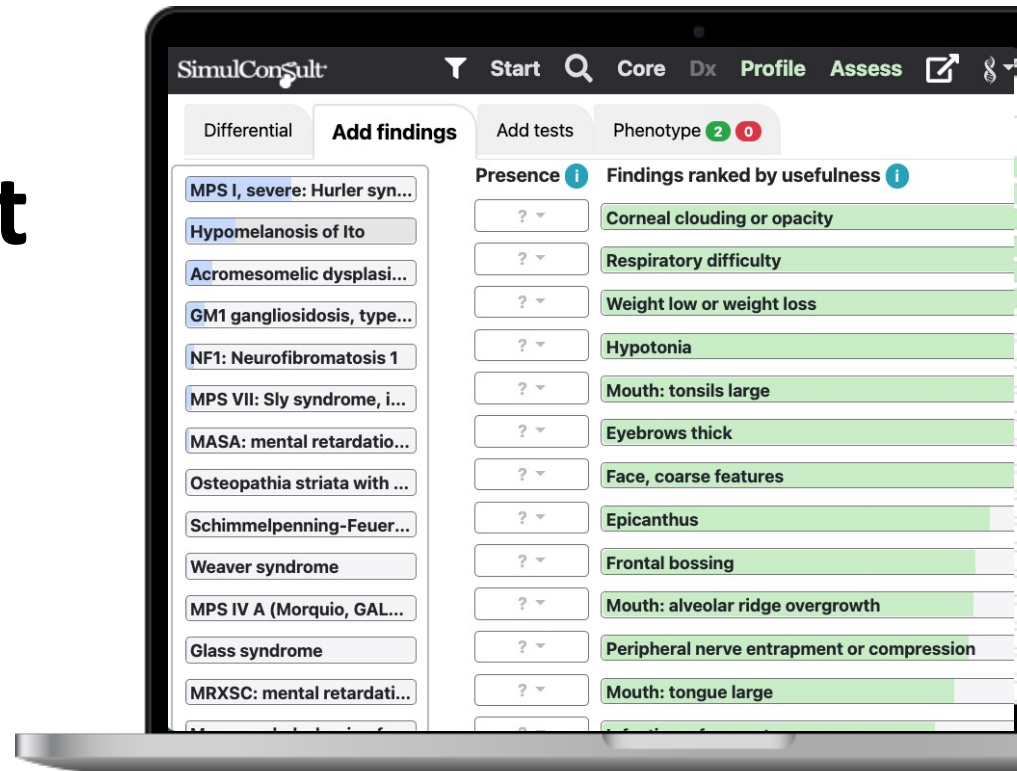


Using SimulConsult in clinical care



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 ...



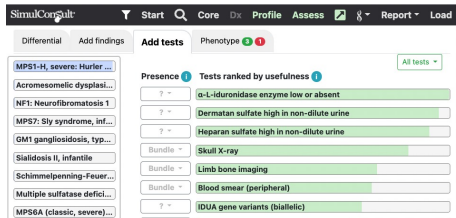
3. Does a history & exam & records findings as suggested by Usefulness algorithm

Add findings from core lists for specialties

☒ Genetics
 ☐ Infectious
 ☐ Metabolic
 ☐ Mitochondrial
 ☐ Neurology
 ☐ Ophthalmology
 ☐ Orthopedics
 ☐ Rheum clinical
 ☐ Rheum tests

Prevalence (%)	Findings in the core list	Prevalence (%)	Findings in the core list	Prevalence (%)	Findings in the core list
3-5	Autistic behavior	3-5	Hearing impairment	3-5	Musical skill, redundant
3-5	Discrete tendency or blending time (Int.)	3-5	Disproportion	3-5	Speech delivery or dyspraxia
3-5	Basic language proficiency	3-5	Hyperarousal	3-5	Positive excitation or catatonia
3-5	Contracts	3-5	Hypertonia / stiffness	3-5	Polysphinctic/during patient's gesture
3-5	✓ Concomit. clouding or specificity	3-5	Hypogonadism or cryptorchidism	3-5	Premature birth
3-5	Digits: polydactyl	3-5	Hypotonia	3-5	Respiratory insufficiency
3-5	Dysarthria or abnormal sound structure	3-5	Infections, frequent	3-5	Scoliosis with or without dyspraxia
3-5	Dysphagia or feeding difficulty	3-5	Intellectual disability (speech delay Int.)	3-5	Skin: areas or lines of pigment or redness
3-5	Eyes: protrusion	3-5	Joint dislocation, unusual	3-5	Skin: eczema like
3-5	Fantasmagoria	3-5	Joint laxity, hyperextensibility	3-5	Skin: hyperpigmented lesions
3-5	Gait disturbance	3-5	✓ Psychosis without psychosis	3-5	Skin: thin, atrophic or fragile
3-5	✓ Speech: nonsynchronous (SS) reflex	3-5	✓ Psychosis	3-5	✓ Stigmata
3-5	✓ Genitalia abnormal	3-5	✓ Microcephaly	3-5	Stomach full
3-5	✓ Hair: abnormal texture	3-5	✓ Motor developmental delay	3-5	✓ Visual impairment despite lens correct.
3-5	✓ Hand: spine	3-5	✓ Mouth: palate cleft	3-5	✓ Weight high or weight gain
3-5	✓ Headache (frequent or severe)	3-5	✓ Myopia, severe	3-5	✓ Weight low or weight loss

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

Prognosis for MPS I, severe: Hurler syndrome classic

At what age do people with this disease have these findings?

[illegible]

Get suggestions on findings using the Core List

Green shading dynamic indicates usefulness, multiple specialty lists

Add findings from core lists for specialties i

☒ **Genetics** ☐ Infectious ☐ Metabolic ☐ Mitochondrial ☐ Neurology ☐ Ophthalmology ☐ Orthopedics ☐ Rheum clinical ☐ Rheum tests

Presence i Findings in the core list

? ▾	Autistic behavior	? ▾	Hearing impairment	? ▾	Nuchal skin, redundant
? ▾	Bleeding tendency or bleeding time inc...	? ▾	Hepatomegaly	? ▾	Optic atrophy or hypoplasia
? ▾	Bone fractures propensity	? ▾	Hypertelorism	? ▾	Pectus excavatum or carinatum
? ▾	Cataracts	? ▾	Hypertonia / stiffness	? ▾	Polyhydramnios during patient's gesta...
✓ ▾	Corneal clouding or opacity	? ▾	Hypogonadism or cryptorchidism	? ▾	Premature birth
? ▾	Digits: polydactyly	? ▾	Hypotonia	? ▾	Respiratory insufficiency
? ▾	Dysarthria or abnormal sound character	? ▾	Infections, frequent	? ▾	Scoliosis with or without kyphosis
? ▾	Dysphagia or feeding difficulty	? ▾	Intellectual disability (speech delay inc...	? ▾	Skin: areas or lines of pigment or redne...
? ▾	Eyes: proptosis	? ▾	Joint dislocations, unusual	? ▾	Skin: cutis laxa
? ▾	Fontanels large	? ▾	Joint laxity, hyperextensibility	? ▾	Skin: hyperpigmented lesions
? ▾	Gait disturbance	Recent ▾	Kyphosis without scoliosis	? ▾	Skin: thin, atrophic or fragile
? ▾	Gastroesophageal (GE) reflux	Recent ▾	Macrocephaly	X ▾	Stature short
? ▾	Genitalia ambiguous	? ▾	Microcephaly	? ▾	Stature tall
? ▾	Hair: abnormal texture	? ▾	Motor developmental delay	? ▾	Visual impairment despite lens correcti...
? ▾	Hair: sparse	? ▾	Mouth: palate cleft	? ▾	Weight high or weight gain
? ▾	Headache (frequent or severe)	? ▾	Myopia, severe	? ▾	Weight low or weight loss

Highest usefulness ↑

Get suggestions on findings using the Dx/Add Findings

Blue = probability of disease

Green = Usefulness of finding

Differential

Add findings

Add tests

Phenotype 2 0

MPS1-H, severe: Hurler syndrome classic

Sjögren-Larsson syndrome

Kabuki syndrome 1

Acromesomelic dysplasia 1

Cockayne syndrome I, moderate or classic

GM1 gangliosidosis, type I (infantile)

Schimmelpenning-Feuerstein-Mims syndr...

MPS7: Sly syndrome, infantile

Sialidosis II, infantile

Amblyopia, common (idiopathic)

MPS6B (intermediate): Maroteaux-Lamy s...

MPS6C (mild): Maroteaux-Lamy syndrome

Multicentric osteolysis nodulosis and arth...

Mucopolidosis II

Multiple sulfatase deficiency, infantile

MPS6A (classic, severe): Maroteaux-Lam...

NF1: Neurofibromatosis 1

MPS7: Sly syndrome, neonatal

Glaucoma, idiopathic

Ichthyosis X-linked

GLC3A: CYP1B1-related primary congenit...

Mucopolidosis III γ

Cockayne syndrome II, severe or early-on...

Achondroplasia

Primary ciliary dyskinesia, AR

Top

< 1 of all diseases >

Presence ⓘ Findings ranked by usefulness ⓘ

? ▾

Hypotonia

? ▾

Face, coarse features

? ▾

Mouth: tonsils large

? ▾

Eyebrows thick

? ▾

Respiratory insufficiency

? ▾

Macrocephaly

? ▾

Epicanthus

? ▾

Stature short

? ▾

Mouth: alveolar ridge overgrowth

? ▾

Peripheral nerve entrapment or compression

? ▾

Lip: vermilion, upper lip, thick

? ▾

Frontal bossing

? ▾

Lip: vermilion, lower lip, thick

? ▾

Mouth: tongue large

? ▾

Weight low or weight loss

? ▾

Atlantoaxial instability or dislocation

? ▾

Infections, frequent

? ▾

Sleep apnea

? ▾

Hernia of the abdominal wall

? ▾

Dolichocephaly

? ▾

Nose: nasal bridge, depressed

? ▾

Motor developmental delay

? ▾

Hair: hirsutism

? ▾

Hand, clenched

Top

< 1 of all findings >

Highest usefulness ↑

Get suggestions on useful tests Dx/Add Tests

Differential

Add findings

Add tests

Phenotype 3 1

All tests ▾

MPS1-H, severe: Hurler syndrome ...

Acromesomelic dysplasia 1

NF1: Neurofibromatosis 1

MPS7: Sly syndrome, infantile

GM1 gangliosidosis, type I (infantile)

Sialidosis II, infantile

Schimmelpenning-Feuerstein-Mim...

Multiple sulfatase deficiency, infan...

MPS6A (classic, severe): Maroteau...

MPS4A (Morquio, GALNS-related)

Sjögren-Larsson syndrome

Kabuki syndrome 1

MPS3A: Sanfilippo syndrome A

Mucopolipidosis III α/β

MPS2A (severe): Hunter syndrome

Hypomelanosis of Ito

Amblyopia, common (idiopathic)

Top



1 of all diseases



Presence ⓘ

Tests ranked by usefulness ⓘ

? ▾

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

Bundle ▾

Blood smear (peripheral)

? ▾

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

Bundle ▾

Head CT scan

? ▾

Neutrophils: granulocytes with inclusions or granules on peripheral blood smear

? ▾

Bone imaging: diaphysis abnormal

Top



1 of all findings



Highest usefulness ↑

Document Findings and share with colleagues in computable form

Includes HPO codes

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

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



SimulConsult

Initial Diagnose Profile Assess Tips

Differential Add findings Add tests Phenotype 2 1 Genotype

Top findings ranked by usefulness in narrowing the differential diagnosis

☒ All findings shown ☐ Only flagged input findings shown

Presence Findings ranked by usefulness

?	Myopia, severe
?	Digits: slender fingers and toes
?	Palpebral fissure downslanted
?	Mouth: palate high arched
?	Teeth: crowded
?	Dolichostenomelia

The finding:
Myopia, severe
was flagged in the following contexts:

On 2000-05-31 (age 8 years) Dr. Rodrigo Mills documented in the EHR
"Eyes : **Wears glasses , Hx of Myopia.** Chest :"

On 2002-10-24 (age 10 years) Dr. Rodrigo Mills documented in the EHR
"Wears glasses. **Hx of myopia.** HEENT :"

NOTE: Information used to flag a finding is processed on the local computer and is not sent to the SimulConsult server.

- 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

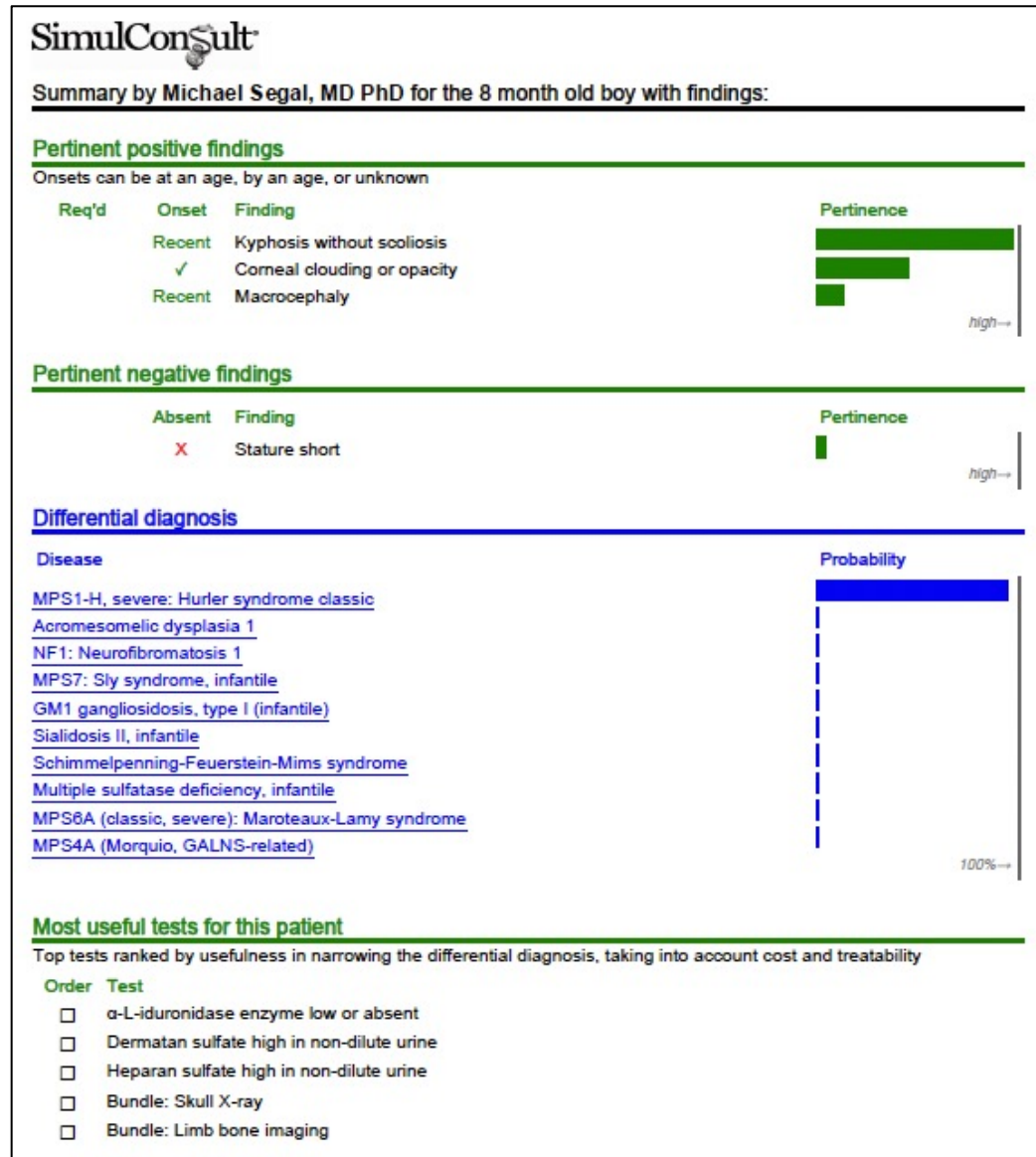
ACMG: short stature evaluation

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



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

Genetics | Metabolic | Musculoskeletal | Neurology | **Neuro clinical** | Neuroimaging

Presence: Findings in the core list

Agitation behavior	++	Hearing impairment	++	Neural tube, redundant	++
Blindness tendency	++	Hemiparesis	++	Optic atrophy or hypoplasia	++
Bone fractures, frequent	++	Hypertonia	++	Palmar creases or cysts	++
Cataracts	++	Hypotonia / stiffness	++	Polyphagia during patient's gestation	++
Central blindness or opacity	++	Hypoglossia or clefts	++	Preterm birth	++
Clonus, irritability	++	Hypotonia	++	Respiratory distress	++
Discrete or abnormal sound character	++	Infections, frequent	++	Scaliness pits or without lymphatics	++
Disphagia or feeding difficulty	++	Intellectual disability (speech delay included)	++	Side areas or lines of pigment or redness	++
Eyes, prominent	++	Joint dislocations, unusual	++	Side note lines	++
Fatigue/large	++	Joint laxity, hypermobility	++	Side hyperpigmented lesions	++
Gait abnormality	++	Repetitive	++	Side thin, atrophic or fragile	++
Gastrointestinal (GI) reflux	++	Syncope without accidents	++	Stature short	++
Genitourinary anomalies	++	Microcephaly	++	Stature tall	++
Global abnormal features	++	Motor developmental delay	++	Visual impairment despite lens correction	++
Hair sparse	++	Mouth, points, cleft	++	Weight high or weight gain	++
Headache (frequent or severe)	++	Myopia, severe	++	Weight low or weight loss	++

Highest usefulness 1

3. Sends findings to genetics, including Computable Patient Summary (REDCap or EHR)

Referral to Genetics

1) Please select your name

2) Your email address

3) Patient name

4) Patient DOB

5) Your consent (compatible patient summary only)

6) Optional comments

Enables a fast Curbside eConsult

Customizing an automated SOAP Report

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

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.