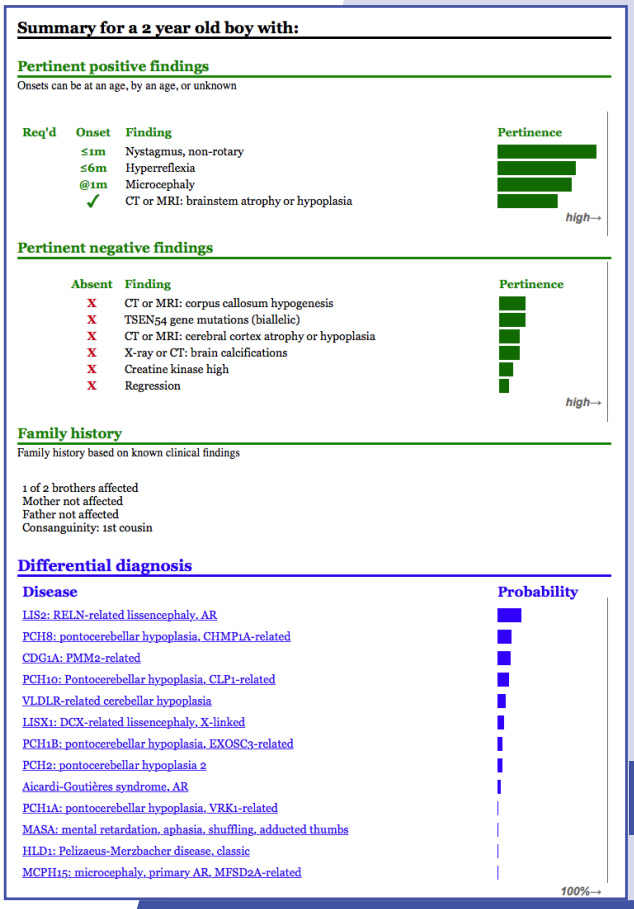
Clinician tools for ordering gene tests

Personalized Panel of useful genes to test for your patient



Summary for a 2 year old boy with:

Pertinent positive findings

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

Req'd	Onset	Finding	Pertinence
	≤1m	Nystagmus, non-rotary	
	≤6m	Hyperreflexia	
	@1m	Microcephaly	
✓		CT or MRI: brainstem atrophy or hypoplasia	

high→

Pertinent negative findings

Absent	Finding	Pertinence
X	CT or MRI: corpus callosum hypogenesis	
X	TSEN54 gene mutations (biallelic)	
X	CT or MRI: cerebral cortex atrophy or hypoplasia	
X	X-ray or CT: brain calcifications	
X	Creatine kinase high	
X	Regression	

high→

Family history

Family history based on known clinical findings

1 of 2 brothers affected
Mother not affected
Father not affected
Consanguinity: 1st cousin

Differential diagnosis

Disease	Probability
LIS2: RELN-related lissencephaly, AR	
PCH8: pontocerebellar hypoplasia, CHMP1A-related	
CDG1A: PMM2-related	
PCH10: Pontocerebellar hypoplasia, CLP1-related	
VLDLR-related cerebellar hypoplasia	
LISX1: DCX-related lissencephaly, X-linked	
PCH1B: pontocerebellar hypoplasia, EXOSC3-related	
PCH2: pontocerebellar hypoplasia 2	
Aicardi-Goutières syndrome, AR	
PCH1A: pontocerebellar hypoplasia, VRK1-related	
MASA: mental retardation, aphasia, shuffling, adducted thumbs	
HLD1: Pelizaeus-Merzbacher disease, classic	
MCPH15: microcephaly, primary AR, MFSD2A-related	

100%→

Key Features

- **"Personalized Panel"** automatically generates a list of the useful genes to test, considering treatability and cost, and also documents the clinical rationale to be used with panels, exomes and exome slices
- **Complete coverage** of described Mendelian disorders, as well as many non-genetic disorders in neurology & rheumatology
- **"Useful Findings"** and **"Useful Tests"** prompts you about clinical findings and test results relevant to your patient
- **"Disease Profile"** and **"Assess Disease"** enable you to evaluate the rationale for the differential diagnosis
- **"Summary"** (shown) allows you to save to your desktop and later jump back in with the prior findings

CONSULT on test plan

Most useful tests for this patient

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

Order	Test
☐	CT or MRI: lissencephaly
☐	Bundle: Isoelectric focusing for transferrin glycosylation
☐	Transferrin hypoglycosylated; type 1 pattern
☐	CT or MRI: pontine atrophy or hypoplasia
☐	RELN gene mutations (biallelic)
☐	MRI: white matter abnormality

Personalized panel: Most useful genes to test for this patient

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

Order	Test
☐	RELN gene mutation (biallelic)
☐	CLP1 gene mutation (biallelic)
☐	CHMP1A gene mutation (biallelic)
☐	PMM2 gene mutation (biallelic)
☐	VLDLR gene mutation (biallelic)
☐	DCX gene mutation (X-linked)
☐	EXOSC3 gene mutation (biallelic)
☐	RNASEH2B gene mutation (biallelic)
☐	L1CAM gene mutation (X-linked)
☐	VRK1 gene mutation (biallelic)
☐	MFSD2A gene mutation (biallelic)
☐	PLP1 gene duplication (X-linked)
☐	TREX1 gene mutation (biallelic)

Key Benefits

Focused on your patient. Uses your patient's pertinent positive and negative findings to generate general and gene test recommendations

Fast. Get a simultaneous consult in seconds

Accurate. Reduces diagnostic errors by helping you consider uncommon presentations and rare diseases, and order the right tests

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