Advanced Phenome with Prognosis Table

Communicate expectations about how a disease will unfold in a simple, clear way for over 6,100 unusual diseases.

SimulConsult

Disease Prognosis Table©

Prognosis for VLDLR-related cerebellar hypoplasia

Signs and Symptoms	At what age do people with this disease have these findings?												
	Birth	1 month	3 months	6 months	1 year	3 years	6 years	10 years	15 years	25 years	40 years	60 years	80 years
Ataxia	Few	Few	Some	Some	Most	Most	Most	Most	Most	Most	Most	Most	Most
Intellectual disability	NA	Few	Few	Some	Most	Most	Most	Most	Most	Most	Most	Most	Most
Motor developmental delay	NA	Few	Few	Some	Some	Most	Most	Most	Most	Most	Most	Most	Most
Gait disturbance	NA	NA	NA	NA	Some	Most	Most	Most	Most	Most	Most	Most	Most
Nystagmus, non-rotary	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Eye movement deficit, horizontal	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Hyperreflexia	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Seizures with abnormal movements	Few	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Foot: pes planus	Few	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
Cataracts	Few	Few	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some
Dysarthria or abnormal sound character	NA	NA	Few	Few	Some	Some	Some	Some	Some	Some	Some	Some	Some
Stature short	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few
Findings detected by laboratory tests													
CT or MRI: pan-cerebellar atrophy or hypoplasia	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most
VLDLR gene mutations (biallelic)	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most
CT or MRI: pontine atrophy or hypoplasia	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
CT or MRI: lissencephaly	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some
CT or MRI: brainstem atrophy or hypoplasia	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some	Some

KEY	None or NA	Few is less than or equal to 30%	Some is more than 30%	Most is more than 85%

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Provider Resources: GeneReviews and OMIM
Patient and Family-Oriented Resources: Genetics Home Reference and Disease-focused patient advocacy organizations

Key Features

- **Clinical and lab findings** in disease
- **Variable expression** shown in an intuitive way
- Shows how **diseases unfold over time**
- Clusters findings in logical way, by frequency (and age of patient)

Key Benefits

PCORI research-tested tool is well liked by patient families, primary care physicians, and specialists. (PMIDs: 26842872 & 26086630)

Answers the key question at diagnosis "what should I expect" completely and intutitively illustrates "variable expression."

SimulConsult®
A Simultaneous Consult On Your Patient's Diagnosis

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