FAMILIES / GENETICS / CLINICIANS

Decode an ASH1L genetic report

A field-by-field companion for preserving what the laboratory actually reported - and understanding what the report cannot predict.

USE THIS WHEN

You are reading, organizing, or discussing a laboratory report.

TIME NEEDED

15 to 20 minutes

TAKE OR SHARE

Bring this with the signed report.

Read the complete report, not a copied variant line.

A portal summary can omit the transcript, testing method, classification basis, inheritance, limitations, or additional findings. The signed laboratory report is the source document.

01 / MAP THE REPORT

A report connects several fields

The synthetic specimen below is not a real person or variant. It shows where the most important fields usually appear and why they must stay together.

GENETIC LABORATORY REPORT - SYNTHETIC EXAMPLE

FOR ORIENTATION ONLY - NOT A REAL REPORT, PERSON, OR VARIANT

1. **GENE AND TRANSCRIPT**
   ASH1L | transcript: laboratory-specified reference
2. **DNA AND PROTEIN NOTATION**
   c.[example] | p.[example]
3. **ZYGOSITY AND VARIANT CLASS**
   heterozygous | consequence: laboratory-defined
4. **CLASSIFICATION**
   likely pathogenic | classification date: [example]
5. **INHERITANCE**
   not yet determined | parental testing: not completed
6. **METHOD AND LIMITATIONS**
   assay type, coverage, confirmation, and detection limits
7. **ADDITIONAL FINDINGS**
   other findings, copy-number findings, or none reported

Keep every field connected.

The same cDNA or protein shorthand can be interpreted differently if the transcript, classification, inheritance, method, or additional findings are missing.

02 / FIELD DECODER

The fields to preserve

Copy exact wording first. Interpretation can be updated later, but a lost transcript, method, or classification date is difficult to reconstruct.

FIELD 01

Gene and transcript

Which reference sequence the laboratory used. Ask whether it is the laboratory's current clinically relevant transcript.

FIELD 02

cDNA and protein lines

How the change is described at DNA and protein level. Preserve both and do not infer a missing protein consequence.

FIELD 03

Variant class

May be truncating, missense, splice, deletion/CNV, complex, or another laboratory-defined consequence.

FIELD 04

Zygosity

Whether one or both copies carry the reported change. Heterozygous does not itself mean pathogenic.

FIELD 05

Classification and date

Pathogenic, likely pathogenic, VUS, likely benign, or benign - with the evidence date and laboratory wording.

FIELD 06

Inheritance

De novo, inherited, unknown, or not assessed. Preserve exactly who was tested and by what method.

FIELD 07

Method and limits

What the assay could and could not detect, including relevant coverage, confirmation, and copy-number limitations.

FIELD 08

Additional findings

Other reported variants or copy-number findings. Keep their separate classifications and uncertainty visible.

03 / CLASSIFICATION AND INHERITANCE

Classification is not prognosis

A laboratory classification addresses whether a variant is considered disease-causing. It does not predict severity, timing, course, or every clinical feature.

CLASSIFICATION

Pathogenic or likely pathogenic

The laboratory considers the variant causative or probably causative under its classification framework. Ask what evidence and date support the classification and whether reinterpretation is recommended.

UNCERTAIN

Variant of uncertain significance (VUS)

Evidence is insufficient or conflicting. A VUS should not be treated as a confirmed molecular diagnosis or used alone for irreversible medical decisions. Family testing or later evidence may change the interpretation.

INHERITANCE

De novo

Not detected in tested parental samples, subject to sample identity and assay limits. This does not eliminate germline mosaicism or settle every recurrence question.

INHERITANCE

Inherited or unknown

Record the tested relative, relationship, result, method, and whether segregation is complete. Unknown is not the same as de novo.

Genetic counseling keeps the result in context.

Questions about recurrence, parental testing, mosaicism, reproductive options, and testing of relatives require the complete family and laboratory context.

04 / MOLECULAR CONSEQUENCE

Variant consequence needs variant-specific interpretation

Variant type is useful organization, not a substitute for classification, transcript review, or functional evidence.

Loss of function / truncating

Expected to reduce or disrupt normal gene product. Not every predicted consequence is display-positioned or proven to have the same biological effect.

Missense

One amino acid is replaced by another. Position and prediction are not enough; classification requires multiple evidence types.

Splice

May alter RNA processing. The protein consequence can remain undefined unless transcript or functional evidence resolves it.

Deletion or CNV

May involve all or part of ASH1L and sometimes neighboring genes. Size, boundaries, inheritance, and other genes matter.

Complex or mixed

Structural or multi-variant configurations must remain visible rather than forced into a simple sequence class.

05 / YOUR REPORT AND CONVERSATION

Copy the report, then ask the next questions

Use this page as a private index. Exact variants can identify a person in a rare-disease community, so circulate the completed page only when needed.

GENE AND TRANSCRIPT

Copy the laboratory wording.

CDNA AND PROTEIN LINES

Include punctuation and transcript-dependent notation.

CLASSIFICATION, DATE, AND INHERITANCE

Record the laboratory, report date, parental testing, and exact result.

METHOD, LIMITATIONS, AND ADDITIONAL FINDINGS

Preserve the relevant wording or page reference.

Questions to take to genetics

- [ ] What exact transcript, cDNA line, protein line, and genome build were used?
- [ ] What is the classification today, and what evidence supports it?
- [ ] Was confirmatory testing performed or considered necessary?
- [ ] What is known about inheritance, parental testing, mosaicism, and recurrence?
- [ ] Were copy-number changes and other findings assessed?
- [ ] When should reinterpretation be requested, and who will initiate it?

Before sharing a report

Remove identifiers the reader does not need. A community introduction or first conversation usually does not require the complete report.

1. ClinGen ASH1L gene-disease validity
   https://search.clinicalgenome.org/kb/genes/HGNC%3A19088
2. ClinGen ASH1L dosage sensitivity
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