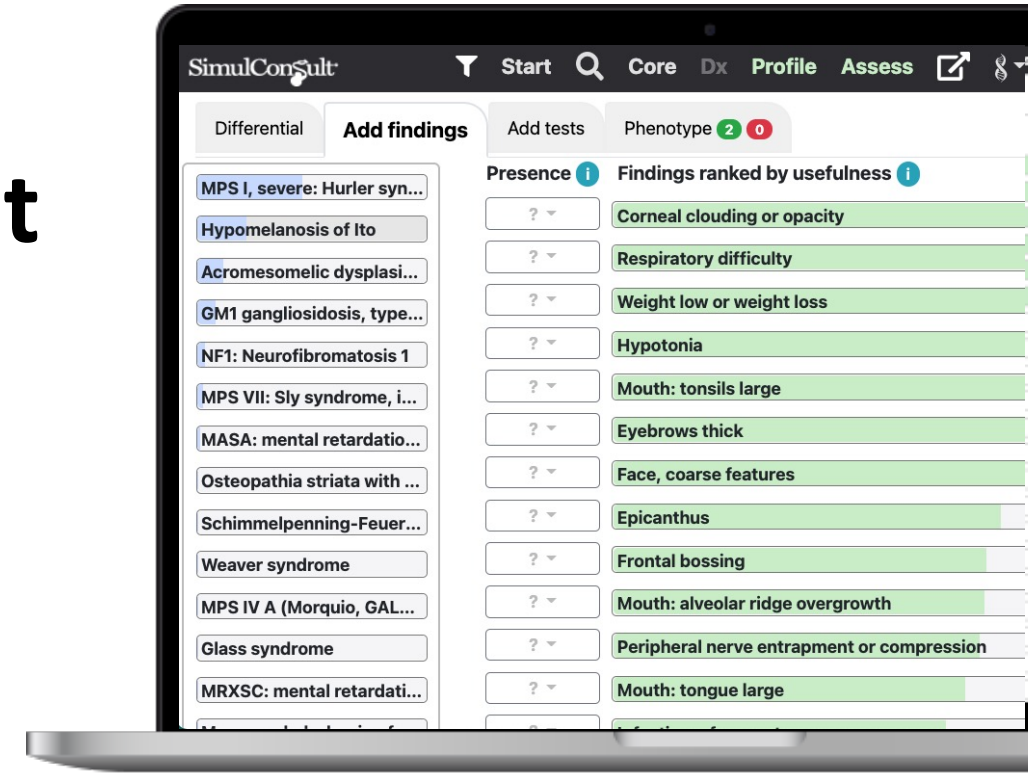
SimulConsult®
Computational Medicine Platform

Using SimulConsult in clinical care



Learning Objectives

- **See how specialist use SimulConsult to diagnose cases and save time**
- See how referring clinicians could use it to obtain curbside consultations & refer for formal consultations

Process Used by Specialists

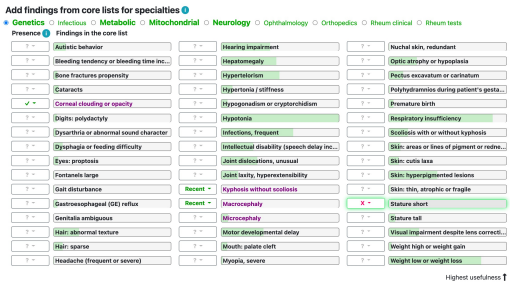
1. Reads referral and chart to summarize what is known

Memo

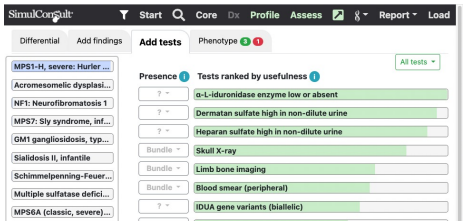
X year old boy with findings of …

2. Specialists sees patient

3. Does a history & exam & records findings as suggested by Usefulness algorithm



4. Prioritizes testing prompted by Usefulness



5. Uses automated Reports to improve collaboration and help with documentation

This is an 8 month old boy with:

Pertinent positive findings

Recent Kyphosis without scoliosis HP:0002808
✓ Corneal clouding or opacity HP:0007759
Recent Macrocephaly HP:0000256

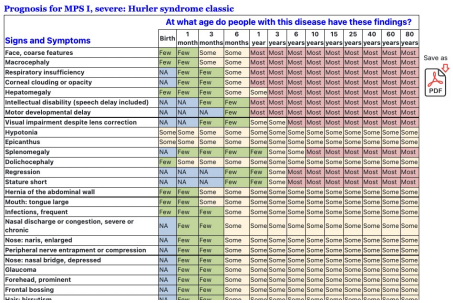
Pertinent negative findings

Stature short HP:0004322

COMPUTABLE PATIENT SUMMARY (can be used in SimulConsult to re-load the findings)

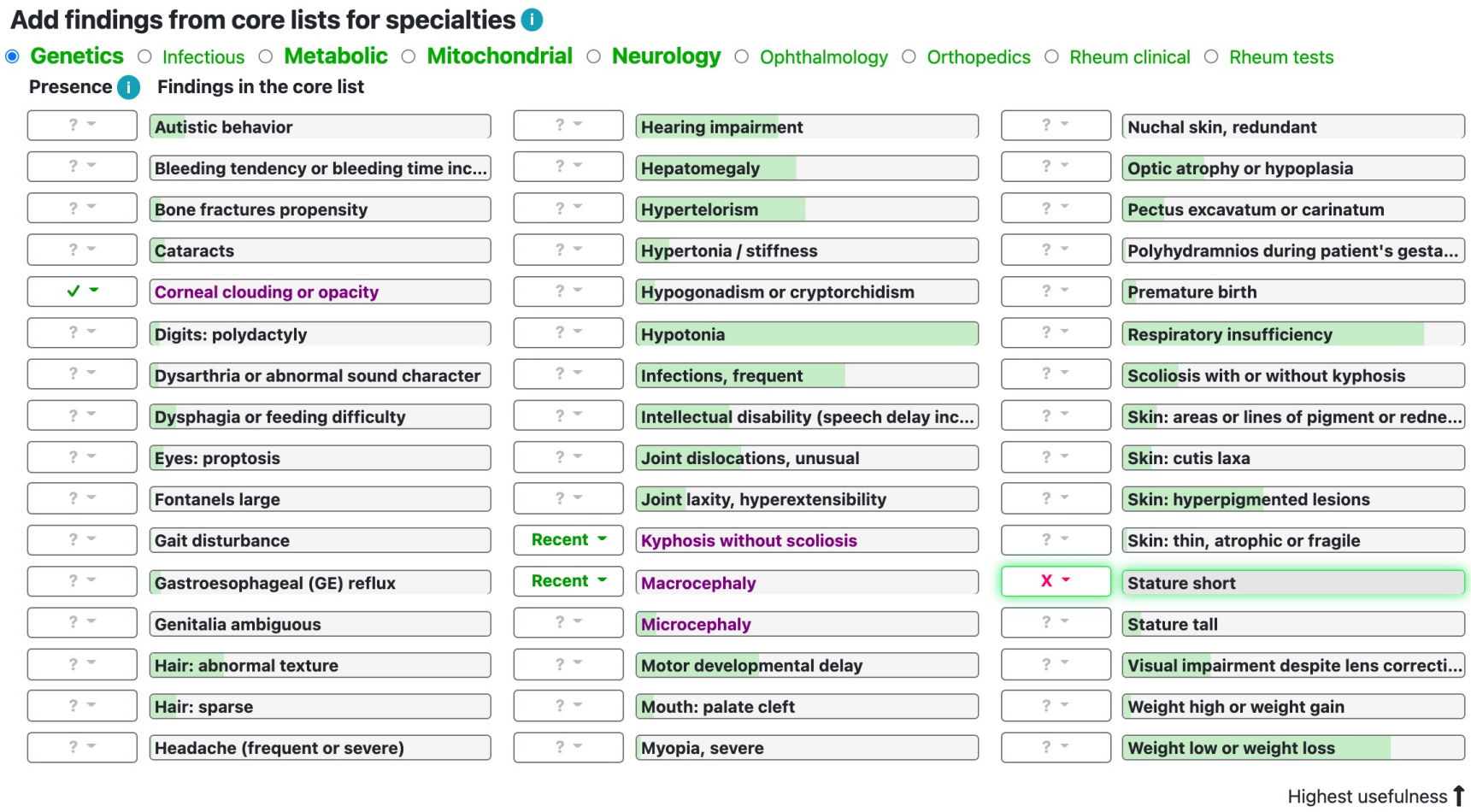
d=243 &u=f0&o=499999 &u=f158&o=399999 &u=c0239117&o=399999 &u=c0022821&o=269 &u=c0221355&o=269 &u=f13&o=499999 &i=1 &t=c

6. Shares Prognosis Table with referring clinician and patient when a diagnosis is confirmed



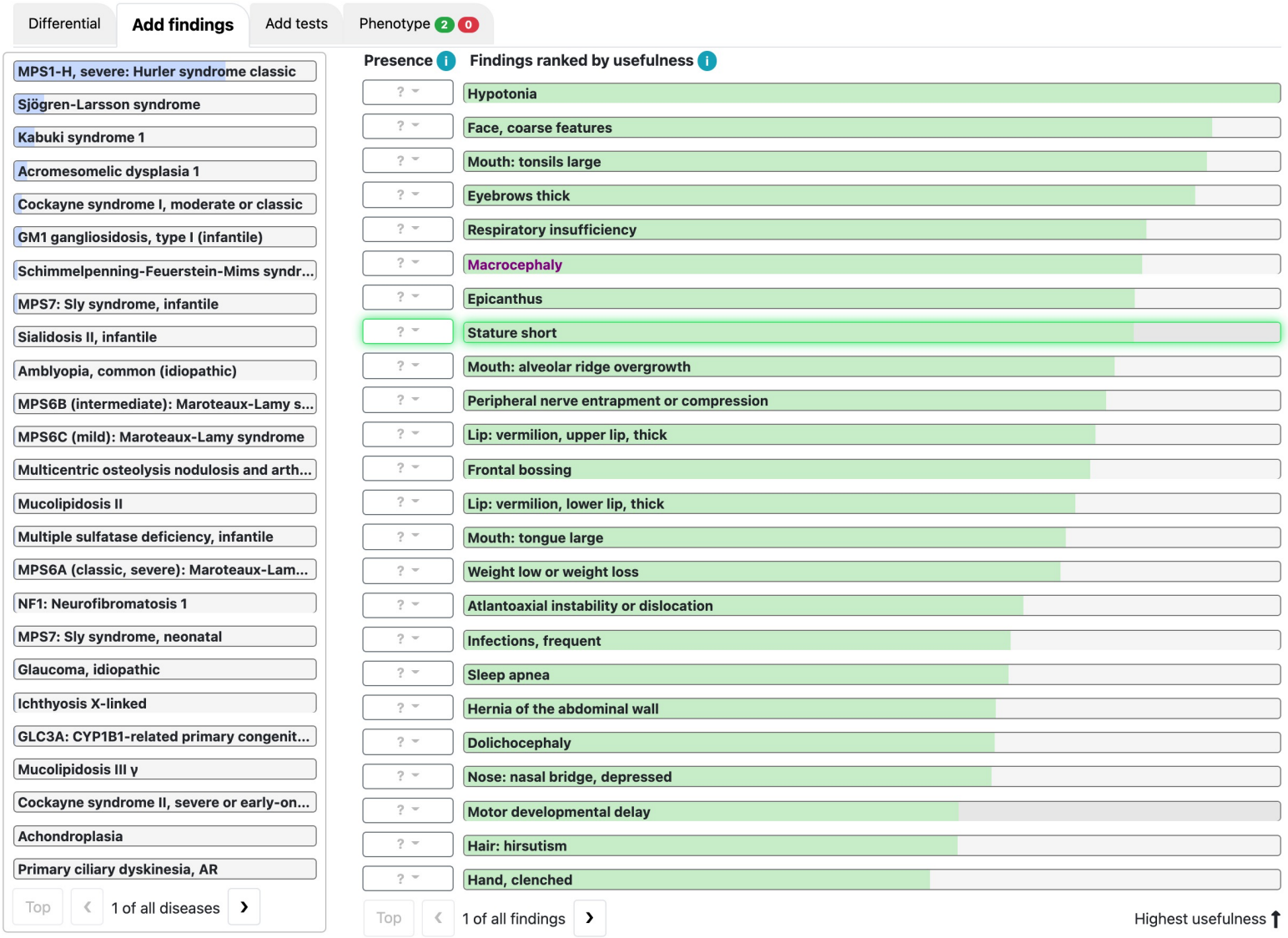
Get suggestions on findings using the Core List

Green shading dynamic indicates usefulness, multiple specialty lists

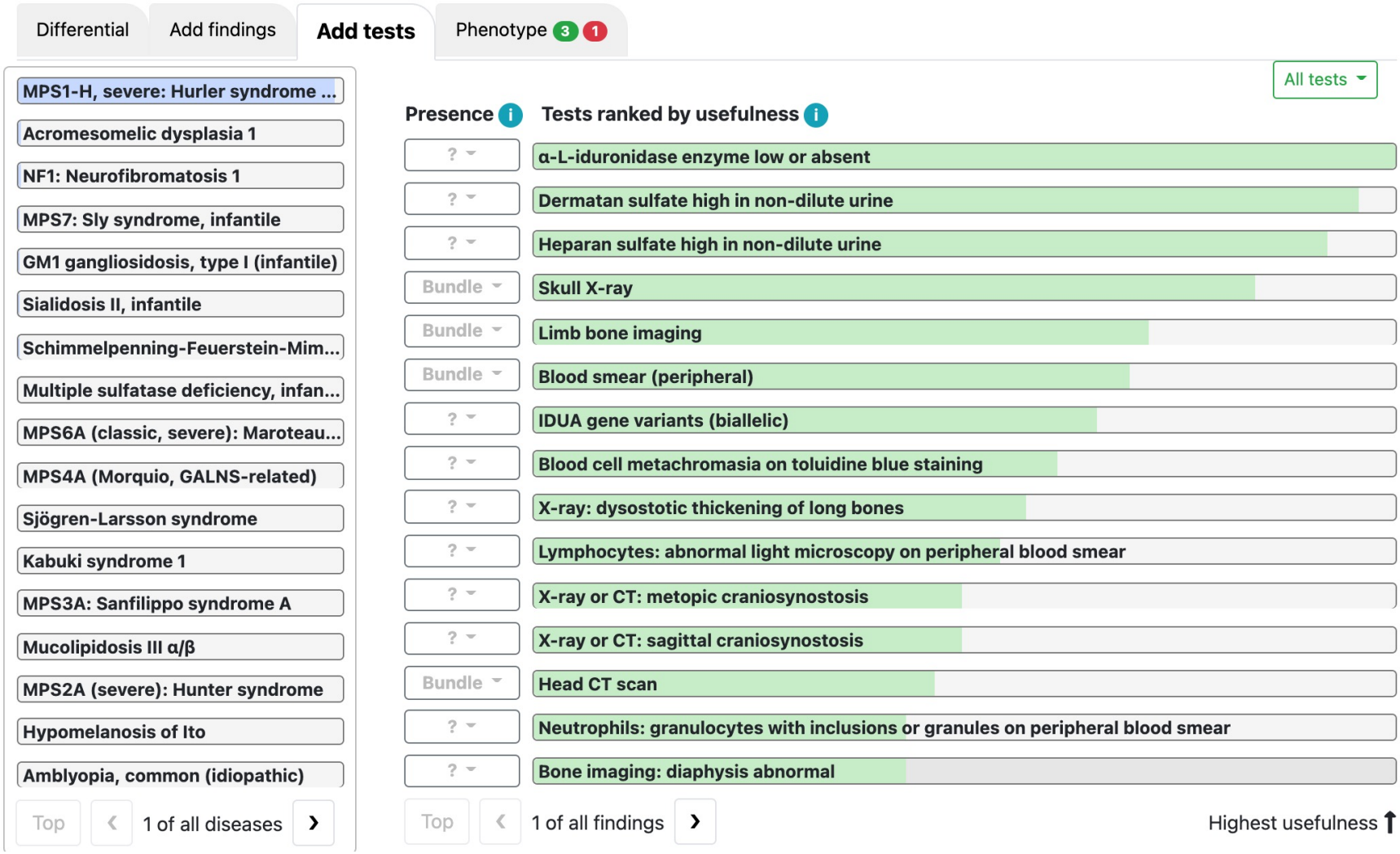


Get suggestions on findings using the Dx/Add Findings

Blue = probability of disease *Green = Usefulness of finding*



Get suggestions on useful tests Dx/Add Tests



Document Findings and share with colleagues in computable form

Includes HPO codes

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Pertinent positive findings

Recent	Kyphosis without scoliosis	HP:0002808
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Document Findings in SOAP note format (editable)

HISTORY OF PRESENT ILLNESS
This is an 8-month-old boy with findings:

Kyphosis without scoliosis, onset at about 6 months old
Macrocephaly, onset at about 6 months old
Corneal clouding or opacity, present now

Growth / development
Macrocephaly, onset at about 6 months old
Stature short, absent

PHYSICAL EXAM
Present

Kyphosis without scoliosis
Corneal clouding or opacity
Macrocephaly

Absent

Stature short

ASSESSMENT
This is an 8-month-old boy with:

Pertinent positives

Kyphosis without scoliosis
Corneal clouding or opacity
Macrocephaly

Impressions / Differential Diagnosis
MPS1-H, severe: Hurler syndrome classic
~~Acromesomelic dysplasia 1~~
~~NF1: Neurofibromatosis 1~~
~~MPS7: Sly syndrome, infantile~~
~~GM1 gangliosidosis, type I (infantile)~~

PLAN FOR TESTING / DISPOSITION
alpha-L-iduronidase enzyme low or absent
Dermatan sulfate high in non-dilute urine
Heparan sulfate high in non-dilute urine
IDUA gene variants (biallelic)
Blood cell metachromasia on toluidine blue staining
X-ray: dysostotic thickening of long bones
Lymphocytes: abnormal light microscopy on peripheral blood smear
X-ray or CT: metopic craniosynostosis
X-ray or CT: sagittal craniosynostosis
Neutrophils: granulocytes with inclusions or granules on peripheral blood smear
Bone imaging: diaphysis abnormal

You can choose to focus on only the most likely by deleting lines you don't want. Similarly, delete the tests you don't plan to order

Share Prognosis Table™ with colleagues and patients

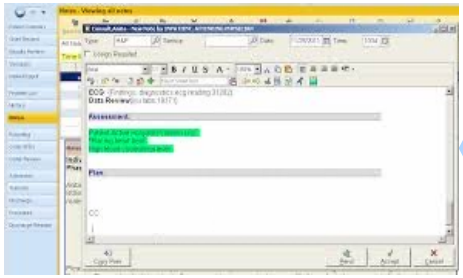
Prognosis for MPS I, severe: Hurler syndrome classic

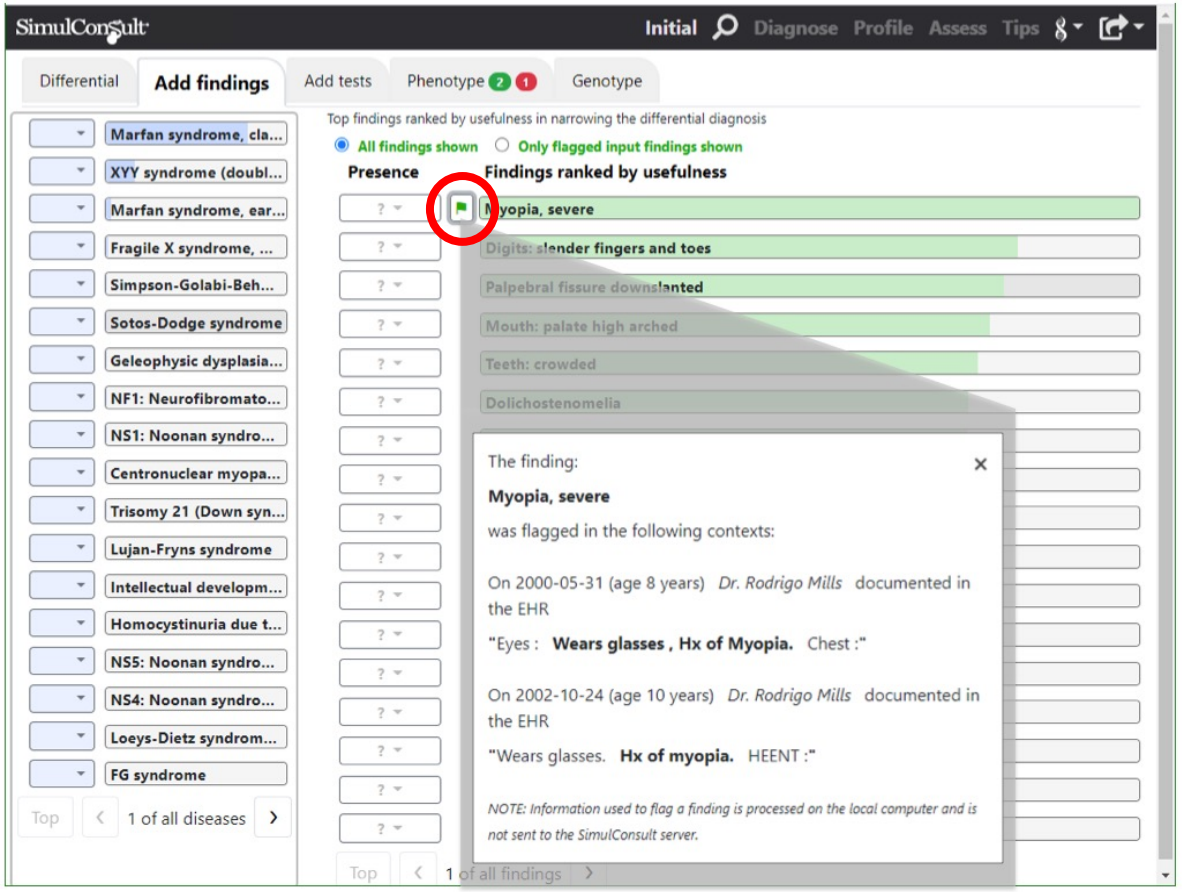
Signs and Symptoms	At what age do people with this disease have these findings?												
	Birth	1 month	3 months	6 months	1 year	3 years	6 years	10 years	15 years	25 years	40 years	60 years	80 years
Face, coarse features	Few	Few	Some	Some	Most	Most	Most	Most	Most	Most	Most	Most	Most
Macrocephaly	Few	Few	Some	Some	Most	Most	Most	Most	Most	Most	Most	Most	Most
Respiratory insufficiency	NA	Few	Few	Some	Most	Most	Most	Most	Most	Most	Most	Most	Most
Corneal clouding or opacity	NA	Few	Few	Some	Most	Most	Most	Most	Most	Most	Most	Most	Most
Hepatomegaly	Few	Few	Few	Some	Some	Most	Most	Most	Most	Most	Most	Most	Most
Intellectual disability (speech delay included)	NA	NA	Few	Few	Most	Most	Most	Most	Most	Most	Most	Most	Most
Motor developmental delay	NA	NA	NA	Few	Most	Most	Most	Most	Most	Most	Most	Most	Most
Visual impairment despite lens correction	NA	NA	Few	Few	Some	Some	Most	Most	Most	Most	Most	Most	Most
Hypotonia	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Epicanthus	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Splenomegaly	NA	Few	Few	Few	Few	Some	Some	Most	Most	Most	Most	Most	Most
Dolichocephaly	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Regression	NA	NA	NA	Few	Few	Some	Most	Most	Most	Most	Most	Most	Most
Stature short	NA	NA	NA	Few	Few	Some	Most	Most	Most	Most	Most	Most	Most
Hernia of the abdominal wall	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Mouth: tongue large	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Infections, frequent	Few	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Nasal discharge or congestion, severe or chronic	NA	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Nose: naris, enlarged	NA	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Peripheral nerve entrapment or compression	NA	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Nose: nasal bridge, depressed	NA	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Glaucoma	NA	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Forehead, prominent	NA	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Frontal bossing	NA	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Hair: hirsutism	NA	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some

Save as

Coming soon: See which findings are in the EHR

EHR





- Commercial version of EHR integration to be released 1Q22.
- Pilot: Clinicians ensure accuracy by selecting the relevant and believable extracted findings. Extraction of findings from the EHR was 100% sensitivity & specificity after clinician selection. Clinicians saved an average of 45 minute per patient by using the system.

External Links about diseases and findings

Links about the active disease and finding

MPS1-H, severe: Hurler syndrome classic

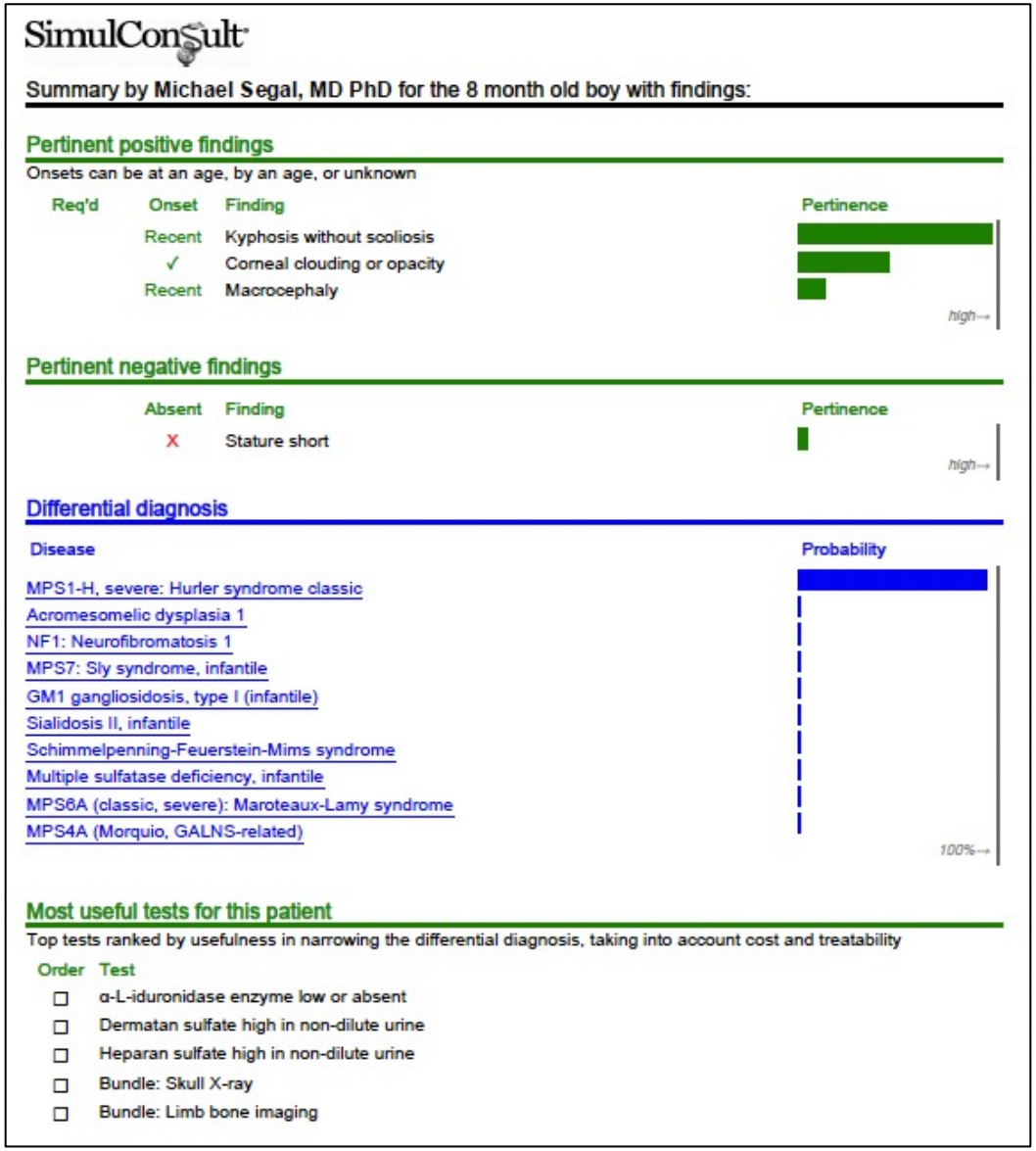
GeneReviews: Mucopolysaccharidosis Type I
Orphanet: Mucopolysaccharidosis I
National MPS Society: Hurler, Hurler-Scheie and Scheie Syndromes
OJRD: MPS I
OMIM article 607014

Stature short

SimulConsult resource: Calculate stature percentile
ACMG: short stature evaluation
Human Phenotype Ontology entry
Unified Medical Language System entry

Are there other links you'd like to include? Submit them, specifying the relevant disease or finding.

Insurers are accepting this Report as evidence of medical necessity



SimulConsult

Summary by Michael Segal, MD PhD for the 8 month old boy with findings:

Pertinent positive findings

Onsets can be at an age, by an age, or unknown

Req'd	Onset	Finding	Pertinence
	Recent	Kyphosis without scoliosis	
	✓	Corneal clouding or opacity	
	Recent	Macrocephaly	

high→

Pertinent negative findings

Absent	Finding	Pertinence
X	Stature short	

high→

Differential diagnosis

Disease	Probability
MPS1-H, severe: Hurler syndrome classic	
Acromesomelic dysplasia 1	
NF1: Neurofibromatosis 1	
MPS7: Sly syndrome, infantile	
GM1 gangliosidosis, type I (infantile)	
Sialidosis II, infantile	
Schimmelpenning-Feuerstein-Mims syndrome	
Multiple sulfatase deficiency, infantile	
MPS6A (classic, severe): Maroteaux-Lamy syndrome	
MPS4A (Morquio, GALNS-related)	

100%→

Most useful tests for this patient

Top tests ranked by usefulness in narrowing the differential diagnosis, taking into account cost and treatability

Order	Test
☐	α-L-iduronidase enzyme low or absent
☐	Dermatan sulfate high in non-dilute urine
☐	Heparan sulfate high in non-dilute urine
☐	Bundle: Skull X-ray
☐	Bundle: Limb bone imaging

Learning Objectives

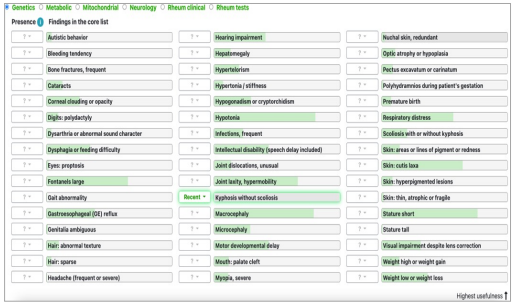
- See how specialist use SimulConsult to diagnose cases and save time
- **See how referring clinicians could use it to obtain curbside consultations & refer for formal consultations**

Process used by referring clinicians

1. Referring clinician sees patient

>

2. Quick collection of Core findings



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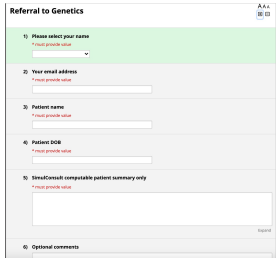
3. Sends findings to genetics, including Computable Patient Summary

(REDCap or EHR)



Enables a fast Curbside eConsult

Customizing an automated SOAP Report

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Impressions / Differential Diagnosis
MPS1-H, severe: Hurler syndrome classic

This seems like a good match for the findings noted.

PLAN FOR TESTING / DISPOSITION
X-ray: dysostotic thickening of long bones

I suggest a formal referral to genetics for further evaluation, with an appointment in the next few days. I will alert scheduling to expect the request. In the interim, please consider ordering the x-ray. Having the results at the first appointment will be very helpful.