

# Loss of Heterozygosity

## Find the pertinent genes and likely diagnosis in seconds

### Key Features

- Automatically correlates the list of patient findings with the genes in chromosomal regions
- Identifies and ranks the **genes by pertinence** for this patient
- Enables a quick call to the ordering physician to focus on additional findings that help reach a conclusion, relying on the “**useful findings**” and “**useful tests**” features

**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	High
	≤6m	Hyperreflexia	High
✓	@1m	Microcephaly	High
		CT or MRI: brainstem atrophy or hypoplasia	High

**Pertinent negative findings**

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

**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**

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, YRK1-related  
MASA: mental retardation, aphasia, shuffling, adducted thumbs  
HLD1: Pelizaeus-Merzbacher disease, classic  
MCPH1: microcephaly, primary, AR, MFSD2A-related

**Probability**

100%→

### CONSULT on genes in regions of loss of heterozygosity

**Differential diagnosis** | **Add findings** | **Add tests** | **Phenotype** | **Genotype** |  Advanced mode

**Diseases**

- VLDLR-related cerebellar hypoplasia
- JBTS25: Joubert syndrome
- Homocystinuria, megalocystinuria
- Hartnup disease, symptomatic
- MRT5: mental retardation
- Adrenoleukodystrophy
- LIS2: RELN-related lissencephaly
- Retinal cone dystrophy
- Multiple sulfatase deficiency
- PCH8: pontocerebellar hypoplasia
- Zellweger syndrome
- CDG1A: PMM2-related congenital disorder of glycosylation
- PCH10: Pontocerebellar hypoplasia
- LISX1: DCX-related lissencephaly
- PCH1B: pontocerebellar hypoplasia
- PCH2: pontocerebellar hypoplasia
- Dubowitz syndrome
- Aicardi-Goutières syndrome
- Senior-Løken syndrome
- PCH1A: pontocerebellar hypoplasia
- Multiple sulfatase deficiency

**From 353 genes in LOH regions:**

**Pertinent gene zygosity**

5	✓	▼	Gene	Finding color key: Pertinence
5	✓	▼	VLDLR gene variants (biallelic)	High
5	✓	▼	CEP104 gene variants (biallelic)	High
5	✓	▼	MTRR gene variants (biallelic)	High
5	✓	▼	SLC6A19 gene variants (biallelic)	High
5	✓	▼	NSUN2 gene variants (biallelic)	High
5	✓	▼	PEX10 gene variants (biallelic)	High
5	✓	▼	KCNV2 gene variants (biallelic)	High
5	✓	▼	SUMF1 gene variants (biallelic)	High
5	✓	▼	NPHP4 gene variants (biallelic)	High
5	✓	▼	GLDC gene variants (biallelic)	High
5	✓	▼	ESPN gene variants (biallelic)	High
5	✓	▼	SLC1A1 gene variants (biallelic)	High
5	✓	▼	CRBN gene variants (biallelic)	High
5	✓	▼	SLC6A3 gene variants (biallelic)	High
5	✓	▼	B3GALT6 gene variants (biallelic)	High
5	✓	▼	TRNT1 gene variants (biallelic)	High
5	✓	▼	GLIS3 gene variants (biallelic)	High
5	✓	▼	PLEKHG5 gene variants (biallelic)	High

**Differential Dx**  
**Incidental genes**  
**Discovery genes**  
**Panel of genes**  
**Loss of heteroz**  
**Prognosis**  
**Assess finding**  
**Profile finding**  
**Database**  
**Search** | **File**  
**Start** | **Help**

**More tips** | **Summary** | **Note**  
**OMIM** | **Gene test search**

**Tip:** The "Summary" and "Note" buttons at right display patient reports

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### Key Benefits

**Focused on your patient.**  
Uses your patient's pertinent positive and negative findings to generate a list of candidate genes in order of pertinence

**Fast.** Analysis in seconds, enables useful discussion with referring physician, when needed

**Accurate.** Reduces errors and inconsistencies

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