About PubCaseFinder

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OVERVIEW

PubCaseFinder (https://pubcasefinder.dbcls.jp) is a phenotype-driven differential diagnosis system that helps clinicians rank rare diseases by comparing signs and symptoms of a patient against those of diseases defined in Orphanet. Users can narrow down the ranked list of rare diseases by specifying the causative genes of rare diseases. Also, the user can compare signs and symptoms of a patient against those included in published case reports to get a ranked list of published case reports. To keep up with new data, PubCaseFinder is equipped with an automatic update system.

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1. DISCLAIMER

The PubCaseFinder is intended to be used by qualified and licensed physicians to assist in reaching the correct diagnosis in patients with hereditary diseases and for use as a teaching aid. The PubCaseFinder does not make diagnoses. Instead, it produces a ranked list of possibilities that can be used by physicians as a part of the diagnostic workup. The PubCaseFinder does not contain information about all possible diagnoses or even all possible hereditary diseases. The PubCaseFinder should not be used to make medical decisions without the advice of a physician. PubCaseFinder provides data in good faith as an investigative tool, but without verifying the accuracy, clinical validity or utility of the data. PubCaseFinder makes no warranty, express or implied, nor assumes any legal liability or responsibility for any purpose for which the data are used. DBCLS shall bear no liability concerning damages incurred by the change, discontinuation or termination of the Service.
2. GETTING DIFFERENTIAL DIAGNOSES

Inputting patient’s signs and symptoms

You can search for the appropriate HPO terms by typing part of the name or HPO ID into the search box. This search box provides autocompletion functionality: Possible matches are shown below, while the query is typed in (Figure 1). If you click the box including a sign or symptom, its detailed information appears on the modal dialog (Figure 2).

![Figure 1](image1.png)

![Figure 2](image2.png)
Getting differential diagnoses

After inputting signs and symptoms, you can get a ranked list of rare diseases by clicking the ‘Get diagnosis’ button (Figure 3). You can get a ranked list of rare diseases by comparing signs and symptoms of a patient against those of diseases defined in Orphanet, and the top-listed diseases represent the most likely differential diagnosis (Figure 4). The matched phenotypes and causative genes are shown in each row. The matched phenotypes mean that those phenotypes are the best matches for the query phenotypes base on the semantic similarity. You can download the results by clicking the “Download Results” button. If you click the box including a phenotype in each row (Figure 4), you can confirm the detailed contextual information which includes both the phenotype and disease in published case reports (Figure 5).
By inputting gene symbols into the search box, you can narrow down the 4,066 diseases to the diseases whose causative genes are the user-specified genes. This search box also provides autocompletion functionality: Possible matches are shown below, while the query is typed in (Figure 6).

After inputting gene symbols, you can narrow down the ranked list of rare diseases by clicking the ‘Get diagnosis’ button (Figure 7). Figure 7 shows that 4,066 diseases have narrowed down to 14 diseases.

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**Figure 5**

**Narrowing down a ranked list of rare diseases**

By inputting gene symbols into the search box, you can narrow down the 4,066 diseases to the diseases whose causative genes are the user-specified genes. This search box also provides autocompletion functionality: Possible matches are shown below, while the query is typed in (Figure 6).

After inputting gene symbols, you can narrow down the ranked list of rare diseases by clicking the 'Get diagnosis' button (Figure 7). Figure 7 shows that 4,066 diseases have narrowed down to 14 diseases.
By using the uploading function, you can quickly input a list of phenotypes (HPO ID) or genes (Entrez Gene ID). In Figure 7, there are two links of “Upload File (HPO ID List)” and “Upload File (Entrez Gene ID List).” You can upload a file including a list of phenotypes (Figure 8) or genes (Figure 9) by clicking them. The comma-separated values format is supported.

**Figure 7**

**Uploading a list of phenotypes or genes**

By using the uploading function, you can quickly input a list of phenotypes (HPO ID) or genes (Entrez Gene ID). In Figure 7, there are two links of “Upload File (HPO ID List)” and “Upload File (Entrez Gene ID List).” You can upload a file including a list of phenotypes (Figure 8) or genes (Figure 9) by clicking them. The comma-separated values format is supported.
3. GETTING A RANKED LIST OF CASE REPORTS

After getting a ranked list of rare diseases, you can check the link for getting a ranked list of published case reports in each row (Figure 7). By clicking the link, a ranked list of published case reports related to the rare disease are shown by comparing signs and symptoms of a patient against those included in published case reports (Figure 10). The matched phenotypes, genes, and mutations included in the published case report are shown in each row. Also, MeSH terms are shown in each row, and you can narrow down published case reports by inputting MeSH term in the box “Narrow down the case reports.” You can download the results by clicking the “Download Results” button. If you click the tabs of “Phenotypes” or “Causative Genes,” you can check the phenotypes or causative genes related to the rare disease (Figure 10).

![Figure 10](image-url)