



## PhenomeCentral - Information for Families

### What is the purpose of PhenomeCentral?

- PhenomeCentral is a restricted access database that securely stores detailed clinical and genetic information on individuals with undiagnosed rare diseases
- PhenomeCentral automatically matches patients with similar features or DNA changes, and connects the clinicians who are treating these patients
  - Enabling faster diagnoses and simpler identification of the genetic cause of disease

### What data will be collected?

*With your consent, clinicians/researchers will be able to enter:*

- Detailed observations and measurements of the affected individual obtained by physical examination (also referred to as a *phenotype*); this is not identifiable information and therefore consent is implied
- Available data obtained from exome or whole genome sequencing (*genotype*)
  - The exome or whole genome sequencing may have been performed as part of a clinical service or within an ethically approved research project
  - Consent for exome or whole genome sequencing was obtained separately as part of a clinical service or an ethically approved research project
- Health information of the affected individual including: prenatal history, medical history, family history
- Medical images

### Who will have access to the data contained within PhenomeCentral?

- No one outside of your healthcare team will be able to view your personal health information, but in a similar fashion to a game of “Go Fish” the database will send out anonymized queries from your data in the hopes of finding another participant with similar phenotype and genotype information

### How will my clinical and genetic information be kept private?

- Information about you/your child will be given a specific code, and personal identifiers will be removed
- The automatic matching is possible without revealing your/or your child’s identity
- Only your doctor will be able to link the personal identifiers back to coded data

### Future contact

- Your doctor may contact you if a match between your/your child’s de-identified data is made with another individual within the **PhenomeCentral** database
- Such a match may increase the chances of finding a diagnosis for you/your child
- You may also be asked if de-identified details can be included in a publication in the medical/scientific literature

### Can I withdraw from PhenomeCentral?

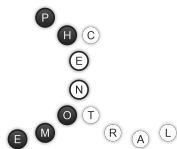
- If in the future you wish to withdraw you/your child’s record from **PhenomeCentral** for any reason, this can be done quite simply by making a request to your healthcare provider who obtained your consent
- Parents/guardians who have given consent for their child’s record to be entered in **PhenomeCentral** should ensure that when the child reaches the age of 18 years he/she is made aware of his/her entry so that he/she can make his/her own choice whether to continue or withdraw the entry

### Other questions

- If you have more questions about **PhenomeCentral** please ask your healthcare provider for further explanation and/or visit <https://phenomecentral.org>

### What do I do next?

- If you are willing to include information about you/your child into **PhenomeCentral**, please complete and sign consent form below



## PhenomeCentral Consent Form

**Please print and keep the signed copy of this form in the patient record (do not send to PhenomeCentral)**

I give permission on behalf of myself/my child ..... (name)

born..... (yyyy/dd/mm) to Dr./Prof..... to

submit de-identified information onto the PhenomeCentral database:

Available exome or whole genome sequencing data ☐ Yes ☐ No

Health information as outlined in the information form ☐ Yes ☐ No

Photographs of me/my child ☐ Yes ☐ No

By signing this consent I confirm that I have read the explanatory form 'PhenomeCentral – Information for Families' and understand that:

- My name/my child's name will not be published
- My/my child's information will be placed on **PhenomeCentral's** secure website (phenomecentral.org)
  - The database will be used by clinicians and researchers and scientists worldwide
- The organisers of **PhenomeCentral** can accept no legal liability for the use or misuse of information

Name of patient/parent/guardian.....

Relationship to patient (e.g. self/mother/father/guardian).....

Signature..... Date.....

Signature of clinician seeking consent..... Date.....

**Thank you for taking the time to read and complete this form. Please return it to your healthcare provider.**