

# Enhanced Whole Genome Sequencing and Whole Exome Sequencing Reports

**Clearer layouts and improved usability designed to deliver actionable insights for confident clinical decisions**

Baylor Genetics is introducing newly enhanced Whole Genome Sequencing (WGS) and Whole Exome Sequencing (WES) reports — designed to deliver greater clarity, usability, and clinical relevance. These improvements reflect feedback from healthcare providers and align with national reporting guidelines, reinforcing Baylor Genetics' commitment to precision diagnostics.

**In the coming weeks**, providers will begin receiving reports in the new format, ensuring timely access to clearer insights for patient care.

## What's New

- A streamlined design that consolidates priority information into a single, easy-to-read view
- An updated organizational framework where findings are sorted by result type: Primary Findings, ACMG Secondary Findings, Incidental Findings, and Genes with No Known Disease Association (candidate genes)
- Prioritization of the most clinically relevant management information and removal of redundant variant details

## Future Updates

- All other report type, including Panels (e.g., Epilepsy Panel), Chromosomal Microarray Analysis (CMA), and Mitochondrial testing (mtDNA)
- Canceled reports, including WGS and WES
- Clinical content such as indication, summary, and disease description

# New Design Highlights

Making Clinical Insights Easier to Interpret

## Clinical Findings

All key clinical information is now presented on the front page for quick, confident assessment.

This includes:

- 1. Clinical Indication
- 2. Results
- 3. Results Summary

**CLINICAL INDICATION**

Based on the submitted clinical information, the patient has microcephaly, highly arched eyebrow, long eyelashes, upslanted palpebral fissure, hearing impairment, anteverted nares, choanal atresia, cleft palate, microretrognathia, oral-pharyngeal dysphagia, chronic lung disease, hypoxemia, hypercapnia, respiratory failure, obstructive sleep apnea, cardiomyopathy, congestive heart failure, tetralogy of Fallot, pulmonary artery stenosis, ventricular septal defect, akinesia, edema, increased body weight, adrenal insufficiency, gastroesophageal reflux, split hand, 1-5 toe syndactyly, eczematoid dermatitis, and mottled pigmentation.

**RESULTS**

**+** **PRIMARY FINDINGS**  
Positive - See Results Summary section below.

**+** **ACMG SECONDARY FINDINGS**  
Positive - See Results Summary section below.

**-** **INCIDENTAL FINDINGS**  
Negative - None Detected.

**-** **GENES WITH NO KNOWN DISEASE ASSOCIATION**  
Negative - None Detected.

**RESULTS SUMMARY**

A de novo heterozygous pathogenic variant in the NIPBL gene was detected, consistent with Cornelia de Lange syndrome 1.

A de novo heterozygous variant of uncertain significance in the KMT2D gene and a paternally inherited intragenic deletion in the BAZ2B gene were detected, possibly related to this patient's phenotype.

## Visual Cues for Faster Interpretation

The new result bars make it easier to interpret findings at a glance, so providers can act quickly and confidently:

This includes:

- Clear shading for results:
  - Dark shading = Positive result
  - Light shading = Indeterminate
  - No shading = Negative or opt-out
- Symbols for clarity:
  - An “i” symbol marks findings in genes with no known disease association
- Opt-in/opt-out indicators:
  - Clearly defined selections, with opt-out shown by a circle and slash

**RESULTS**

**+** **PRIMARY FINDING**  
Positive - See Results Summary section below.

**+** **ACMG SECONDARY FINDING**  
Positive - See Results Summary section below.

**+** **PRIMARY FINDING**  
Indeterminate - See Results Summary section below.

**i** **GENES WITH NO KNOWN DISEASE ASSOCIATION**  
Variant Detected - For details see Genes with No Known Disease Association table.

**-** **INCIDENTAL FINDING**  
Negative - None Detected.

**⊘** **ACMG SECONDARY FINDING**  
Opt-Out

For questions about this update, your Baylor Genetics regional account executive is ready to help. You may also contact Client Services at 1-800-411-4363 or [info@baylorgenetics.com](mailto:info@baylorgenetics.com).

New Informational Column

To make reports easier to navigate, the first page now includes a dedicated informational column with key testing details—so providers can access critical information quickly and confidently.

This column highlights:

- The preferred RNA sequencing (RNAseq) option
- Comparator information for family-based testing

Optional Testing

RNAseq ..... Opt-Out

Comparators

Mother ..... Lab#1234567

Secondary Findings ..... Opt-Out

Father ..... Lab#1234567

Secondary Findings ..... Opt-Out

Variant Findings Table

Prioritizing Actionable Insights

To support faster clinical interpretation, the second page now includes a consolidated Variant Findings Table. This table organizes and prioritizes all actionable findings for efficient review, including:

- Primary Findings
- ACMG Secondary Findings
- Incidental Findings

Additionally, variants are listed in order of pathogenicity.

VARIANT FINDINGS

Disease	Inheritance Pattern	Gene / Variant	Genotype	Variant Type	Inherited From	Variant Classification
PRIMARY FINDINGS						
Cornelia De Lange Syndrome 1	Autosomal Dominant	NIPBL c.2479del, p.R87Gfs*20	Heterozygous	Sequence Variant	De Novo	Pathogenic
KMT2D-Related Disorders	Autosomal Dominant	KMT2D: c.1952C>G, p.S651W	Heterozygous	Sequence Variant	De Novo	Variant of Uncertain Significance
Neurodevelopmental Disorder	Autosomal Dominant	BAZ2B:HG38 chr2:159470492-159491775 del(21.28kb)	Heterozygous	Sequence Variant	De Novo	Variant of Uncertain Significance
ACMG SECONDARY FINDINGS						
KCNQ1-Related Arrhythmias	Autosomal Dominant	KCNQ1: c.1552C>T, p.R518*	Heterozygous	Sequence Variant	N/A	Pathogenic
INCIDENTAL FINDINGS						
Metaphyseal Chondrodysplasia, Schmid Type	Autosomal Dominant	COL10A1 (NM_000493.4):c.2011T>C (p.S671P)	Heterozygous	Sequence Variant	Mother	Likely Pathogenic

Guideposts for Easier Navigation & Standardized Nomenclature

To make reports more clear, each variant page now includes clear *guideposts* that indicate whether a finding is primary, ACMG secondary, or incidental.

Inclusion of reference sequence identifiers to support easier variant look-up and ensure consistency across the field. These enhancements streamline interpretation and reinforce Baylor Genetics' commitment to precision and clarity.

PRIMARY FINDINGS			NIPBL (NM_133433.4) c.2479del, p.R87Gfs*20			
Disease	Inheritance Pattern	Gene / Variant	Genotype	Variant Type	Inherited From	Variant Classification
Cornelia De Lange Syndrome 1	Autosomal Dominant	NIPBL c.2479del, p.R87Gfs*20	Heterozygous	Sequence Variant	De Novo	Pathogenic
ACMG SECONDARY FINDINGS			KCNQ1 (NM_000218.3):c.1552C>T, p.Arg518*			
Disease	Inheritance Pattern	Gene / Variant	Genotype	Variant Type	Inherited From	Variant Classification
KCNQ1-Related Arrhythmias	Autosomal Dominant	KCNQ1: c.1552C>T, p.Arg518Ter	Heterozygous	Sequence Variant	N/A	Pathogenic

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