



SOFTWARE ENGINEERING PROJECT

ON

“CLINICAL DIAGNOSIS iOS APP - THE PHENOMIZER”

Submitted to

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PROJECT DESCRIPTION

The project is about building and implementing an iOS App "The phenomizer" to facilitate the disease prediction from a set of phenotypes. The App is intended for medical doctors and performs computational clinical diagnosis to return the results to the doctor. Through the App, the doctor can search or select one or more phenotype(s) from a list and these phenotype(s) are queried against a database of known diseases that are associated with the selected phenotype(s). The diseases are then ranked upon the likelihood of them being caused by the queried phenotype(s). Results are then displayed to the doctor.

MUST-HAVE¹

1. Welcome screen. The welcome screen will give the following options:
 - (a) Update Database
 - (b) New Query
 - (c) Help
2. Help page. This will guide the user through using the App.
3. New query feature allowing user to select new features and request the diagnosis.
4. Search option for searching the phenotype. Search will be applied in two ways:
 - (a) Search features: In the features screen, search can be performed either by name or HPO id of the phenotype.
 - (b) Search diseases: In the diseases screen, search can be performed by name of the disease.
5. Features screen. This will provide a list of features in lexicographically sorted order corresponding to the Human Phenotype Ontology (HPO).
6. Diseases screen. This will provide list of diseases in lexicographically sorted order available in the Phenomizer database.
7. Selected features screen. Provide a list of currently selected features with an option for removing a feature before proceeding with the diagnosis. From this screen a button "Perform Diagnosis" starts the diagnosis with the selected features.
8. Diagnosis screen. Here a list of all the diseases annotated to the selected features will be provided. The list will be sorted according to the p-value with the most related disease being listed at the top.
9. App will also allow users to view a disease's annotated features by selecting diseases in the diseases screen.
10. Update database. This option will allow the user to update their Phenomizer database to correspond with the most recent one on the web server. User will be notified if no internet connection is available for updating the database.

¹These functionalities would definitely be part of the App.

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11. User will be notified in the diagnosis screen if no internet connection is available to send the query and perform the diagnosis.

MAY-HAVE²

1. Auto-complete functionality when searching in features screen.
2. Updating the list with the matching features when input is changed in search box in the features screen.
3. Checking database version when the App starts. Notifying the user if a new version of database is available.
4. Enable encrypted communication with the web server.
5. Landscape viewing of the App.
6. Ontology screen. Navigate through the DAG of the ontology available in the Phenomizer database.
7. Selecting phenotypes from the ontology screen.
8. In the selected features screen, showing the path from where the phenotype was added i.e. whether the phenotype was added from features, diseases, or ontology screen.
9. Auto-complete functionality when searching in diseases screen.
10. Updating the list with the matching diseases when input is changed in search box in the diseases screen.
11. Option to save a query.
12. Load query screen. This will be reachable as an option from the welcome screen. Listing all the saved queries. Option to load or delete the already saved query(ies).
13. Loaded query screen. Listing of features saved in the query and possibly option to update the query.

MUST-NOT-HAVE³

1. Anything neither present in the Phenomizer Android App nor expressly described in the Must-Have or May-Have section of this document.
2. Ontology screen, any visualization that is more complicated than traversing the ontology DAG through sequence of clicks.
3. Manual update of data from user in any way except through the update database function.

²These functionalities may be part of the App if time permits.

³These functionalities will not be a part of the App.

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4. Storing/saving diagnosis results.
 5. Storing saved queries in encrypted format.
 6. Sending results through e-mail.
 7. Searching on features and diseases by using synonyms.
 8. Any additional algorithm, e.g. locality based filtering of diseases.
 9. Providing statistical information about user's activity.
 10. Providing a billing functionality.

Dr. Marcel Schulz

M. Schulz 27.11.15

(signature & date)

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(signature & date)