SimulConsult®
A Simultaneous Consult On Your Patient's Diagnosis

Basic features of SimulConsult

Diagnosis is one of the most complex intellectual tasks in medicine. Diagnosis requires taking knowledge about findings (symptoms, signs and test results) learned as "disease X has these findings" and inverting it to instead ask "with this group of findings, what diseases are likely". The process of diagnosis is iterative – the diseases under consideration determine the most useful questions to ask next, the answers to which in turn affects the probability of the diseases under consideration in the differential diagnosis.

The SimulConsult Diagnostic Decision Support System (**DDSS**) aims to make each clinician ***their best diagnostician***, while saving clinician time, (including accounting for the time to use the tool)**.**

To do this, SimulConsult has curated medicine into a computational database. The database currently covers ~7,500 diseases, including genetic diseases with germline changes convincingly associated with human disease and all convincing chromosomal disorders. It also includes many nongenetic diseases, with particular strength in neurology and rheumatology.

The fundamental tasks performed by the DDSS are as follows:

- **Helps you collect findings.** Enables quick entry of patient findings – both pertinent positives and negatives (Figures 1 & 2).
- **Computes differential diagnosis.** Compares the findings you enter about your patient with every disease in the database and computes a measure of fit resulting in a probability-weighted list of possible diagnoses called a differential diagnosis.
- **Suggests useful findings.** Computes which among its ~10,000 findings would most change the differential diagnosis and displays these findings in rank order of Usefulness. When you specify the presence of absence of a finding, the software recalculates the differential diagnosis and useful findings iteratively each time a finding is entered.

Note: The DDSS ***does not*** use machine learning, but rather manual curation by expert clinicians. Machine learning is well-known to be bad at ***unlearning,*** which is a problem because our understanding of medicine continues to change. In addition, many subtle facts about rare diseases have never been written down and are only known to specialists.

Time course of findings. Critical to the performance of the DDSS is the fact that the database has the time course of each finding in each disease, and you specify the time course in your patient when you enter findings, when known. The DDSS then uses time-based Bayesian logic to do a pattern match.

Pertinent negatives. Diagnoses of exclusion and pertinent negatives are a critical part of genetic diagnosis; the DDSS allows you to specify that a finding is absent.

The main "Dx" screen. After you enter the patient age and sex, and an initial finding or findings from search or core lists, you will go to the main screen, "Dx" for diagnose. It has four tabs:

- **Differential.** This shows the evolving differential diagnosis (Figure 3) with the blue bars indicating probability
- **Add findings.** This displays clinical findings not yet commented on, in order of usefulness (Figures 4). The green shading indicates the finding's relative usefulness.
- **Add tests.** This displays test results not yet commented on, in order of usefulness. Instead of "All tests" (Figure 5) you can select to see only one kind of test. Some test "bundles" are shown (e.g., bone imaging in Figure 5) to combine the usefulness of all the results in the test.
- **Rule in or out.** By clicking "Rule in or out selected disease" in the Add findings or Add tests the usefulness assessment can focus on a particular disease (Figure 6).
- **Phenotype.** These are the findings you have entered about the patient, both pertinent positives and negatives.

Getting started using the software

Task	Description
Share button	The rightmost button on the top black navigation bar allows you to load or save a patient using either a PDF file that is both human and machine readable, or a Text string (Figure 7).
Info buttons	These explain features and conventions in the software.
Hover over a disease or finding	If you hover over a disease or finding it will display the synonyms and explanatory terms.
Links	With a disease or a finding highlighted, click Links on the black top navigation bar to get links to relevant resources.
Two key colors	Green is associated with findings and blue with diseases

Entering information about the patient

Task	Description
Basic demographics	On the initial screen, you must enter the age and chromosomal sex of the patient to get started.
Family history	On the Initial screen, this feature allows you to specify if others in the family are affected, if known (Figure 8).
Primary, specialist or tertiary	On the initial screen, this feature allows you to ignore the incidence of the disease and is useful for when a patient has already had many commonly diagnosed conditions ruled out.
"⌕" Search	Click "⌕" on the top black navigation bar and then the Finding search tab. Then begin typing the name of a finding. A list of findings matching that term will appear. (Figure 2)
Core Lists	Click **Core** on the top black navigation bar. The lists of 48 core findings are available for several specialties, but most helpful in specialties for which patients are well described by 48 core findings, such as neurology or rheumatology. (Figure 1)
Presence / Absence	To the left of each finding is a pull-down menu with a "?" (once you've entered an age and sex for the patient). This is where you specify presence or absence. Include onset ***at*** an age or onset ***by*** an age to get maximum use of available data.
Required finding	Occasionally, a finding that is relatively common is so abnormal you want to be sure every disease you consider has that finding. The asterisk to the left of the presence display (when on the Search results and the Phenotype tab of the Dx screen) allows you to specify that a finding is required. Leaving a finding not required means the DDSS will consider the possibility of that finding being incidental to the diagnosis.

Assessing possible diseases

Task	Description
Differential diagnosis	All the tabs on the Dx screen show the differential diagnosis. The Phenotype tab on Dx shows the patient findings entered.
Profile	Click the button of the name of the disease and then click "Profile" up on the black bar to see all the findings in this disease, and click younger or older to see how they emerge over time
Assess	***With a disease selected***, click "Assess" up on the black bar to see the fit between the patient and the information in the database about the disease – this is the reveal of the logic of the ranking in the differential diagnosis. Research has shown that DDSSs are only adopted if the physician can assess the logic, because no clinician wants to be in the position of answering the question "why did you diagnose x" with "the computer said so". ***With a finding selected***, click "Assess" up on the black bar to see the frequency of the finding in the diseases in the differential diagnosis.

Saving and Reports

Task	Description
Text string	Automatically generate a text string that includes all the patient data (Figure 7). Pasting the text string into the Load field allows reopening the patient where you left off.
Summary report (findings only)	Assembles the pertinent positives and negatives and displays the HPO codes. The PDF is also machine readable, so you can pick up where you left off. The file can be generated from the Report menu (Figure 9) or the Share screen. It can be uploaded from the Share screen only (Figure 10).
Summary report (full)	Assembles the pertinent positives and negatives, differential diagnosis, and most useful tests into a graphical display. Document is also machine readable, so you can pick up where you left off. The file can be generated from the Report menu (Figure 11) or the Share screen. It can be uploaded from the Share screen only (Figure 12).
Prognosis table	Useful for Patient Education and communication between specialists and primary care. Characterizes the natural progression of the selected disease (without treatment).
SOAP note (*Subjective, Objective, Assessment and Plan*)	Automatically outputs a SOAP note format with the data you entered as plain text that can be copied and pasted to the electronic health record

Table of Figures

Figure 1: Genetics Core List

Notice that after commenting on the findings the green shading indicating usefulness will recalculate and lead you to the iteratively next most useful findings based on the differential.

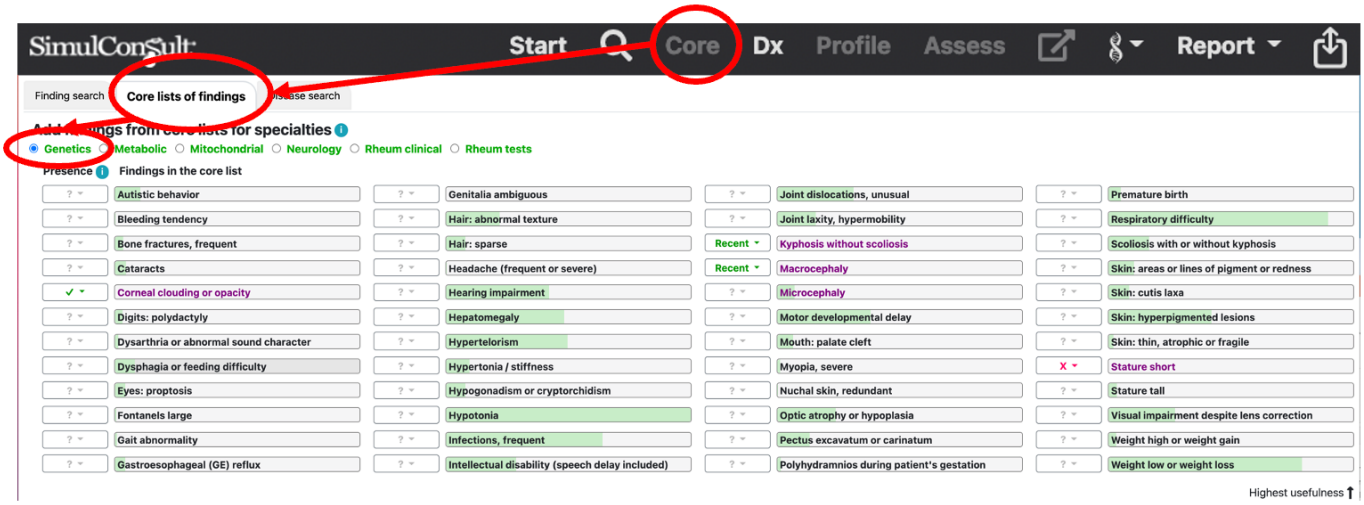

Figure 2: Search for finding

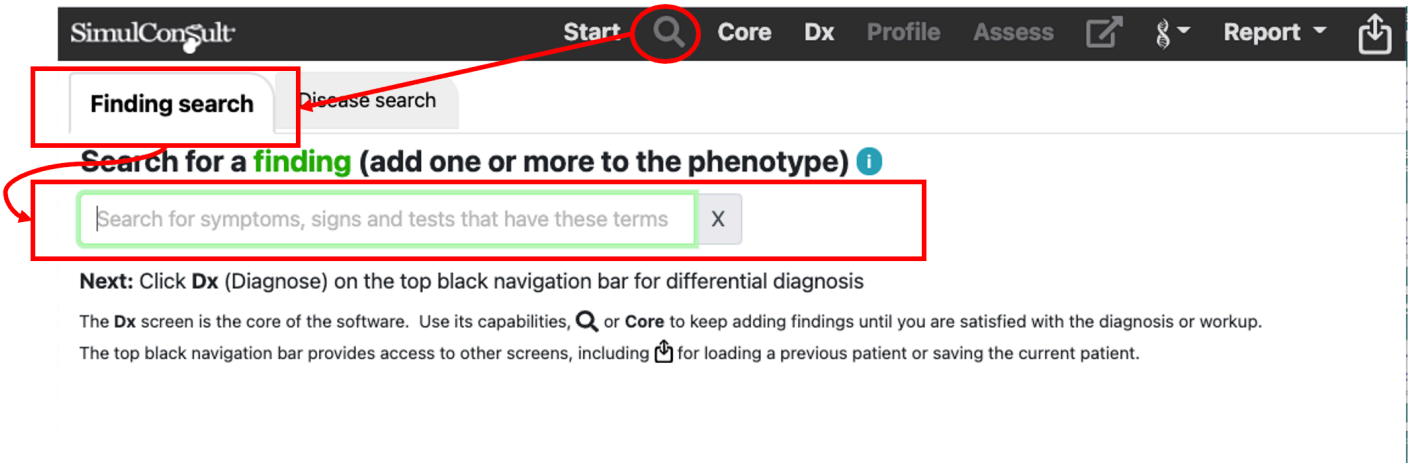

Figure 3: Dx/Differential

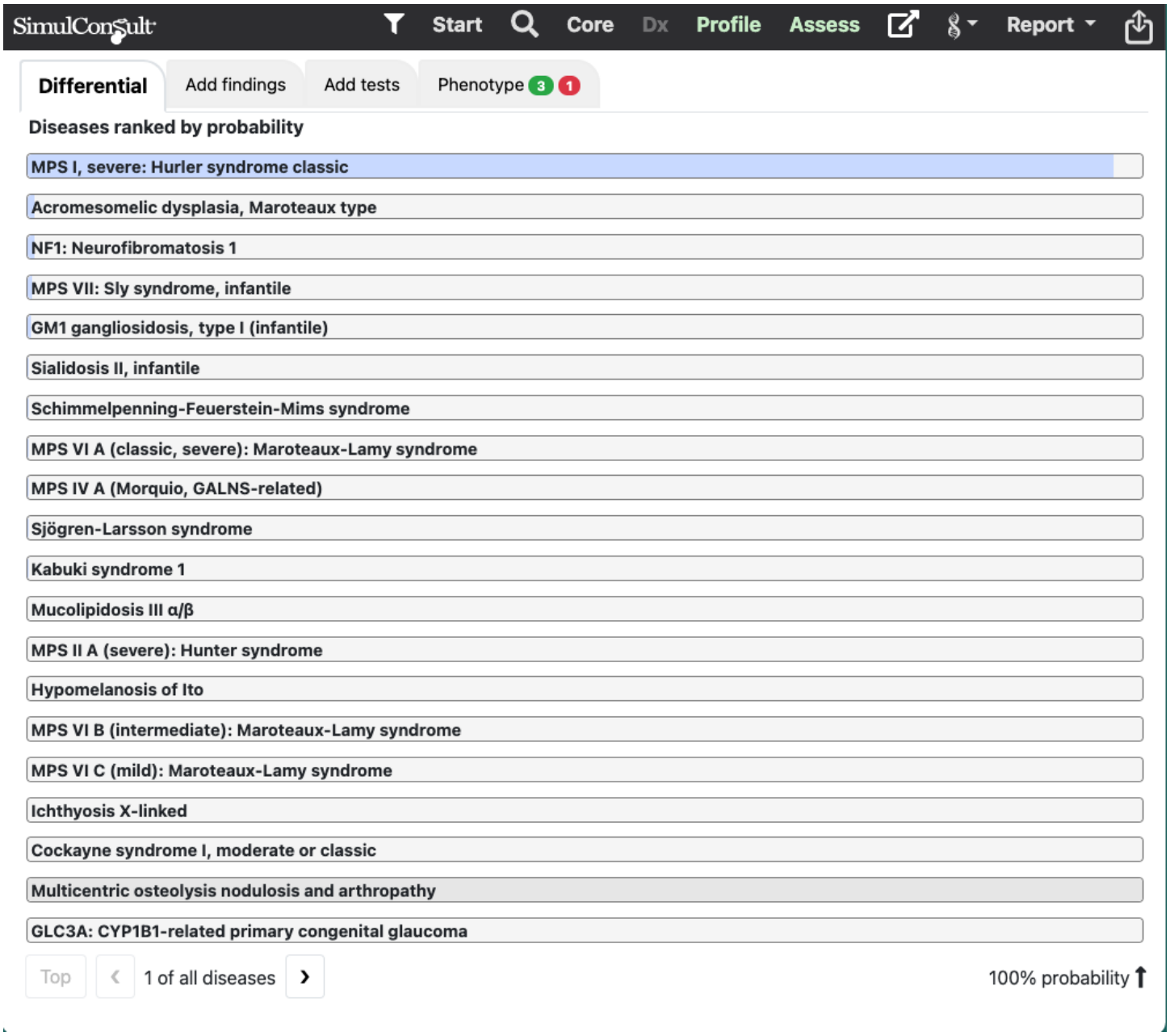

Figure 4: Add findings by usefulness

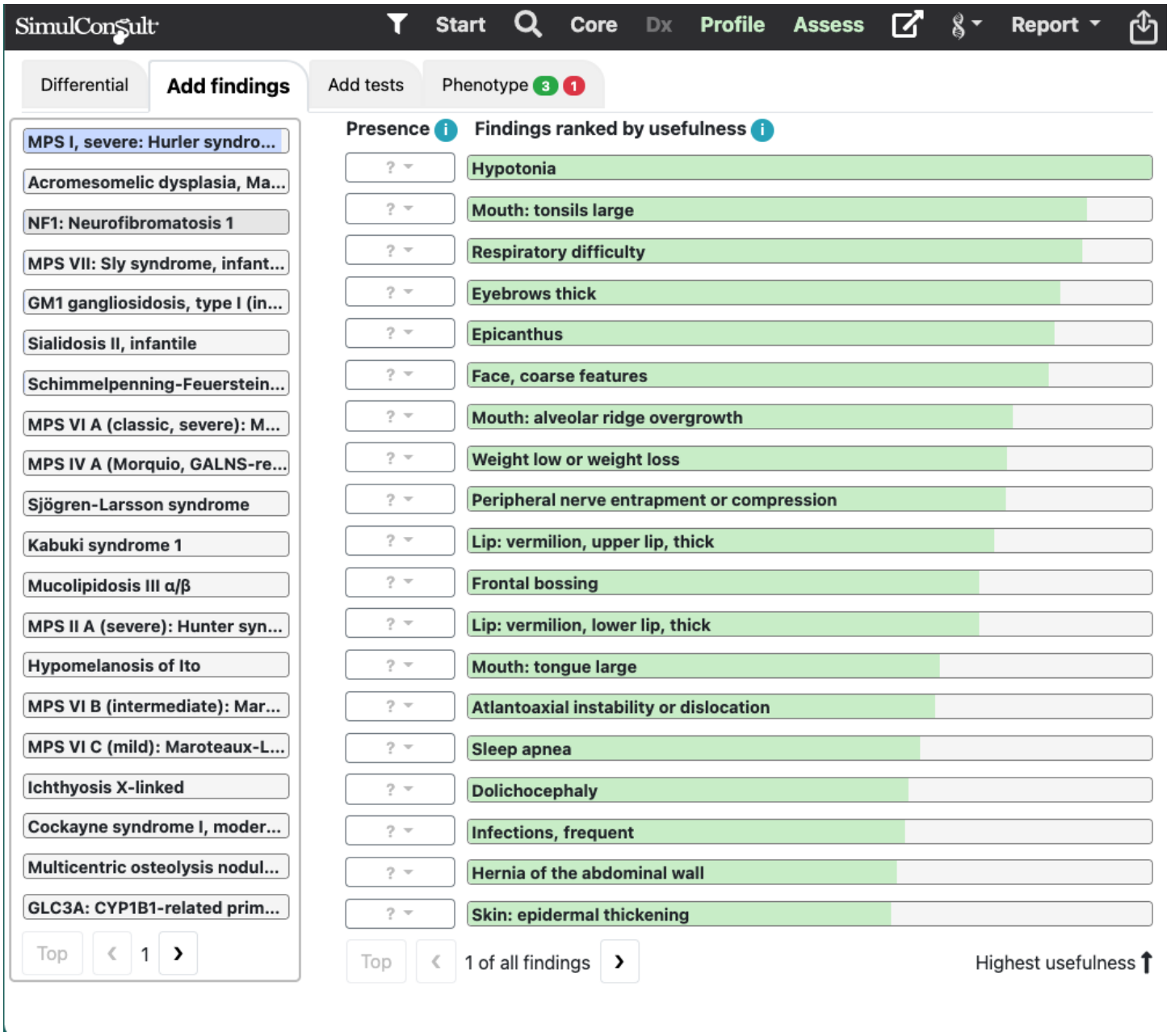

Figure 5: Add tests by usefulness

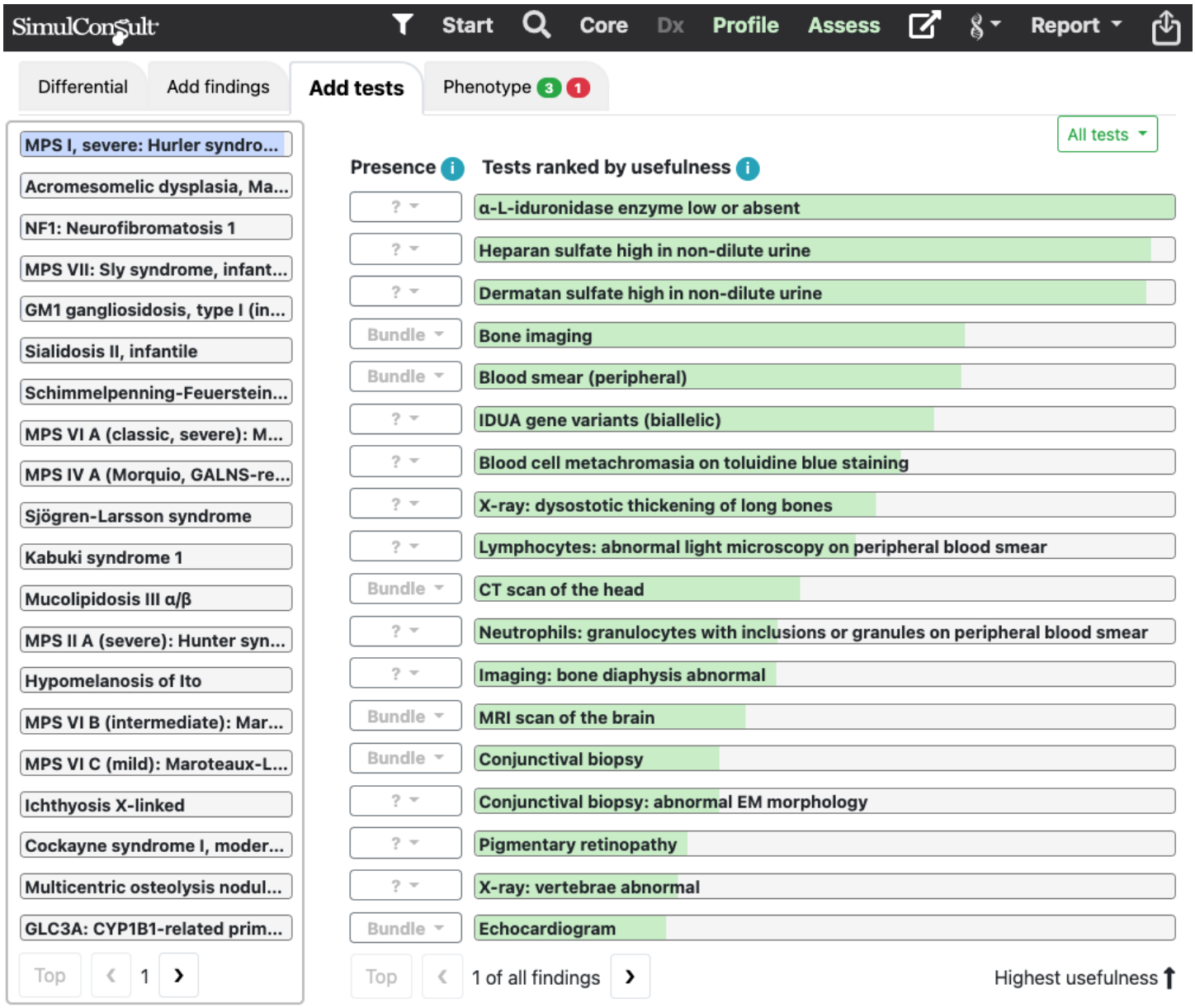

Figure 6: Rule in or Rule out a disease

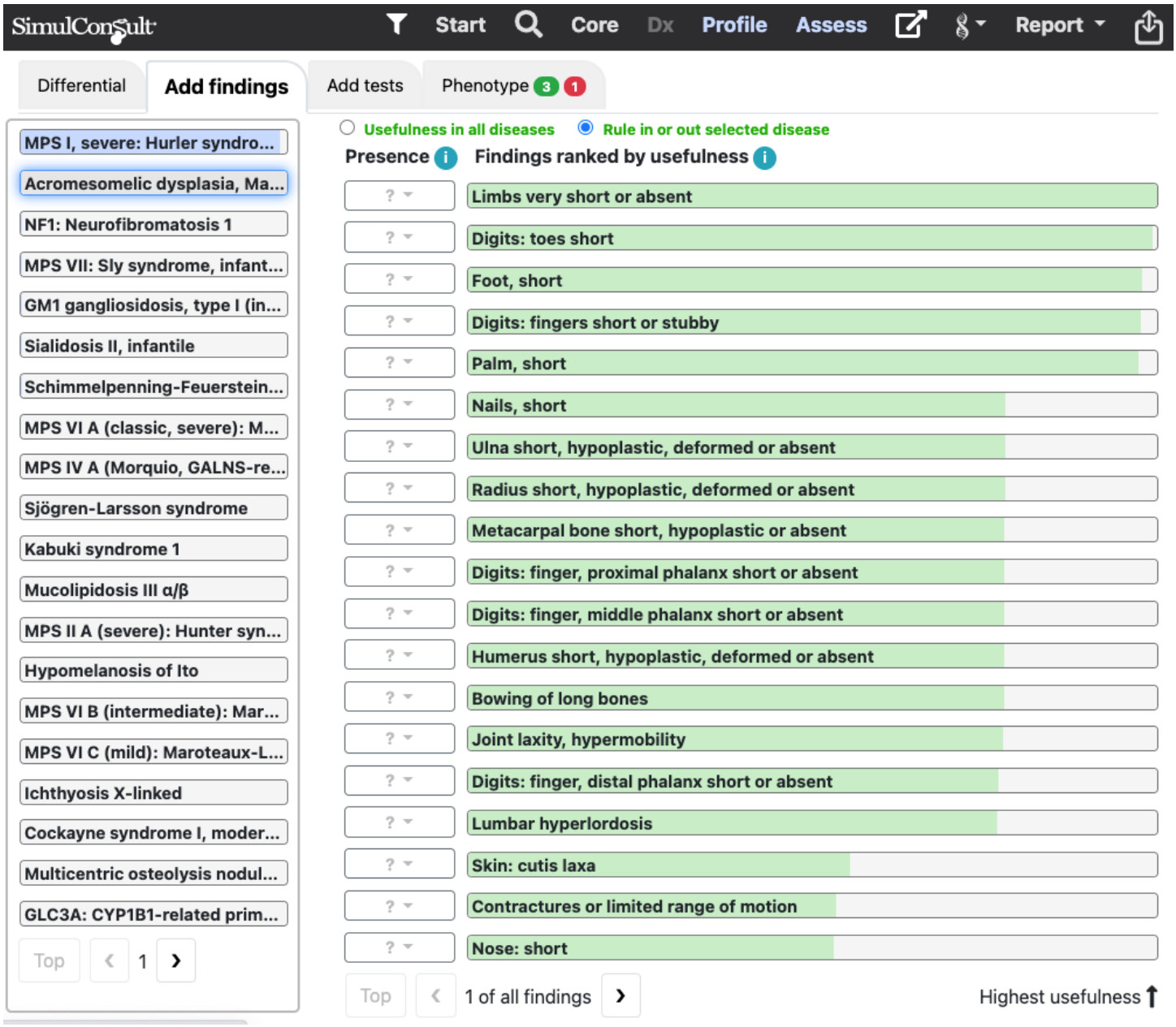

Figure 7: Patient Summary Text String

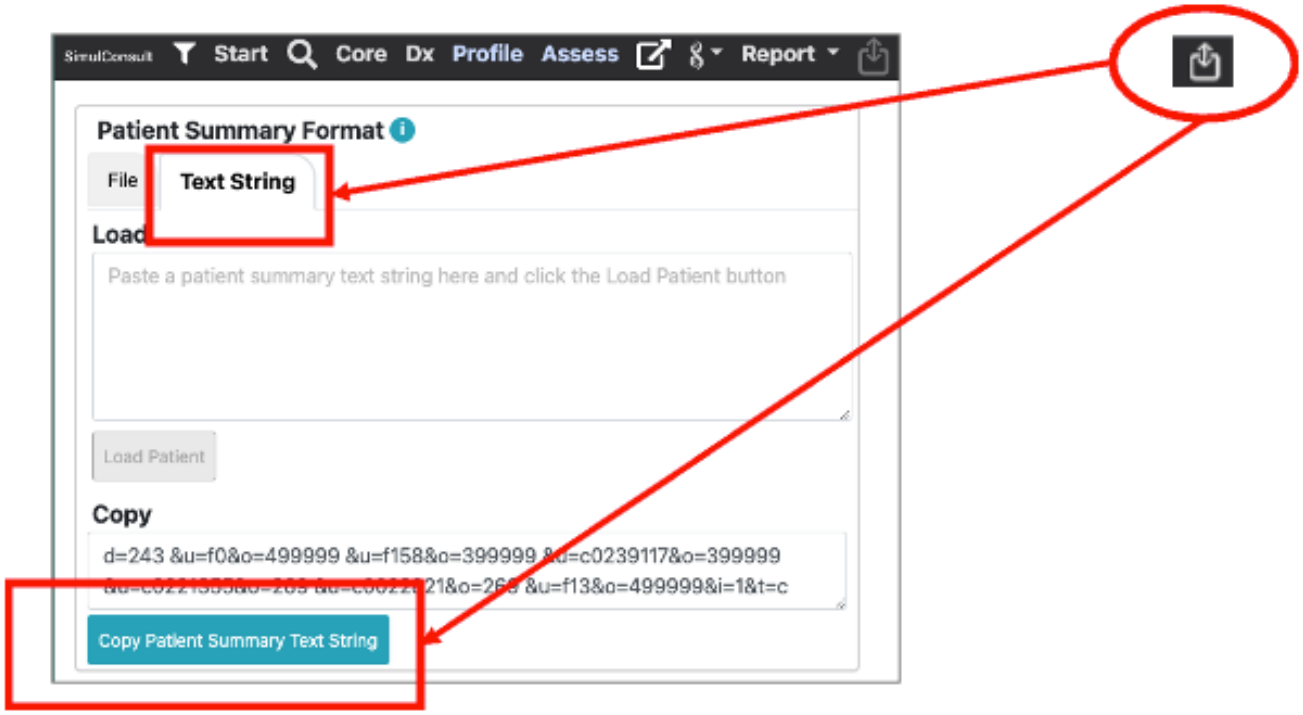

Figure 8: Add family history

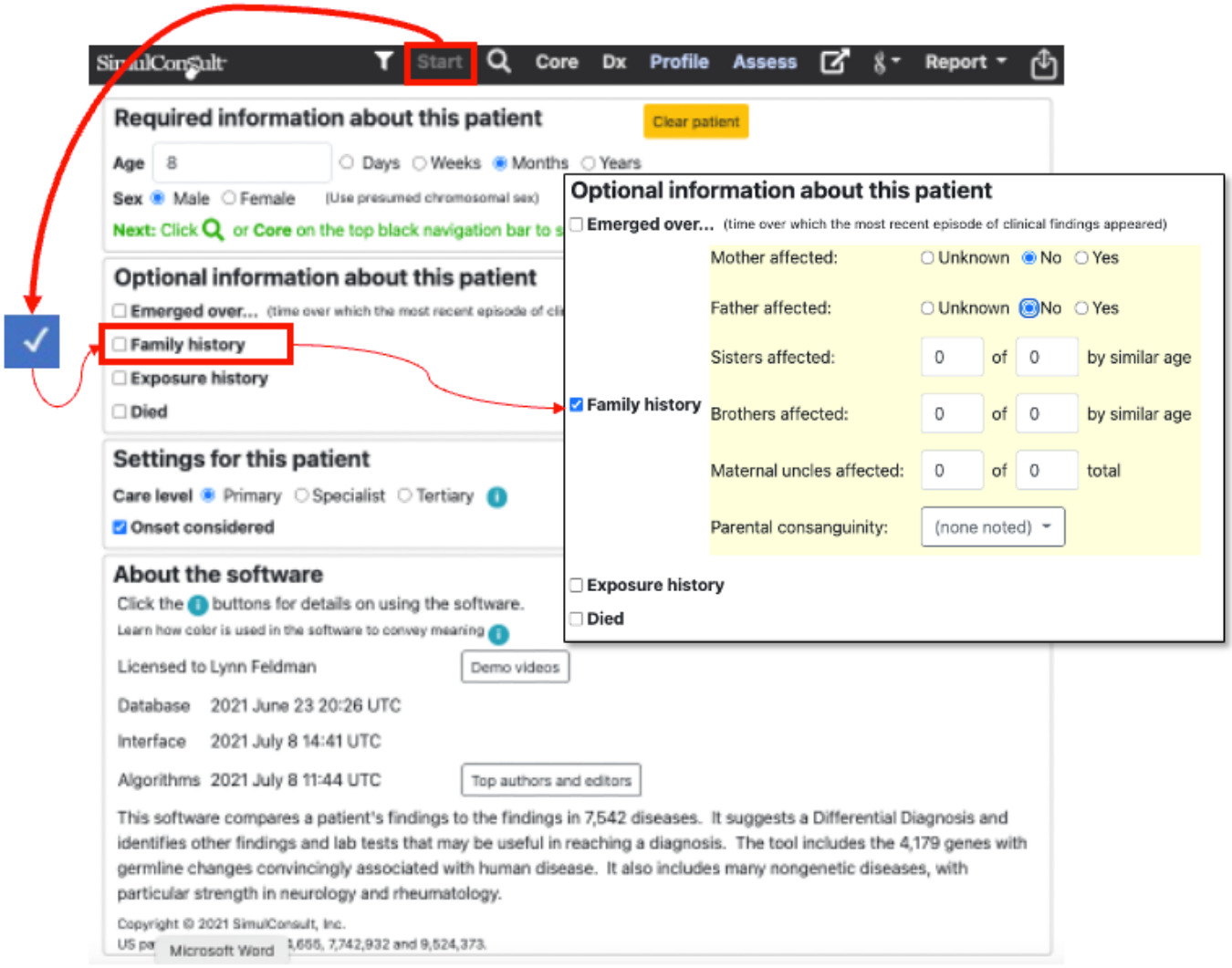

Figure 9: Patient Summary (Findings only) to generate PDF and save

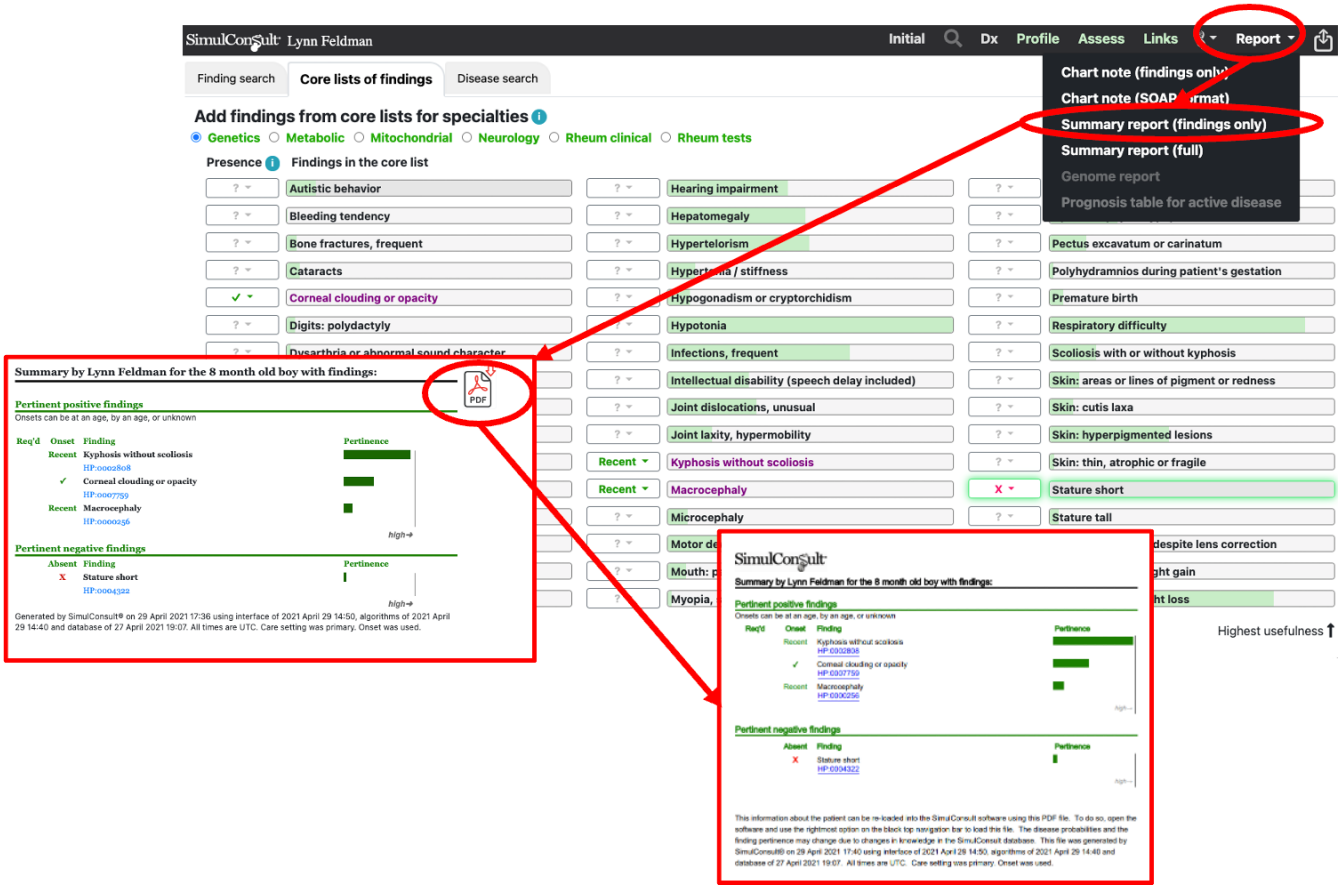

Figure 10: Patient Summary (Findings only) upload

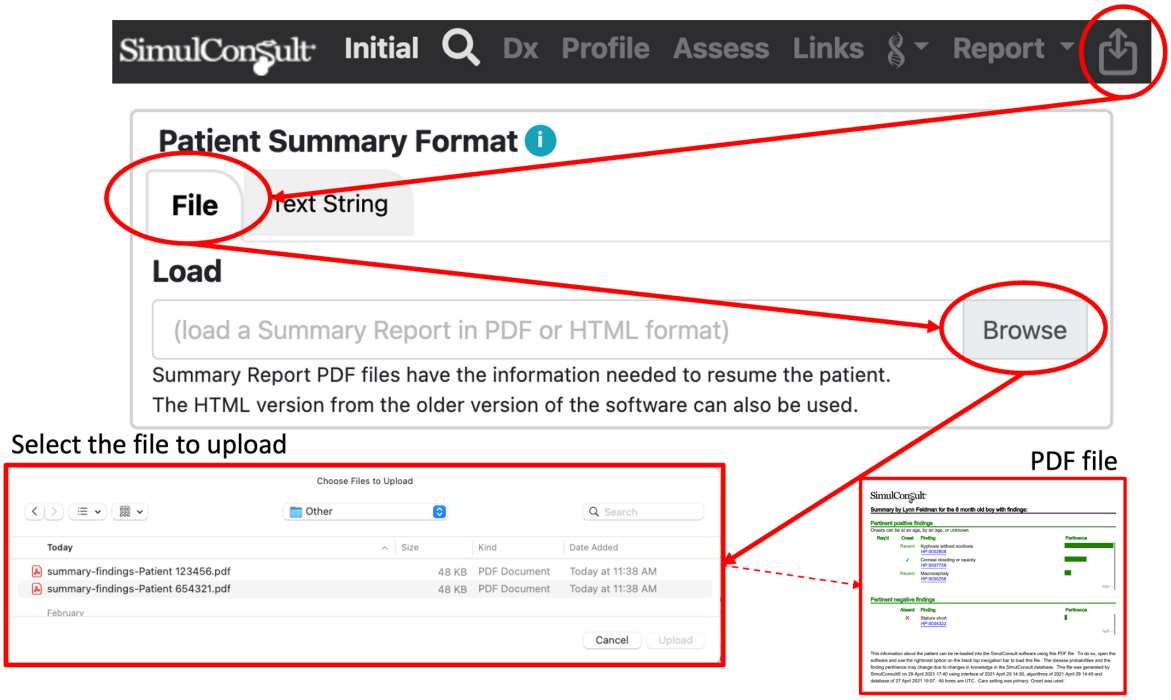

Figure 11: Patient Summary (Full) generate and save

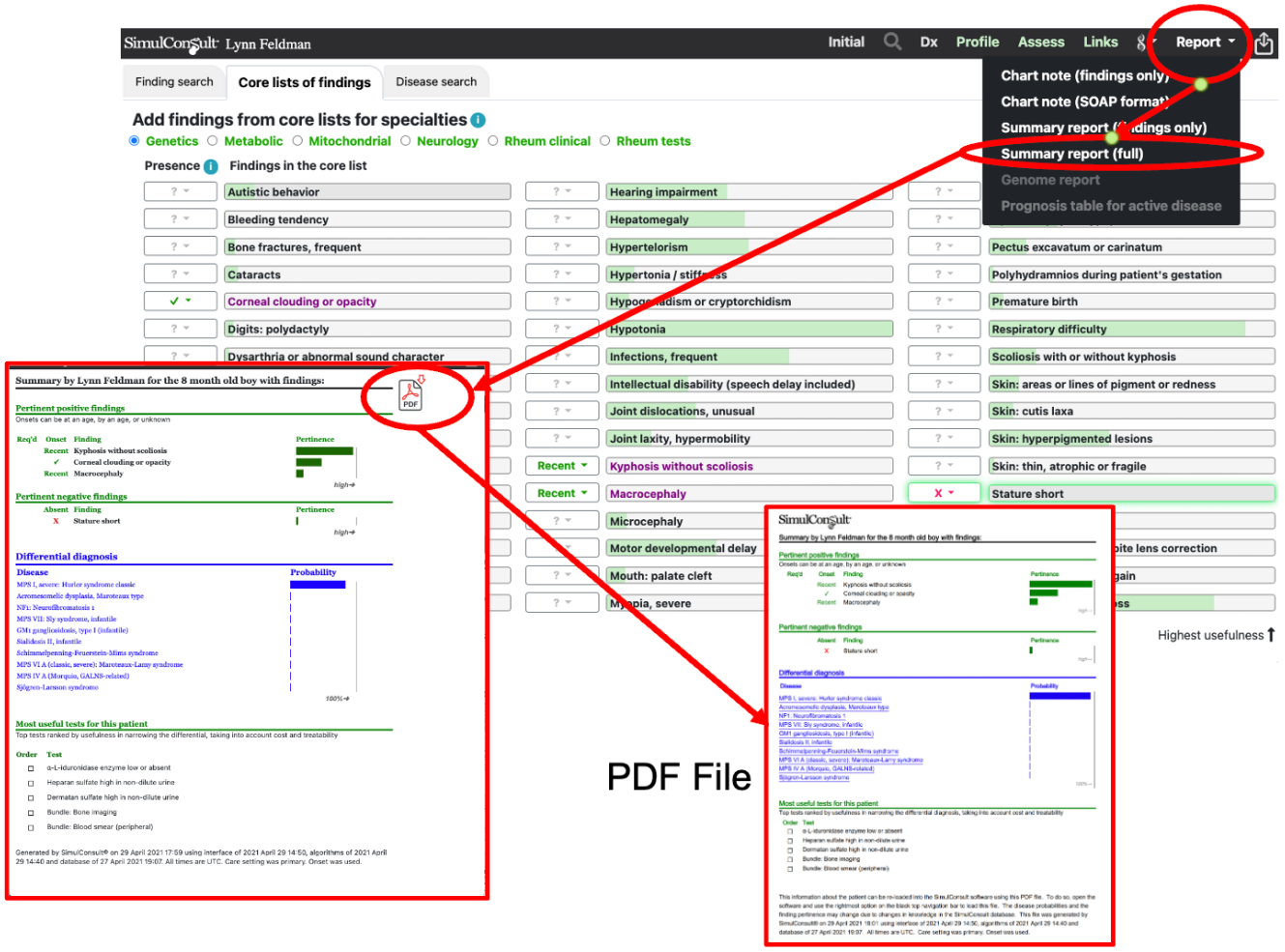

Figure 12: Patient Summary (Full) upload