Instructions for PhenoTips data entry in MYO-SEQ

As part of the MYO-SEQ project, we have set up each of the partners with an account in the clinical database at PhenoTips (more information can be found here: http://phenotips.org/).

Partners in MYO-SEQ will be expected to enter the phenotypic data which has been collected for each of the patients whose samples are being sent for sequencing as part of the project.

In order to do this you need to follow the instructions below:

How to enter your clinical data into the MYO-SEQ database

Go to: https://rdconnect.ccm.sickkids.ca

LOG-IN
1. Enter username and password - supplied to you by Katherine Johnson
2. Click on the PhenoTips logo on top left of the screen - this ensures you are on the correct page to begin

CHANGING A PASSWORD:
3. The first time you log-in, you should change your password to something only you will remember. To do this, click on your name on the top right of the screen, select 'Preferences' on the left hand menu and choose 'Change Password'.

VIEWING USERGROUPS:
4. Whenever you want to return to the starting page, click the PhenoTips logo. From here you can view the usergroups – and you can see who is a member of each group. This is important for when you come to share your data with others (this could be implemented in the future, not relevant for the moment).

ENTERING PATIENT RECORDS:
5. To enter patient data click on 'New patient record' on the right of the screen (it appears in two places, either is fine).
6. Select the disease form you wish to use and click 'Create' - a blank form will appear.
7. Confirm that consent has been taken for NGS and for clinical data sharing by checking the boxes at the top of the form - if the consent is not in place, data can still be entered but the patient should be re-consented and the boxes checked once this has been done (very important, sample will not be sequenced if box not ticked)
8. Enter the MYO-SEQ ID (QR code) which goes with your sample in the 'Identifier' field
9. If there are any additional identifying codes which belong to this data, you can add these too by clicking +NEW ENTRY (make sure you enter your own code)
10. Select Male or Female
11. Add ethnicity descriptions - these are currently free text
12. Choose Y or N for Clinical Status (Y- affected; N- unaffected)
13. Under 'Family Study' click +NEW ENTRY if this patient is linked to others in the database - you can then select the type of relative and patient ID of linked people. You can do this as many times as you need to. (Not currently working, will be implemented in the future).
14. Now work through the Inheritance, Progression and Onset data. Indented descriptions are sub-categories of those above. If you click on the little 'info' icon, a definition will pop-up.
15. Work through the rest of the form - click on big blue headings to reveal each section.

16. Where a blue triangle exists beside an observed phenotype, clicking on it will reveal sub-categories.
17. You can add any additional phenotypic information not included in the form by typing a feature in the ‘Quick Phenotype Search’ box. Suggestions from the ontology will appear and you can choose the most appropriate one.
18. Under Diagnosis you can type in any confirmed diagnosis and choose from the pop-up OMIM descriptions. You may also select from the suggested diagnoses which have been generated based on your entered phenotype data.
19. Click on the 'Save' button to save your record.

SETTING PERMISSIONS FOR ACCESS: (this could be implemented in the future, not relevant at the moment).
20. You now need to decide who can see/edit your record: At the top of the form, on the right, it will say, 'This case is owned by You, it is Private and it is shared with 0 collaborators' Click on the spanner icon next to this statement.
21. Leave “Ownership and Global Visibility” as they are (default is Private) but under Collaborators, type the name of either your working group (MYO-SEQ) into the bottom box and select the appropriate pop-up. YOU SHOULD ONLY DO THIS IF YOU INTEND TO SHARE YOUR RECORD WITH ALL MYO-SEQ PARTNERS.
22. Use the drop down menu to give permission to those with whom you shared data to either view OR view and edit the record.
23. If you wish to give any other working groups permissions too, or allow individual investigators to access the record you can also do this here.
24. Click 'Update'.

VIEWING DATA ENTERED
25. When you log-in you will see a list of the data to which you have access listed on the right of the screen. It is split between the data which you ‘own’ because you entered it and data to which you have been granted access by other partners.
26. Clicking on one of these records will open it up. If you own it and wish to edit it, select the pencil ‘edit’ icon. ALWAYS RE-SAVE YOUR RECORD IF YOU MAKE ANY CHANGES
27. To search for particular data, select ‘view all data’ and then choose, ‘Advanced data filters’ from the top left. This allows you to search records based on a number of filters.