USER GUIDE

DATABASE CENTER FOR LIFE SCIENCE,
JOINT SUPPORT-CENTER FOR DATA SCIENCE RESEARCH,
RESEARCH ORGANIZATION OF INFORMATION AND SYSTEMS

About PubCaseFinder

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OVERVIEW

PubCaseFinder is a search system for medical professionals to search genetic diseases, rare diseases, causative genes, and published cases in order of relevance by using symptoms and signs as queries.

CONTENTS

1. DISCLAIMER

The PubCaseFinder is intended to be used by qualified and licensed physicians to assist in the research about rare and genetic diseases and for use as a teaching aid. 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. HOW TO SEARCH DISEASES

1) Inputting 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

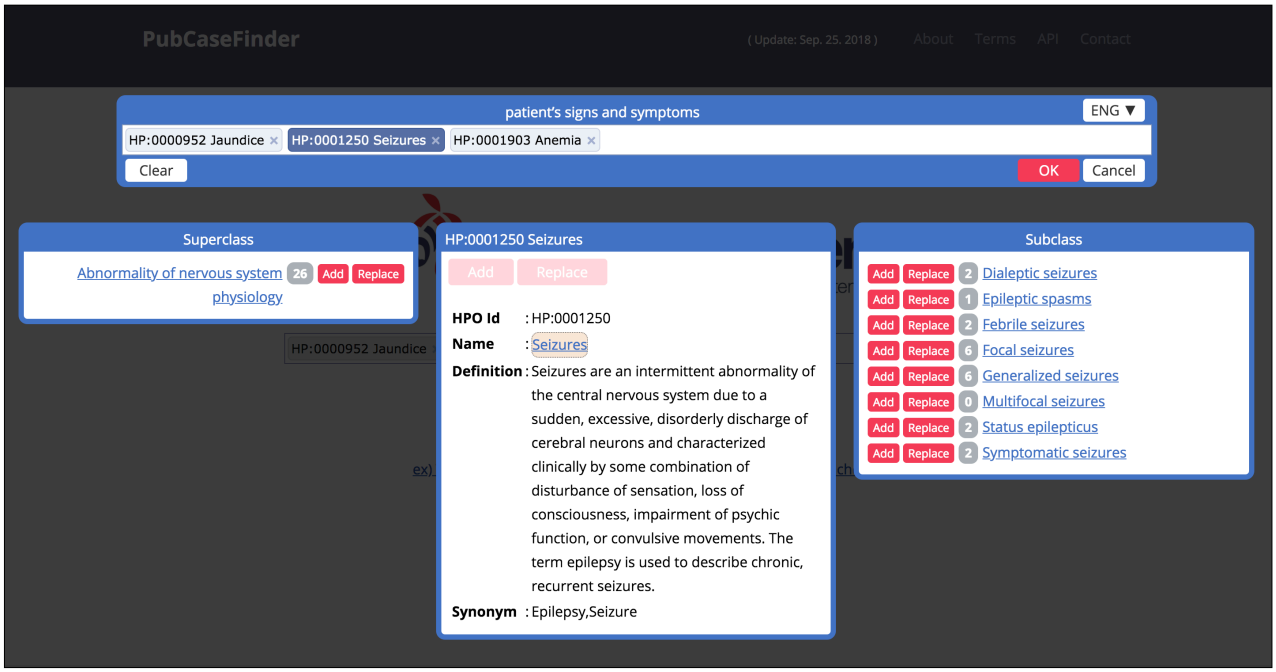


Figure 2

2) Searching rare and genetic diseases

After inputting signs and symptoms, you can get a ranked list of rare and genetic diseases by clicking the 'Find diseases' button (Figure 3). You can get a ranked list of rare diseases defined in Orphanet (Figure 4A) and genetic diseases defined in OMIM (Figure 4B) by comparing signs and symptoms of an affected individual against those of diseases defined in Orphanet and OMIM. 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 summary of results which includes top 10 diseases of both Orphanet and OMIM by clicking the "Download Summary of 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).

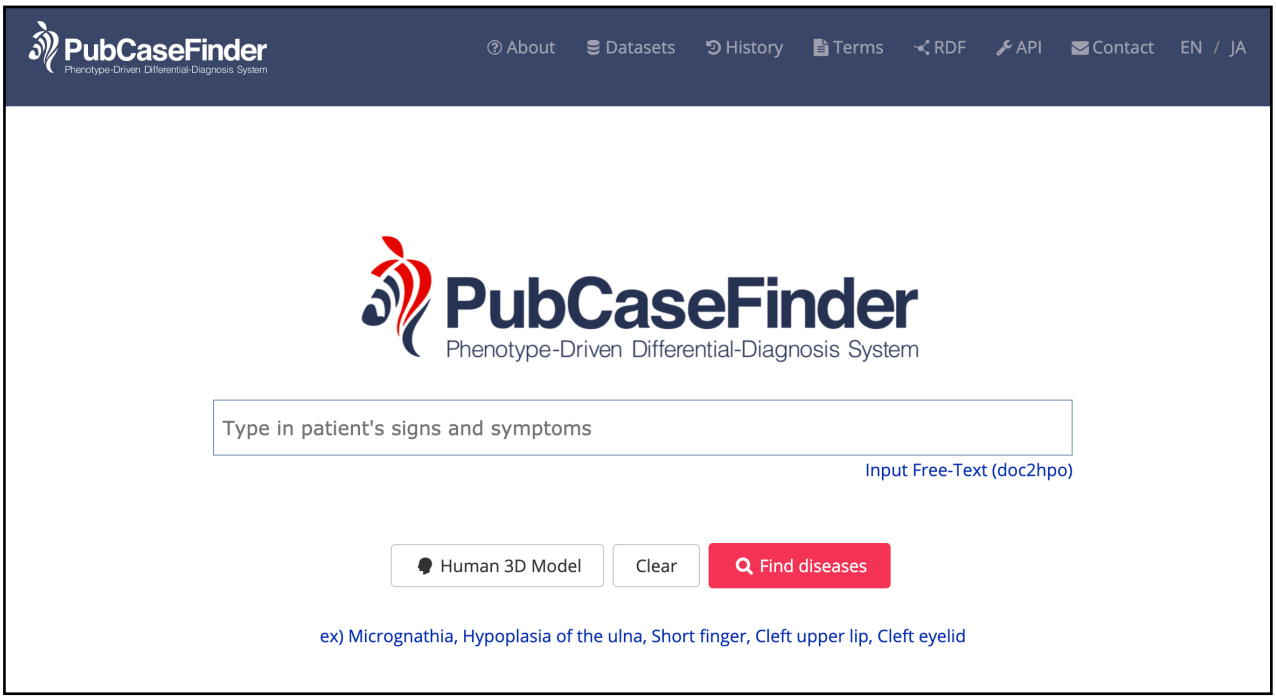


Figure 3

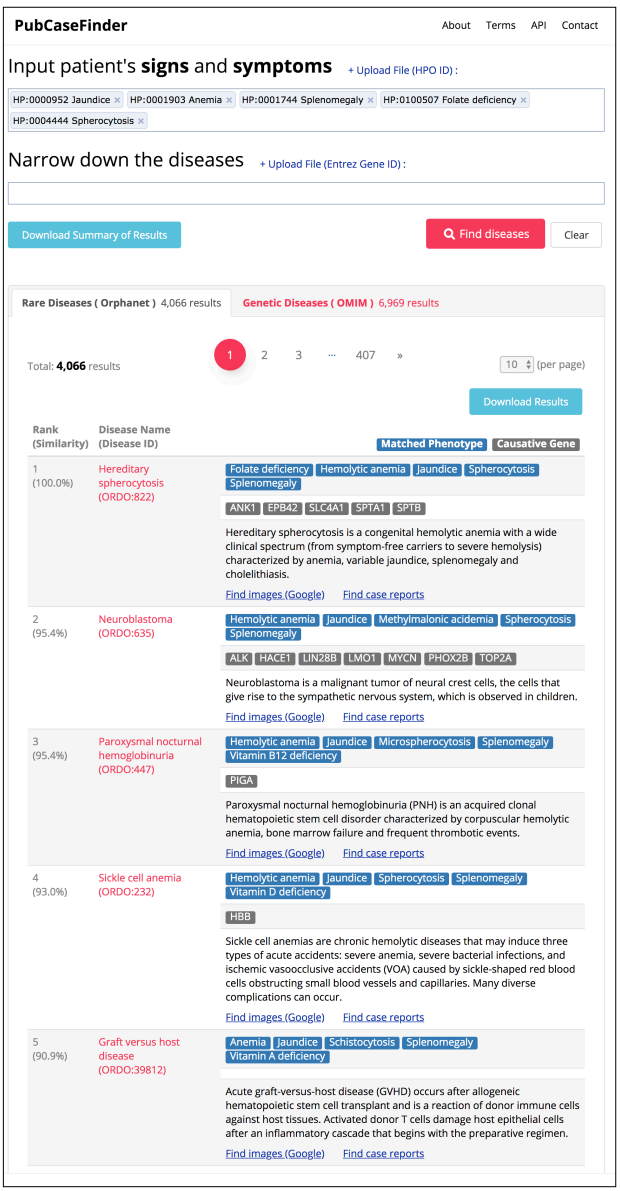


Figure 4A

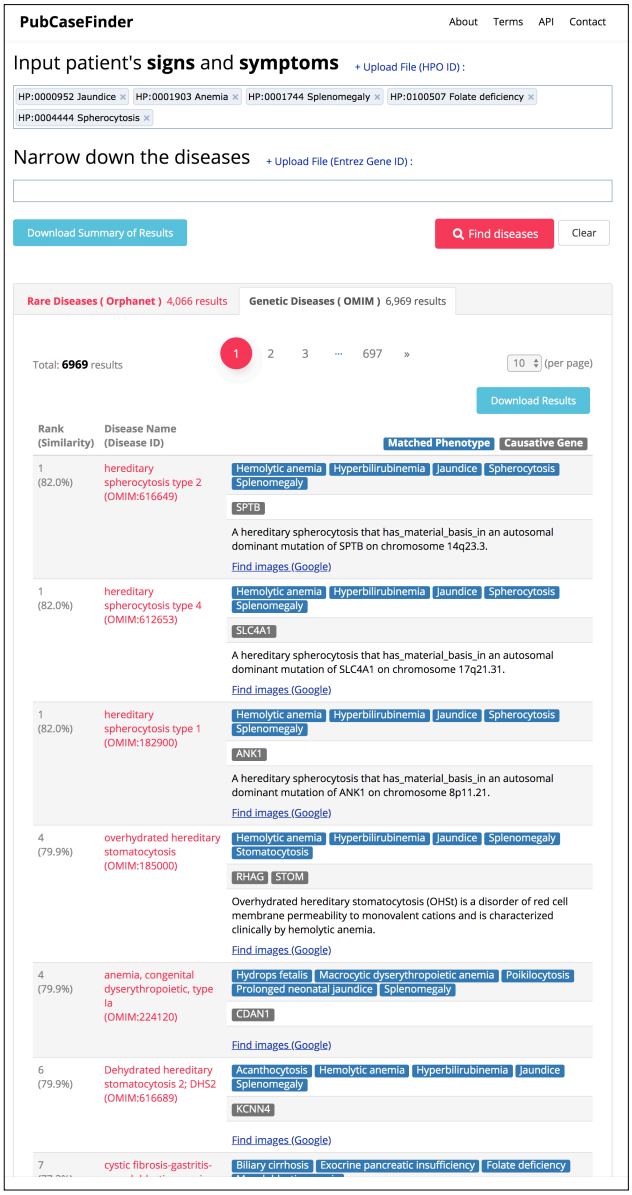
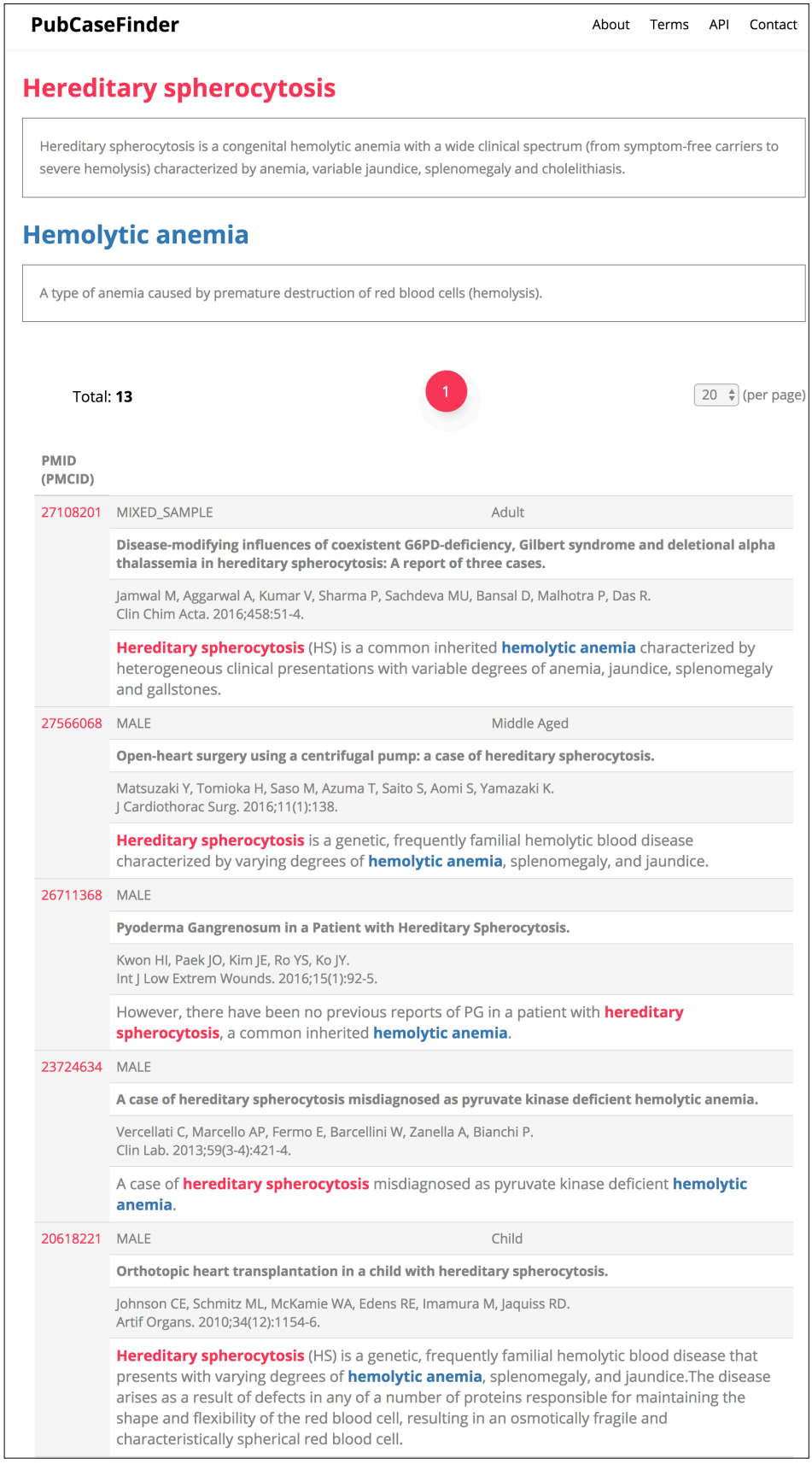


Figure 4B


PubCaseFinder
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Hereditary spherocytosis
Hereditary spherocytosis is a congenital hemolytic anemia with a wide clinical spectrum (from symptom-free carriers to severe hemolysis) characterized by anemia, variable jaundice, splenomegaly and cholelithiasis.
Hemolytic anemia
A type of anemia caused by premature destruction of red blood cells (hemolysis).
Total: 13
1
20 (per page)
PMID (PMCID)
27108201 MIXED_SAMPLE Adult
Disease-modifying influences of coexistent G6PD-deficiency, Gilbert syndrome and deletional alpha thalassemia in hereditary spherocytosis: A report of three cases.
Jamwal M, Aggarwal A, Kumar V, Sharma P, Sachdeva MU, Bansal D, Malhotra P, Das R.
Clin Chim Acta. 2016;458:51-4.
Hereditary spherocytosis (HS) is a common inherited hemolytic anemia characterized by heterogeneous clinical presentations with variable degrees of anemia, jaundice, splenomegaly and gallstones.
27566068 MALE Middle Aged
Open-heart surgery using a centrifugal pump: a case of hereditary spherocytosis.
Matsuzaki Y, Tomioka H, Saso M, Azuma T, Saito S, Aomi S, Yamazaki K.
J Cardiothorac Surg. 2016;11(1):138.
Hereditary spherocytosis is a genetic, frequently familial hemolytic blood disease characterized by varying degrees of hemolytic anemia, splenomegaly, and jaundice.
26711368 MALE
Pyoderma Gangrenosum in a Patient with Hereditary Spherocytosis.
Kwon HI, Paek JO, Kim JE, Ro YS, Ko JY.
Int J Low Extrem Wounds. 2016;15(1):92-5.
However, there have been no previous reports of PG in a patient with hereditary spherocytosis, a common inherited hemolytic anemia.
23724634 MALE
A case of hereditary spherocytosis misdiagnosed as pyruvate kinase deficient hemolytic anemia.
Vercellati C, Marcello AP, Fermo E, Barcellini W, Zanella A, Bianchi P.
Clin Lab. 2013;59(3-4):421-4.
A case of hereditary spherocytosis misdiagnosed as pyruvate kinase deficient hemolytic anemia.
20618221 MALE Child
Orthotopic heart transplantation in a child with hereditary spherocytosis.
Johnson CE, Schmitz ML, McKamie WA, Edens RE, Imamura M, Jaquiss RD.
Artif Organs. 2010;34(12):1154-6.
Hereditary spherocytosis (HS) is a genetic, frequently familial hemolytic blood disease that presents with varying degrees of hemolytic anemia, splenomegaly, and jaundice.The disease arises as a result of defects in any of a number of proteins responsible for maintaining the shape and flexibility of the red blood cell, resulting in an osmotically fragile and characteristically spherical red blood cell.

Figure 5

3) Narrowing down rare and genetic diseases

By inputting gene symbols into the search box, you can narrow down the 4,066 rare diseases and 6,969 genetic 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 'Find diseases' button (Figure 7). Figure 7 shows that 4,066 rare diseases have narrowed down to 3 diseases, and 6,969 genetic diseases have narrowed down to 2 diseases.

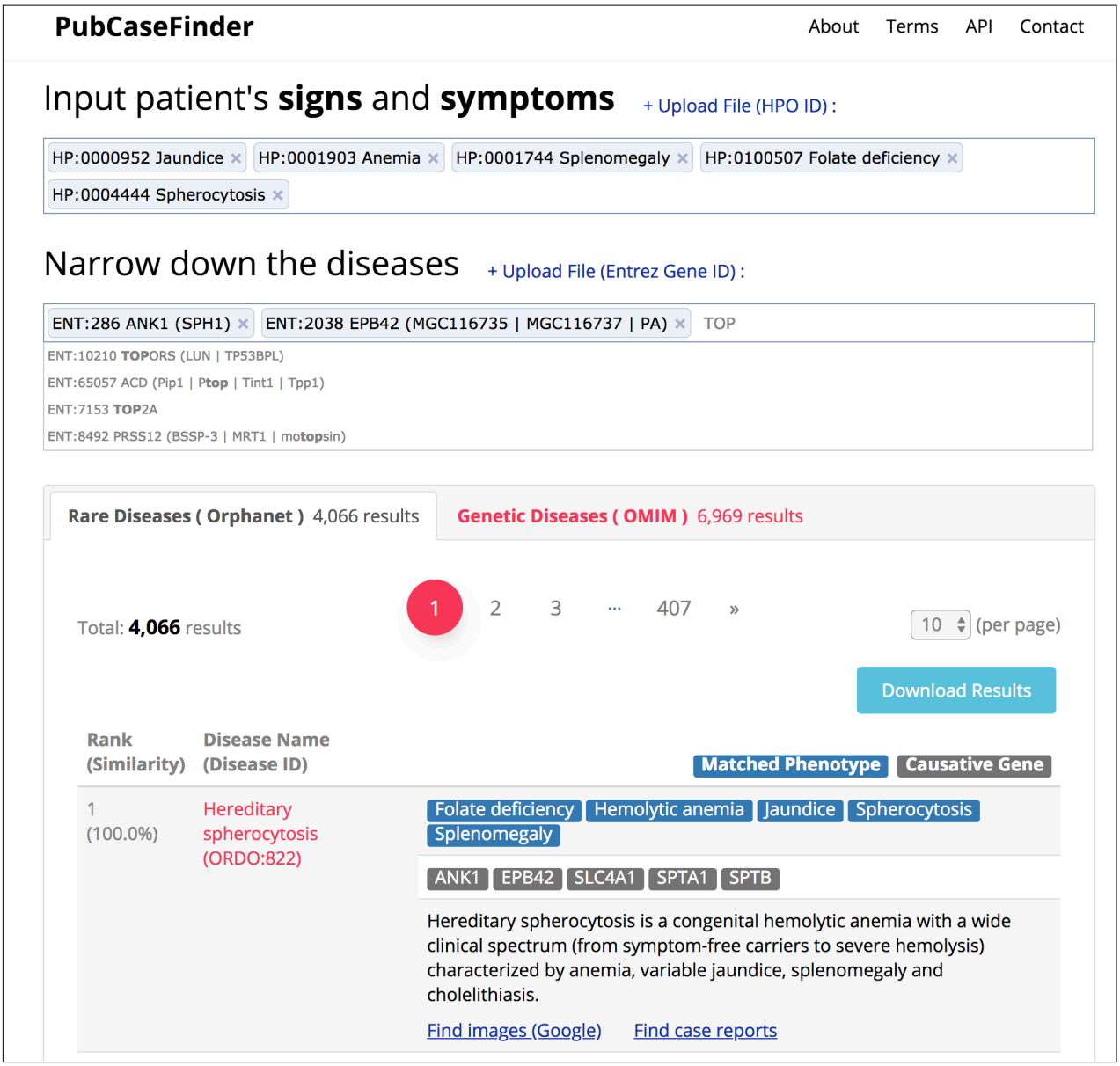
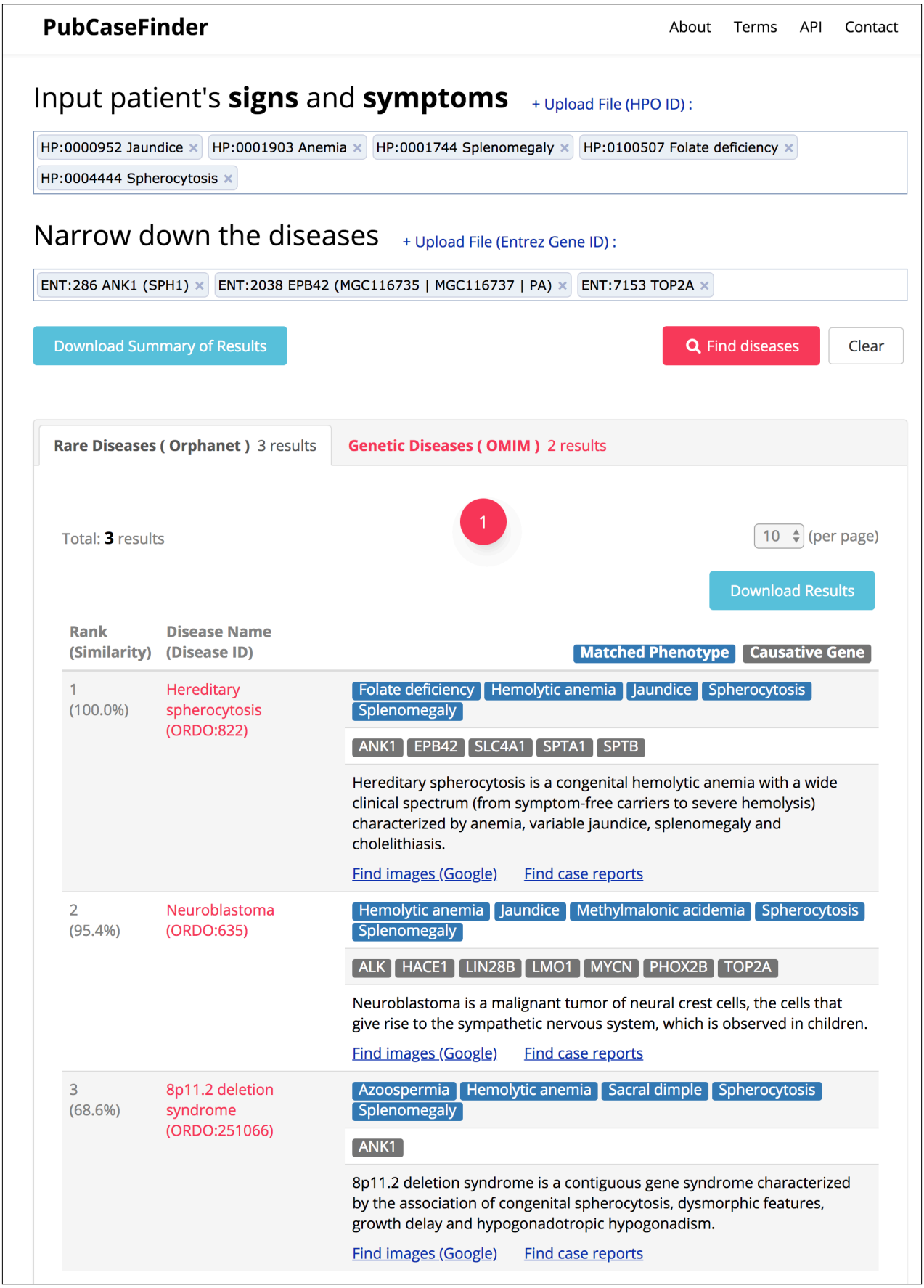


Figure 6


PubCaseFinder
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Input patient's signs and symptoms
+ Upload File (HPO ID) :
HP:0000952 Jaundice
HP:0001903 Anemia
HP:0001744 Splenomegaly
HP:0100507 Folate deficiency
HP:0004444 Spherocytosis
Narrow down the diseases
+ Upload File (Entrez Gene ID) :
ENT:286 ANK1 (SPH1)
ENT:2038 EPB42 (MGC116735 | MGC116737 | PA)
ENT:7153 TOP2A
Download Summary of Results
Find diseases
Clear
Rare Diseases (Orphanet) 3 results
Genetic Diseases (OMIM) 2 results
Total: 3 results
1
10 (per page)
Download Results
Rank (Similarity)
Disease Name (Disease ID)
Matched Phenotype
Causative Gene
1 (100.0%)
Hereditary spherocytosis (ORDO:822)
Folate deficiency
Hemolytic anemia
Jaundice
Spherocytosis
Splenomegaly
ANK1
EPB42
SLC4A1
SPTA1
SPTB
Hereditary spherocytosis is a congenital hemolytic anemia with a wide clinical spectrum (from symptom-free carriers to severe hemolysis) characterized by anemia, variable jaundice, splenomegaly and cholelithiasis.
Find images (Google)
Find case reports
2 (95.4%)
Neuroblastoma (ORDO:635)
Hemolytic anemia
Jaundice
Methylmalonic acidemia
Spherocytosis
Splenomegaly
ALK
HACE1
LIN28B
LMO1
MYCN
PHOX2B
TOP2A
Neuroblastoma is a malignant tumor of neural crest cells, the cells that give rise to the sympathetic nervous system, which is observed in children.
Find images (Google)
Find case reports
3 (68.6%)
8p11.2 deletion syndrome (ORDO:251066)
Azoospermia
Hemolytic anemia
Sacral dimple
Spherocytosis
Splenomegaly
ANK1
8p11.2 deletion syndrome is a contiguous gene syndrome characterized by the association of congenital spherocytosis, dysmorphic features, growth delay and hypogonadotropic hypogonadism.
Find images (Google)
Find case reports

Figure 7

4) 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 8, 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 9) or genes (Figure 10) by clicking them. The comma-separated values format is supported.

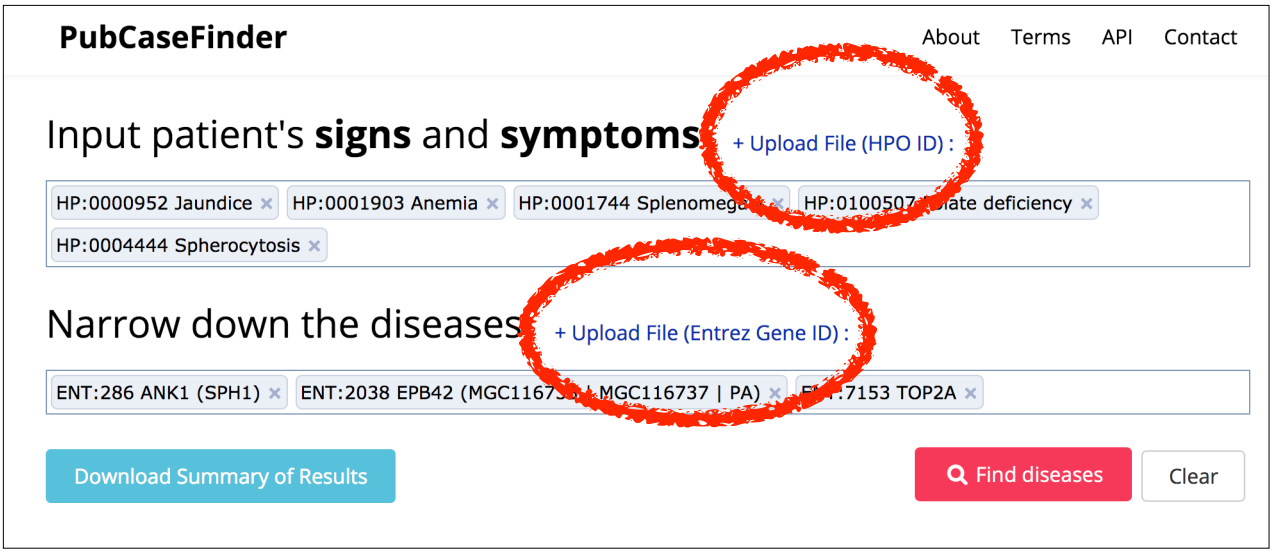

Figure 8

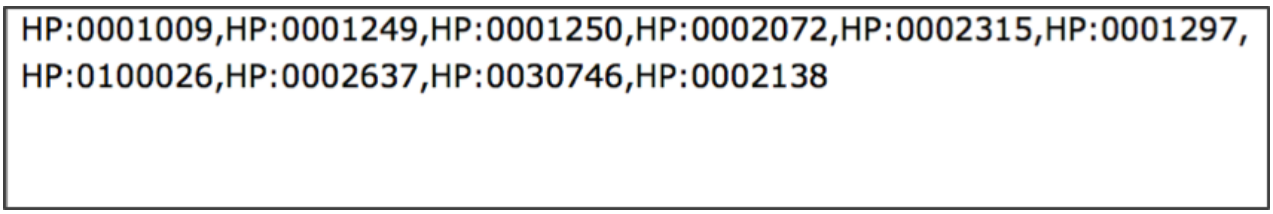

Figure 9

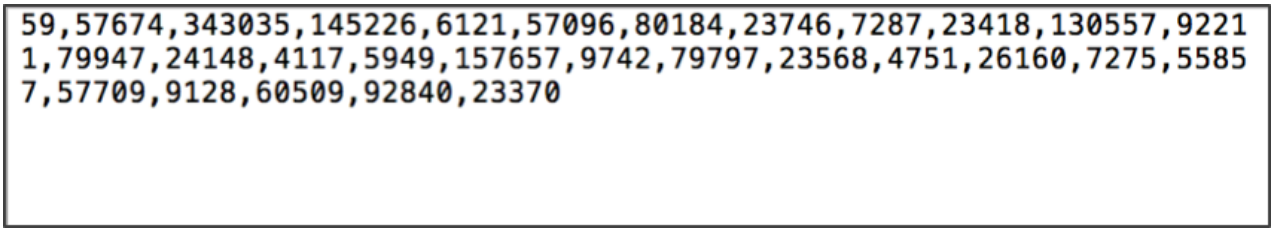

Figure 10

5. HOW TO SEARCH CASE REPORTS

After getting a ranked list of rare and genetic 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 an affected individual against those included in published case reports (Figure 11). 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

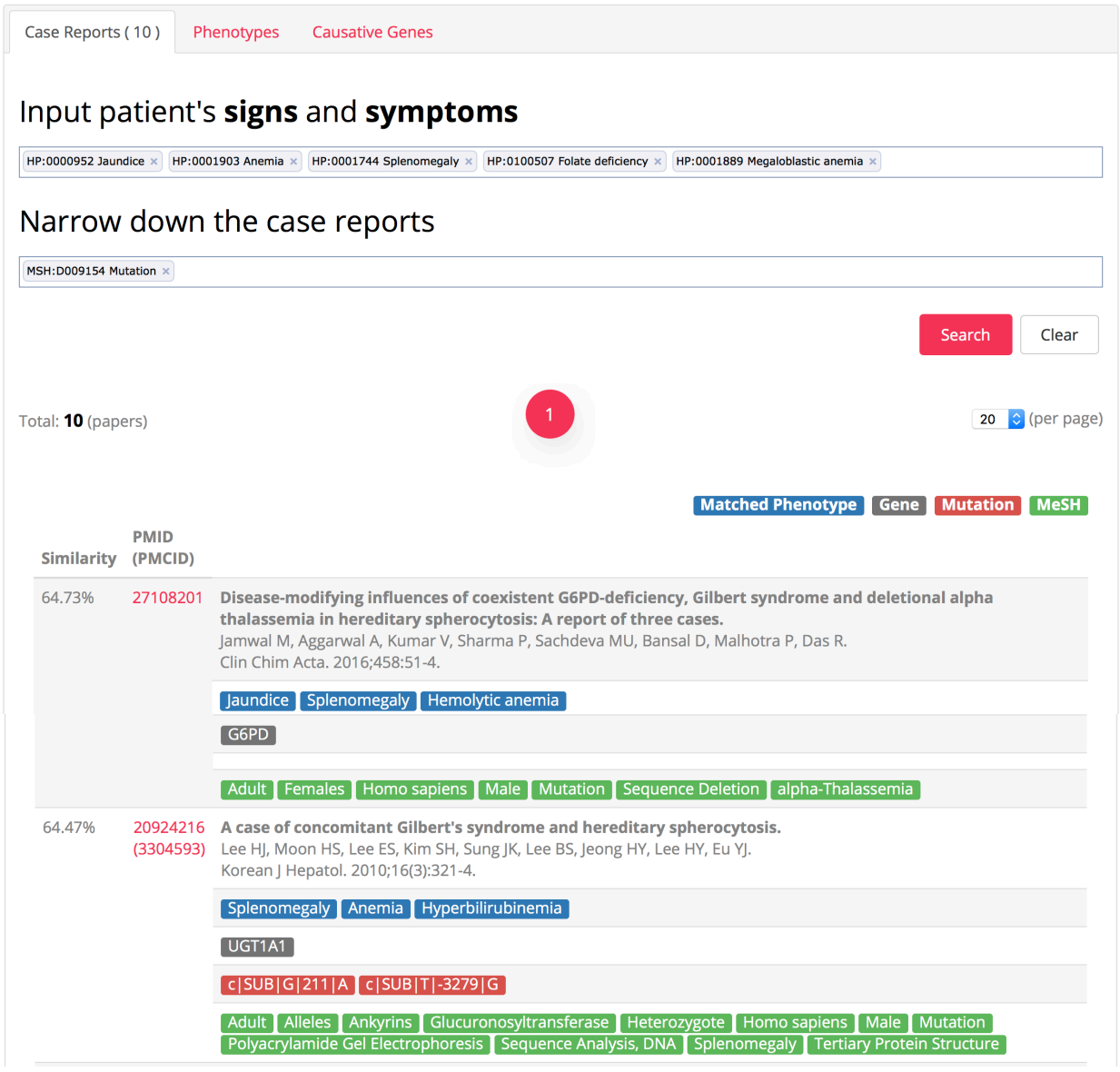

Figure 11

“Phenotypes“ or “Causative Genes,” you can check the phenotypes or causative genes related to the rare disease (Figure 11).