Deliverable

D1.3 Training modules, guidance document and online help module for collection of phenotypes	
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Lead beneficiary	CNAG-CRG (Sergi Beltran)
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Explanation according to GA Annex I:

Training and good practice guidance will be provided in order to ensure the quality of data at the source, based on users' guides already in place around HPO and PhenoTips.

Abstract:

In order to engage users with Solve-RD and facilitate their contribution of phenotypic data to the project, we have developed training materials and have carried out training activities, which will continue in time. We have set up a practical guide to PhenoTips data entry (https://rd-connect.eu/phenotips-guide/), provided an in-depth YouTube video with instructions (https://www.youtube.com/watch?v=wvTJXxtuI8Y), organized our first training webinar for ERNs (available at https://www.youtube.com/watch?v=56omyLC0wQw&t=13s) and created an option for uploading phenotype data using an Excel template. In addition, further support to researchers is provided by one-to-one tele-conferences for PhenoTips usage and by answering any queries they have through help@rd-connect.eu.

Introduction:

Phenotypic data upload can be very time-consuming and tedious especially for Solve-RD beneficiaries and associates with large, heterogeneous datasets. For this purpose, we have produced a number of training modules and guidance documents to facilitate phenotypic entry via our user-friendly PhenoTips instance. Upon registration to RD Connect, each partner will be also granted access to PhenoTips where they can record all their clinical data collected for each affected and unaffected individual. PhenoTips has been linked to our RD-Connect Genome-Phenome Analysis Platform and can be accessed at https://platform.rd-connect.eu/phenotips/. Here we outline information regarding training modules, guidance documents and online help modules for the collection of phenotypes for the Solve-RD project. These modules and guidance documents have been set up to provide in-depth instructions for the benefit of the user to facilitate phenotype upload in a precise, time-efficient manner.

Report:

All clinical data will be collected by the RD-Connect's PhenoTips instance that can be accessed at https://platform.rd-connect.eu/phenotips/. We have prepared a number of online training modules and guidelines to help researchers to use PhenoTips for their clinical patient data collection. Below, we briefly outline.

1) Online PhenoTips guidelines

We have prepared step-by-step online guidelines on how to access and manage the PhenoTips instance through the RD Connect platform. Users can access these guidelines at https://rd-connect.eu/phenotips-guide/ (Fig1) where they will be guided through patient record entry, creating family pedigrees, reporting clinical symptoms and findings as well as setting permissions for data access. These guidelines also explain the steps users are expected to follow once cases are solved and how they can share their findings.

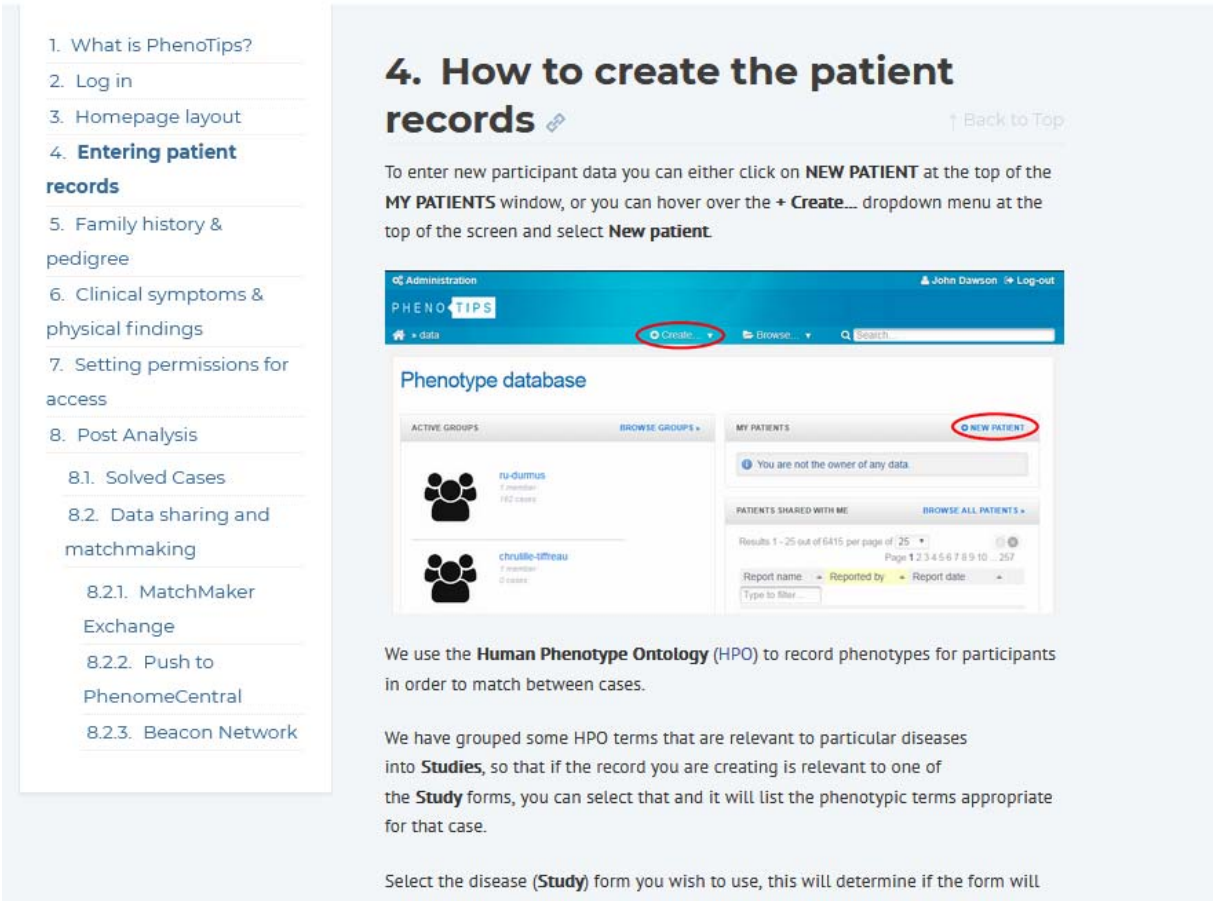
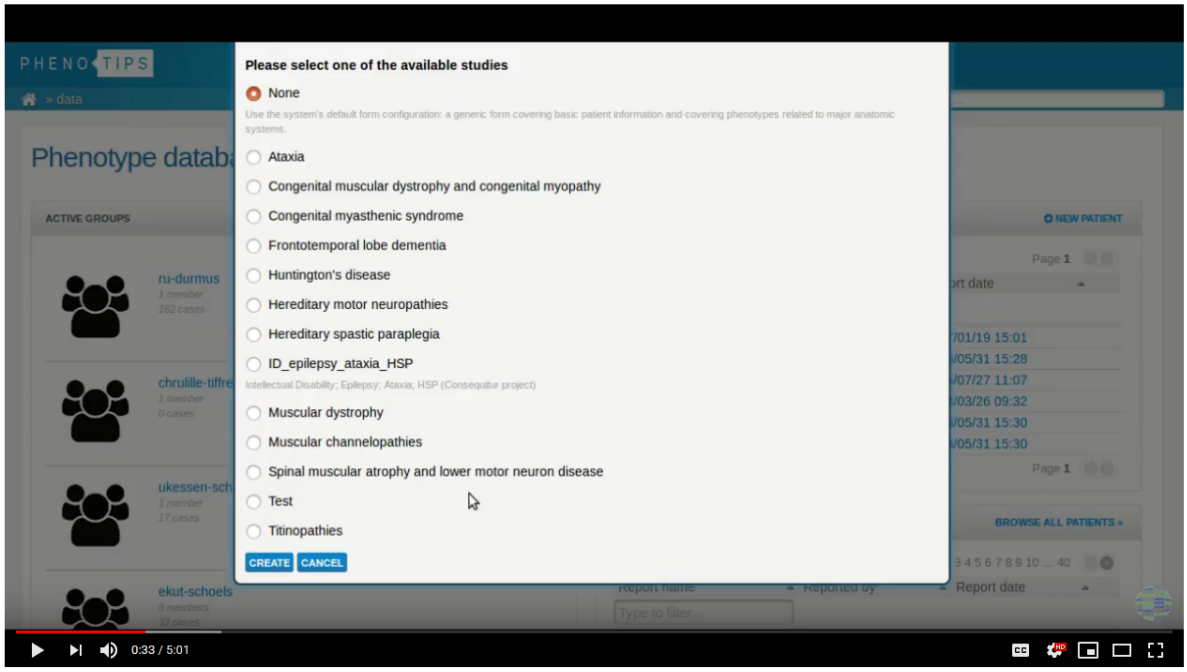


Fig1. Screenshot of RD-Connect PhenoTips guidelines available at https://rd-connect.eu/phenotips-guide/

2) Video tutorial available at Youtube

Users can also learn how to correctly enter clinical data into PhenoTips with our YouTube video entitled “Entering clinical data in PhenoTips for the RD-Connect Genome-Phenome Analysis Platform”. The video can be found here: https://www.youtube.com/watch?v=wvTJXxtuI8Y. We demonstrate with real examples, how PhenoTips access and data entry is achieved. A screenshot of the video is shown in Fig2.



Entering clinical data in PhenoTips for the RD-Connect Genome-Phenome Analysis Platform

Fig2. Screenshot of tutorial video available at https://www.youtube.com/watch?v=wvTJXxtuI8Y

3) Online webinars and teleconferences

We hold regular webinars to provide assistance to all PhenoTips users. Our last webinar took place on 6th September, 2018 where 39 users registered. The webinar was subsequently uploaded to YouTube and can be found here: (https://www.youtube.com/watch?v=56omyLC0wQw&t=13s). We have a second webinar scheduled for the 16th of January 2019. Many Solve-RD participants require an initial one-to-one teleconference to get started with the PhenoTips instance which we provide by a case-by-case manner.

We also have a help-desk email to provide case-by-case support at: help@rd-connect.eu.

Conclusion:

We have established a multitude of online training modules, guidance documents and help tools to aid all Solve-RD beneficiaries and associated partners for the efficient, high quality collection of patient phenotypes. We will continue organizing webinars and providing support to Solve-RD data contributors and will adapt training materials and documentation as requirements evolve and new needs and issues are identified.