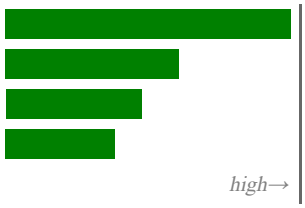
SimulConsult

Summary for the 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	
	@1m	Microcephaly	
	✓	CT or MRI: brainstem atrophy or hypoplasia	
	≤6m	Hyperreflexia	



Pertinent negative findings

Absent	Finding	Pertinence
X	Regression	

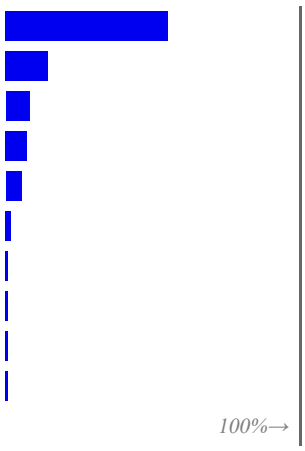
Family history

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
PCH2: pontocerebellar hypoplasia 2	
PCH8: pontocerebellar hypoplasia 8	
PCH10: pontocerebellar hypoplasia 10	
Vici syndrome	
Aicardi-Goutières syndrome, AR	
LIS2: RELN-related lissencephaly, AR	
VLDLR-related cerebellar hypoplasia	
PCH1B: pontocerebellar hypoplasia, EXOSC3-related	
CDG1A: PMM2-related	
Muscular dystrophy-dystroglycanopathy B6	



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: MRI scan of the brain
☐	Bundle: CT scan of the head
☐	TSEN54 gene variants (biallelic)
☐	Creatine kinase high
☐	CHMP1A gene variants (biallelic)

Generated by SimulConsult® on 29 March 2019 15:55 using interface of 2019 March 15 18:13, algorithms of 2019 March 15 18:16 and database of 14 March 2019 20:53. All times are UTC. Care setting was primary. Onset was used.