Deliverable

D5.1 Bespoke Phenotips frontends for associated ERNs and undiagnosed disease programmes	
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Explanation according to GA Annex I:

Bespoke Phenotips frontends for collection of clinical information of unsolved patients from associated ERNs and undiagnosed disease programmes will be created.

Abstract:

This report provides information on the new clinical submission forms designed and implemented in GPAP-PhenoStore, the phenotypic module of the RD-Connect Genome-Phenome Analysis Platform (GPAP, https://platform.rd-connect.eu/). These templates have been designed in collaboration with clinical experts from Solve-RD WP1 and in alignment with Genomics England data models. This implementation facilitates the collation and future portability of structured clinical information of unsolved patients from associated ERNs and undiagnosed disease programs.

Introduction:

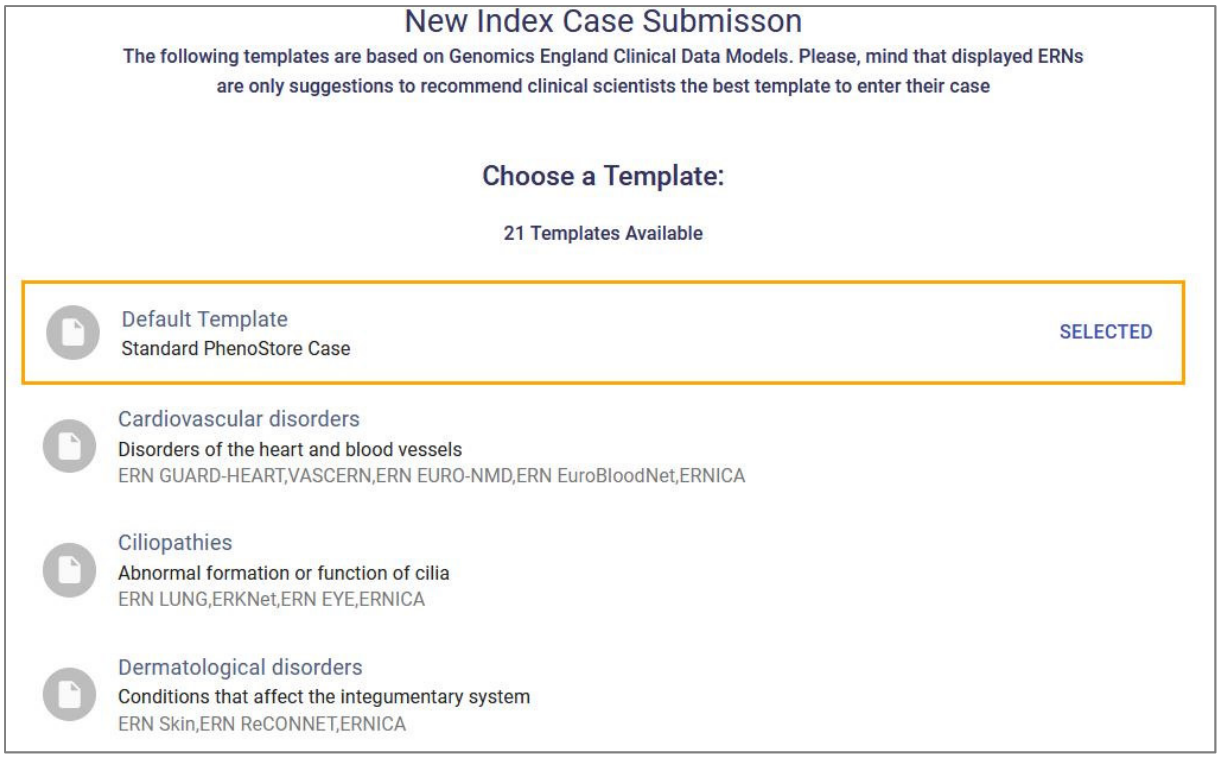
The collation of structured phenotypic data using standards and ontologies such as HPO (*https://hpo.jax.org*), OMIM (*https://www.omim.org*) and ORDO (*http://www.orphadata.org*) can be an important workload for clinicians and researchers. To facilitate phenotypic data entry, the RD-Connect GPAP offers a user-friendly phenotypic data entry tool, GPAP-PhenoStore (https://playground.rd-connect.eu/phenostore/), developed in the context of the EJP-RD project.

Herein and in collaboration with Solve-RD WP1 and clinicians from the core ERNs and undiagnosed rare disease programs, we have designed specific disease forms to facilitate data entry in the system. These forms are based on the Genomics England data model for Rare Diseases (*https://www.genomicsengland.co.uk/?wpdmdl=5500*) and provide a tailored list of relevant signs and symptoms for describing the patients' phenotypes in 20 types of disorders such as neurology and neurodevelopmental disorders or tumour syndromes. Specific disease-related fields such as the number of different types of polyps for ERN-GENTURIS have been included to corresponding forms. Disorders from all ERNs submitting data within Solve-RD are represented and mapped in these templates.

The reported system and design are interoperable and very flexible, enabling new additions and customizations by Solve-RD partners if required in the future. Data from all Solve-RD participants entering the system will then be exported into phenopackets (*http://phenopackets.org*) and used for downstream (meta)analyses.

Report:

Phenotypic data in PhenoStore is collected through available forms for users. These forms are customizable and can be adapted to ease the introduction of information for certain diseases. Below is shown the main page with the disease forms to choose from:



Through the Solve-RD project, and in collaboration with WP1, we were able to create new forms to facilitate the collection of data by clinicians/researchers from ERNs. Specifically, forms covering disorders for ERNs GENTURIS, ITHACA, NMD and RND were created.

We introduced mandatory fields to all these forms (see image below), which is necessary to provide a standard and robust phenotypic database.

In addition, we were able to provide a pre-defined list of mapped HPOs to the different diseases by using the data models generated by Genomics England, accessible at "Rare Disease Conditions Clinical Data Models". Then, with the collaboration of the ERNs the HPOs were chosen and introduced in each form specific for each ERN.

Individual information

Local ID*

Enter the ID or code you use to refer to this individual at your own centre, but do NOT use personally identifying information (no initials or date of birth)

Vital status

Alive

Country of birth

Unknown

Date of birth*

If day and month are left blank,it will take the 1st of January

year | month | day

Sex*

Male Female Unknown

Disease status*

Affected Unaffected Unknown

Examples shown below are different forms for “Cardiovascular disorders” and “Endocrine disorders” with their respective specific disease types HPO trees to select from.

Template Selected: **Cardiovascular disorders**

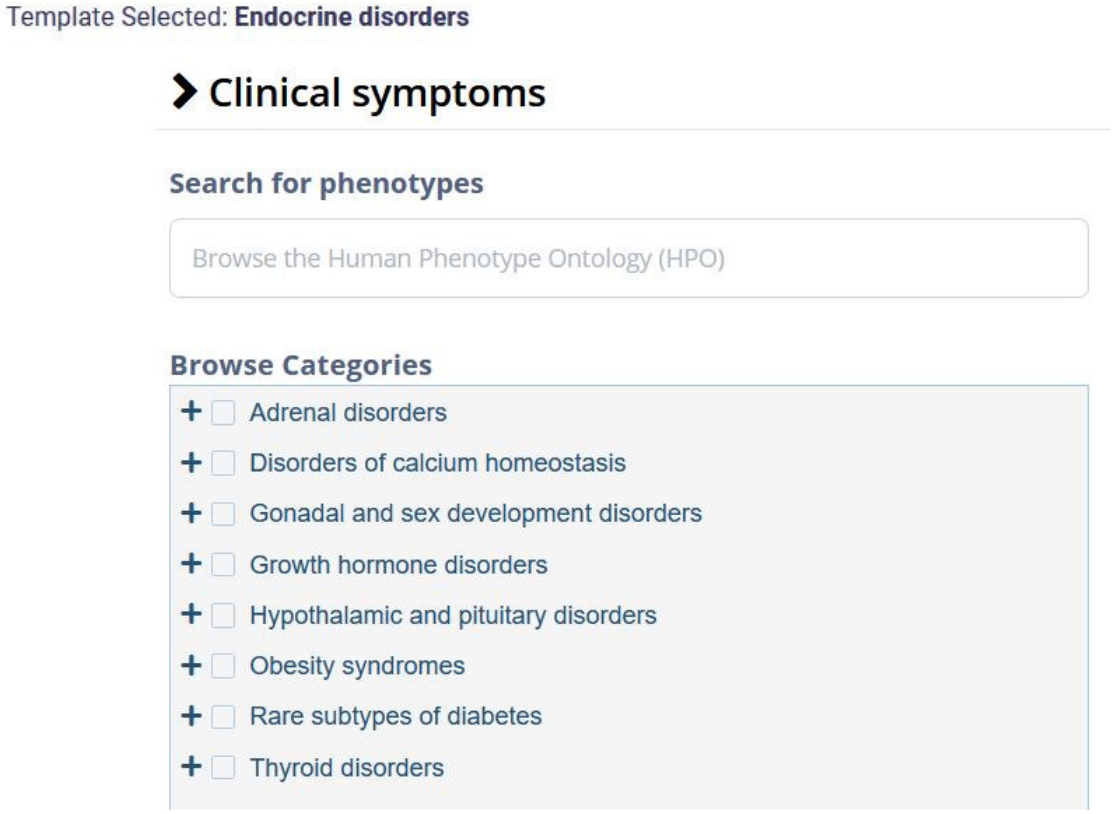
Clinical symptoms

Search for phenotypes

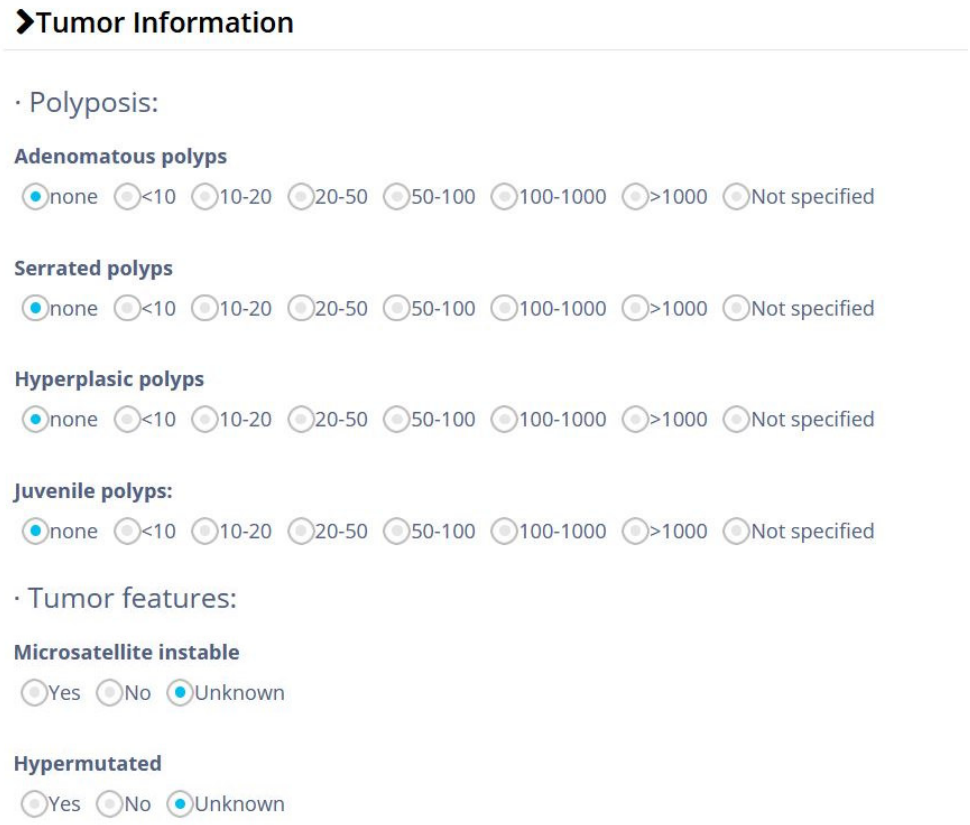
Browse the Human Phenotype Ontology (HPO)

Browse Categories

- Arteriopathies
- Connective Tissues Disorders and Aortopathies
- Cardiac arrhythmia
- Cardiomyopathy
- Congenital heart disease
- Lymphatic disorders
- Pulmonary heart disease



Finally, a specific field was created (see below) with collaboration of ERN-GENTURIS, which allows submitters to introduce numbers of developed tumors in participants, a very important feature to improve the analyses performed for ERN-GENTURIS diseases.



All these new features will be released in the new GPAP-PhenoStore platform. However, they are already accessible through the RD-Connect GPAP Phenostore Playground (https://playground.rd-connect.eu/phenostore/#/).

It is important to note that other forms outside Solve-RD project were also developed previously through other projects or under the EJP-RD project for other ERNs.

Conclusion:

We successfully introduced new phenotypic forms specific for diseases related to the ERNs working in the Solve-RD project. These new forms will facilitate the collation of the participants' HPOs and other disease-specific relevant information and will improve phenotypic description and consistency of the introduced traits across the project.