

Phenome Dashboard

Streamlined diagnostic workflow for experts

The screenshot displays the Phenome Dashboard interface. At the top, there are tabs for 'Differential', 'Add findings', 'Add tests', and 'Phenotype'. The 'Phenotype' tab is active, showing a count of 5 green and 1 red finding. Below the tabs, there are two main sections: 'All findings' and 'Flagged findings'. The 'All findings' section is selected, showing a list of findings with a 'Presence' column (indicated by a question mark and a dropdown arrow) and a 'Findings' column (indicated by a green bar). The findings listed are: Modifier: Spasticity character to hypertonia, Syncope, presyncope or extreme lightheadedness, Lethargy, malaise, fatigue, daytime sleeping, Voice or cry: absent, weak or low volume, Blinking reduced, Psychotic features, Hallucinations, Apathy, Erectile dysfunction, and Temperature instability. At the bottom, there is a 'Top' button, a navigation bar showing '1 of all findings', and a 'Maximum usefulness' indicator with an upward arrow.

**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

Presence 1 Findings in the workup list

X	Alcohol abuse (fetal exposure included)	?	Dysphagia or feeding difficulty	?	HIV (AIDS) antibody test positive	?	RPR positive in blood
?	Apathy	?	Dystonia	?	Hyperglycemia (diabetes mellitus histor...	X	Seizure movements
?	Ataxia	Bundle	EEG (electroencephalogram)	?	Hyperreflexia	?	Skin: erythematous rash, current or rec...
Bundle	Basic electrolytes and renal (Chem 7)	?	ESR high (≥ 20)	?	Hypertonia / stiffness	?	Strokes as assessed clinically
?	Bradykinesia	?	Fever	?	Hypothyroidism	?	Syncope, presyncope or extreme ligh...
Bundle	Calcium in blood	?	Folate low in blood or erythrocytes	Recent	Intellectual / cognitive disability (demen...	?	Transaminases high in blood
Bundle	CBC: complete blood count with differe...	Recent	Gait disturbance	?	Lethargy, malaise, fatigue, daytime slee...	?	Tremors of limbs, trunk or head
?	Choreaathetosis	?	Hallucinations	?	Modifier: Recurrent episodes	Bundle	UA (urinalysis)
?	Confusion or delirium	Bundle	Head CT scan	?	Muscle weakness	X	Unilateral location or asymmetry
?	Depression	Bundle	Head MRI scan	X	Personality change or disturbance	?	Urinary incontinence past infancy
?	Disinhibition of behavior	?	Headaches (frequent or severe)	?	Psychotic features	?	Urine dark color (non-yellow)
?	Dysarthria or abnormal sound character	Bundle	Heavy metals in blood	Recent	Regression	?	Vitamin B12 low in blood

Maximum usefulness ↑

2 Use text input

Flagged Findings generator

Use Natural Language Processing (NLP) of your text to flag the findings recognized. Adding more text later combines their information. Although the text is processed on the server, your information is not retained.

Shuffling gait. Cognitive disability. Regression

Load & go to Dx

All findings 1 Flagged findings 2

Presence 1 Findings 1

?	Gait disturbance
?	Intellectual / cognitive disability (dementia, speech delay included)
?	Regression

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

Profile disease Parkinson disease, idiopathic View younger View older

Presence 1 Findings in this disease

?	Dopamine agonist response good	Frequency @60 yo	Gone	Later
Recent	Gait disturbance			
✓	Male sex (phenotypically)			
Recent	Regression			
?	Modifier: Rigidity character to hypertonia			
?	Bradykinesia			
?	Hypertonia / stiffness			
?	Tremors of limbs, trunk or head			
?	Face, expressionless			
?	Sleep disturbances			
?	Posture stooped			
?	Blinking reduced			
?	Neuroleptics, adverse reaction, acute			
?	DaTscan showing decreased Ioflupane ...			
?	Unilateral location or asymmetry			
?	Olfactory impairment, generalized			
?	Micrographia			
?	Modifier: Rest character to tremor			
?	Dysarthria or abnormal sound character			
?	Modifier: Recurrent episodes			
X	Death, premature			

Top 1 of 3 100% frequency ↑

Check the differential diagnosis (DDx)

Diseases ranked by probability

	Parkinson disease, idiopathic
*	Dementia with Lewy bodies
	Alzheimer disease, sporadic
	Wernicke-Korsakoff syndrome
	Stroke, ischemic, idiopathic, in adult
	FTD: frontotemporal dementia, idiopathic
*	Vitamin B12 deficiency or ineffectiveness
	Hypothyroidism, auto-immune, adult onset
	Corticobasal degeneration

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

The screenshot shows the 'Add findings' interface. On the left, a list of diseases is shown, including Parkinson disease, idiopathic; Dementia with Lewy bodies; Alzheimer disease, sporadic; Wernicke-Korsakoff syndrome; Vitamin B12 deficiency or ineffectiveness; FTD: frontotemporal dementia, idiopathic; Stroke, ischemic, idiopathic, in adult; Corticobasal degeneration; Hypothyroidism, auto-immune, adult onset; Spinal cord tumor; Hashimoto encephalopathy; Aqueductal stenosis, nonsyndromic; Glioma, high-grade or malignant; Osteoarthritis (degenerative); Marchiafava-Bignami disease; Alzheimer disease 2, APOE-epsilon4; and others. On the right, a table of findings ranked by usefulness is shown. The findings are categorized by 'Usefulness in all diseases' and 'Rule in or out active disease'. The findings include: Unilateral location or asymmetry (Present, recent onset (50-69 years of age), Present, onset @ -, Present, onset < -, Present, onset unknown (✓), Absent (X), Not specified (deleted from phenotype)); Blinking reduced; Tremors of limbs, trunk or head; Hypertonia / stiffness; Syncope, presyncope or extreme lightheadedness; Modifier: Spasticity character to hypertonia; Vibration or joint position sense decreased; Micrographia; and Neuropathic pain. Each finding has a green bar indicating its usefulness and a dropdown menu for 'Presence'.

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.

The screenshot shows the '69 year old man' interface. On the left, a list of diseases is shown, including Dementia with Lewy bodies; Parkinson disease, idiopathic; Corticobasal degeneration; FTD: frontotemporal dementia, idiopathic; Alzheimer disease 2, APOE-epsilon4; PARK8: LRRK2-related Parkinsonism; Alzheimer disease, sporadic; Alzheimer disease 1, APP-related; Alzheimer disease 3, PSEN1-related; Alzheimer disease 4, PSEN2-related; Creutzfeldt-Jakob disease; Multiple system atrophy; PARK18: EIF4G1-related Parkinsonism; and IRGC1: basal ganglia calcification. On the right, a table of findings ranked by pertinence is shown. The findings are categorized by 'Pertinent positive findings' and 'Pertinent negative findings'. The findings include: REM sleep behavior disorder by history (acts out dreams); Intellectual / cognitive disability (dementia, speech delay included); Regression; Gait disturbance; Tremors of limbs, trunk or head; Bradykinesia; Unilateral location or asymmetry; and Spasticity character to hypertonia. Each finding has a green bar indicating its pertinence and a dropdown menu for 'Presence'.

Decide on useful test plan

Differential

Add findings

Add tests

Phenotype 6 2

* Dementia with Lewy bo...

Parkinson disease, idio...

Corticobasal degenera...

FTD: frontotemporal d...

Alzheimer disease 2, A...

PARK8: LRRK2-related ...

Alzheimer disease, spo...

Alzheimer disease 1, A...

☒ Usefulness in all diseases

☐ Rule in or out active disease

All tests ▾

Presence ⓘ

Tests ranked by usefulness ⓘ

Bundle ▾

EEG (electroencephalogram)

Bundle ▾

Head MRI scan

Bundle ▾

Head CT scan

? ▾

CT or MRI: cerebral cortex atrophy or hypoplasia

? ▾

EEG: slowing, generalized

? ▾

REM sleep behavior disorder on sleep EEG

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) [HP:0001249](#)

Recent Intellectual / cognitive disability (dementia, speech del... [HP:0002376](#)

Recent Regression [HP:0001288](#)

Recent Gait disturbance [HP:0001337](#)

Recent Tremors of limbs, trunk or head [HP:0002067](#)

Recent Bradykinesia

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=25202&u=f0&o=4999998&u=f158&o=399999&u=segal.2207061504358&o=25568&u=segal.0207261425149&o=25566&u=segal.021218145709&o=4999998&u=f18&o=25566&u=f250&o=25566&u=f195&o=25566&u=f253&o=25566&u=segal.030225183914&o=4999998&i=1&t=c>

Patient Summary

SimulConsult

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

Pertinent negative findings

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

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	

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