



Guide to Use of SimulConsult's Phenome Software

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WELCOME!

Narrative medical resources enable medical professionals to start with a disease and learn about its findings (symptoms, signs and tests). Patients, of course, appear the other way: with findings that medical professionals need to use to deduce the diagnosis from thousands of known disease phenotypes. The group of all such disease phenotypes is called the “Phenome”. SimulConsult built its Phenome version to help solve this diagnostic problem, enabling clinicians to start with findings and get to the correct disease phenotype. In doing so, it offers a “differential diagnosis” consisting of a list of likely diseases with some indication of their relative probability.

Figure 1: SimulConsult's core functionality: finding-based Diagnosis (the layman's view)

A layman's view of diagnosis...

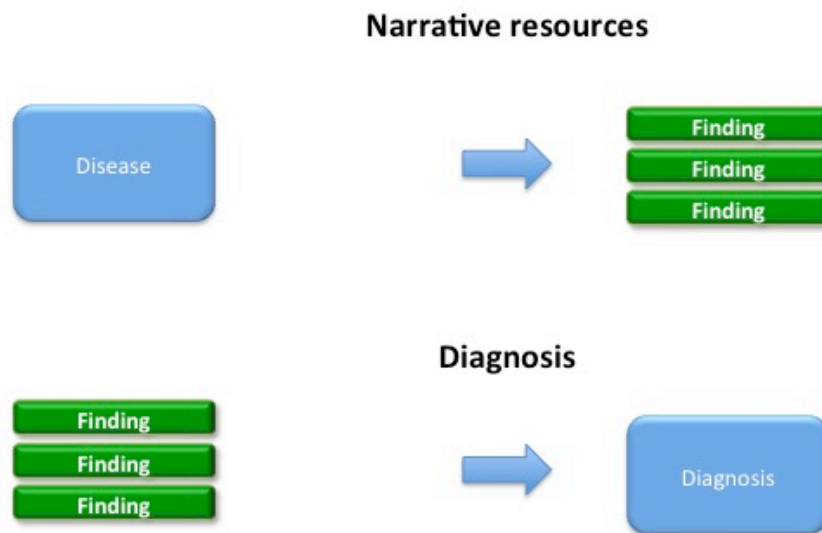
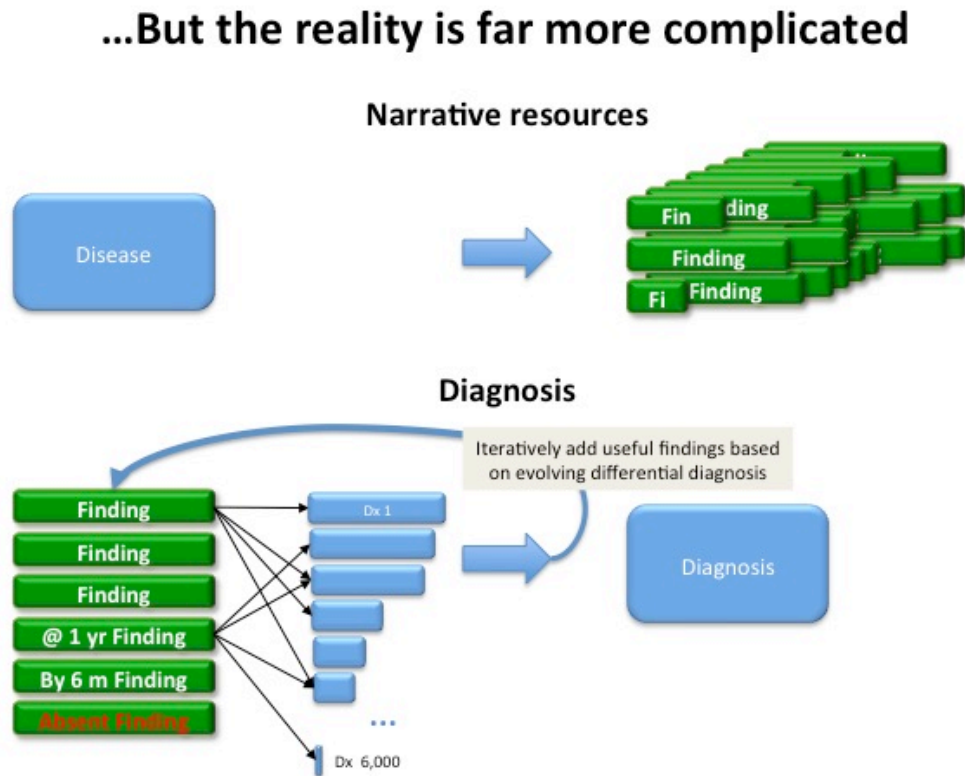


Figure 2: SimulConsult's core functionality: finding-based diagnosis that handles the full complexity



INTRODUCTION TO A FEW SIMULCONSULT CONVENTIONS

Colors and their meaning

Diseases are **blue**. Findings are **green**. Visited text turns **purple**, using the web convention for visited links. *Note: There are other colors used in the authoring and editing sections of the database.*

Contextual links

When you click on either the disease or finding you will see links to various resources at the bottom. Illustrated here is a link to a GeneReviews article and to OMIM. (Note: for Macintosh users: to get these outside links to work you'll need to follow a Macintosh link on the opening page of the software for instructions on settings needed).

Figure 3: The user can click on a disease to show disease-related links

The screenshot shows a web application interface for differential diagnosis. At the top, there are four tabs: "Differential diagnosis" (selected), "Add findings", "Add tests", and "Patient". To the right of these tabs are two buttons: "Advanced mode" (unchecked) and "To home screen". Below the tabs, there is a section titled "Diseases" with three checkboxes: "Incidence" (checked), "Onset" (checked), and "Genome" (unchecked). To the right of these checkboxes is a column header "Relative Probability". A list of diseases is displayed, each with a corresponding blue bar representing its relative probability. The diseases listed are: PCH2: pontocerebellar hypoplasia type 2, Aicardi-Goutières syndrome, PMM2-CDG (CDG-Ia), LIS2: Lissencephaly 2: microlissencephaly, Norman-Roberts type (R), VLDLR-associated cerebellar hypoplasia, MDC1D: muscular dystrophy, congenital, 1D, PEHO-like syndrome, Dekaban-Arima coloboma chorioretinal cerebellar vermis aplasia, HLD4: Leukodystrophy, hypomyelinating, 4, JBTS1: INPP5E Joubert syndrome, Mevalonic aciduria, infantile (mevalonate kinase deficiency), Mucopolipidosis IV (sialolipidosis), typical form, Twinkle (C10orf2)-related mitochondrial DNA depletion (hepatocere), Leukoencephalopathy, cystic, without megalencephaly, and Marinesco-Sjögren syndrome. At the bottom right of the list, there is a button labeled "Highest probability" with a right-pointing arrow. Below the list, there is a yellow box containing a tip: "Tip: [GeneReviews: Pontocerebellar Hypoplasia 2 & 4](#)". To the right of this tip are two buttons: "More tips" and "OMIM".

Diseases	Relative Probability
PCH2: pontocerebellar hypoplasia type 2	High
Aicardi-Goutières syndrome	Medium
PMM2-CDG (CDG-Ia)	Low
LIS2: Lissencephaly 2: microlissencephaly, Norman-Roberts type (R)	Low
VLDLR-associated cerebellar hypoplasia	Low
MDC1D: muscular dystrophy, congenital, 1D	Low
PEHO-like syndrome	Low
Dekaban-Arima coloboma chorioretinal cerebellar vermis aplasia	Low
HLD4: Leukodystrophy, hypomyelinating, 4	Low
JBTS1: INPP5E Joubert syndrome	Low
Mevalonic aciduria, infantile (mevalonate kinase deficiency)	Low
Mucopolipidosis IV (sialolipidosis), typical form	Low
Twinkle (C10orf2)-related mitochondrial DNA depletion (hepatocere)	Low
Leukoencephalopathy, cystic, without megalencephaly	Low
Marinesco-Sjögren syndrome	Low

Tip: [GeneReviews: Pontocerebellar Hypoplasia 2 & 4](#)

More tips OMIM

Regular sources include GeneReviews, OMIM (Online Mendelian Inheritance in Man), Orphanet, as well as many individual books and articles. Links to disease associations, and various useful calculators are also included. When there are many links, the "More tips" button will light up.

Similarly, when clicking on a finding, here **Nystagmus**, one gets tips related to the finding.

Figure 4: The user can click on a finding (clinical, test or gene) to show finding-based links

Differential diagnosis
Add findings
Add tests
Patient

☐ Advanced mode
[To home screen](#)

Diseases

PCH2: pontocerebellar
Aicardi-Goutières synd
PCH8: pontocerebellar
PCH10: Pontocerebellar
LIS2: RELN-related liss
CDG1A: PMM2-related
VLDLR-related cerebel
PCH1B: pontocerebellar
Muscular dystrophy-dy
PCH9: pontocerebellar
PEHO-like syndrome
PCH1A: pontocerebellar
PCH3: pontocerebellar
Microcephaly, postnata
Dekaban-Arima colobor
LIS1: Lissencephaly, isc

Patient: 2 year old boy
Change initial information

Pertinent positive findings

*
≤ 1m
Nystagmus, non-rotary

*
≤ 6m
Hyperreflexia

*
✓
CT or MRI: brainstem atrophy or hypoplasia

*
@1m
Microcephaly

✓
History of a similar disorder in family or contacts

Pertinent negative findings

x
Regression

x
Early death if undiagnosed

Tip: [Orphanet article on Congenital nystagmus](#)
More tips

Tip: [NeuroExam article on Extraocular Movements](#)
OMIM

Summary
Note

Order gene test

Contextual buttons in the advanced mode

When the advanced mode is selected (using the check box in the top right corner of the screen), more capabilities are available.

These buttons are contextual. If you click on a disease, you will be offered choices related to the disease selected. In this case, clicking on the blue button for the disease “PCH2: pontocerebellar ...”, the “**Assess disease**” and “**Profile disease**” buttons will turn blue to indicate they relate to a disease, here PCH2.

Figure 5: Disease-related buttons (in advanced mode)

The screenshot displays the 'Patient' tab in an advanced mode interface. The top navigation bar includes 'Differential diagnosis', 'Add findings', 'Add tests', and 'Patient', with 'Patient' being the active tab. A checkbox in the top right corner is labeled 'Advanced mode' and is checked. The main content area is divided into a left sidebar and a central panel. The sidebar, titled 'Diseases', lists various conditions, with 'PCH2: pontocerebellar' highlighted in blue. The central panel shows patient information: 'Patient: 2 year old boy' and a 'Change initial information' button. Below this, there are sections for 'Pertinent positive findings' and 'Pertinent negative findings'. The 'Pertinent positive findings' section lists several findings with associated dropdown menus: 'Nystagmus, non-rotary' (≤ 1m), 'Hyperreflexia' (≤ 6m), 'CT or MRI: brainstem atrophy or hypoplasia' (✓), 'Microcephaly' (@1m), and 'History of a similar disorder in family or contacts' (✓). The 'Pertinent negative findings' section lists 'Regression' (✗) and 'Early death if undiagnosed' (✗). A 'Finding color key' indicates that green represents 'Pertinence'. On the right side of the interface, there is a vertical stack of buttons: 'Differential Dx', 'Gene discovery', 'Assess disease' (highlighted in blue), 'Profile disease' (highlighted in blue), 'Database', 'Search', 'File', 'Home', and 'Help'. At the bottom, there is a yellow bar with a tip: 'Tip: GeneReviews: Pontocerebellar Hypoplasia 2 & 4'. To the right of the tip are buttons for 'More tips', 'OMIM', 'Summary', 'Note', and 'Order gene test'.

Differential diagnosis **Add findings** **Add tests** **Patient** ☒ Advanced mode

Diseases

- PCH2: pontocerebellar
- Aicardi-Goutières synd
- PCH8: pontocerebellar
- PCH10: Pontocerebella
- LIS2: RELN-related liss
- CDG1A: PMM2-related
- VLDLR-related cerebel
- PCH1B: pontocerebella
- Muscular dystrophy-dy
- PCH9: pontocerebellar
- PEHO-like syndrome
- PCH1A: pontocerebella
- PCH3: pontocerebellar
- Microcephaly, postnata
- Dekaban-Arima colobor
- LIS1: Lissencephaly, isc

Patient: 2 year old boy [Change initial information](#)

Pertinent positive findings

Finding color key: Pertinence

- * ≤ 1m Nystagmus, non-rotary
- * ≤ 6m Hyperreflexia
- * ✓ CT or MRI: brainstem atrophy or hypoplasia
- * @1m Microcephaly
- ✓ History of a similar disorder in family or contacts

Pertinent negative findings

- ✗ Regression
- ✗ Early death if undiagnosed

Assess disease **Profile disease**

Database **Search** **File** **Home** [Help](#)

Tip: [GeneReviews: Pontocerebellar Hypoplasia 2 & 4](#) [More tips](#) [OMIM](#) **Summary** **Note** [Order gene test](#)

Similarly, if you click on a finding, you will be offered choices related to the finding selected. By clicking on Nystagmus the “Assess finding” and “Profile finding” buttons will turn green to indicate that they relate to a finding, here Nystagmus.

Figure 6: Finding-related buttons (under advanced mode)

[Differential diagnosis](#)
[Add findings](#)
[Add tests](#)
[Patient](#)

☒ Advanced mode

Diseases

- PCH2: pontocerebellar
- Aicardi-Goutières synd
- PCH8: pontocerebellar
- PCH10: Pontocerebellar
- LIS2: RELN-related liss
- CDG1A: PMM2-related
- VLDLR-related cerebel
- PCH1B: pontocerebella
- Muscular dystrophy-dys
- PCH9: pontocerebellar
- PEHO-like syndrome
- PCH1A: pontocerebella
- PCH3: pontocerebellar
- Microcephaly, postnata
- Dekaban-Arima colobom
- LIS1: Lissencephaly, isc
- HLD4: Leukodystrophy

Patient: 2 year old boy
[Change initial information](#)

Pertinent positive findings

Finding color key:

Pertinence

*	≤ 1m	Nystagmus, non-rotary
*	≤ 6m	Hyperreflexia
*	✓	CT or MRI: brainstem atrophy or hypoplasia
*	@1m	Microcephaly
✓		History of a similar disorder in family or contacts

Pertinent negative findings

✗		Regression
✗		Early death if undiagnosed

[Differential Dx](#)
[Gene discovery](#)
[Assess finding](#)
[Profile finding](#)

[Database](#)

[Search](#)
[File](#)

[Home](#)
[Help](#)

Tip: [Orphanet article on Congenital nystagmus](#)
[More tips](#)

Tip: [NeuroExam article on Extraocular Movements](#)
[OMIM](#)

[Summary](#)
[Note](#)

[Order gene test](#)

PATIENT CASE USED FOR ILLUSTRATIONS

This VLDLR case was published by Dixon and Salazar in 2012 and will be used throughout for illustrating the points.

A 2-year old boy from Middle East

- **Family history:**
 - Neither parent affected
 - 1 of 2 brothers affected
 - Parent consanguinity: first cousins
- **At 1 month:** microcephaly
- **By 1 month:** Nystagmus, non-rotary
- **By 6 months:** hyperreflexia
- **Absent:** regression
- **MRI:** pan cerebellar hypoplasia

GET ACCESS

1. Individual Cloud-accessed version

Click “Go to Login” at www.SimulConsult.com and then either:

Individual log-in with last name and password

Figure 7: Individual Access

SimulConsult®
A Simultaneous Consult On Your Patient's Diagnosis

Diagnostic Decision Support

This software compares a patient's findings to the findings in 6,080 diseases. It suggests a "differential diagnosis" and identifies other findings and lab tests that may be useful in reaching a diagnosis. It uses detailed information about the patient to assist with diagnosis, so it is best to watch one of the **demo videos** before using it.

The tool includes the 3,320 genes with convincing germline human disease phenotypes. It also includes many nongenetic diseases, with particular strength in neurology and rheumatology.

The software may be used only by medical professionals (doctors, nurse practitioners, genetic counselors, bioinformaticists, medical librarians, laboratory technician, or research and development scientists developing clinical laboratory processes and supervised trainees in these professions). When the software has loaded in the box below, you will be able log-in, or enter using **institutional access**. By entering the software you signify that you are a medical professional and that you accept the **Limitations and Terms of Use** agreement for use of the program and database.

Last name Password

US patents 6,212,519, 6,754,655 and 7,742,932.

Troubleshoot: Windows Macintosh **Demo videos** **Phenome version manual**

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Note that on the above page we publish the number of genes and diseases currently in the database.

2. Institutional access as a result of your IP address being recognized

In a library subscription, the software is made available through IP access.

3. Enterprise version (Health system or Laboratory): Access from within EHR or LIM

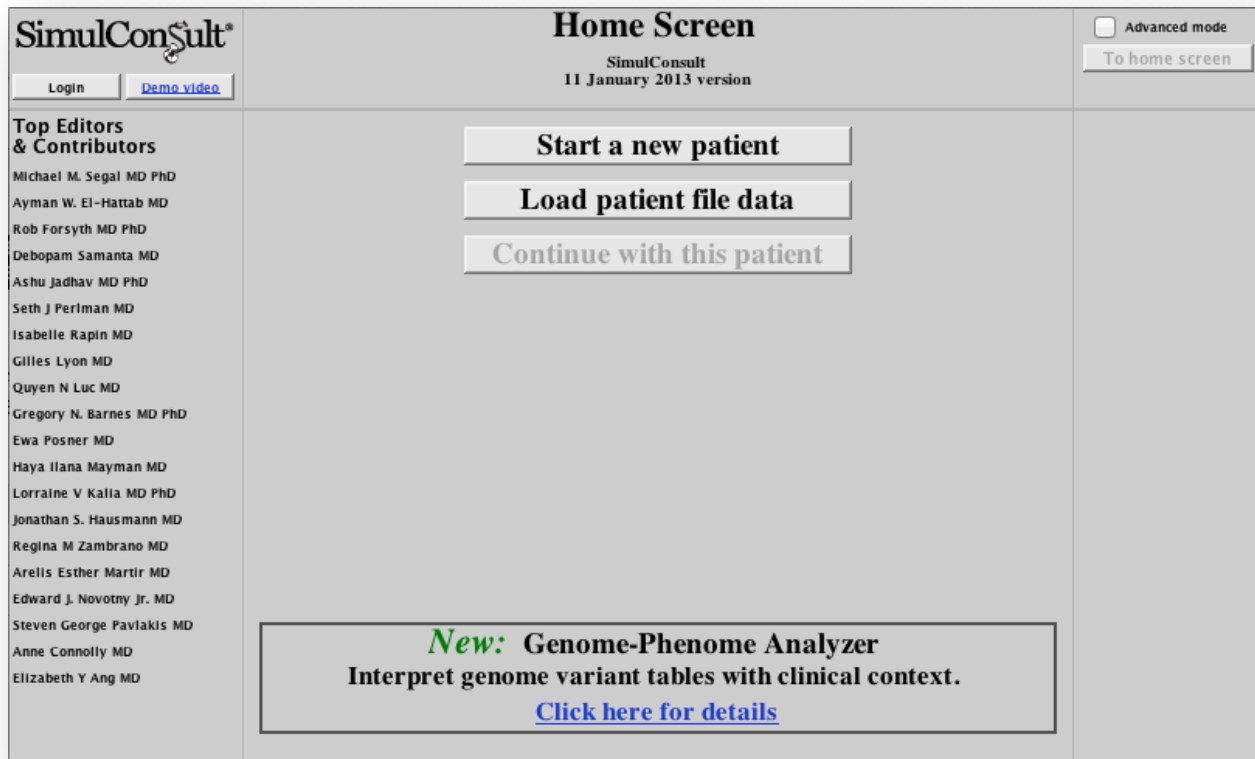
*If you have the **electronic health record or laboratory information system integrated version**, please see the companion document for how to launch SimulConsult from within your system.*

BEGIN BY ENTERING THE PATIENT AGE AND SEX

From Cloud version, start a new patient, then enter age and gender

Click the “Start a new patient” button.

Figure 8: Start a new patient



The first step is to enter the age and gender. Once that is entered, choices will appear for how to enter findings.

Figure 9 a & b: Entering the patient age and gender then offers choices for how to enter findings

The screenshot shows the 'Start a Patient' form. At the top right, there are links for 'Advanced mode' and 'To home screen'. The 'Required initial information:' section contains an 'Age now:' field with a dropdown menu set to 'years' (radio buttons for days, weeks, months, and years are present), and a 'Gender:' section with radio buttons for 'male' and 'female'. Below this is a 'Choose one or more findings:' section. It includes a 'Text search (one or more findings)' input field with a magnifying glass icon, a 'QuickLists' section with a list of categories (Family history, Ancestry, Genetics, Metabolic, Mitochondrial, Neurology, Rheumatology, Size & Vitals), and a 'Search category for findings' section with a 'Category list' button. A 'Differential diagnosis' button is also visible on the right. At the bottom, a yellow tip box states: 'Tip: Choose one or more unusual or prominent findings to begin the patient description'.

Starting from the enterprise version

The age and gender information comes in automatically and you start here.

This screenshot shows the 'Start a Patient' form with the 'Age now:' field pre-filled with '2' and the 'Gender:' section pre-selected for 'male'. The 'Choose one or more findings:' section is highlighted with a green border. The 'Text search (one or more findings)' input field, the 'QuickLists' section, and the 'Search category for findings' section are all highlighted in green. The 'Category list' button is also highlighted in green. The 'Differential diagnosis' button remains unhighlighted. The yellow tip box at the bottom is the same as in the previous screenshot.

ENTER INITIAL KEY FINDINGS, USING DIFFERENT SEARCH MODES

The process of entering clinical findings (signs, symptoms) and test results has been optimized for speed, including prompting the clinician to comment on findings most helpful in narrowing the evolving differential diagnosis (the probability weighted list of potential diagnoses relevant for this patient based on findings entered).

The importance of onset information

Most genetic diseases unfold over time, and as a result, using information available about the onset of particular findings is helpful in narrowing the differential diagnosis

The importance of pertinent negatives

Because many genetic diseases share multiple findings, geneticists usually make heavy use of the absence of pertinent findings during the process of diagnosis. The software also supports this option.

The importance of requiring certain findings

The default in the software is to take into account the possibility that a finding is not related to the primary diagnosis; the software has extensive probability information to make these assessments. However, when a common finding is particularly notable, such as a very high creatine kinase level, you have the ability to specify that you require it to be a finding in the diagnosis.

How to enter a finding: onset, presence or absence

To enter a finding, click to the left of the finding where you see the “+/- and the down arrow.” The menu will appear. Click on the selection you want.

1. Pertinent positive options

- Onset **at** a particular age
- Present **by** a particular age, onset unknown (not as informative as option “a” but sometimes all that is known)
- Present, onset unknown

2. Pertinent negative option

- Absent now

Figure 10: How to enter a finding

The screenshot displays the 'Neurology QuickList' interface. It features two columns of findings, each with a dropdown menu icon (a small square with a downward arrow) to its left. The findings listed include: +Distal location of motor or sensory deficit, +Proximal location of motor or sensory deficit, +Recurrent exacerbations, +Unilateral location or asymmetric, Ataxia, Attention deficit (ADHD), Autistic, Choreoathetosis, Contractures or limited range of motion, Cramps, frequent or severe, Creatine kinase high, Deafness, Dysphagia or feeding difficulties, Dystonia, Eye movement deficit, horizontal or vertical, and Gait disturbance. A dropdown menu is open for the finding 'Microcephaly <3rd %ile', showing options for onset (e.g., 'Onset at ~ 1 week old', 'Onset at ~ 1 month old', 'Onset at ~ 3 months old', 'Onset at ~ 6 months old', 'Onset at ~ 1 year old', 'Onset at ~ 3 years old') and presence (e.g., 'Present by ~ 1 week old, onset unknown', 'Present by ~ 1 month old, onset unknown', 'Present by ~ 3 months old, onset unknown', 'Present by ~ 6 months old, onset unknown', 'Present by ~ 1 year old, onset unknown', 'Present by ~ 3 years old, onset unknown', 'Present now - onset unknown', 'Absent now', 'Not specified'). To the right of the findings, there are buttons for 'Differential Dx', 'Search modes', and 'QuickLists:'. Below the 'QuickLists:' button, there are buttons for 'Family history', 'Ancestry', 'Genetics', 'Neurology', and 'Immunology'.

Modes for entering initial findings

You have several search modes for locating **the initial** findings, each of which is illustrated below.

1. **QuickLists** (common findings in genetics, neurology, rheumatology and ancestry, as well as an interface for family history)
2. **Categories** (e.g., Cardiac & vascular)
3. **Search** (one or more terms)

Note: Once you have an initial differential diagnosis, the Useful findings and Useful tests tabs will often be the most convenient way to add findings.

Enter using “QuickLists”

Using the “Neurology QuickLists” and “Family History”, you can add a set of pertinent positives and negatives.

Figure 11: Entering findings using the Neurology QuickLists

Neurology QuickList				Sort options
<input type="checkbox"/> +/-	Ataxia	<input type="checkbox"/> +/-	+Location distal: motor /joint	<input checked="" type="radio"/> Alphabetical
<input type="checkbox"/> +/-	Attention deficit	<input type="checkbox"/> +/-	+Location: proximal motor or senso	<input type="radio"/> Usefulness
<input type="checkbox"/> +/-	Autistic behavior	<input type="checkbox"/> +/-	Motor developmental delay	<input type="button" value="Recalculate usefulness"/>
<input type="checkbox"/> +/-	Choreoathetosis	<input type="checkbox"/> +/-	Muscular atrophy or hypoplasia	QuickLists:
<input type="checkbox"/> +/-	Contractures or passive limited rang	<input type="checkbox"/> +/-	Myoclonus	<input checked="" type="button" value="Family history"/>
<input type="checkbox"/> +/-	Cramps, frequent or severe	<input type="checkbox"/> * <input type="checkbox"/> ≤ 1m	Nystagmus, non-rotary	<input type="button" value="Ancestry"/>
<input type="checkbox"/> +/-	Creatine kinase high	<input type="checkbox"/> +/-	Optic atrophy or hypoplasia	<input type="button" value="Genetics"/>
<input type="checkbox"/> +/-	Deafness	<input type="checkbox"/> +/-	Protein high in CSF	<input type="button" value="Metabolic"/>
<input type="checkbox"/> +/-	Dysarthria or abnormal sound chara	<input type="checkbox"/> +/-	+Recurrent exacerbations	<input type="button" value="Mitochondrial"/>
<input type="checkbox"/> +/-	Dysphagia or feeding difficulty	<input type="checkbox"/> +/-	Regression	<input type="button" value="Neurology"/>
<input type="checkbox"/> +/-	Dystonia	<input type="checkbox"/> +/-	Scoliosis with or without kyphosis	<input type="button" value="Rheumatology"/>
<input type="checkbox"/> +/-	Eye movement deficit, horizontal	<input type="checkbox"/> +/-	Seizures with abnormal movements	<input type="button" value="Size & Vitals"/>
<input type="checkbox"/> +/-	Gait disturbance	<input type="checkbox"/> +/-	Sleep disturbance	<input type="button" value="Search"/>
<input type="checkbox"/> +/-	Headache (frequent or severe)	<input type="checkbox"/> +/-	Somnolence or lethargy	<input type="button" value="Differential diagnosis"/>
<input type="checkbox"/> * <input type="checkbox"/> ≤ 6m	Hyperreflexia	<input type="checkbox"/> +/-	Stereotypies	<input type="button" value="Home"/>
<input type="checkbox"/> +/-	Hypertonia	<input type="checkbox"/> +/-	Strokes as assessed clinically	
<input type="checkbox"/> +/-	Hypoalgesia	<input type="checkbox"/> +/-	Tremor of limbs, trunk or head	
<input type="checkbox"/> +/-	Hyporeflexia	<input type="checkbox"/> +/-	+Unilateral location or asymmetry	
<input type="checkbox"/> +/-	Hypotonia	<input type="checkbox"/> +/-	Vertigo, significant	
<input type="checkbox"/> +/-	Intellectual disability	<input type="checkbox"/> +/-	Visual impairment	
<input type="checkbox"/> +/-	Irritability or agitation, pronounced	<input type="checkbox"/> +/-	Weakness, significant	

Tip: [NINDS Spasticity Information Page](#)

Tip: [NeuroExam article on testing reflexes](#)

Notice that the list can be sorted alphabetically, as shown here by selecting the sort option in the top right, or in order of usefulness of these findings in narrowing the differential diagnosis, which is relevant once some findings have been entered.

QuickLists are developed as a set of up to 46 findings most often used in the referrals to the specialty. QuickLists allow the user increase the speed with which the software focuses on the relevant subset of diseases. To ensure the QuickList is most useful, the curation process for new and updated disease reviews the findings in the QuickList to ensure complete coverage.

Figure 12: Entering family history using the QuickList

Specify family history:

Mother affected	<input type="radio"/> Yes	<input checked="" type="radio"/> No	<input type="radio"/> Unknown
Father affected	<input type="radio"/> Yes	<input checked="" type="radio"/> No	<input type="radio"/> Unknown
Sisters affected by age patient affected:	<input type="text" value="0"/>	of	<input type="text" value="0"/> determinable
Brothers affected by age patient affected:	<input type="text" value="1"/>	of	<input type="text" value="2"/> determinable
Maternal uncles affected:	<input type="text" value="0"/>	of	<input type="text" value="0"/> total
Other nearby individuals affected:	<input type="text" value="0"/>	of	<input type="text" value="0"/> total
Parental consanguinity:	<input type="text" value="1st cousin"/>		
<input type="button" value="Clear"/>		<input type="button" value="Help"/>	<input type="button" value="Finish"/>

Enter using “Categories” of findings

Select “Lab: radiology” in the “Category of findings”.

Figure 13: Using categories (advanced mode)

Pick a category of findings, then press "Expand category"

Lab: clinical neurophysiology: evoked responses

Lab: electrocardiogram (EKG, ECG)

Lab: endoscopy and exploratory surgery

Lab: gene or DNA tests

Lab: hematology

Lab: pathology: biopsies & morphology

Lab: radiology

Lab: serology & immunological tests

Lab: stool tests

Lab: ultrasound & echocardiogram

Lab: urine tests

Mental status, mental development & sleep

Motor, motor development & muscle

Mouth & throat

Nose

Peripheral nerve, sensory, pain & headaches

Psychiatric & behavioral

Reflexes

Seizures & other paroxysmal disturbances

Skin and mucosa

Teeth

Expand category

Search

Differential Dx

Gene discovery

Assess finding

Profile finding

Database

Search

File

Home

Help

More tips

Summary

Note

OMIM

Order gene test

By clicking on “**Lab: radiology**” and then “**Expand category**”, we can scroll down to find “CT or MRI: brainstem atrophy or hypoplasia” and enter that it is present with onset unknown.

Figure 14: Entering findings using Categories (advanced mode)

Pick from "Lab: radiology"

☐ Advanced mode
[To home screen](#)

+/-	CT or MRI of spine: syringomyelia (syrinx of spine)
+/-	CT or MRI: 4th ventricle enlargement, major
+/-	CT or MRI: Arnold-Chiari malformation
+/-	CT or MRI: Wormian bones (extra bones in sutures)
+/-	CT or MRI: abdominal wall mass
+/-	CT or MRI: anencephaly or hydranencephaly
+/-	CT or MRI: anterior chamber mass or cyst
+/-	CT or MRI: aprosencephaly
+/-	+CT or MRI: aqueductal stenosis (sparing of 4th ventricle in hydrocephalus)
+/-	CT or MRI: arrhinencephaly
+/-	CT or MRI: basal ganglia abnormalities
+/-	CT or MRI: brain abscesses
+/-	CT or MRI: brain cysts or cavities
+/-	CT or MRI: brain edema
+/-	CT or MRI: brain lipomas
* ▾	✓ CT or MRI: brainstem atrophy or hypoplasia
+/-	CT or MRI: brainstem lucencies or lesions
+/-	CT or MRI: brainstem structural lesion
+/-	CT or MRI: caudate nuclei fused
+/-	CT or MRI: cavum septum pellucidum
+/-	CT or MRI: cerebral cortex atrophy or hypoplasia, focal or generalized

Differential diagnosis

Search

More tips

Summary

Note

Tip: The "Patient summary" button at right pops up a patient report

OMIM

Order gene test

Enter using search

You can always search on terms, and SimulConsult supports many synonyms. You can put more than one search term in the box at a time, separate them with a space. Often it is best to use a word fragment if different endings are used, such as “nyst” for nystagmus.

Figure 15: Entering findings using search (basic mode)

Start a Patient

Advanced mode
To home screen

Required initial information:

Age now: 2 days weeks months years

Gender: male female

Choose one or more findings:

Text search (one or more findings) hyperreflex nyst

QuickLists

Family history

Ancestry

Genetics

Metabolic

Mitochondrial

Neurology

Rheumatology

Size & Vitals

Search category for findings

Category list

Differential diagnosis

Tip: Choose one or more unusual or prominent findings to begin the patient description

Figure 16: Selecting from the search results to define onset (basic mode)

Pick from findings with "hyperreflex nyst"

Advanced mode
To home screen

FRMD7 gene mutation (X-linked)

Hyperreflexia

Nystagmus, non-rotary

Nystagmus, rotary

Toes upgoing without hyperreflexia

Differential diagnosis

Search

Tip: Orphanet article on Congenital nystagmus

More tips

Summary

Note

Tip: NeuroExam article on Extraocular Movements

OMIM

Order gene test

View and use synonyms

Each disease and finding name may have synonyms, which the software also checks in search mode. To see the synonyms for a particular finding or disease hover your mouse over the button. The results display in a yellow “tool tip” box as shown here.

Figure 17: Viewing synonyms (basic mode)

Pick from findings with "hyperreflex nyst"

+/-

FRMD7 gene mutation (X-linked)

*

≤ 6m

Hyperreflexia

*

≤ 1m

Nystagmus, non-rotary

+/-

Nystagmus, rotary

+/-

Toes upgoing without hyperreflexia

Synonyms: Jerk or pendular (jerky saccades included), abnormal head posture, head tilt, visual null point

Advanced mode

To home screen

Differential diagnosis

Search

Tip: [Orphanet article on Congenital nystagmus](#)

More tips

Summary

Note

Tip: [NeuroExam article on Extraocular Movements](#)

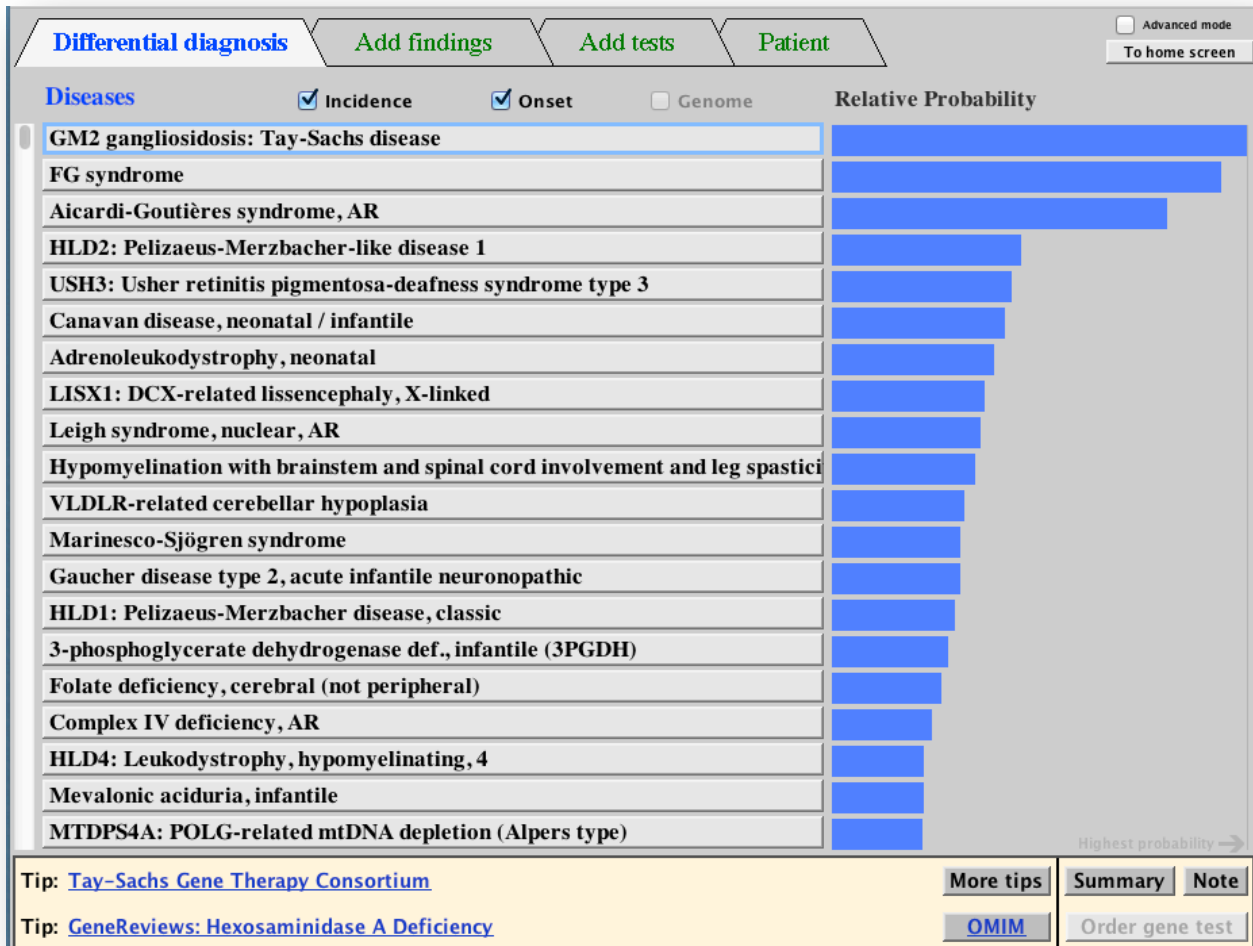
OMIM

Order gene test

VIEW THE DIFFERENTIAL DIAGNOSIS

After entering the initial findings, you can go to the differential diagnosis screen by pressing the “**Differential diagnosis**” button (see previous figure). From there you can add further findings to narrow the differential diagnosis.

Figure 18: Differential Diagnosis



VIEW THE FINDINGS ALREADY ENTERED

To see the findings we have entered about this case, and their pertinence to the differential, click the **“Patient”** tab from the **Differential Diagnosis** section. Note that by clicking the onset, one can always “un-specify” the finding to eliminate it from the patient findings by choosing “Not specified”.

A mathematical representation of the common medical concept of “Pertinence” (as used in “pertinent positives and negatives”) is represented by the green shading overlay on the patient’s findings.

Figure 19: The Patient Tab

Differential diagnosis **Add findings** **Add tests** **Patient** ☐ Advanced mode [To home screen](#)

Diseases

- GM2 gangliosidosis: Ta
- FG syndrome
- Aicardi-Goutières synd
- HLD2: Pelizaeus-Merzh
- USH3: Usher retinitis pi
- Canavan disease, neona
- Adrenoleukodystrophy,
- LISX1: DCX-related lis
- Leigh syndrome, nuclea
- Hypomyelination with b
- VLDLR-related cerebel
- Marinesco-Sjögren synd
- Gaucher disease type 2,
- HLD1: Pelizaeus-Merzh
- 3-phosphoglycerate deh
- Folate deficiency, cereb
- Complex IV deficiency,
- HLD4: Leukodystrophy
- Mevalonic aciduria, inf
- MTDPS4A: POLG-relat

Patient: 2 year old boy [Change initial information](#)

Pertinent positive findings **Finding color key:**
Pertinence

- * ≤ 1m Nystagmus, non-rotary
- * ≤ 6m Hyperreflexia
- ✓ History of a similar disorder in family or contacts

Pertinent negative findings

- ✗ Early death if undiagnosed

Tip: [Orphanet article on Congenital nystagmus](#) [More tips](#) **Summary** **Note**

Tip: [NeuroExam article on Extraocular Movements](#) [OMIM](#) [Order gene test](#)

TIP

The higher the pertinence of the entered findings, the more important it is for you to be sure that the finding is reliably determined – since the high pertinence findings are (by definition) driving the diagnosis.

ADD FINDINGS TO NARROW THE DIFFERENTIAL DIAGNOSIS

After entering the initial findings you have more methods to add findings to narrow the differential diagnosis, illustrated below:

1. **Add findings** and **Add tests** tabs, which provide a list of findings ranked in order of which will be **most useful** in narrowing the evolving differential diagnosis.
 - a. When adding tests, you have the choice to view a “bundle” of test results that could result from ordering a single test, such as an MRI.
2. The **Disease profile** (select the Advanced mode to get this option) to enter findings about the disease you suspect
3. **Search** button in the Add findings or Add tests tabs to return to the options available under the initial findings

Use suggested findings and tests

Click on the **Add findings** tab to get a suggestion of findings with which to narrow the differential diagnosis.

TIP

Notice the light blue shading representing the differential diagnosis overlay on the diseases.

Figure 20: Entering findings using the 'Add (useful) findings' tab

The screenshot displays the 'Add findings' tab of a clinical decision support system. The top navigation bar includes 'Differential diagnosis', 'Add findings' (active), 'Add tests', and 'Patient'. A 'To home screen' button is also present. The left sidebar lists various diseases, with 'GM2 gangliosidosis: Tay-Sachs disease' selected. The main area shows a list of findings with a dropdown menu for 'Regression' open, displaying options for onset and present-by age. The 'Absent now' option is highlighted in blue. The bottom of the screen features a footer with tips, a 'More tips' button, and a 'Summary' button.

Findings by usefulness	Recalc	Search
+/- Weakness, significant		
+/- Regression		
Onset at ~ birth		
Onset at ~ 1 month old		
Onset at ~ 3 months old		
Onset at ~ 6 months old		
Onset at ~ 1 year old		
Onset at ~ 3 years old		
Present by ~ birth, onset unknown		
Present by ~ 1 month old, onset unknown		
Present by ~ 3 months old, onset unknown		
Present by ~ 6 months old, onset unknown		
Present by ~ 1 year old, onset unknown		
Present by ~ 3 years old, onset unknown		
Present now - onset unknown		
Absent now		
Not specified		
+/- Spasticity character to hypertonia		
+/- Gait disturbance		
+/- Seizures with abnormal movements		
+/- Eye movement deficit, horizontal		
+/- Bundle: Gastrointestinal dysmotility		

Tip: [Tay-Sachs Gene Therapy Consortium](#)

Tip: [GeneReviews: Hexosaminidase A Deficiency](#)

More tips

Summary

Note

OMIM

Order gene test

TIP

The differential diagnosis automatically recalculates every time a new finding is entered. By contrast, “**Add findings**” and “**Add tests**” recalculate manually. When you click the “**Recalc**” button, the commented findings move to the “**Patient**” tab.

Using the **Add tests**, we can comment on the results of the scan

Figure 21: Entering findings using the 'Add (useful) tests' tab

Differential diagnosis
Add findings
Add tests
Patient

☐ Advanced mode
[To home screen](#)

Diseases

Tests by usefulness
All
Recalc
Search

PCH2: pontocerebellar	+/-	Bundle: MRI scan of the brain
Aicardi-Goutières synd	+/-	Bundle: CT scan of the brain
PCH8: pontocerebellar	+/-	MRI: white matter abnormality
PCH10: Pontocerebella	+/-	CT or MRI: pan-cerebellar atrophy or hypoplasia
LIS2: RELN-related liss	+/-	X-ray or CT: brain calcifications
CDG1A: PMM2-related	+/-	CT or MRI: cerebral cortex atrophy or hypoplasia, focal or generalized
VLDLR-related cerebel	+/-	WBC high in CSF
PCH1B: pontocerebella	+/-	Transaminases (LFTs) high
Muscular dystrophy-dy	+/-	Pigmentary retinopathy
PCH9: pontocerebellar	* ✓	CT or MRI: brainstem atrophy or hypoplasia
PEHO-like syndrome	+/-	CT or MRI: pontine atrophy or hypoplasia
PCH1A: pontocerebella	+/-	Bundle: MR spectroscopy
PCH3: pontocerebellar	+/-	Bundle: Organic acids in urine
Microcephaly, postnata	+/-	CT or MRI: lissencephaly
Dekaban-Arima colobom	+/-	Interferon-α in the CSF elevated
LIS1: Lissencephaly, isc	+/-	Metabolic acidosis
HLD4: Leukodystrophy	+/-	+MRI: hypomyelination type of white matter abnormality
Muscular dystrophy-dy	+/-	Lactate high in serum
JBTS1: INPP5E Joubert	+/-	CT or MRI: corpus callosum hypogenesis
Mevalonic aciduria, inf		

More tips
Summary
Note

Tip: The "Patient summary" button at right pops up a patient report
OMIM
Order gene test

Use test “bundles”, to find relevant value for tests with multiple outputs

Here we re-enter the finding of “CT or MRI: brainstem atrophy or hypoplasia” to show that there are multiple ways to navigate to the same finding.

Figure 22: Entering test results using the ‘Bundles’ feature

Findings in		Bundle: CT scan of the brain	
		Bundle: \$1000	Finding: \$1000
+/-	Pan-cerebellar atrophy or hypoplasia	Sort options <input checked="" type="radio"/> Usefulness <input type="radio"/> Alphabetical	Recalculate usefulness Set unmarked absent Clear all <div>Done</div>
+/-	Brain calcifications		
+/-	Cerebral cortex atrophy or hypoplasia, focal or generalized		
* ▾	<input checked="" type="checkbox"/> Brainstem atrophy or hypoplasia		
+/-	Pontine atrophy or hypoplasia		
+/-	Lissencephaly		
+/-	Corpus callosum hypogenesis		
+/-	+Basal ganglia nature to brain calcifications		
+/-	Verml cerebellar atrophy or hypoplasia		
+/-	Basal ganglia abnormalities		
+/-	Brain cysts or cavities		
+/-	Subcortical bands of gray matter		
+/-	Hydrocephalus, not ex-vacuo		
+/-	Thick cortex		
+/-	Skull thick		
+/-	4th ventricle enlargement, major		
+/-	Heterotopias or subependymal nodules		
+/-	+Periventricular or subependymal nature to intracerebral calcifications		
+/-	Sagittal craniosynostosis		
+/-	Sella turcica empty or pituitary absent, underdeveloped, or dysplastic		
+/-	Holoprosencephaly		
Tip: The "Patient summary" button at right pops up a patient report		More tips OMIM	Summary Note Order gene test

Get back to the “QuickLists” and other search modes

To return to QuickLists or any of the other methods typically used for entering initial findings, click the green “Search modes” button.

Figure 23: Getting back to the QuickLists

Differential diagnosis
Add findings
Add tests
Patient

☐ Advanced mode
[To home screen](#)

Diseases

Tests by usefulness
All
Recalc
Search

PCH2: pontocerebellar	+/-	Bundle: MRI scan of the brain
Aicardi-Goutières synd	+/-	Bundle: CT scan of the brain
PCH8: pontocerebellar	+/-	TSEN54 gene mutations (biallelic)
PCH10: Pontocerebella	+/-	X-ray or CT: brain calcifications
LIS2: RELN-related liss	+/-	WBC high in CSF
CDG1A: PMM2-related	+/-	Interferon-α in the CSF elevated
VLDLR-related cerebel	+/-	Bundle: MR spectroscopy
PCH1B: pontocerebella	+/-	CT or MRI: pontine atrophy or hypoplasia
Muscular dystrophy-dy	+/-	Creatine kinase high
PCH9: pontocerebellar	+/-	CHMP1A gene mutations (biallelic)
PEHO-like syndrome	+/-	CT or MRI: cerebral cortex atrophy or hypoplasia, focal or generalized
PCH1A: pontocerebella	+/-	CT or MRI: corpus callosum hypogenesis
PCH3: pontocerebellar	+/-	MR Spectroscopy: lactate high in brain
Microcephaly, postnata	+/-	+Basal ganglia nature to brain calcifications
Dekaban-Arima colobom	+/-	MRI: white matter abnormality
LIS1: Lissencephaly, iso	+/-	CLP1 gene mutations (biallelic)
HLD4: Leukodystrophy	+/-	Neopterin high in CSF
Muscular dystrophy-dy	+/-	MR Spectroscopy: NAA low in brain
JBTS1: INPP5E Joubert	+/-	CT or MRI: vermal cerebellar atrophy or hypoplasia
Mevalonic aciduria, inf		

More tips
Summary
Note

Tip: The "Patient summary" button at right pops up a patient report
OMIM
Order gene test

Require a finding

One or more findings may be so striking or unusual that you only want to see diseases with that finding. By default, the software includes the possibility a finding may be incidental. To override this default, you can require the finding, by selecting the box next to the onset box and selecting Required, which displays a "*" to the left of the onset, as is shown below for hyperreflexia. This is most useful for a common finding that is very striking in a patient.

Figure 24: How to require a finding

Differential diagnosis
Add findings
Add tests
Patient

☐ Advanced mode
[To home screen](#)

Diseases

Patient: 2 year old boy
Change initial information

Pertinent positive findings

*

≤ 1m

Nystagmus, non-rotary

*

≤ 6m

Hyperreflexia

*

✓

CT or MRI: brainstem atrophy or hypoplasia

*

@1m

Microcephaly

✓

History of a similar disorder in family or contacts

Pertinent negative findings

✗

Regression

✗

Early death if undiagnosed

More tips
Summary
Note

Tip: The "Patient summary" button at right pops up a patient report
OMIM
Order gene test

Advanced Mode: Use the “Profile disease”

To use this feature, click the “**Advanced**” button in the top right of your screen, then click on a button for a particular disease, then click the Profile disease button to view or comment on findings in that disease, including adding a set of pertinent negatives.

TIP

Use the disease profile when you suspect a disease.

Figure 25: Using Advanced mode and the disease profile

The screenshot displays the 'Advanced mode' interface. At the top, there are four tabs: 'Differential diagnosis' (selected), 'Add findings', 'Add tests', and 'Patient'. A 'Differential diagnosis' sub-tab is also visible. Below the tabs, there are checkboxes for 'Incidence' (checked), 'Onset' (checked), and 'Genome' (unchecked). The main area shows a list of diseases with corresponding 'Relative Probability' bars. The diseases listed are:

- PCH2: pontocerebellar hypoplasia 2
- Aicardi-Goutières syndrome, AR
- PCH8: pontocerebellar hypoplasia, CHMP1A-related
- PCH10: Pontocerebellar hypoplasia, CLP1-related
- LIS2: RELN-related lissencephaly, AR
- VLDLR-related cerebellar hypoplasia
- PCH1B: pontocerebellar hypoplasia, EXOSC3-related
- Muscular dystrophy-dystroglycanopathy B6
- PCH9: pontocerebellar hypoplasia, AMPD2
- PEHO-like syndrome
- PCH1A: pontocerebellar hypoplasia, VRK1-related
- PCH3: pontocerebellar hypoplasia 3
- Microcephaly, postnatal progressive, with seizures and brain atroph
- LIS1: Lissencephaly, isolated
- HLD4: Leukodystrophy, hypomyelinating, 4
- Mevalonic aciduria, infantile
- PCH4: pontocerebellar hypoplasia, TSEN54-related
- LISX1: DCX-related lissencephaly, X-linked
- Mucopolipidosis IV (sialolipidosis), typical form
- Hemorrhagic destruction of the brain, subependymal calcification, a

On the right side, there is a sidebar with buttons: 'Differential Dx', 'Gene discovery', 'Assess disease', 'Profile disease', 'Database', 'Search', 'File', 'Home', and 'Help'. At the bottom, there is a bar with buttons: 'More tips', 'Summary', 'Note', 'OMIM', and 'Order gene test'. A tip at the bottom left reads: 'Tip: [GeneReviews: Pontocerebellar Hypoplasia 2 & 4](#)'.

In addition to using the “**Profile disease**” screen as another vehicle to enter the same findings, the screen can be used to understand how a disease unfolds over time. Use the purple “**Scroll age**” button at the top right to see which findings will be present now (black), at each age, and also, how frequently overall that finding is present in that disease, but will emerge later (purple).

Figure 26: Entering findings using the ‘Disease profile’ list (shown at the patient’s current age)

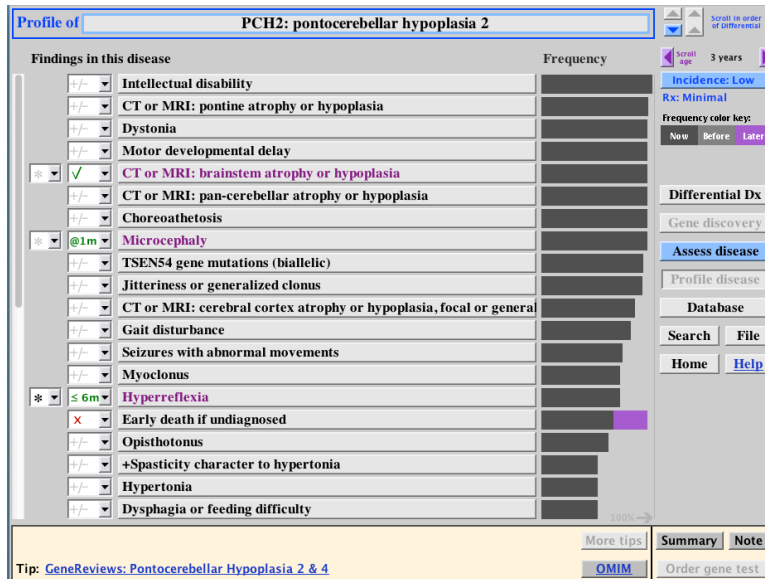
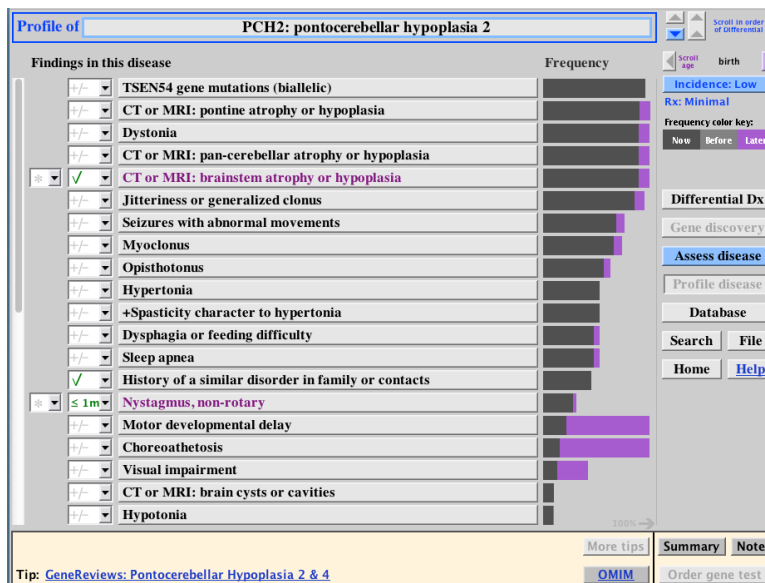


Figure 27: The same 'Disease Profile' for PCH2 shown at birth



Advanced Mode: Use the “Assess disease”

When dictating a note about the rationale for the diagnosis, click on the top diseases in the differential diagnosis and select the **Assess Disease** button.

Figure 28: The Differential Diagnosis

TIP

Use the Assess Disease when dictating your note to explain the rationale for your differential diagnosis.

The screenshot displays the 'Differential diagnosis' tab in a software interface. The top navigation bar includes 'Differential diagnosis', 'Add findings', 'Add tests', and 'Patient'. The 'Advanced mode' checkbox is checked. The main area shows a list of diseases with columns for 'Incidence', 'Onset', and 'Relative Probability'. The first disease, 'PCH2: pontocerebellar hypoplasia 2', is highlighted. A blue arrow labeled 'Select Disease' points to this entry. Another blue arrow labeled 'Assess Disease' points to the 'Assess disease' button in the right-hand sidebar. The sidebar also contains buttons for 'Differential Dx', 'Incidental genes', 'Discovery genes', 'Panel of genes', 'Loss of heteroz', 'Prognosis', 'Profile disease', 'Database', 'Search', 'File', 'Start', and 'Help'. At the bottom, there are tips and a search bar.

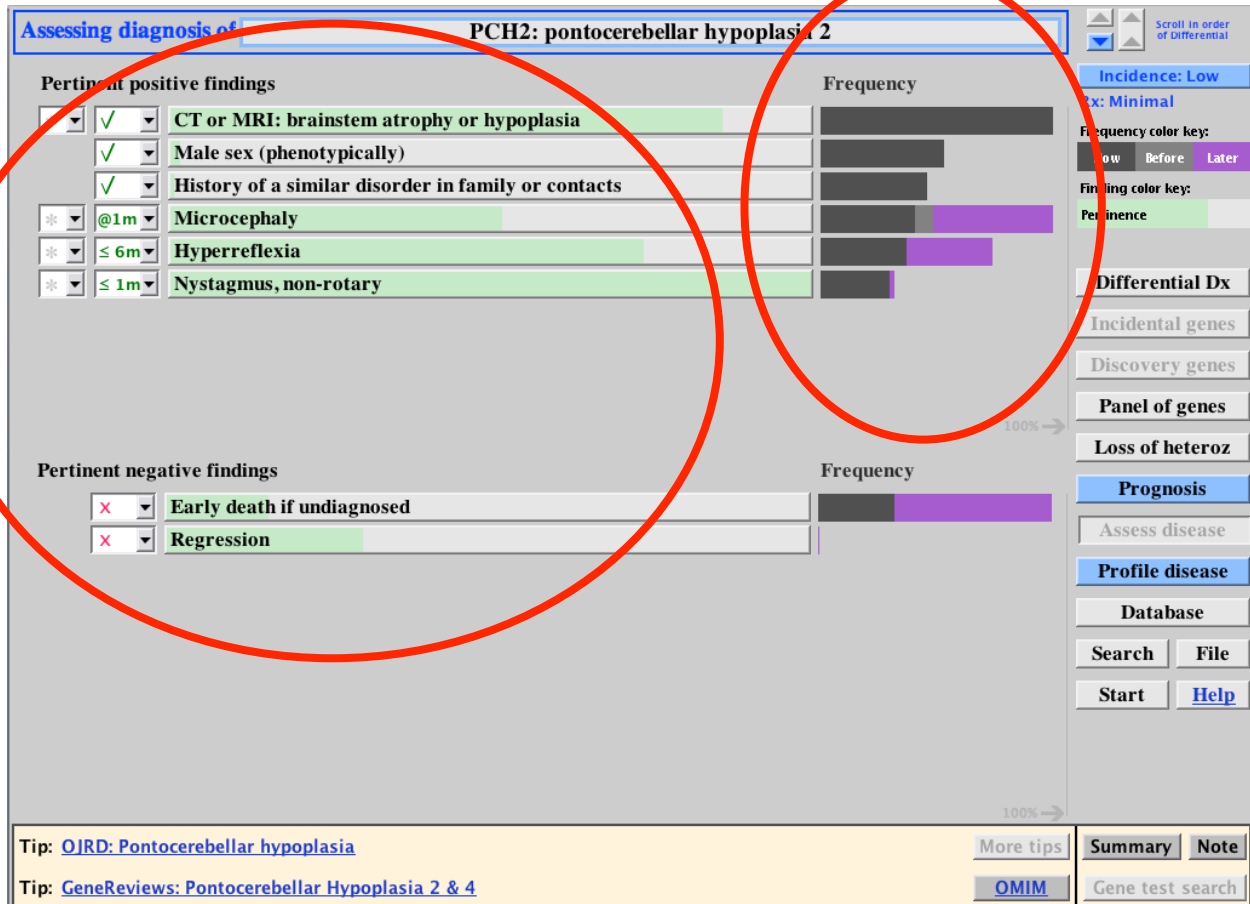
Tip: [OJRD: Pontocerebellar hypoplasia](#)

Tip: [GeneReviews: Pontocerebellar Hypoplasia 2 & 4](#)

More tips | [OMIM](#) | [Summary](#) | [Note](#) | [Gene test search](#)

You will see a screen that super-imposes the patient findings on the frequency of those findings in the disease, in this case PCH2. The length of the frequency bar is the overall frequency of that finding in patients with PCH2. The color of the bar indicates the onset. Black for onset now, versus, onset at an earlier age in gray or later in purple.

Figure 29: Assess disease screen for PCH2



In general, a “good” fit is a lot of black on top and very little on the bottom. To see why Aicardi Goutieres syndrome is a less good fit, use the down arrow at the page to tab through the differential diagnosis diseases to see the fit.

Figure 30: Tabbing through the differential diagnosis using Assess Disease

Assessing diagnosis of **PCH2: pontocerebellar hypoplasia 2**

Pertinent positive findings

		Frequency
* ✓	CT or MRI: brainstem atrophy or hypoplasia	[Bar]
✓	Male sex (phenotypically)	[Bar]
✓	History of a similar disorder in family or contacts	[Bar]
* @1m	Microcephaly	[Bar]
* ≤ 6m	Hyperreflexia	[Bar]
* ≤ 1m	Nystagmus, non-rotary	[Bar]

Pertinent negative findings

		Frequency
x	Early death if undiagnosed	[Bar]
x	Regression	[Bar]

Right Sidebar:

- Incidence: Low
- Rx: Minimal
- Frequency color key: Now Before Later
- Finding color key: Pertinence
- Differential Dx
- Incidental genes
- Discovery genes
- Panel of genes
- Loss of heteroz
- Prognosis
- Assess disease
- Profile disease
- Database
- Search File
- Start Help
- Summary Note
- Gene test search

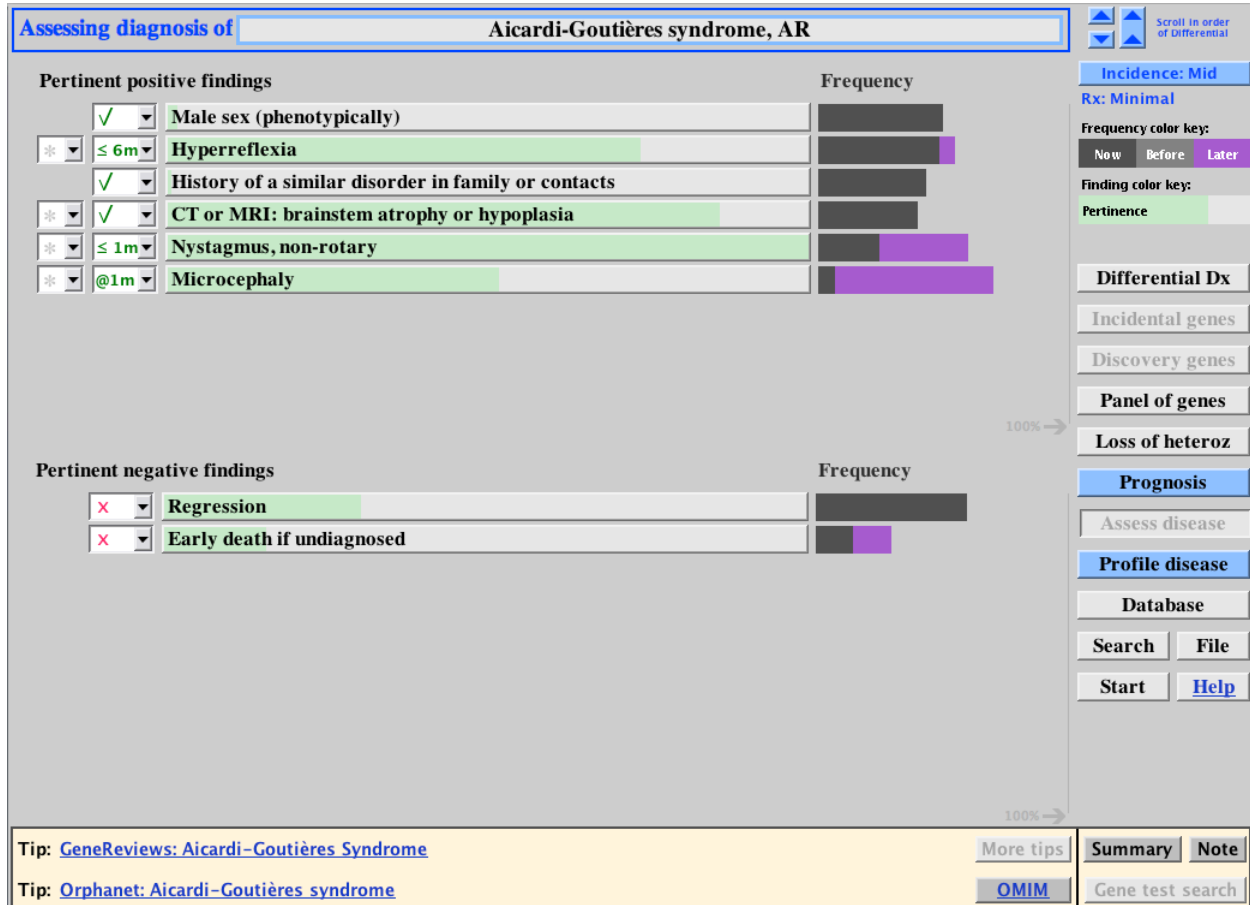
Bottom:

Tip: [OJRD: Pontocerebellar hypoplasia](#) [More tips](#)

Tip: [GeneReviews: Pontocerebellar Hypoplasia 2 & 4](#) [OMIM](#)

Notice that in Aicardi-Goutieres syndrome roughly 60% of patients would have regression and all would have it by now, and in addition, perhaps 10% of patients would already have died. This makes it somewhat less likely that Aicardi-Goutieres is the diagnosis, as reflected in the differential diagnosis.

Figure 31: Fit of the #2 disease in Diff Dx

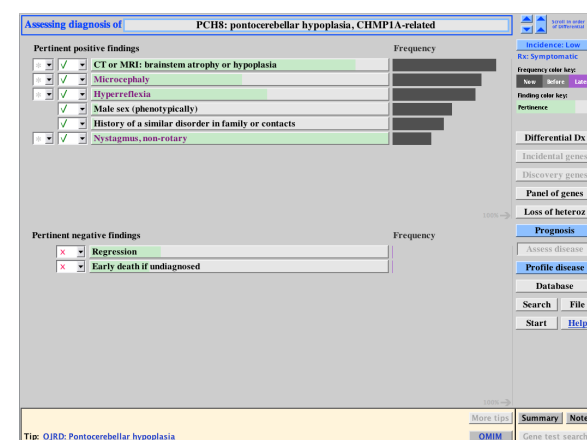
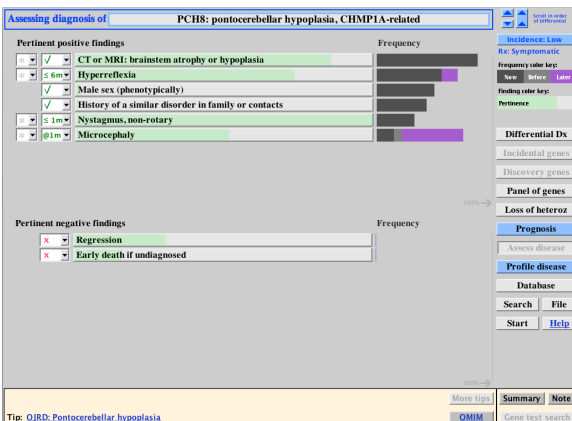
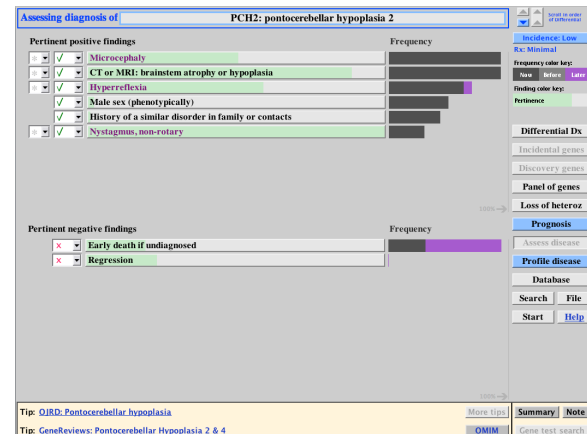
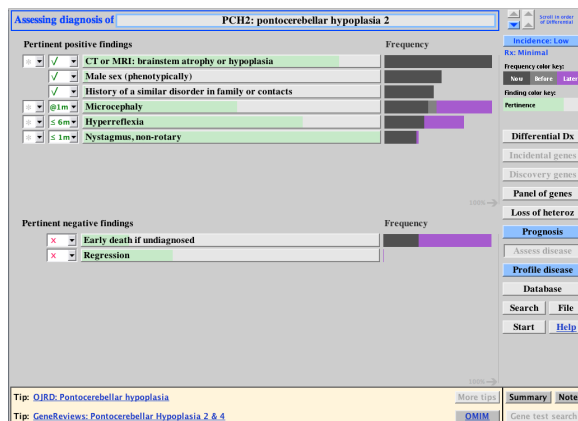
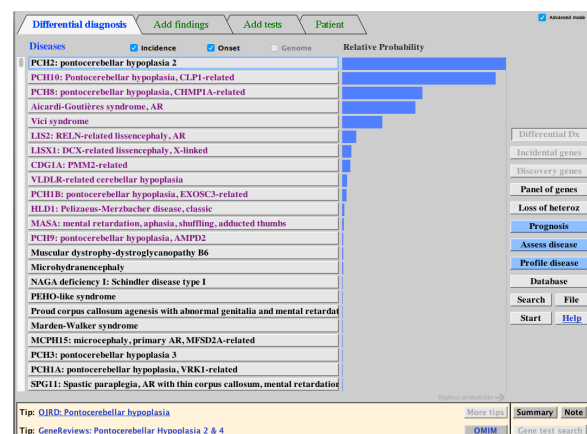
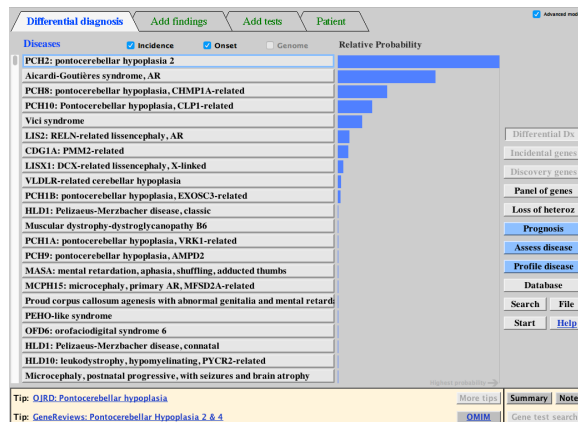


“Assess disease” and the importance of onset information

A simplistic notion of diagnosis simply specifies presence or absence, but this significantly reduces the ability to differentiate among diseases in the differential diagnosis. This is also obvious when looking at the Assess Disease, where the top diseases all seem like good fits.

Not using onset leaves a less tight differential and makes the diseases seem equally probable. (Left is with onset, right is without.)

Figure 32: Differential and Assessment with and without onset



SAVE AND REOPEN PATIENT HISTORY

Output options

The software has three key outputs:

1. **Summary:** The Summary, a human readable “Informatics Lab Report” that provides a snapshot of the patient’s findings as they were entered and run against the database with a particular date stamp. The display includes the pertinence of each finding and the likelihood of each disease in the Differential Diagnosis, as well as recommended tests.
2. **Query string:** SimulConsult has created special codes (in the form of a query string) that allow you to reopen SimulConsult with the previously entered findings already present. (Note: In the version of SimulConsult accessed from the cloud, the Summary is machine-readable because it has the query string embedded in it).
3. **Note:** The Note is in the classic “subjective, objective, assessment, and plan” (SOAP) note format used in clinical medicine. In the version of SimulConsult accessed from the cloud, it outputs a file that can be copied and pasted into a medical record as plain text and then edited. It also contains the query string that can be saved. Note: since only medical professionals may have direct access to SimulConsult it is not advised to put the query string in the note, if notes are open to patient inspection, such as through Open Notes.

In addition, some coded outputs are possible within the Enterprise offerings.

Save the “Summary” with the “Finding Code”

There are two ways to save the patient findings, depending on your purpose.

1. Click on the “**Summary**” button and then save the HTML page using the save command in your browser. Note: this produces a version that is both human readable and also can be reopened and read by the software.

Figure 33: Generating the HTML Patient Summary

The screenshot shows a web application interface for patient management. At the top, there are tabs for 'Differential diagnosis', 'Add findings', 'Add tests', and 'Patient'. The 'Patient' tab is selected. Below the tabs, there is a section for 'Patient: 2 year old boy' with a 'Change initial information' link. To the left is a list of diseases, including 'PCH2: pontocerebellar', 'Aicardi-Goutières synd', 'PCH8: pontocerebellar', 'PCH10: Pontocerebellar', 'LIS2: RELN-related liss', 'CDG1A: PMM2-related', 'VLDLR-related cerebel', 'PCH1B: pontocerebella', 'Muscular dystrophy-dy', 'PCH9: pontocerebellar', 'PEHO-like syndrome', 'PCH1A: pontocerebella', 'PCH3: pontocerebellar', 'Microcephaly, postnata', 'Dekaban-Arima colobor', 'LIS1: Lissencephaly, isc', 'HLD4: Leukodystrophy', 'Muscular dystrophy-dy', 'JBTS1: INPP5E Joubert', and 'Mevalonic aciduria, inf'. The main area displays 'Pertinent positive findings' and 'Pertinent negative findings'. The positive findings include 'Nystagmus, non-rotary', 'Hyperreflexia', 'CT or MRI: brainstem atrophy or hypoplasia', 'Microcephaly', and 'History of a similar disorder in family or contacts'. The negative findings include 'Regression' and 'Early death if undiagnosed'. On the right side, there are buttons for 'Differential Dx', 'Gene discovery', 'Assess finding', 'Profile finding', 'Database', 'Search', 'File', 'Home', and 'Help'. At the bottom right, there are buttons for 'Summary' and 'Note'. A blue arrow points to the 'Summary' button. A tip at the bottom left states: 'Tip: The "Summary" and "Note" buttons at right display patient reports'.

Advanced mode

Differential diagnosis Add findings Add tests Patient

Diseases

Patient: 2 year old boy Change initial information

Pertinent positive findings

Finding color key: Pertinence

* ≤ 1m Nystagmus, non-rotary

* ≤ 6m Hyperreflexia

* √ CT or MRI: brainstem atrophy or hypoplasia

* @1m Microcephaly

√ History of a similar disorder in family or contacts

Pertinent negative findings

x Regression

x Early death if undiagnosed

Differential Dx

Gene discovery

Assess finding

Profile finding

Database

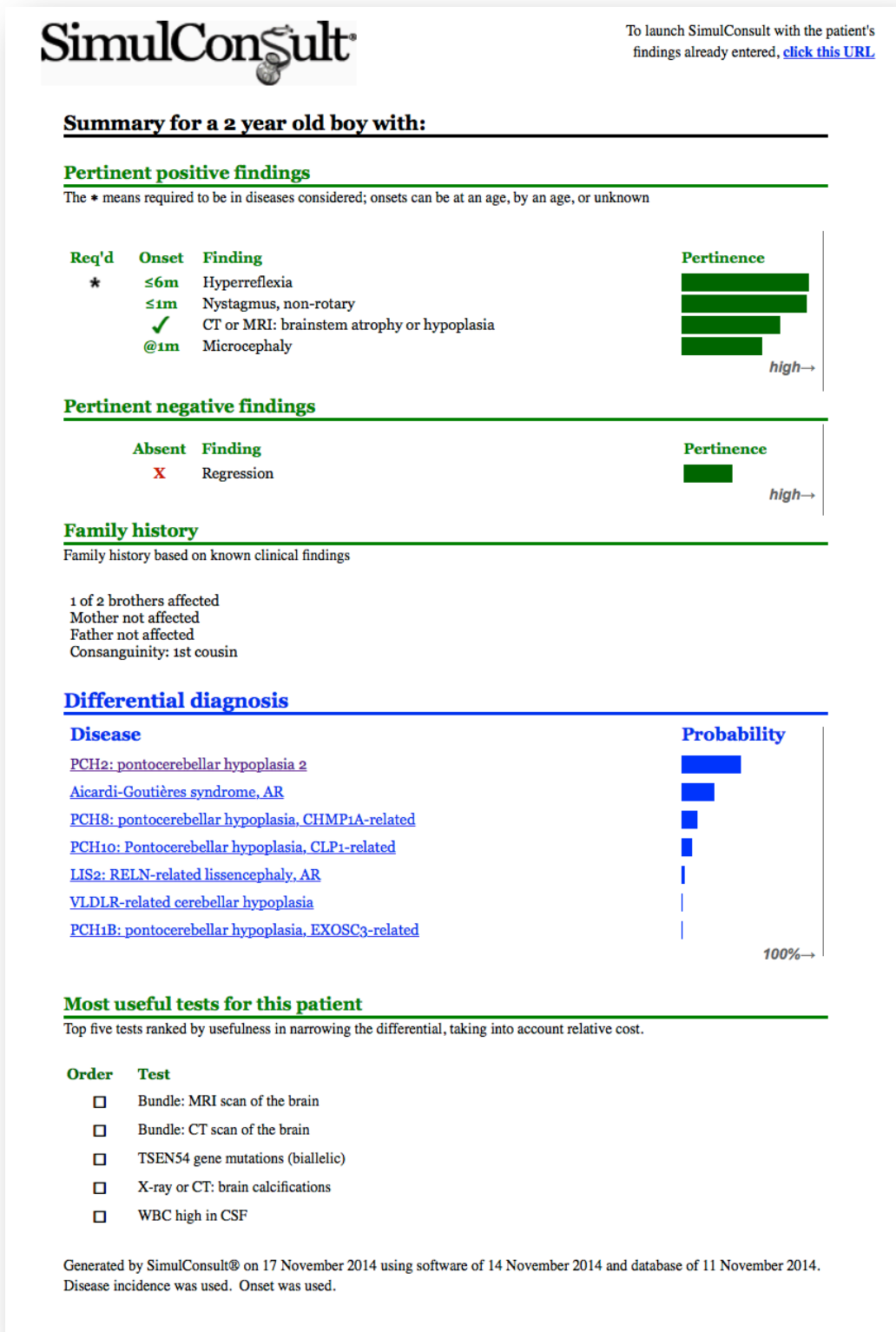
Search File

Home Help

Summary Note

Tip: The "Summary" and "Note" buttons at right display patient reports

Figure 34: The HTML Patient Summary (can be saved, printed, or printed to PDF)



- The second approach to saving the patient information uses the “**File**” button visible in the **advanced mode**.

Figure 35: Save the Patient Summary using the advanced mode and file button

The screenshot shows the OMIM (Online Mendelian Inheritance in Man) interface in 'Advanced mode'. The top navigation bar includes 'Differential diagnosis', 'Add findings', 'Add tests', and 'Patient'. The 'Advanced mode' checkbox is checked in the top right corner. The main content area displays a list of diseases under the 'Differential diagnosis' tab, with columns for 'Incidence', 'Onset', 'Genome', and 'Relative Probability'. The diseases listed include PCH2: pontocerebellar hypoplasia 2, Aicardi-Goutières syndrome, AR, PCH8: pontocerebellar hypoplasia, CHMP1A-related, PCH10: Pontocerebellar hypoplasia, CLP1-related, LIS2: RELN-related lissencephaly, AR, VLDLR-related cerebellar hypoplasia, PCH1B: pontocerebellar hypoplasia, EXOSC3-related, Muscular dystrophy-dystroglycanopathy B6, PCH9: pontocerebellar hypoplasia, AMPD2, PEHO-like syndrome, PCH1A: pontocerebellar hypoplasia, VRK1-related, PCH3: pontocerebellar hypoplasia 3, Microcephaly, postnatal progressive, with seizures and brain atrophy, LIS1: Lissencephaly, isolated, HLD4: Leukodystrophy, hypomyelinating, 4, Mevalonic aciduria, infantile, PCH4: pontocerebellar hypoplasia, TSEN54-related, LISX1: DCX-related lissencephaly, X-linked, Mucopolysaccharidosis IV (sialolipidosis), typical form, and Hemorrhagic destruction of the brain, subependymal calcification, a. The 'File' button is located in the bottom right corner of the interface, next to the 'Database' button. A blue arrow points to the 'File' button, and another blue arrow points to the 'Advanced mode' checkbox.

Advanced mode

File button

Differential diagnosis

Add findings

Add tests

Patient

Advanced mode

Diseases

Incidence

Onset

Genome

Relative Probability

PCH2: pontocerebellar hypoplasia 2

Aicardi-Goutières syndrome, AR

PCH8: pontocerebellar hypoplasia, CHMP1A-related

PCH10: Pontocerebellar hypoplasia, CLP1-related

LIS2: RELN-related lissencephaly, AR

VLDLR-related cerebellar hypoplasia

PCH1B: pontocerebellar hypoplasia, EXOSC3-related

Muscular dystrophy-dystroglycanopathy B6

PCH9: pontocerebellar hypoplasia, AMPD2

PEHO-like syndrome

PCH1A: pontocerebellar hypoplasia, VRK1-related

PCH3: pontocerebellar hypoplasia 3

Microcephaly, postnatal progressive, with seizures and brain atrophy

LIS1: Lissencephaly, isolated

HLD4: Leukodystrophy, hypomyelinating, 4

Mevalonic aciduria, infantile

PCH4: pontocerebellar hypoplasia, TSEN54-related

LISX1: DCX-related lissencephaly, X-linked

Mucopolysaccharidosis IV (sialolipidosis), typical form

Hemorrhagic destruction of the brain, subependymal calcification, a

Differential Dx

Gene discovery

Assess disease

Profile disease

Database

File

Home

Help

More tips

Summary

Note

OMIM

Order gene test

Tip: [GeneReviews: Pontocerebellar Hypoplasia 2 & 4](#)

Figure 36: Saving the Patient Summary

The screenshot shows the '1. Findings' tab of a software interface. The main area is titled 'Use a File or Text String for patient findings'. It contains a 'File name: Summary.html' field. Below this are two buttons: 'Open file' (labeled 'Open a Patient Summary (.html format)') and 'Save file' (labeled 'Save a Patient Summary (.html format)'). A blue arrow points to the 'Save file' button, with the text 'Save File button' next to it. Below the file section is a 'Text String representation of patient findings' section. It includes a text input field, a 'Paste' button, and a 'Load patient' button. Below the input field is a text string: '4=1&t5=2&t8=1&t9=1&t10=1&i=t&t=c'. To the right of this string is a 'Copy current patient' button, with the text '(Copy to the system clipboard)' below it. Below the copy button is a 'Clear current patient' button. On the right side of the interface, there is a vertical list of buttons: 'Differential Dx', 'Incidental genes', 'Discovery genes', 'Panel of genes', 'Loss of heteroz', 'Prognosis', 'Assess finding', 'Profile finding', 'Database', 'Search', 'File', 'Start', and 'Help'.

Save the “Finding Code” directly

You can also choose to save the find code, which will allow you to reopen the patient. This is often useful in a shared exam room where you can not save to the desktop. Use the copy current patient button and paste the code in an email to yourself.

Figure 37: Save the finding code directly

The screenshot shows the '1. Findings' tab of a software interface, similar to Figure 36. The main area is titled 'Use a File or Text String for patient findings'. It contains a 'File name: Summary.html' field. Below this are two buttons: 'Open file' (labeled 'Open a Patient Summary (.html format)') and 'Save file' (labeled 'Save a Patient Summary (.html format)'). Below the file section is a 'Text String representation of patient findings' section. It includes a text input field, a 'Paste' button, and a 'Load patient' button. Below the input field is a text string: '4=1&t5=2&t8=1&t9=1&t10=1&i=t&t=c'. To the right of this string is a 'Copy current patient' button, with the text '(Copy to the system clipboard)' below it. A blue arrow points to the 'Copy current patient' button, with the text 'Save Finding Code' next to it. Below the copy button is a 'Clear current patient' button. On the right side of the interface, there is a vertical list of buttons: 'Differential Dx', 'Incidental genes', 'Loss of heteroz', 'Prognosis', 'Assess finding', 'Profile finding', 'Database', 'Search', 'File', 'Start', and 'Help'.

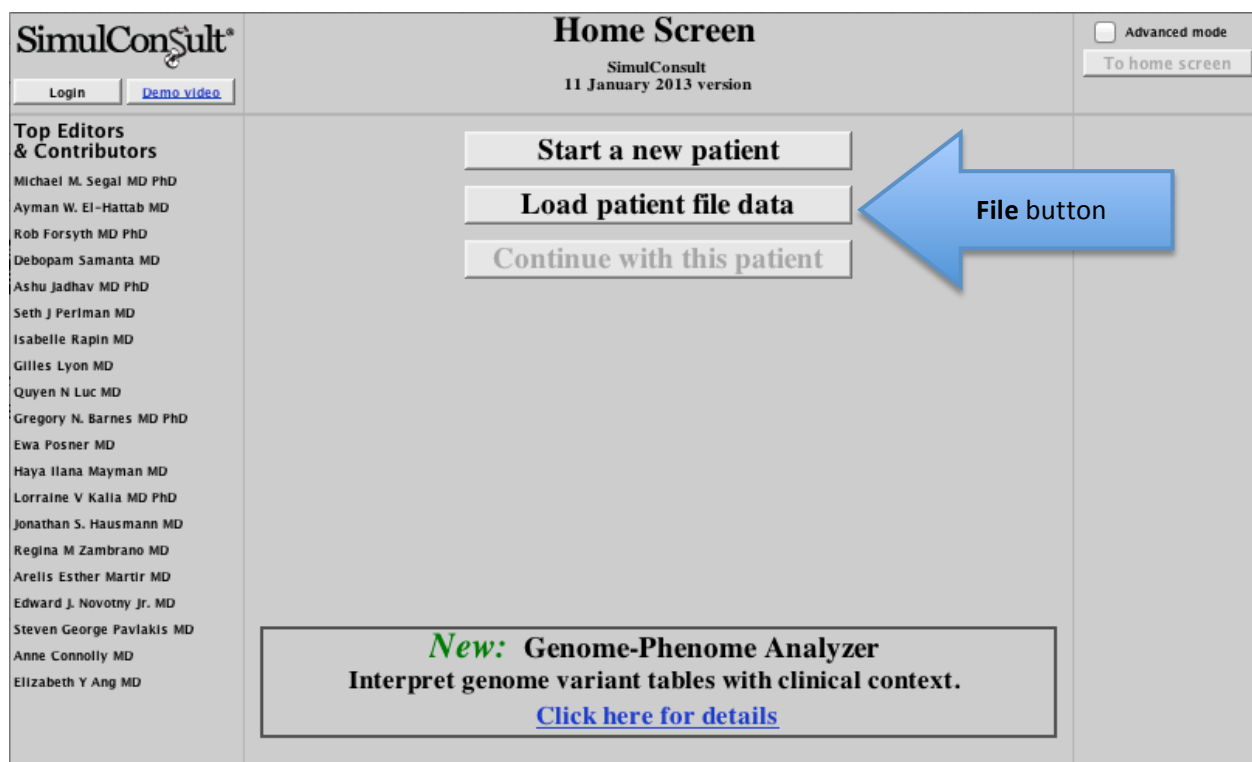
Open previously saved findings

Some users separate the task of collecting and entering clinical findings from subsequent interpretation or sharing with the lab. The tasks may be divided among people or by time.

In some radiology and genomics labs, some users are exploring ways to get the referring clinician to submit the Patient Summary as part of the referral, so that the clinical picture can include the most robust information about findings. In any of these scenarios, as well as one where you just need to save your work and come back later, you can reopen a patient summary easily. In **advanced mode**, use the **“File”** button for to open the previously saved file. If you have an enterprise version, use the links described in the companion guide for launching and saving in your environment.

Note: those with Genome-Phenome privileges also can save a step and load a previously saved patient file from the Home screen.

Figure 38: Load patient data



Note: For those with Genome-Phenome privileges, you must enter clinical findings directly or open a previously saved patient file of clinical findings before you can open a variant table.

Figure 39: Open patient file

The screenshot shows the '1. Findings' tab of a software interface. At the top, there are three tabs: '1. Findings' (active), '2. Variants', and '3. Pedigree'. Below the tabs, the main area is titled 'Use a File or Text String for patient findings'. It contains a 'File name: Summary.html' label. Below this, there are two buttons: 'Open file' and 'Open a Patient Summary (.html format)'. A blue arrow points to the 'Open file' button with the text 'Open File button'. Below these buttons, there is a 'Save file' button and a 'Save a Patient Summary (.html format)' button. Further down, there is a section titled 'Text String representation of patient findings' with a text input field, a 'Paste' button, and a 'Load patient' button. Below the input field, there is a text string 'i4=1&t5=2&t8=1&t9=1&t10=1&i=t&t=c' and a 'Copy current patient' button with the subtext '(Copy to the system clipboard)'. At the bottom of this section is a 'Clear current patient' button. On the right side of the interface, there is a vertical menu with buttons: 'Differential Dx', 'Incidental genes', 'Discovery genes', 'Panel of genes', 'Loss of heteroz', 'Prognosis', 'Assess finding', 'Profile finding', 'Database', 'Search', 'File', 'Start', and 'Help'.

Figure 40: Open using the Finding Code

The screenshot shows the '1. Findings' tab of a software interface, similar to Figure 39. A blue arrow points to the 'Paste' button in the 'Text String representation of patient findings' section with the text 'Paste a previously saved finding code here to reopen that'. The rest of the interface is identical to Figure 39, including the tabs, buttons, and vertical menu on the right.

Saving the “Note”

The Note can be saved using the Note button.

Figure 41: Note button

The screenshot displays a web-based clinical decision support system interface. At the top, there are tabs for 'Differential diagnosis', 'Add findings', 'Add tests', and 'Patient'. The 'Patient' tab is currently selected. Below the tabs, the patient information is shown as 'Patient: 2 year old boy' with a 'Change initial information' link. The interface is divided into two main sections: 'Pertinent positive findings' and 'Pertinent negative findings'. The 'Pertinent positive findings' section lists several findings with associated severity levels (indicated by asterisks and timeframes) and a 'Finding color key' for 'Pertinence'. The 'Pertinent negative findings' section lists findings with associated severity levels. On the right side, there is a sidebar with buttons for 'Differential Dx', 'Gene discovery', 'Assess finding', 'Profile finding', 'Database', 'Search', 'File', 'Home', and 'Help'. At the bottom right, a blue arrow points to a 'Note' button, which is labeled 'Note button'. A tip at the bottom left states: 'Tip: The "Summary" and "Note" buttons at right display patient reports'.

Differential diagnosis **Add findings** **Add tests** **Patient** ☒ Advanced mode

Diseases

- PCH2: pontocerebellar
- Aicardi-Goutières syndr
- PCH8: pontocerebellar
- PCH10: Pontocerebellar
- LIS2: RELN-related liss
- CDG1A: PMM2-related
- VLDLR-related cerebell
- PCH1B: pontocerebellar
- Muscular dystrophy-dys
- PCH9: pontocerebellar
- PEHO-like syndrome
- PCH1A: pontocerebellar
- PCH3: pontocerebellar
- Microcephaly, postnata
- Dekaban-Arima colobom
- LIS1: Lissencephaly, isc
- HLD4: Leukodystrophy
- Muscular dystrophy-dys
- JBTS1: INPP5E Joubert
- Mevalonic aciduria, inf

Patient: 2 year old boy [Change initial information](#)

Pertinent positive findings **Finding color key:**
Pertinence

* ≤ 1m	Nystagmus, non-rotary
* ≤ 6m	Hyperreflexia
* ✓	CT or MRI: brainstem atrophy or hypoplasia
* @1m	Microcephaly
✓	History of a similar disorder in family or contacts

Pertinent negative findings

x	Regression
x	Early death if undiagnosed

Note button **Note**

Tip: The "Summary" and "Note" buttons at right display patient reports

OMIM [Order gene test](#)

The output of the Note is organized into traditional groups. In some implementations, it outputs as a single unit of text. In others, it has been broken into sections and can be placed within an existing note template.

Figure 42: SOAP Note output in plain text

HISTORY OF PRESENT ILLNESS

This is a 2 year old boy with
Nystagmus, non-rotary, onset by 1 month old
Microcephaly, onset at 1 month old
Hyperreflexia, onset by 6 months old
CT or MRI: brainstem atrophy or hypoplasia, present now

Regression, absent

Growth / development

Microcephaly, onset at 1 month old

FAMILY HISTORY

1 of 2 brothers affected
Mother not affected
Father not affected
Consanguinity: 1st cousin

PHYSICAL EXAM

Present

Hyperreflexia
Nystagmus, non-rotary
Microcephaly

LAB / STUDIES

Present

CT or MRI: brainstem atrophy or hypoplasia, present now

ASSESSMENT

This is a 2 year old boy with:

Pertinent positives

Hyperreflexia
Nystagmus, non-rotary
CT or MRI: brainstem atrophy or hypoplasia
Microcephaly

Pertinent negatives

Regression

Differential Diagnosis

PCH2: pontocerebellar hypoplasia 2
Aicardi-Goutieres syndrome, AR
PCH8: pontocerebellar hypoplasia, CHMP1A-related
PCH10: Pontocerebellar hypoplasia, CLP1-related
LIS2: RELN-related lissencephaly, AR
VLDLR-related cerebellar hypoplasia
PCH1B: pontocerebellar hypoplasia, EXOSC3-related

PLAN

Most useful tests for this patient

Bundle: MRI scan of the brain
Bundle: CT scan of the brain
TSEN54 gene mutations (biallelic)
X-ray or CT: brain calcifications
WBC high in CSF

Some of this data was entered using the Simulconsult Diagnostic Decision Support software. To re-launch the software with the patient's findings as used in this session, copy and paste this URL into a browser window:

http://www.simulconsult.com/run/?d=730&u=ftemp1&o=499999&u=ftemp20&o=59&u=ftemp158&o=399999&u=ftemp220&o=b59&u=ftemp270&o=rb269&u=segal_020614172829&o=

You have the option to select the diseases you want to include in the differential diagnosis before viewing the note.

Figure 43: Select diseases to include in Note before generating Note

Choosing diseases

- ☒ 1: PCH2: pontocerebellar hypoplasia 2
- ☒ 2: Aicardi-Goutières syndrome, AR
- ☒ 3: PCH8: pontocerebellar hypoplasia, CHMP1A-related
- ☒ 4: PCH10: Pontocerebellar hypoplasia, CLP1-related
- ☒ 5: Vici syndrome
- ☒ 6: LIS2: RELN-related lissencephaly, AR
- ☒ 7: CDG1A: PMM2-related
- ☒ 8: LISX1: DCX-related lissencephaly, X-linked
- ☒ 9: VLDLR-related cerebellar hypoplasia
- ☐ 10: PCH1B: pontocerebellar hypoplasia, EXOSC3-related
- ☐ 11: HLD1: Pelizaeus-Merzbacher disease, classic
- ☐ 12: Muscular dystrophy-dystroglycanopathy B6
- ☐ 13: PCH1A: pontocerebellar hypoplasia, VRK1-related
- ☐ 14: PCH9: pontocerebellar hypoplasia, AMPD2
- ☐ 15: MASA: mental retardation, aphasia, shuffling, adducted thumbs

If you select a subset of the diseases, the note reflects it.

Figure 44: Note with subset of selected diagnoses in Diff Dx

HISTORY OF PRESENT ILLNESS

This is a 2 year old boy with
Nystagmus, non-rotary, onset by about 1 month old
Microcephaly, onset at about 1 month old
Hyperreflexia, onset by about 6 months old
CT or MRI: brainstem atrophy or hypoplasia, present now

Regression, absent

Growth / development

Microcephaly, onset at about 1 month old

FAMILY HISTORY

1 of 2 brothers affected
Mother not affected
Father not affected
Consanguinity: 1st cousin

PHYSICAL EXAM

Present

Nystagmus, non-rotary
Hyperreflexia
Microcephaly

LAB / STUDIES

Present

CT or MRI: brainstem atrophy or hypoplasia, present now

ASSESSMENT

This is a 2 year old boy with:

Pertinent positives

Nystagmus, non-rotary
CT or MRI: brainstem atrophy or hypoplasia
Hyperreflexia
Microcephaly

Pertinent negatives

Regression

Differential Diagnosis

PCH2: pontocerebellar hypoplasia 2
Aicardi-Goutieres syndrome, AR
PCH8: pontocerebellar hypoplasia, CHMP1A-related
PCH10: Pontocerebellar hypoplasia, CLP1-related
Vici syndrome
LIS2: RELN-related lissencephaly, AR
CDG1A: PMM2-related
LISX1: DCX-related lissencephaly, X-linked
VLDLR-related cerebellar hypoplasia

PLAN

Most useful tests for this patient

Bundle: MRI scan of the brain
Bundle: CT scan of the brain
Personalized panel: 13 most useful genes
X-ray or CT: brain calcifications
TSEN54 gene mutations (biallelic)

Finally, if you have a relationship with Intelligent Medical Objects, we can enable automatic coding of the diseases using ICD10.

PREMIUM UPGRADE OPTIONS: THE PROGNOSIS TABLE

SimulConsult currently offers an upgrade option to the Phenome version to have access to the Prognosis Table for all 6,000-plus diseases. When returning results and a diagnosis to patients and their primary care physicians, it is often helpful to have an intuitive way to convey the expectations about how the disease will unfold. Research has shown the Prognosis table needs no explanation.

Figure 45: Prognosis Table

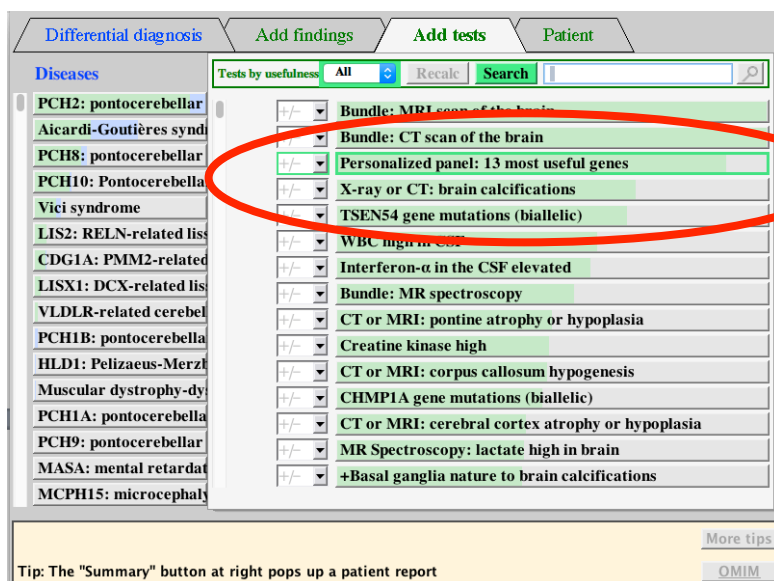
Prognosis for PCH2: pontocerebellar hypoplasia 2													
At what age do people with this disease have these findings?													
Signs and Symptoms	Birth	1 month	3 months	6 months	1 year	3 years	6 years	10 years	15 years	25 years	40 years	60 years	80 years
Dystonia	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most
Jitteriness or generalized clonus	Some	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most
Microcephaly	Few	Some	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most
Choreoathetosis	Few	Some	Some	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most
Motor developmental delay	Few	Some	Some	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most
Intellectual disability	NA	Few	Few	Some	Most	Most	Most	Most	Most	Most	Most	Most	Most
Spasticity character to hypertonia	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Myoclonus	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Opisthotonus	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Hypertonia / stiffness	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Sleep apnea	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Seizures with abnormal movements	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Dysphagia or feeding difficulty	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Nystagmus, non-rotary	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Visual impairment despite lens correction	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Hyperreflexia	NA	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Gait disturbance	NA	NA	NA	NA	Some	Some	Some	Some	Some	Some	Some	Some	Some
Contractures or passive limited range of motion	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
Irritability or agitation, pronounced	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
Stature short	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
Heat, fever, or systemic infection triggers attacks or aggravates findings	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
Ataxia	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
Scollis with or without kyphosis	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
Hypotonia	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
Findings detected by laboratory tests													
CT or MRI: brainstem atrophy or hypoplasia	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most
CT or MRI: pontine atrophy or hypoplasia	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most
CT or MRI: pan-cerebellar atrophy or hypoplasia	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most
TSEN54 gene mutations (biallelic)	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most
Creatine kinase high	Few	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
CT or MRI: cerebral cortex atrophy or hypoplasia	NA	NA	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
MRI: white matter abnormality	NA	NA	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some
Myoglobinuria	Few	Few	Few	Few	Few	Few	Few	Few	Some	Some	Some	Some	Some
CT or MRI: brain cysts or cavities	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
X-ray: osteopenia	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
Renal failure or severe dysfunction	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
TSEN34 gene mutations (biallelic)	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
TSEN2 gene mutations (biallelic)	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
SEPS3 gene mutations (biallelic)	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
VPS53 gene mutations (biallelic)	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
KEY	None or NA			Few is less than or equal to 30%			Some is more than 30%			Most is more than 85%			

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PREMIUM UPGRADE OPTIONS: PERSONALIZED PANEL

The Personalized Panel provides suggestions of the most important genes to order in the initial round of testing. If the list is long, it indicates consideration of large panels or an exome may be warranted. If short, Sanger-sequencing a few genes may be an inexpensive route to an answer for this patient. The personalized panel also provides a list of the genes that must be well covered. For this reason, it can be helpful both to those ordering the tests and to the labs.

Figure 46: Personalized Panel in the Suggested Tests list



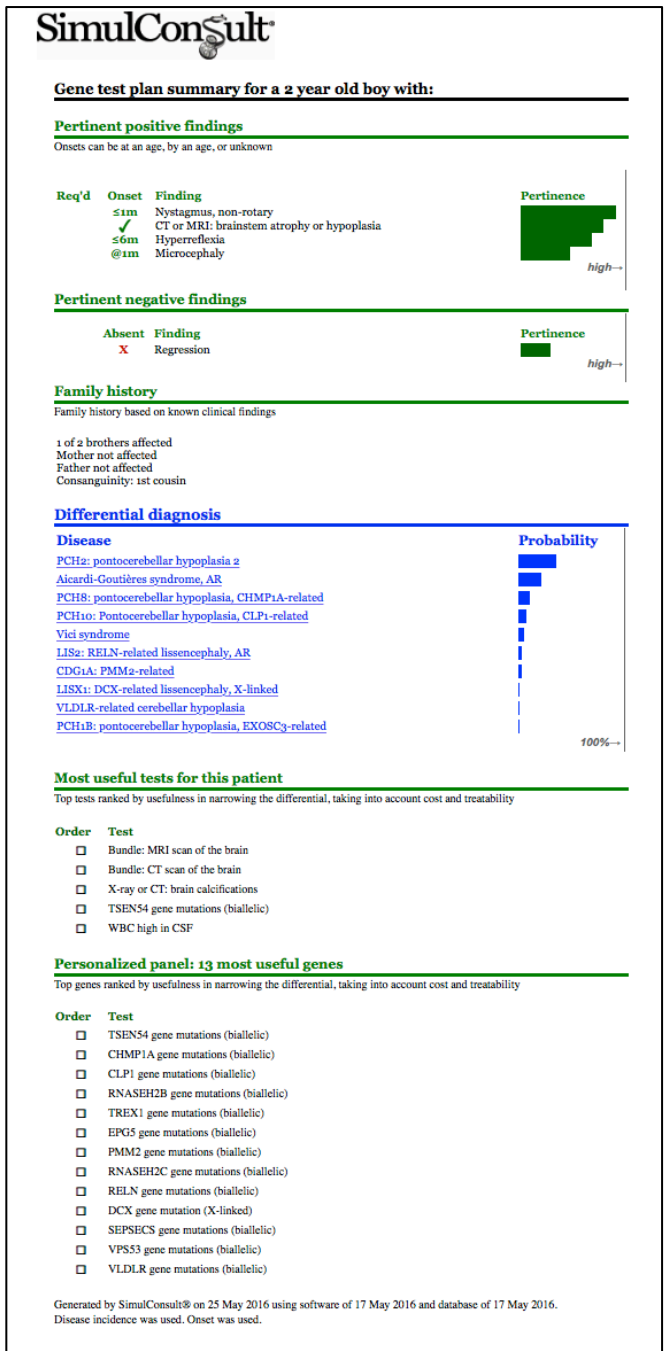
Clicking on the onset button shows the gene list.

Figure 47: Personalized Panel

Findings in		Personalized panel: 13 most useful genes
+/-	▼	TSEN54 gene mutations (biallelic)
+/-	▼	CHMP1A gene mutations (biallelic)
+/-	▼	CLP1 gene mutations (biallelic)
+/-	▼	RNASEH2B gene mutations (biallelic)
+/-	▼	TREX1 gene mutations (biallelic)
+/-	▼	EPG5 gene mutations (biallelic)
+/-	▼	PMM2 gene mutations (biallelic)
+/-	▼	RNASEH2C gene mutations (biallelic)
+/-	▼	RELN gene mutations (biallelic)
+/-	▼	DCX gene mutation (X-linked)
+/-	▼	VPS53 gene mutations (biallelic)
+/-	▼	SEPSECS gene mutations (biallelic)
+/-	▼	VLDLR gene mutations (biallelic)

The Personalized Panel is also added to the Patient Summary.

Figure 48: Patient Summary with Personalized Panel



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