SOFTWARE ENGINEERING PROJECT

ON

CLINICAL DIAGNOSIS IOS APP – THE PHENOMIZER

SUBMITTED TO

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Abstract

In this report we describe the features of Phenomizer – The clinical diagnosis app and guide the user through the Phenomizer workflow for getting the differential diagnosis. In first section we illustrate how to find and enter the patient's clinical features. Then in the second section, we will show how to get the differential diagnosis using the patient's selected features.

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1. Disclaimer

The *Phenomizer* is intended for the qualified and licensed physicians as an aiding tool for reaching the correct diagnosis in patients with hereditary diseases and for use as a teaching aid. The *Phenomizer* does not make diagnoses, rather, it provides a ranked list of possibilities that can be used by the physicians in diagnosis. The *Phenomizer* does not contain information on all the possible diagnoses or even all potential hereditary diseases. The *Phenomizer* must not be used for making medical decisions without the consultation of a physician.

2. How to query the Phenomizer

The medical terms available to the *Phenomizer* are based on the nomenclature of the Human Phenotype Ontology (HPO) [1]. The *Phenomizer* offers two different methods to find the suitable term for a phenotypic aberration, shown by the two tabs on the bottom of the screen: *Features* and *Diseases.*

2.1 Entering medical data of a patient

Under the *Features* tab (Figure 1) HPO terms can be searched either by typing the name or the HPO-identifier into the search box. The search box offers auto-update functionality: possible matches are displayed while the query is typed in. After pressing the search icon, features of the HPO, best agreeing with the query are displayed in a table. An HPO term can added to the *Patient's features* list by simply clicking on it. Number of selected features is shown in the upper left corner of the screen.

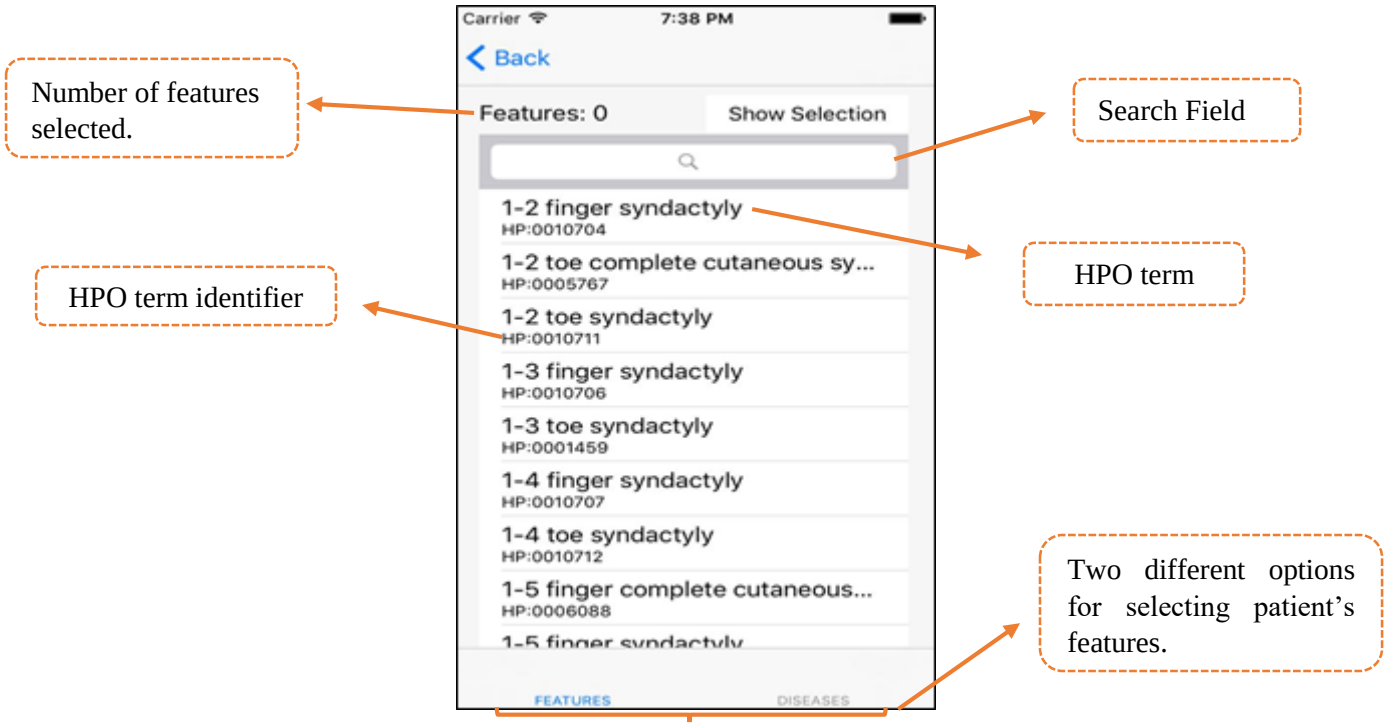


Figure 1: Selecting patient's clinical features from the *Features* screen.

1 http://www.human-phenotype-ontology.org

If your patient shares some phenotypic anomalies with a different genetic disorder you are aware of, the *Diseases* tab offers a quick way to select features (Figure 2). In this tab you search for a syndrome by typing in the name of syndrome in the search field. Selecting a syndrome will lead to a new screen displaying a list of all the features used to describe the syndrome. From here you can add features to the patient's features list. For unselecting a feature, simply click on the selected feature again and it will be unselected and removed from the patient's features list.

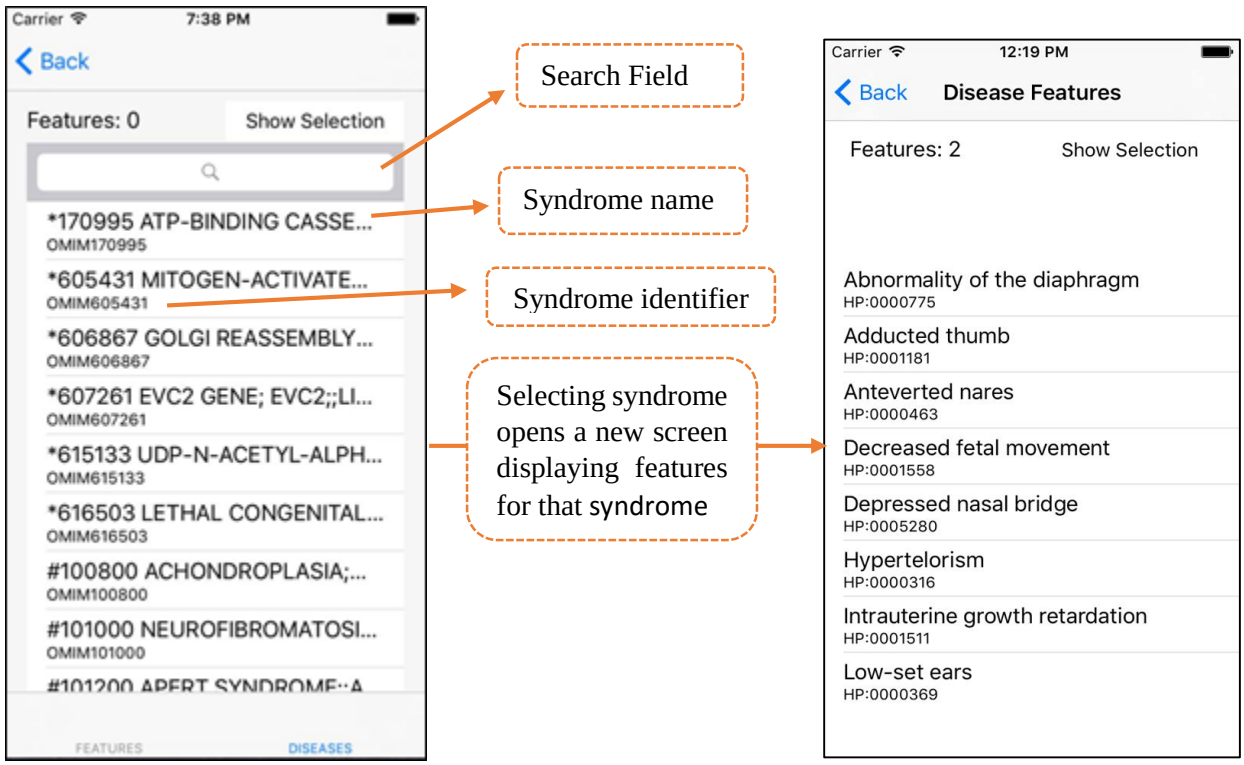


Figure 2: Selecting patient's features from diseases screen. Selecting a syndrome guides the user to a new screen displaying a list of features used to describe the syndrome. Features can be selected from this screen.

2.2 Patient's features

The features selected above can be viewed by selecting the *Show Selection* option. Generally, it is best to select as many clinical features of the patient as possible in order to increases the specificity of the differential diagnosis offered by the *Phenomizer*. Nevertheless, it is also possible to add features one by one for discovering the phenotypic space or to learn about the diseases that are categorized by cliques of phenotypic features.

2.2.1 Adding/deleting features

More features can be added to the current features list by directly going back (through back button) to the previous screen and selecting more features. Selected feature can be deleted by selecting delete option. Delete option is displayed when a feature is clicked (Figure 3.).

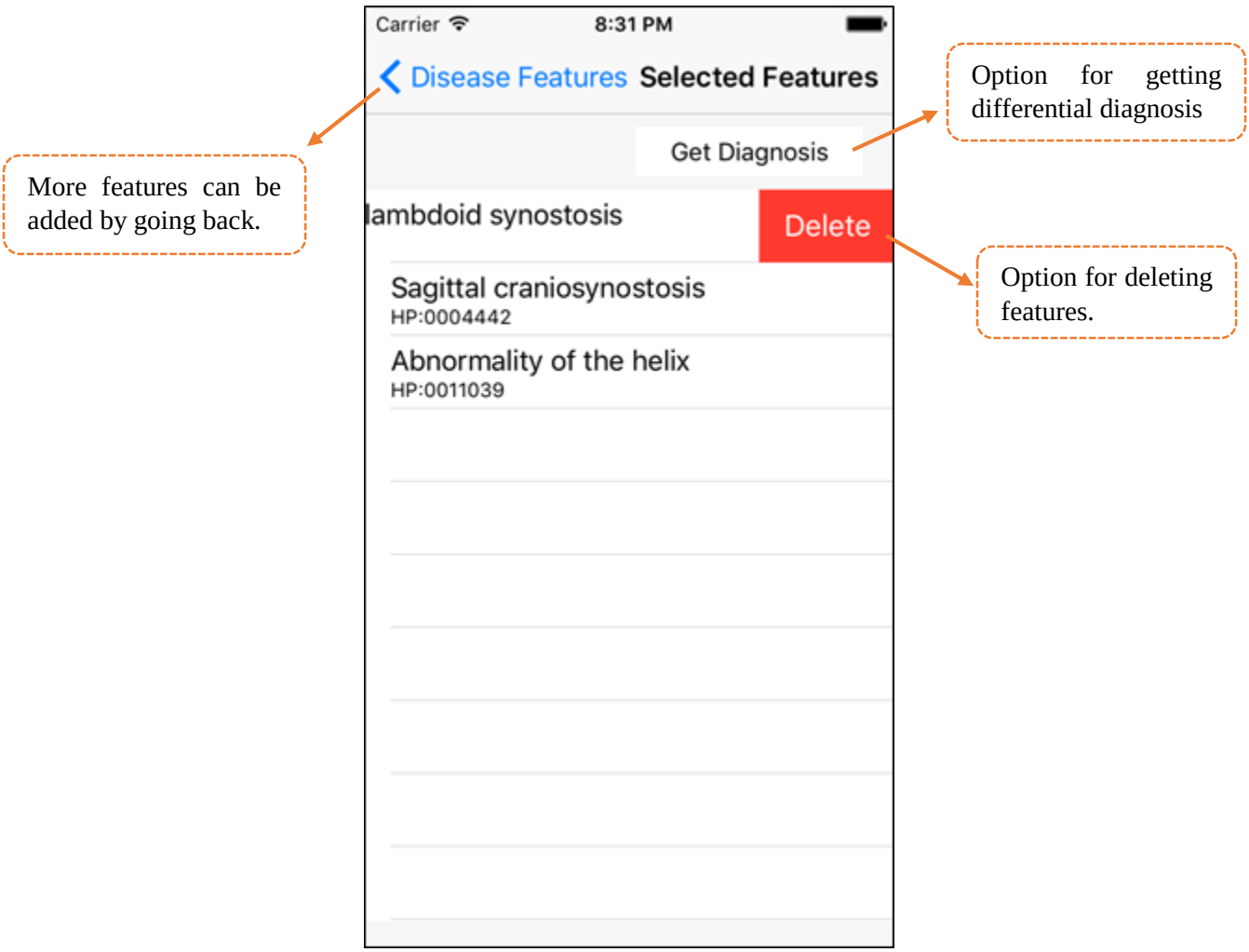
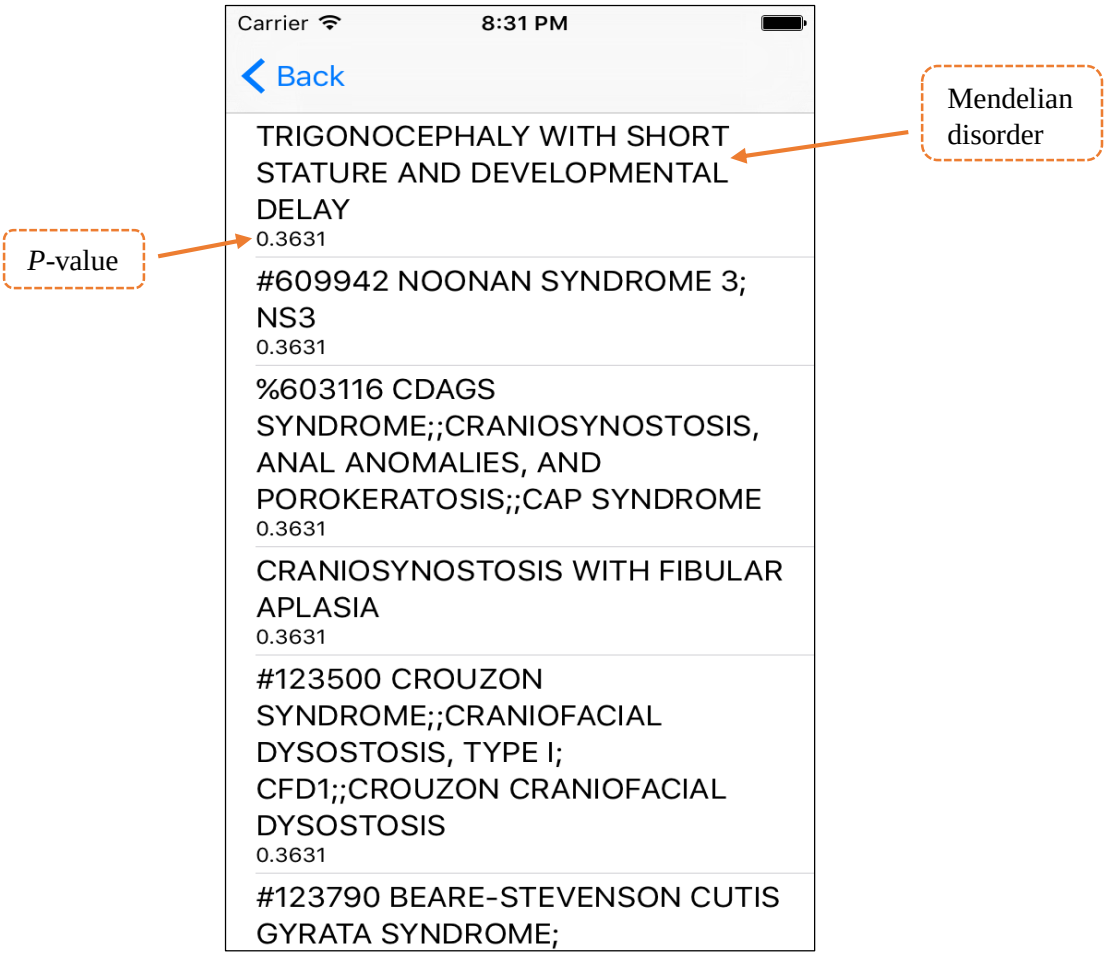


Figure 3: Show Selection option displays list of selected features with option to add more features or get diagnosis for the selected features.

3. Get diagnosis

Click the *Get Diagnosis* button for getting the Phenomizer to compute the list of a differential diagnoses ranked by their *P*-value (Figure 4.). A significant *P*-value does not mean that diagnosis is complete. We recommend that it can be taken to mean that diagnosis is at least reasonable and the physician should consider statistically significant diagnosis sensibly. On the contrary, if no disease is found to have a significant *P*-value for the features entered, we would take it to mean that the combination of the clinical features is not ample to make the diagnosis. It may also mean that the disease which the patient has is not in the database being used by the program (which presently comprises mainly the Mendelian diseases listed in OMIM).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TRIGONOCEPHALY WITH SHORT STATURE AND DEVELOPMENTAL DELAY
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0.3631
CRANIOSYNOSTOSIS WITH FIBULAR APLASIA
0.3631
#123500 CROUZON SYNDROME;;CRANIOFACIAL DYSOSTOSIS, TYPE I; CFD1;;CROUZON CRANIOFACIAL DYSOSTOSIS
0.3631
#123790 BEARE-STEVENSON CUTIS GYRATA SYNDROME;
Mendelian disorder
P-value