PedAM Guide

Contents

1.	The function of PedAM	2
1.1	Disease query	3
1.2	Disease network	5
1.2.1	Phenotype network	6
1.2.2	Gene network	8
1.3	Submit case	11
2.	Contact information	

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.

2

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):

HOME	QUERT	NETWORK -	SUBMIT CASE -	HELP&FAQ +
Precise Disease Search			Query keywords:	
Please enter a <i>pediatric disea</i> : annotation of the certain pediatric rett syndrome	se name and press Search, then you c disease. Sea	u will gain the related	Pediatrics Disease Rett syndrome ; Achondroplasia ; Polvothemia vera ; Cystinuria ; Pitvriasis rubra pilaris	
Fuzzy Disease Search				
Please enter a likely <i>pediatric d</i> then you will gain the related ann	fisease name (more than 2 characte lotation of the likely pediatric disease. Sea	rs) and press Search, rch		
The First 2000 pediatric dis	sease			
About 4542 Results.			*	
ID	Disease	9		
1	polycythemia	a vera		
2	multiple mye	loma		
3	huntington di	sease		
4	hypoplastic left hea	rt syndrome		
5	pityriasis rubra	pilaris		
6	wilson dise	ase		
7	retinoblast	oma		
8	neuroblast	oma		
9	myasthenia	pravis		
10	biotinidase de	ficiency		
11	achondropi	asia		
12	rett syndro	me		
13	fanconi ane	emia		
14	essential thromb	ocythemia		
15	moyamoya di	sease		
16	polycystic ovary	syndrome		
17	sickle cell an	iemia		
18	amyloido:	sis		
19	cystic fibro	sis		
20	galactose	mia		
21	epidermodysplasia	verruciformis		
22	primary ciliary de	vskinesia		
23	leigh syndr	ome		
24	cystinuri	<u>a</u>	*	
Page: 1 2 3 4 5 6 7 8 9 10 11 12 13	<u>14 15 16 17 18 19 20</u>			

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).





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.

Contraction of the local division of the loc	1 - Stars					Frida	too and the second		~
HOME		QUERY		NETWORK+		SUDA	IT CASE +	HELPWO	Q.+
vilson disease									
	Diseaster		Constant	11	Quantum	12	140	101 Days	
General	Phenoty		Symptom	M	Genotype	M	WP-	Diog	
	HP:0001878	Hemolytic and	mia		MP 00028	e 10 i m	icrocific anemia		
Manned by lexical matching	HP:0001399	Hepatic failur			MP:00001	21 1	aure of tooth eruption	n	
	HP:0016554	Acute hepatic	failure		MP:00020	28 h	epatic steatosis		
	HP:0010838	High nonceru	oplasmin-bound	serum copper	MP:00053	49 1 0	ecreased circulating	copper level	
	HPO I Name				MP Nam				
	HP:0002300	Nutism			MP.00000	10 a	bnormal abdominal	fat pad morphology	
	HP:0002758	Osteoarthritis			MP:02000	13 a	bnormal adipose tis:	sue distribution	
	HP:0200122	Adypical or pro	longed hepatitis		MP:00021	1 <u>52</u> a	bnormal brain morph	hology	
	HP.0002307	Drooling			MP:02054	102 a	bnormal action poter	ntial	
	HP:0013355	Aminoacidurii	1		MP:00110	19 al	bnormal adaptive the	ermogenesis	
	HP:0010934	Chendrecalci	aisor		MP:02041	185 a	bnormal adipocyte g	lucose upłake	
	HP:0002040	Esophageal v	arix		MP:00008	3218	bnormal adrenal gla	nd morphology	
	HP:0010939	Osteoporosis			MP:00000	10 a	bnormal abdominal	fat pad morphology	
	HP:0010124	Renal tubular	dysfunction		MP.00082	198 a	bnormal adrenal con	tex morphology	
	HP:0001260	Dysarthria			MP:00104	65 8	berrant origin of the r	right subclavian artery	
	HP:0002240	Hepatomegal	1		MP:00112	49 al	bdominal situs inven	sus	
	HP:0001878	Hemolytic and	rnia		MP:00024	1 <u>20</u> a	bnormal adaptive im	munity	
	HP:0010725	Dementia			MP:02000	10 a	bnormal abdominal	fat pad morphology	
	HP:0014448	Fulminant he	satic failure		MP:00017	77 8	bnormal body tempe	rature homeostasis	
	HP:0200032	Kayser-Fleisc	her ring		MP:00021	152 8	bnormal brain morph	hology	
	HP:0012115	Hepatitis			MP:00024	122 a	bnormal adaptive im	munity	
	HP:0002275	Poor motor ci	ordination		MP:00021	52 8	bnormal brain morph	hology	
	HP:0001337	Tremor			MP.00000	10 a	bnormal abdominal	fat pad morphology	
	HP:0001332	Dystonia			MP.00000	10 a	bnormal abdominal	fat pad morphology	
Hannad by homologous near	HP:0001399	Hepatic failur			MP:00000	10 a	bnormal abdominal i	fat pad morphology	
subbre of minimpers free	HP:0001394	Cinhosis			MP:00112	42 al	bdominal situs inver-	sus	
	HP:0003040	Athropathy			MP:00000	160 a	bnormal angiogenes	sis	
	HP:0002749	Osteomalacia			MP:02041	18 <u>9</u> a	bnormal alveolar pro	cess morphology	
	HP.0003109	Hyperphosph	aturia		MP:00095	42 8	bnormal blood home	eostasis	
	HP:0002150	Hypercalciurii			MP:00082	45 8	bnormal alveolar ma	crophage morpholog	
	HP:0010751	Personality cf	anges		MP:02000	12 2	bnormal abdominal	fat pad morphology	
	HP:0010007	Autosomal re	cessive inheritanc	e	MP:00108	5918	bdominal aorta aneu	irysm	
	HP:0007327	Mixed demyel	nating and axonal	polyneuropat	iy <u>MP-00021</u>	52 a	bnormal brain morph	hology	
	HP/0001903	Atemia			MP:00112	50 at	odominal situs ambi	guus	
	HP:0002015	I Dysphagia			MP:00104	2218	serrant organ of the r	ngnt subcrawan artery	
	HP.0100550	Dysionesia			MP:00054	EQZ M	onormai action poter	nsai	
	HP:0016554	Acute negatic	failure.		MP:00053	<u>552</u> a	onormal amino acid	Nevel	
	HP3010043	Proteinuna			MP:000.5	2/12	snormal assorminal	wall morphology	
	HP.3001271	Pogneuropat	ly Marine		MP:00024	522 S	onormai adaptive im	munity	
	HP.0010829	i mypoparathys	a casim		MP:00104	102 8	serrant ongit of the r	ngnt suodaklan artery	
	HP:3010928	High nonceru	oprasmith-bound	serum copper	MP.00021	22 3	onormai prain morph	nology	
	HP.0003075	i Grycośuna			MP.00053	34 8	prormai amino acid	rever	
	m*.0024787	rumnanthe	cares		MP: 00054	102 8	enerital CD4-positio	re, arpha-beta T cell p	iyal ology
	HP:0001382	Joint hyperma	D B B		MP:00033	DZ A	prormar abdominal	wati morphology	
	HP30010787	reephrolithias	9		MP:00103	11 91	onormai acute phase	e protein level	

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).

		unimd.org	٢	
PedAM	2.0		Search And	SY /
ediatric Disease Annotations &	VonCines		the state	
HOME	QUERY	NETWORK -	SUBMIT CASE -	HELP&FAQ -
Disease-Phenotype Netwo	ork (For a better performan	ce, if the number of items more	than 70, then only show the overla	ap items)
biotinidase deficiency	◆ S	earch		
Disease Phenotype of Dise	ease I 🛛 😑 Overlap Phenotype 🧧	Phenotype of Disease II		

Fig.6 Phenotype network page

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

biotinidase deficiency Search Click the below link to show the methods Rank By Jul Phenotype-based Disease Similarity answardisease (0.134) Rank By JII Phenotype-based Disease Similarity asstynic sconstructure (0.237) metachromatic leukodystrophy (0.118) https://disease.sci.nllllllllllllllllllllllllllllllllllll	
central neurosciparta (U.189) pediatric non-hodgon / hypothoma (0.159) neurosciparta draws / mytotoma (0.157) draws / mytotoma (0.157) henrosflaru, central diabetes miscipau, (0.089) diabetis, anoshaloaathy, (0.148) henrosflaru, (0.153) henrosflaru, central diabetes miscipau, (0.099) draws / mytotoma (0.157) henrosflaru, (0.153) wincers statomis (0.099) ring chromosome, 22 (0.145) wincers (0.153) wincers statomis (0.091) central intervolve (0.153) bet vitelliform macular dystochy (0.085) bet vitelliform macular dystochy (0.085) canavar, disease (0.136) citulinemia (0.083)	

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).

			unimd.org	Ċ		_ ∂ +
·	PedAM Pediatric Disease Annotations & to	ntinos				
	HOME	QUERY	NETWORK -	SUBMIT CASE -	HELP&FAQ -	
	Disease-Gene Network (Fe	or a better performance, if the	number of items more than 7	0, then only show the overlap items	;)	
	marfan syndrome	♦ Searce	h			
	A Disease 🔵 Gene of Disease I	Overlap Gene Of Dise	ase II			

Fig.9 Gene network page

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

PedAM		unimd.org	5	SY	
Podiatric Disease Annotations & Mortfüln HOME	QUERY	NETWORK +	SUBMIT CASE -	HELP&FAQ+	
Disease-Gene Network (For a li imarfan syndrome Rank By Curated Gene-based Disease 5 actic disease (0.551) turner syndrome (0.475) thonacic actic anexysm(0.378) asabhysia neoratorum (0.38) inguinal hernia (0.324) mass syndrome (0.321) asabration anewmonia (0.306) extraise allergic alweoldis (0.287) multiple chemical sensibivity (0.287)	Jetter performance, if the num	ber of items more than 70 Citick the below link to a Rank By All Gene thronic and ane entire disease () entire and the second second enter-stando smore heat valve disease infra cranial answor- mistical answor- mistical answor- renal antery disease actic valve lineoffs	then only show the overlap iow the network beased Disease Similarity system (0.335) 92) 1.543) forme (0.016) e (0.143) sm (0.122) balancies (0.124) e (0.143) sign (0.122)	items)	
Clisease Gene of Disease I 😐 O	verlap Gene of Disease I				

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.