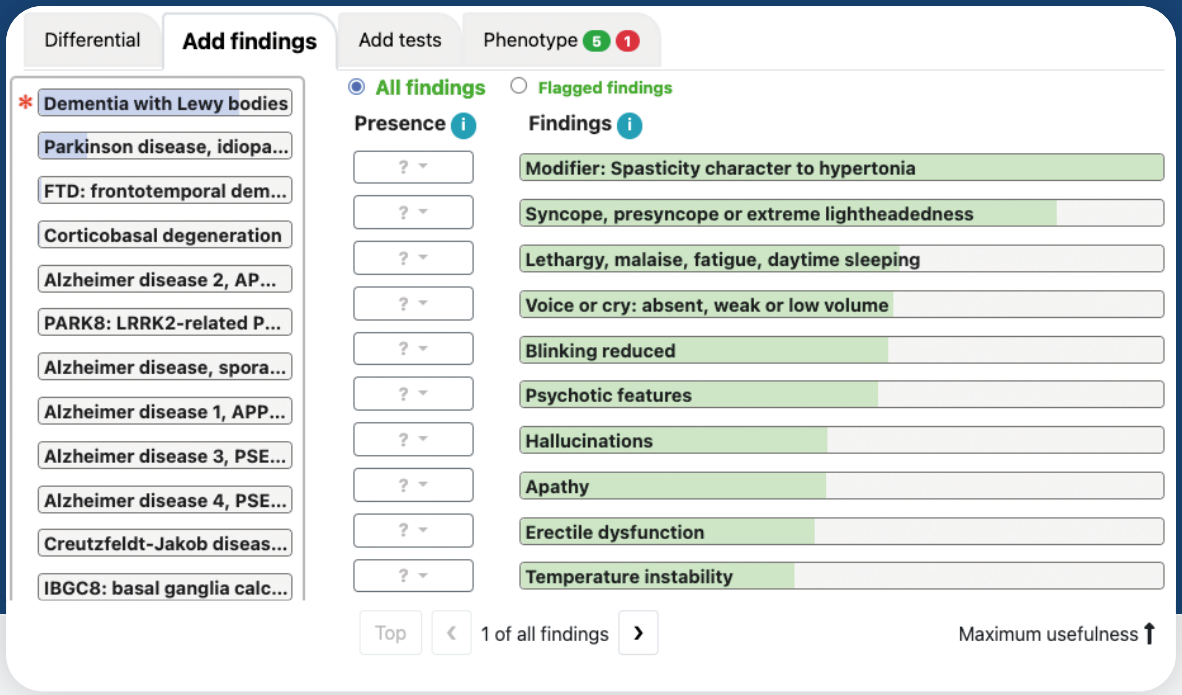
Phenome Dashboard

Streamlined diagnostic workflow for experts



Now it reads your text and notes. It also outputs an editable SOAP note.

Coverage

Covers >9,600 diseases: chromosomal abnormalities, genes with germline changes associated with human disease and their clinical & lab findings, as well as general pediatrics and adult medicine.

Clear logic

You assess the rationale of the fit between your patient and the disease. It achieves “explainable artificial intelligence” using a human-curated database, it is not a black box.

Embedded warnings

Red astericks provide information about risks to patients with certain diseases and cautions about findings, to help you avoid errors in care.

Designed for reliable, accurate diagnosis

Use pertinent positive and negative findings to generate a differential diagnosis and get suggestions on useful clinical findings to add and tests to order.

Saves you time

Reads and interprets medical notes instantly. Get a simultaneous consult in seconds. Editable SOAP notes and easy preparation for lab orders, with automated HPO codes.

Streamlines teamwork

Helps nurse practitioners, genetic counselors, and residents practice “at the top of their license”. Easy handoffs, no rework.

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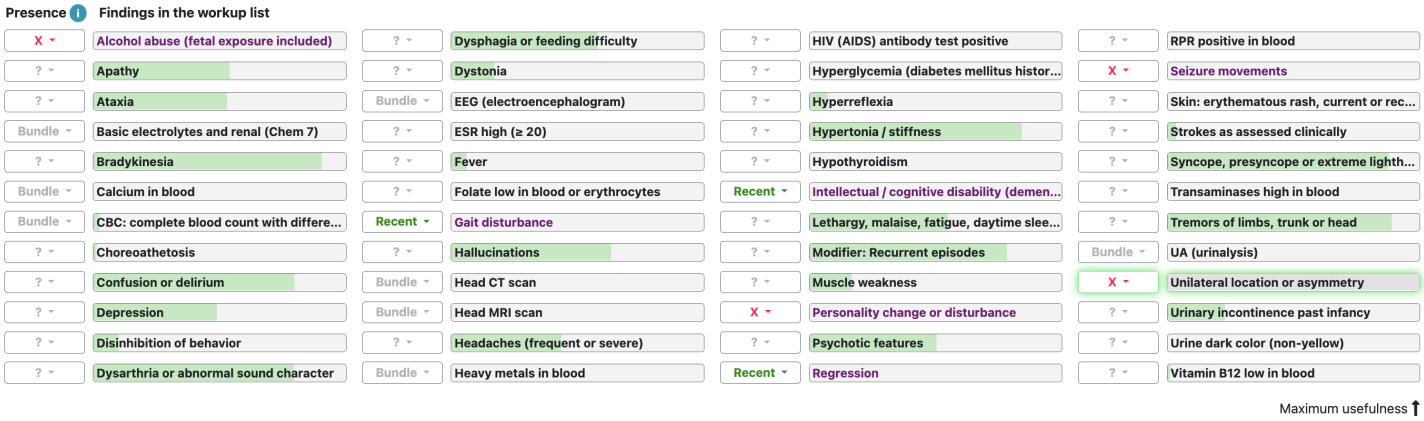
Supports all 3 cognitive modes in diagnosis

1. **WORKUP: Use a workup list of findings** by specialty or problem
2. **TEXT: Start with a few striking findings** from a note or your observations
3. **DISEASE: Begin with a likely candidate disease** and the findings that make you think of it

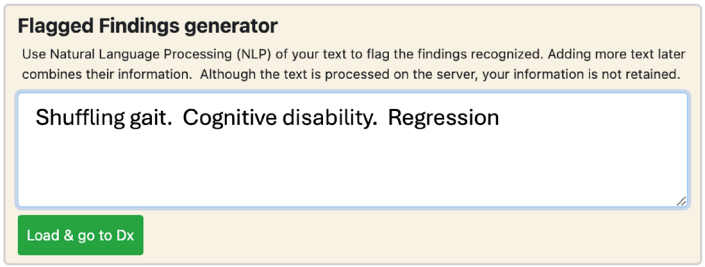
Then use those to iteratively generate a differential diagnosis and what findings would be most useful

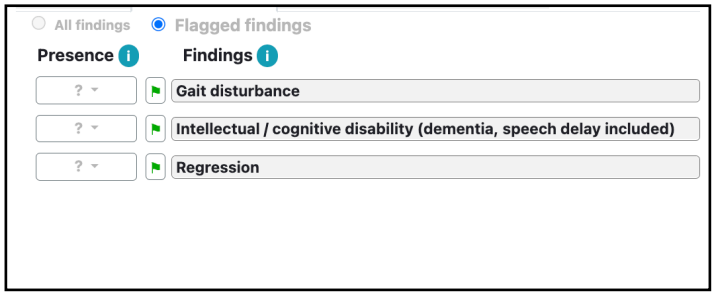
1 Workup Example: Dementia

The green shading changes dynamically as you add findings to indicate what's most useful next



2 Use text input

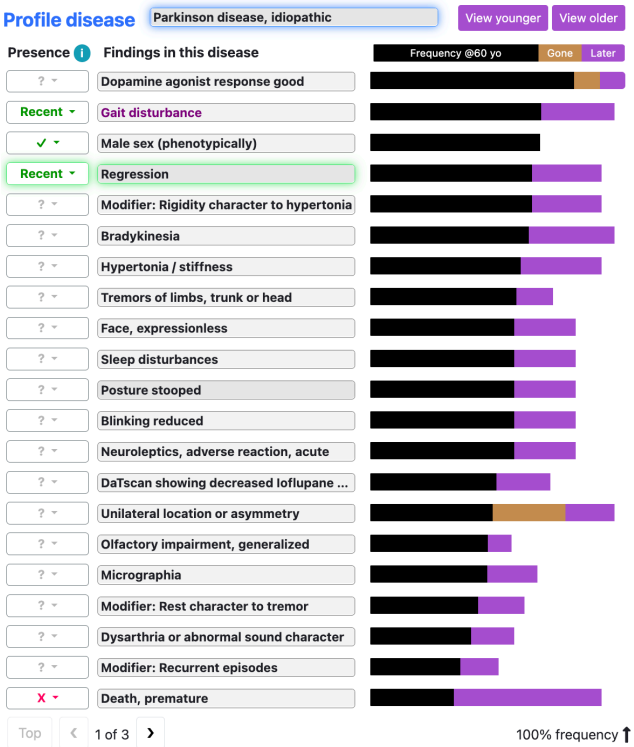




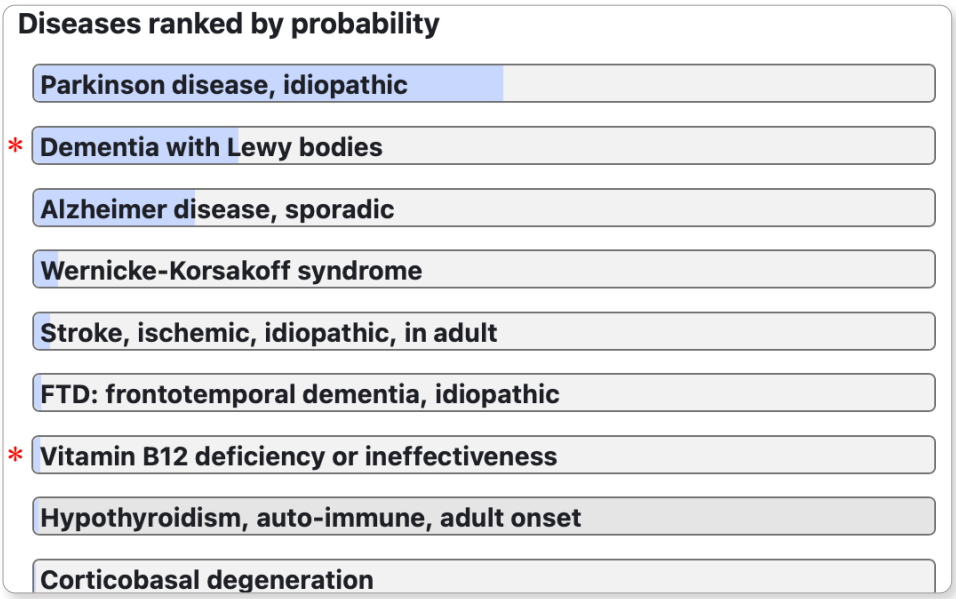
Confirm that the natural language processing found the findings you meant and comment on presence

3 Start with a disease

Comment on several findings that made you think this disease was a good candidate



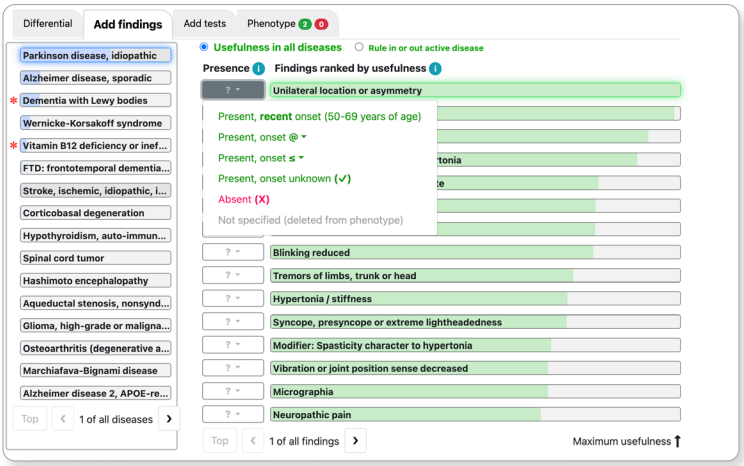
Check the differential diagnosis (DDx)



The blue shading represents the relative probability.

The red asteriks have warnings about specific risks and how to avoid causing harm during the diagnosis and treatment.

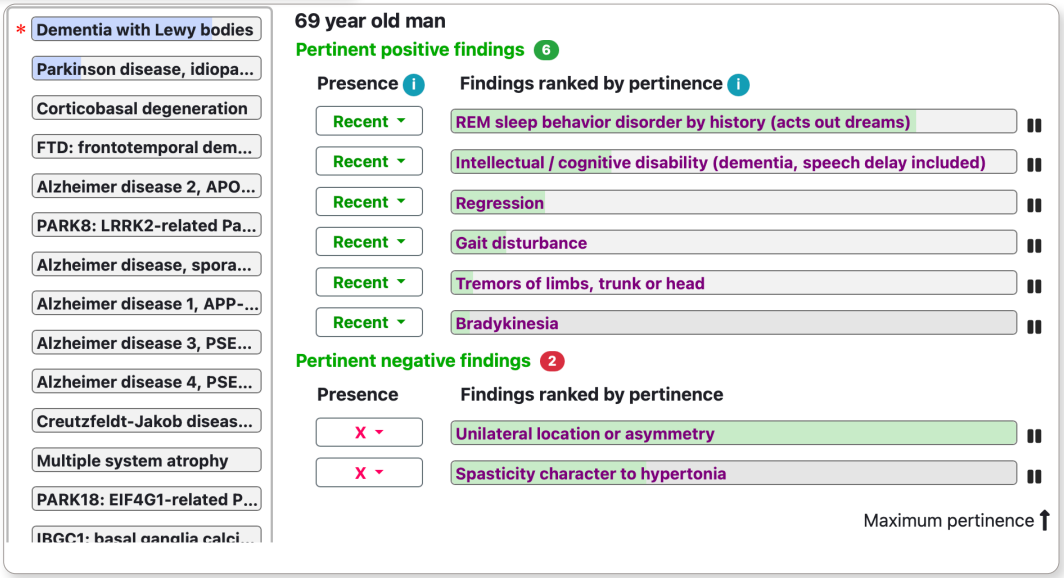
Iteratively add findings, prompted by the green shading for usefulness



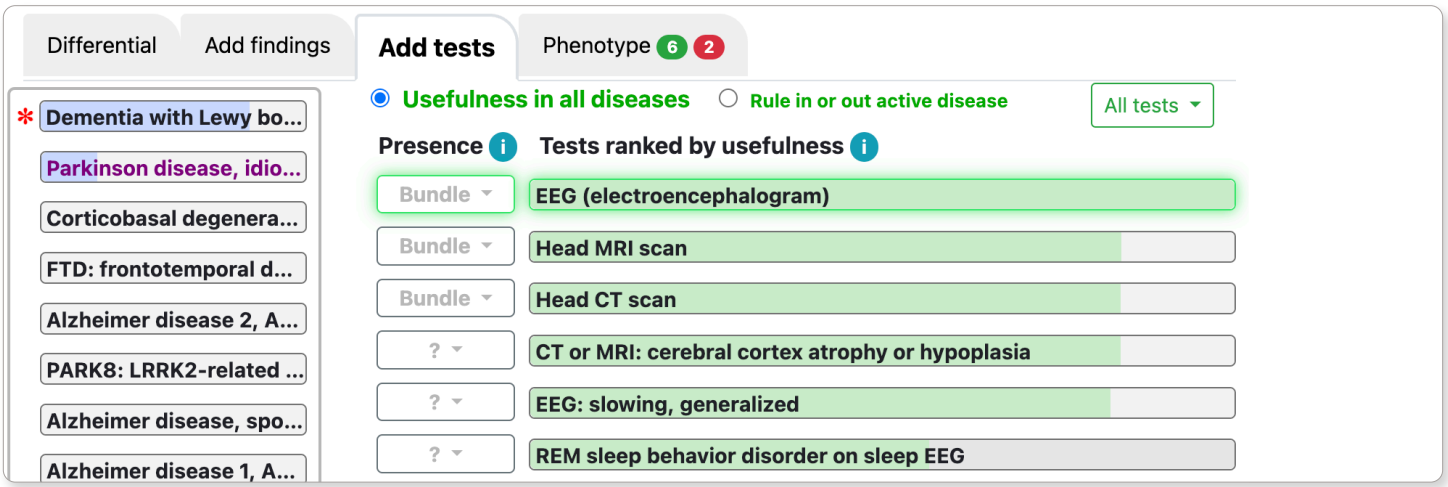
Make use of pertinent negatives as well as positives to narrow the differential diagnosis. Note that the pulldown is what we call the “Presence menu” and allows you to specify the timing of onset when known, or absence.

The differential diagnosis automatically recomputes with the addition of each new finding.

Note: you can use the black pause button at the right of each finding in the Phenotype to toggle it on and off, without losing the finding.



Decide on useful test plan



Document and collaborate

Coded findings

The patient is a 69-year-old man.

Pertinent positive findings

Recent	REM sleep behavior disorder by history (acts out dreams)	
Recent	Intellectual / cognitive disability (dementia, speech del...	HP:0001249
Recent	Regression	HP:0002376
Recent	Gait disturbance	HP:0001288
Recent	Tremors of limbs, trunk or head	HP:0001337
Recent	Bradykinesia	HP:0002067

Pertinent negative findings

Unilateral location or asymmetry	HP:0012833
Spasticity character to hypertonia	HP:0001257

SOAP Note

SUBJECTIVE
The patient is a 69-year-old man.

History: REM sleep behavior disorder by history (acts out dreams) (onset at ~60 years old) and Regression (onset at ~60 years old).

OBJECTIVE
On physical exam the positive findings are Intellectual / cognitive disability (dementia, speech delay included); Gait disturbance; Tremors of limbs, trunk or head and Bradykinesia. There is absence of Unilateral location or asymmetry and Spasticity character to hypertonia.

ASSESSMENT
The patient is a 69-year-old man with pertinent positives of REM sleep behavior disorder by history (acts out dreams); Intellectual / cognitive disability (dementia, speech delay included) and Regression and pertinent negatives of Unilateral location or asymmetry and Spasticity character to hypertonia.

Differential Diagnosis
Dementia with Lewy bodies
Parkinson disease, idiopathic
Corticobasal degeneration
FTD: frontotemporal dementia, idiopathic
Alzheimer disease 2, APOE-related

PLAN

Findings to assess
EEG: slowing, generalized
Amyloid deposition in visceral tissues
CT or MRI: cerebral cortex atrophy or hypoplasia
REM sleep behavior disorder on sleep EEG
tau and Abeta amyloid 42 markers abnormal in CSF
Brain biopsy: Lewy bodies
14-3-3 protein high in CSF
PFT: Reduced maximal expiratory pressure
APOE gene variant (monoallelic)
Brain imaging: loss of dopaminergic neurons in substantia nigra

COMPUTABLE PATIENT SUMMARY (click or load from within SimulConsult)

https://simulconsult.com/diagnose?d=252028u=f08o=4999998u=f1588o=3999998u=segal_220706150439&o=25566&u=segal_020726142514&o=25566&u=segal_091218145709&o=499999&u=f18o=25566&u=f250&o=25566&u=f195&o=25566&u=f253&o=25566&u=segal_030225183914&o=499999&i=1&t=c

Patient Summary

SimulConsult

Summary by Lynn Feldman for the 69-year-old man with with findings

Pertinent positive findings
Onsets can be at an age, by an age, or unknown

Onset	Finding	Pertinence
Recent	REM sleep behavior disorder by history (acts out dreams)	
Recent	Intellectual / cognitive disability (dementia, speech delay included)	
Recent	Regression	
Recent	Gait disturbance	
Recent	Tremors of limbs, trunk or head	
Recent	Bradykinesia	

high→

Pertinent negative findings

Absent	Finding	Pertinence
X	Unilateral location or asymmetry	
X	Spasticity character to hypertonia	

high→

Differential diagnosis

Disease	Probability
Dementia with Lewy bodies	
Parkinson disease, idiopathic	
Corticobasal degeneration	
FTD: frontotemporal dementia, idiopathic	
Alzheimer disease 2, APOE-related	
PARK8: LRRK2-related Parkinson disease, AD	
Alzheimer disease, sporadic	
Alzheimer disease 1, APP-related	
Alzheimer disease 3, PSEN1-related	
Alzheimer disease 4, PSEN2-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
☐	Bundle: EEG (electroencephalogram)
☐	Bundle: Head MRI scan
☐	Bundle: Head CT scan
☐	CT or MRI: cerebral cortex atrophy or hypoplasia
☐	EEG: slowing, generalized

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