PedAM Guide

Contents

1. The function of PedAM

Our website PedAM provides three functions: disease query, disease network and submit case.

In disease query functional module, we offer standardized 8,528 pediatric disease terms (containing 4,542 unique disease concepts and 3,986 synonyms) with 8 annotation fields for each disease, including definition synonyms, gene, symptom, cross-reference (Xref), human phenotypes, and its corresponding phenotypes in the mouse (MPO).

In disease network functional module, users can draw the disease network through query their interested disease pair. Currently, we provide three disease network models to users, they are disease-phenotype network, disease-gene network and disease pair network. Users can choose one of them to draw the network they are interested in.

In submit case functional module, we provide a way to upload medical records. We will select some cases with the comprehensive clinical data of patient's pedigree and provide free service for related disease diagnosis, which eventually will improve treatments and better medical services to patients. We will keep strictly confidential for those submitted data. We greatly appreciate people's contribution for their support and cooperation.

1.1 Disease query

First, users should click the “QUERY” button in the navigation bar on the home page (Fig.1).

Fig.1 “QUERY” button.

Then the following page will show up (Fig.2):

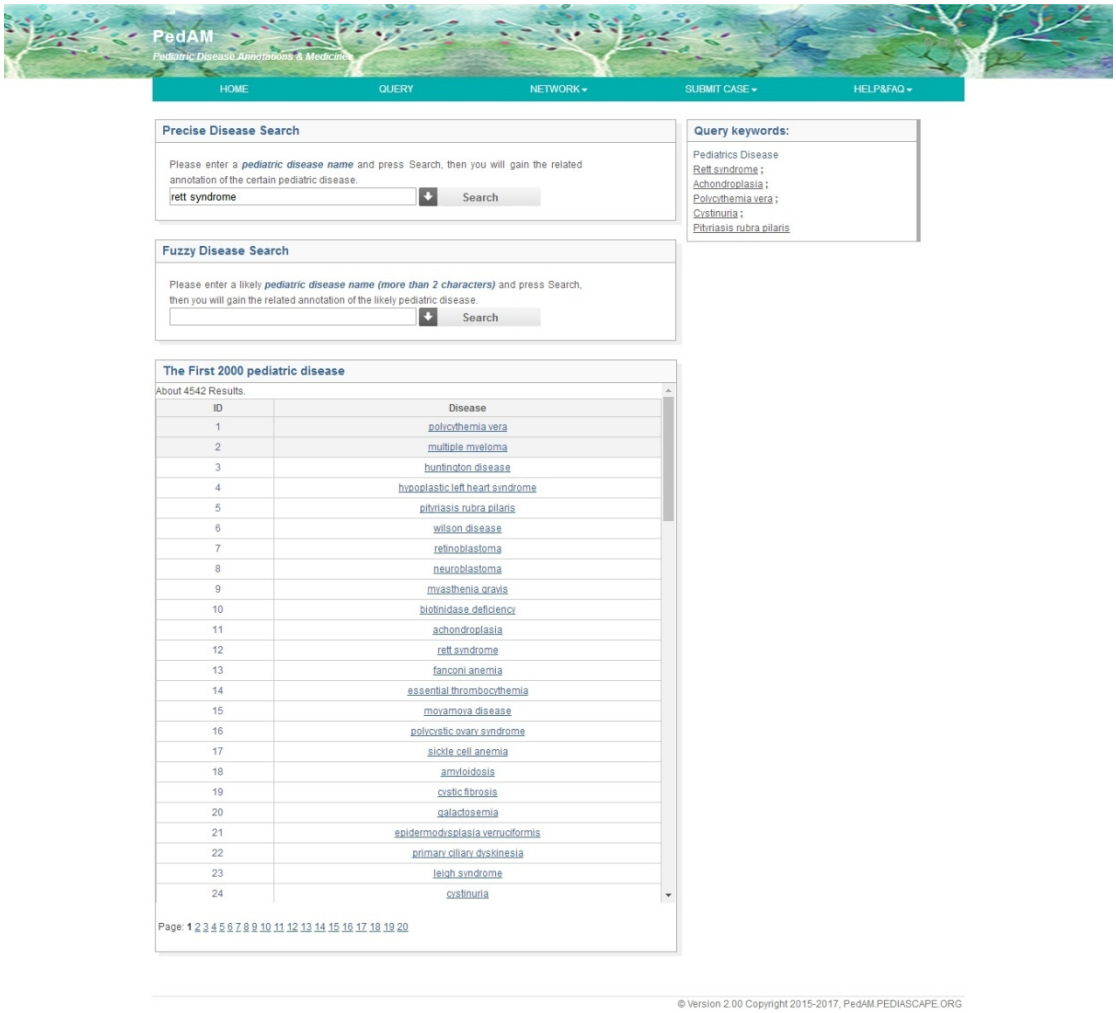

Fig.2 “QUERY” page.

On this page, there are four modules: precise disease search, fuzzy disease search, the first 2000 pediatric diseases and query keywords. Precise disease search and fuzzy disease search are presented to meet different inquiry demands.The first 2000 pediatric diseases provide 2000 most common pediatric disease for reference. Query keywords provides 5 of the most frequently queried keywords for reference.

Here, let's take the illness "Wilson disease" as an example to introduce a specific disease page (Fig.3).

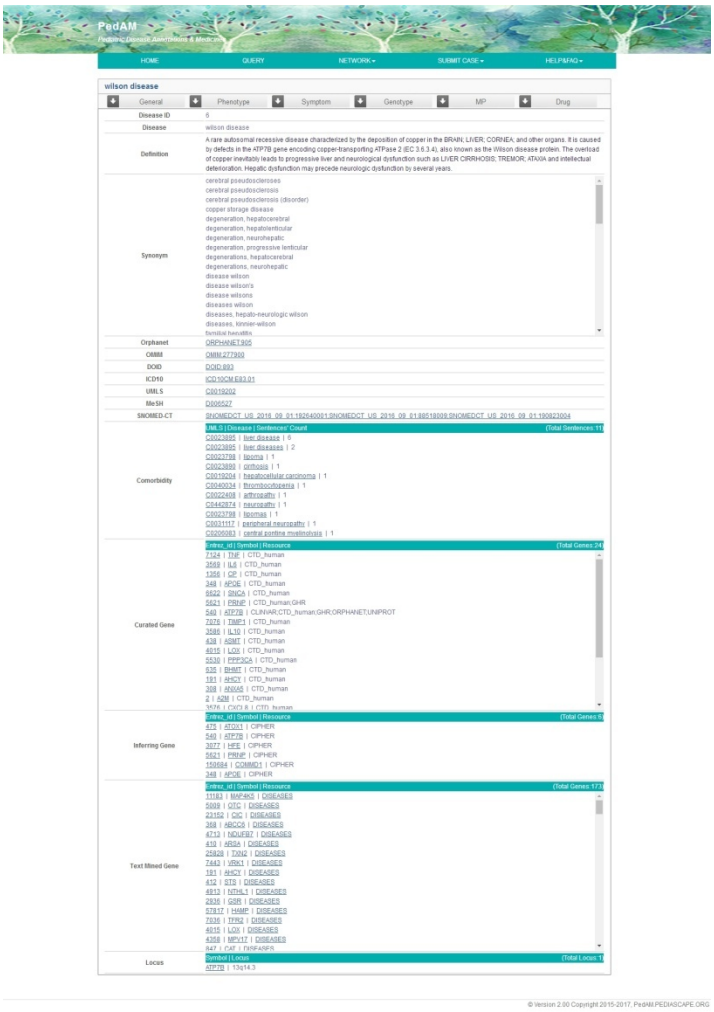

Fig.3 Wilson disease page.

There are three ways to get to the page of ‘willson disease’. First, users

can input “Wilson disease” in the precise disease search; second, users can input “Wilson” in the fuzzy disease search, but users can only get a simple page about this disease with this way; third, users can click the “Wilson disease” in the list of the first 2000 pediatric diseases.

On the “Wilson disease” page, there are six buttons: “General”, “Phenotype”, “Symptom”, “Genotype”, “MP” (Fig.4) and “Drug” that each contains the information of phenotype, symptom, genotype, MPO and Drug, respectively. Clicking the “General” button can display the whole page.

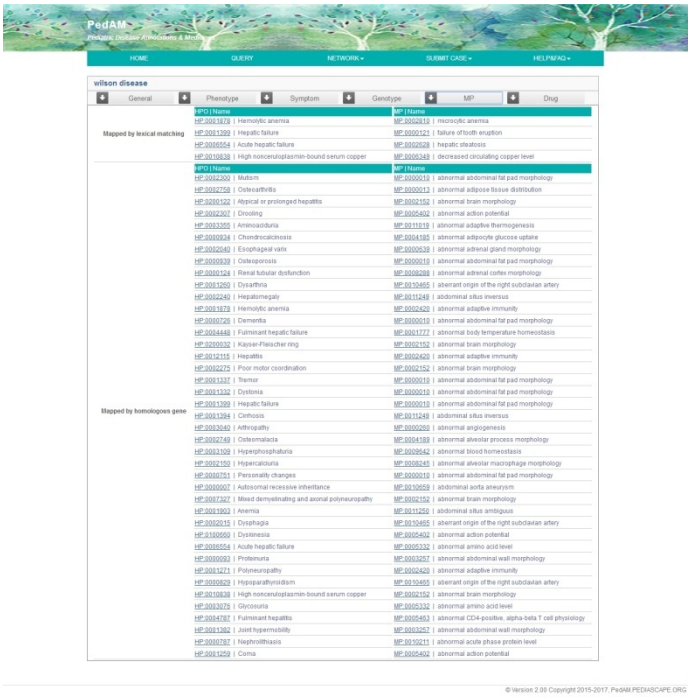

Fig.4 MP of Wilson disease

1.2 Disease network

First, users need move the cursor to the “NETWORK” button in the

navigation bar on the home page (Fig.5).

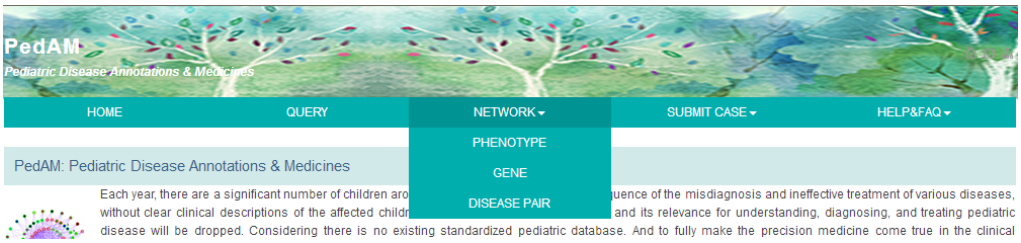

Fig.5 Network button

Our website provides three different networks to draw, they are follows:

1.2.1 Phenotype network

Users can get to the phenotype network page by clicking the 'PHENOTYPE' button (Fig.6).

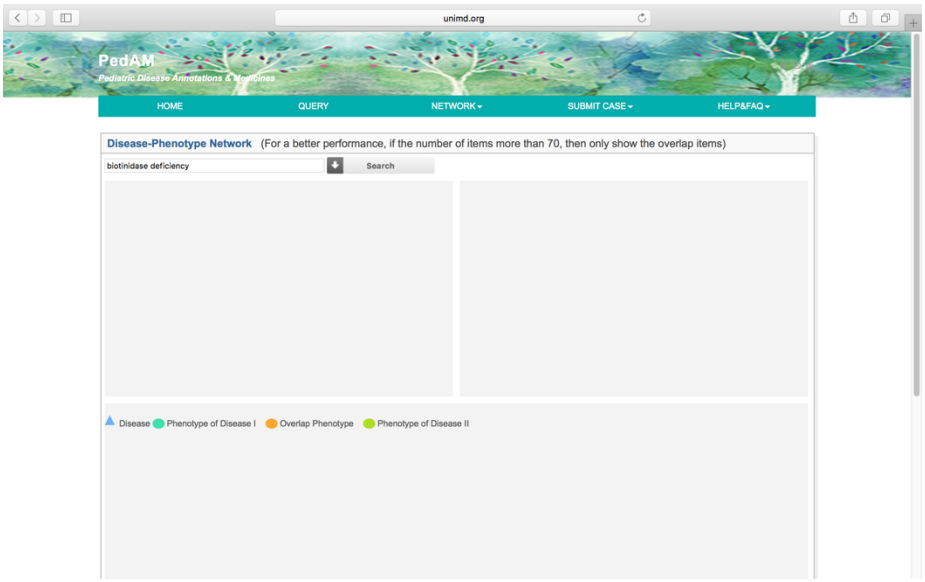

Fig.6 Phenotype network page

On this page, users can query their interested disease with 'Search' button (Fig.7).

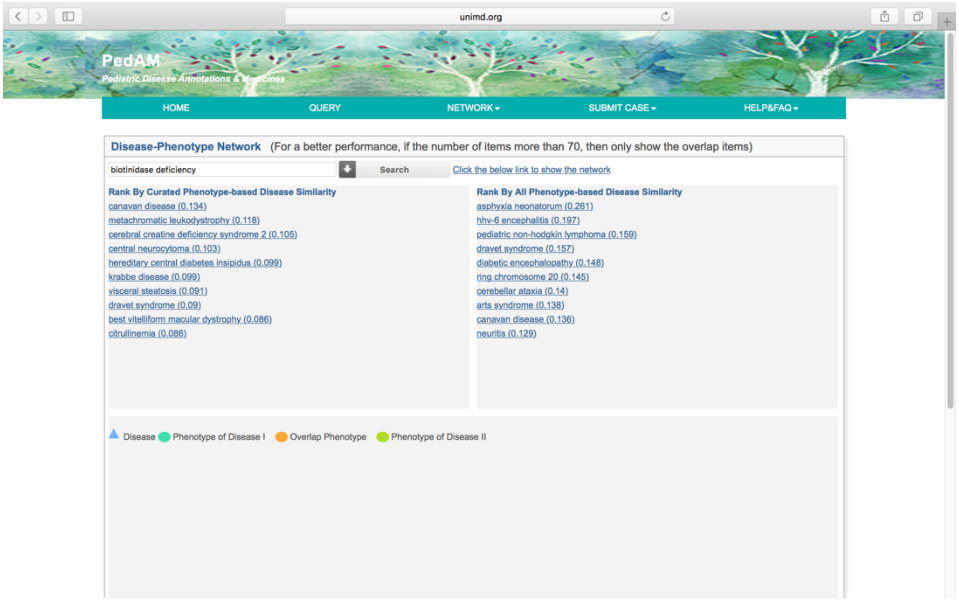

Fig.7 Biotinidase deficiency page

The returned search results contain two parts: the diseases ranked by curated phenotype-based disease similarity displayed on the left of the webpage and the disease ranked by all phenotype-based disease similarity displayed on the right of the webpage. Users can choose the entry for the phenotype they want to research.

After users click an entry in the two lists, the disease-phenotype network will be displayed at the bottom half of the webpage (Fig.8).

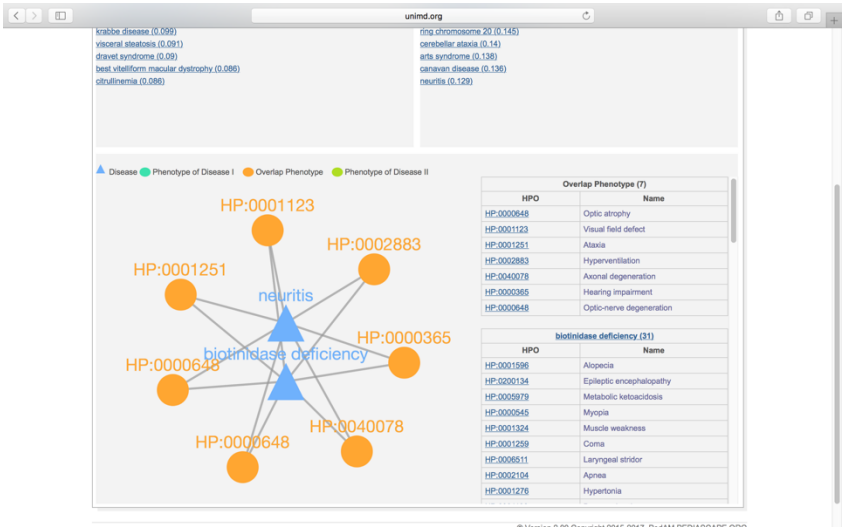

Fig.8 Network forbiotinidase deficiency and neuritis

1.2.2 Gene network

Clicking the 'GENE' button will lead users into gene network page (Fig.9).

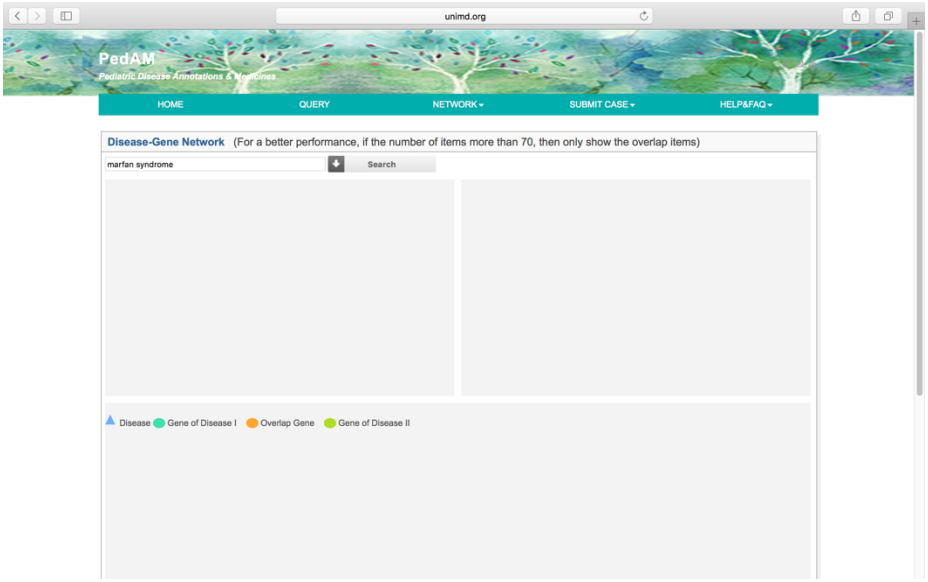

Fig.9 Gene network page

On this page, users can query their interested disease with 'Search' button (Fig.10).

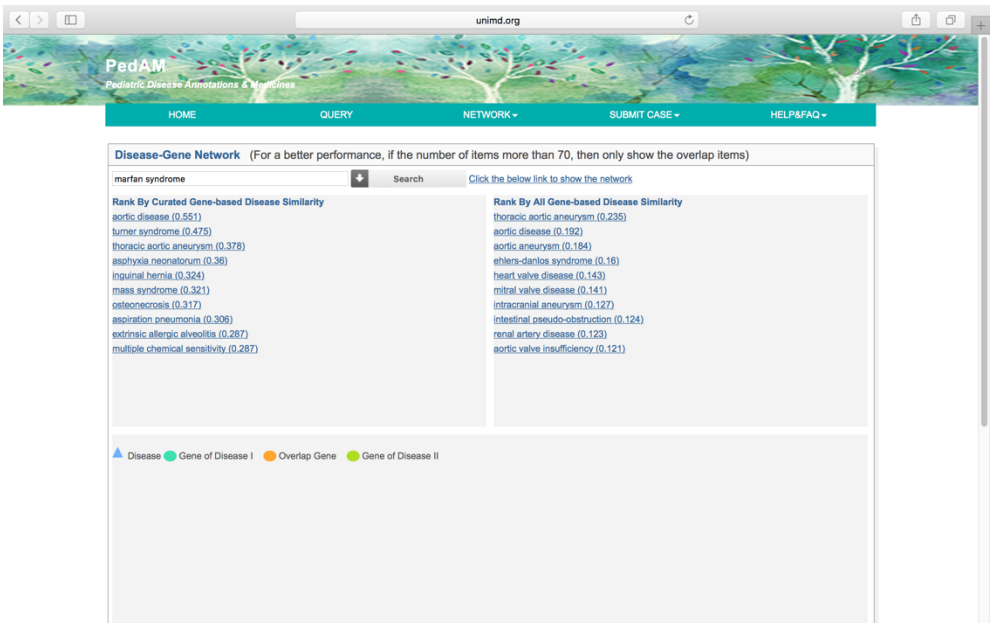

Fig.10 Marfan syndrome page

The returned search results contain two parts: the diseases ranked by curated gene-based disease similarity displayed on the left of the

webpage and the disease ranked by all gene-based disease similarity displayed on the right of the webpage. Users can choose the entry for the gene they want to research.

After users click an entry in the two lists, the disease-gene network will be displayed at the bottom half of the webpage (Fig.11).

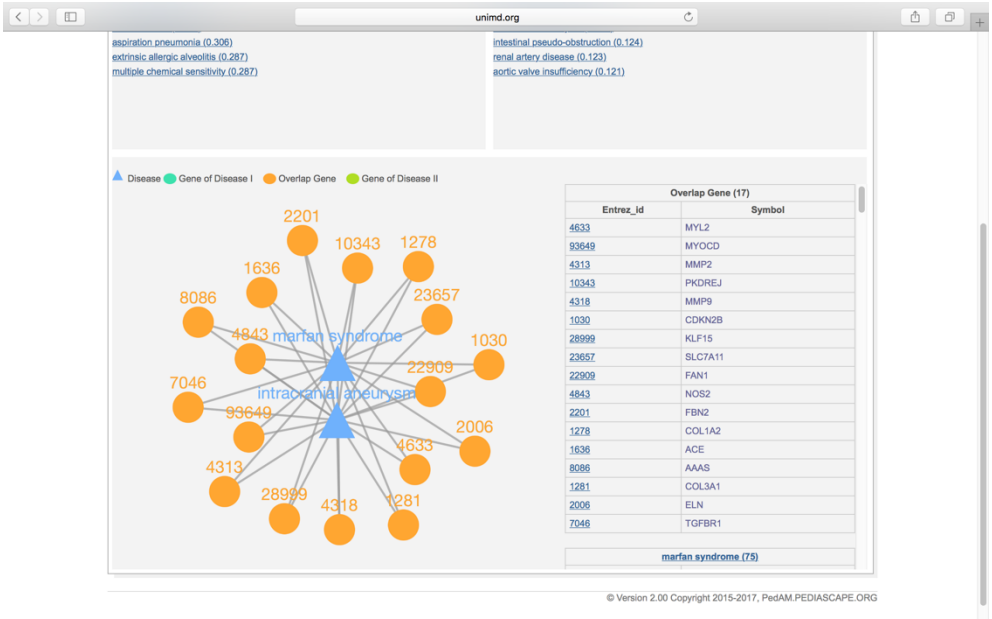

Fig.11 Network for marfan syndrome and intracranial aneurysm

1.2.3 Disease Pair Network

The “DISEASE PAIR” button will let users get into the disease pair network page (Fig.12).

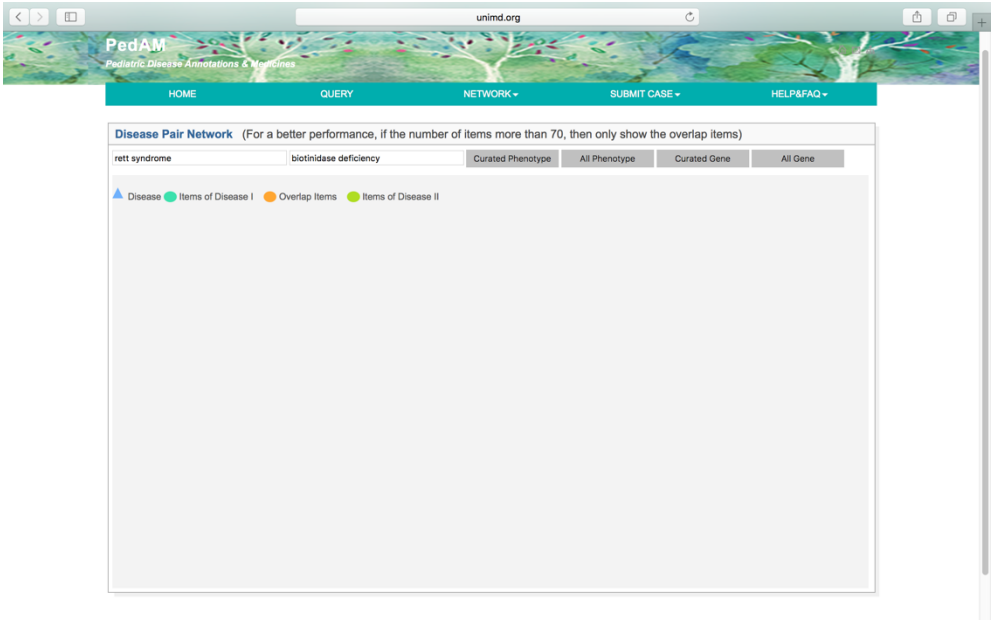

Fig.12 Disease pair network page

There are two search boxes on this page, users can input their interested disease pair in those two boxes. Then users can click each of the four buttons on the right of those two boxes to view the network that they are interested in. The button “Curated phenotype” will show the curated phenotypes that are related to the disease pair. The button “All phenotype” will show the overlapped phenotypes that related to the disease pair.

Button “Curated gene” will show the curated genes that are related to the disease pair. The button “ALL GENE” will show the overlapped genes that are related to the disease pair (Fig.13).

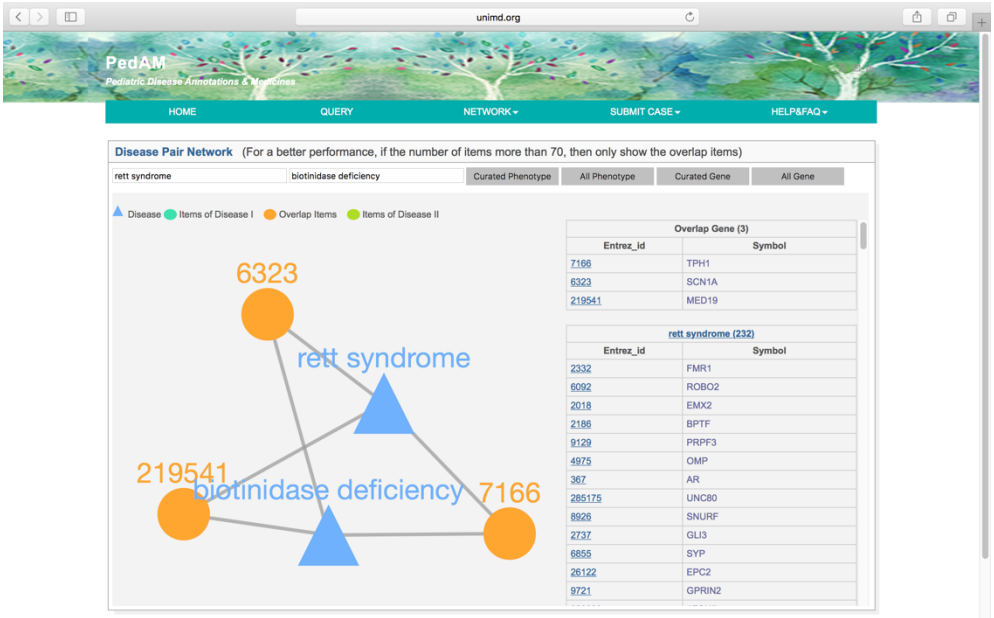

Fig.13 Overlapped genes of rett syndrome and biotinidase deficiency

1.3 Submit case

Clicking the “SUBMIT CASE” button in the navigation box below on the home page will open a new page shown as Fig.15.

Fig.14 Submit case button

Then the following page will show up (Fig.15):

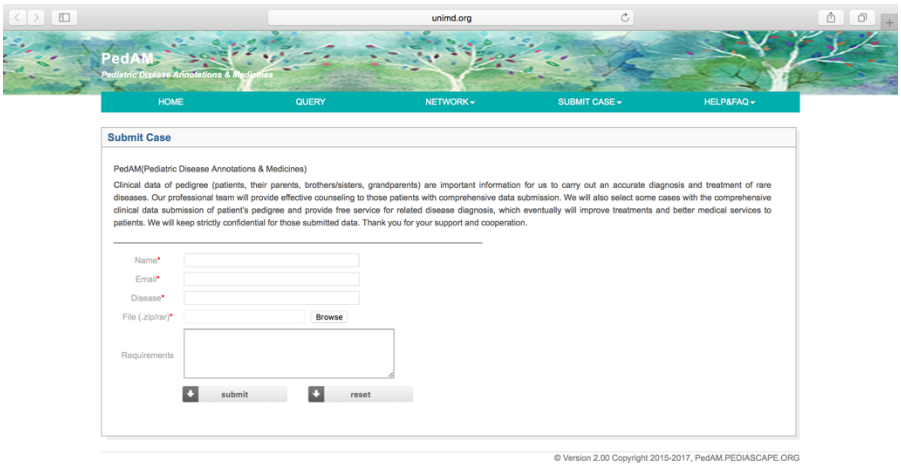

Fig.15 Submit case page

Users can upload their cases into our system through this page. The users must input your name, E-mail, disease and case files and the requirements bar is an option part.

Users can click the “Browse” button to choose the file she/he wants to upload.

2. Contact information

If users have any questions about the use of the website, please contact us with the E-mail: pedam_web@126.com.