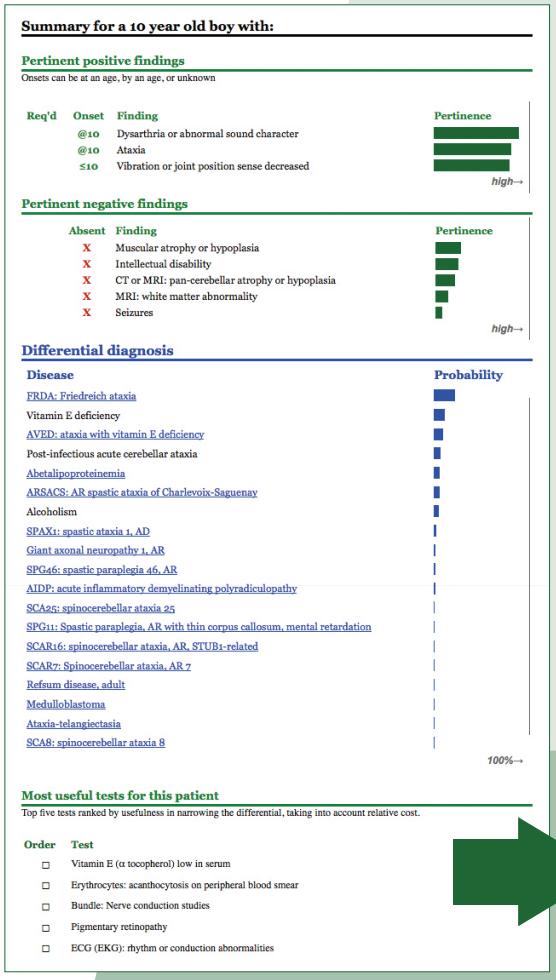
Classic Version

Diagnostic Decision Support

The Classic version is free. It is accessed from the web but runs locally on your computer.



Summary for a 10 year old boy with:

Pertinent positive findings

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

Req'd	Onset	Finding	Pertinence
	@10	Dysarthria or abnormal sound character	
	@10	Ataxia	
	≤10	Vibration or joint position sense decreased	

high→

Pertinent negative findings

Absent	Finding	Pertinence
X	Muscular atrophy or hypoplasia	
X	Intellectual disability	
X	CT or MRI: pan-cerebellar atrophy or hypoplasia	
X	MRI: white matter abnormality	
X	Seizures	

high→

Differential diagnosis

Disease	Probability
FRDA: Friedreich ataxia	
Vitamin E deficiency	
AVED: ataxia with vitamin E deficiency	
Post-infectious acute cerebellar ataxia	
Abetalipoproteinemia	
ARSACS: AR spastic ataxia of Charlevoix-Saguenay	
Alcoholism	
SPAX1: spastic ataxia 1, AD	
Giant axonal neuropathy 1, AR	
SPG46: spastic paraplegia 46, AR	
AIDP: acute inflammatory demyelinating polyradiculopathy	
SCA25: spinocerebellar ataxia 25	
SPG11: Spastic paraplegia, AR with thin corpus callosum, mental retardation	
SCAR16: spinocerebellar ataxia, AR, STUB1-related	
SCAR7: Spinocerebellar ataxia, AR 7	
Refsum disease, adult	
Medulloblastoma	
Ataxia-telangiectasia	
SCA8: spinocerebellar ataxia 8	

100%→

Most useful tests for this patient

Top five tests ranked by usefulness in narrowing the differential, taking into account relative cost.

Order	Test
☐	Vitamin E (α tocopherol) low in serum
☐	Erythrocytes: acanthocytosis on peripheral blood smear
☐	Bundle: Nerve conduction studies
☐	Pigmentary retinopathy
☐	ECG (EKG): rhythm or conduction abnormalities

Key Features

- **"Useful Findings"** prompts you to add clinical findings relevant to your patient
- **"Useful Tests"** prompts you about tests relevant to your patient
- **"Disease Profile"** and **"Assess Disease"** help you evaluate the rationale for the differential diagnosis
- **"Summary"** (shown) allows you to save to your desktop and later jump back in with all the findings entered
- **Complete coverage** of described Mendelian disorders, as well as many non-genetic disorders in neurology & rheumatology

REVISED personalized test protocol when Vitamin E found to be normal

Most useful tests for this patient

Top five tests ranked by usefulness in narrowing the differential, taking into account relative cost.

Order	Test
☐	Bundle: Nerve conduction studies
☐	FXN genes >90 GAA repeats (biallelic)
☐	ECG (EKG): rhythm or conduction abnormalities
☐	Nerve conduction: NCV slow, motor
☐	ABR abnormal

Key Benefits

Focused on your patient. Uses your patient's pertinent positive and negative findings, including onset ages, to generate recommendations

Fast. Get a simultaneous consult in seconds. Pertinence calculation for findings focuses the assessment on what's important

Accurate. Reduces diagnostic errors by sugggesting uncommon presentations and rare diseases

SimulConsult®

Sign up free at
www.SimulConsult.com