o Indiviudal ID 1
- o Individual ID 2
- o Family Relationship
- o Date of Birth
- o Medical Record Number
- o Opt Out of De-Identified Data
- o Opt Out of Incidental Findings
- o Individual Enrolled Date

Once the filter criteria has been selected, click *Search* to update the grid with the criteria
- **NOTE**: results displayed within the grid span across studies when the user has access to multiple studies
  - o To remove the search criteria, click the *Clear Search* button located next to the search bar, on the right side of the screen
- The Details Grid is populated with a row for each sample for the individuals enrolled in the project(s) / study where the user has access
  - o The case / family will be grouped together
- Below the grid count of items returned are pagination controls to allow the user to update the number of items being returned which include 5, 10, 15, 20, or 25 items with the default as 5 items
  - o On the right side of the grid are paging controls that allow navigating to the Next page or Previous page to review details

Downloading results reports
- To download test results, navigate to the Order Dashboard. If there is a report available, there will be a link you can click called 'Reports Available – Download Reports'

- NOTE: A warning is presented to ensure that the appropriate location for the download is selected as the reports can contain PHI. Once acknowledged, a ZIP archive file containing the report(s) for the test will be downloaded by the browser and can be moved to an appropriate location.

Right Panel

- If you click the + icon on any row, there will be a details panel with the following information for the individual you selected
    - Individual details (see image below)
    - Clinical information (see image below)
    - Sample details of each sample added for the individual (see image below)
    - You can also download reports, clinical notes, and raw genomic data from here if available
    - To edit information, click the Edit Individual button which takes you back to the workflow where you can make the edits
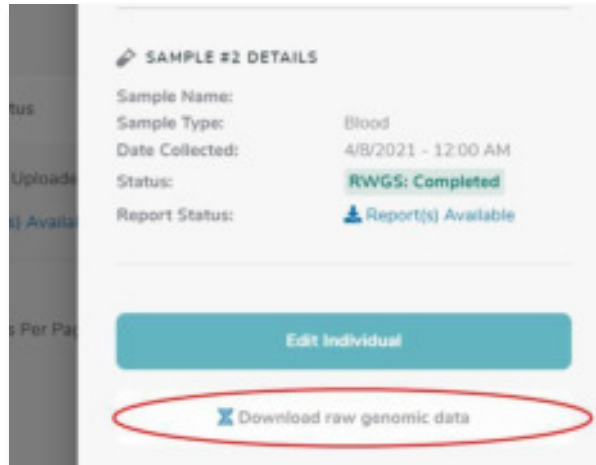

Download raw genomic data

- If there is raw genomic data available to download, you will see a button labelled 'Download Raw Genomic Data' in the right panel

- A window is opened which lists the filename, description and download URL to be able to save the raw genomic data files.
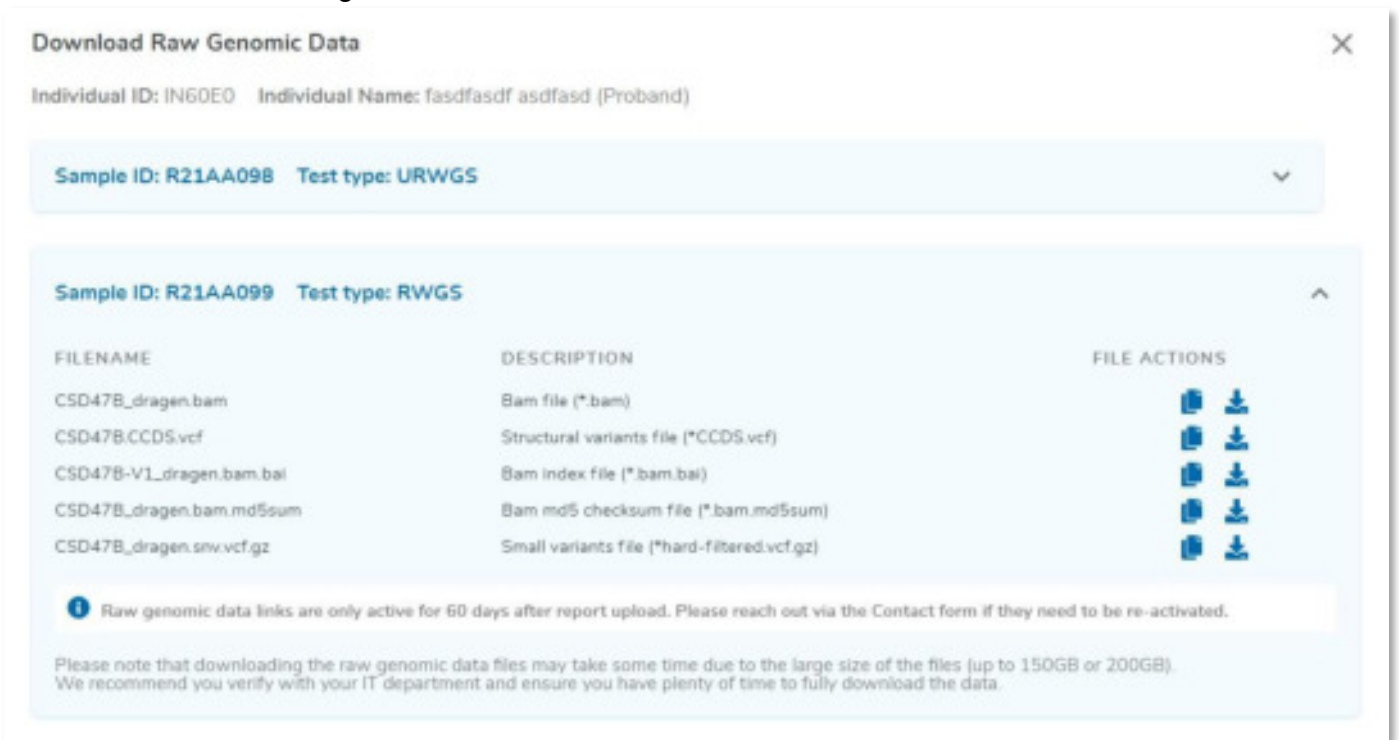
- The links are active for up to 60 days after the final results are available for download

NOTE: Downloading the raw genomic data files may take some time due to the large size of the files (up to 150GB or 200GB). We recommend you verify with your IT department and ensure you have plenty of space to fully download the data

Appendix A – Notifications

The table below outlines which rWGS Portal events trigger email notifications and the recipients.

Recipients:
- Submitter – person performing the action/event.
- Genetic Counselors - individuals identified in the workflow document and added when the project/study was created
- Principal Investigator(s) – individual(s) identified in the workflow document and added when the project/study was created
- Physician(s) – individual(s) identified in the workflow document and added when the project/study was created
- Proband Physician – identified during proband enrollment

Event	Email Recipients
Patient Enrollment	✓ Submitter ✓ Genetic Counselors ✓ Principal Investigator(s) ✓ Physician(s)
Sample Submission	✓ Submitter
Test Addition	✓ Submitter
Result Upload (Preliminary, Final, Amended, Corrected)	✓ Submitter ✓ Genetic Counselors ✓ Principal Investigator(s) ✓ Physician(s) ✓ Proband Physician
Study Creation	✓ Genetic Counselors ✓ Principal Investigator(s)
Sample Accessioning	✓ Genetic Counselors ✓ Principal Investigator(s) ✓ Physician(s)
Reject Accessioning	✓ Submitter ✓ Principal Investigator(s) ✓ Physician(s)

If modifications are needed for the list of Genetic Counselors, Principal Investigator(s) or Physician(s) that were setup for a project/study, please contact RCIGM to make the needed update.