3billion

Your one answer

# Clinical Genome / Exome Based Genetic Testing for Rare Disease Patients



# Sample to Clinical Report

| | 3B-GENOME | 3B-EXOME | 3B-VARIANT |
|---|---|---|---|
| Scope | Genome | Exome | Targeted Region(s) |
| mtDNA analysis [1] | ○ | ○ | N/A |
| Type | Proband, Duo, Trio | Proband, Duo, Trio | N/A |
| Variant type [2] | SNV, INDEL, SV [3] | SNV, INDEL, CNV ( >3 consecutive exons ) | SNV, INDEL, CNV [4] |
| TAT | 4 weeks | 4 weeks | 2 weeks |
| Depth-of coverage | ~30x | ~100x | Available for: • 3billion 3B-EXOME/3B-GENOME tests • External test results with known variant positions |
| Secondary finding | ○ | ○ | |
| Reanalysis | Included | Included | |

[1] Only SNV/INDEL (>10% heteroplasmic level) are called within the mitochondrial genome.
[2] Only low-quality variants are confirmed by Sanger sequencing. Whole exome sequencing covers all variants, while whole genome sequencing includes SNV and INDEL.
[3] Genome SV: variants with exact breakpoints identified are confirmed by Sanger sequencing.
[4] For CNVs, 3B-VARIANT is only available for variants with exact breakpoints identified.

**CAP #** 8750906
AU-ID: 2052626

**CLIA ID #** 99D2274041

## Acceptable sample types

| Type | Whole Blood | Extracted gDNA [2] | Dried Blood Spot Card | Buccal Swab [3] |
|---|---|---|---|---|
| Collection | Min. 1mL of blood in an EDTA tube | **Exome:** Min. 2ug of intact gDNA **Genome:** Min. 5ug of intact gDNA | Collect with 3billion DBS card | Collect with 3billion buccal swab kit |
| Shipping Conditions | Room temperature within 1 day [1] | Cold packs/dry ice in the package | Room temperature within 2 weeks | Room temperature within 1 week |

[1] If it is not shipped within a day, we recommend that you to include cold packs or dry ice in the box/package.
[2] Intact Genomic DNA (gDNA) must be extracted from a CLIA-certified laboratory or a laboratory appropriately qualified for extracting DNA for clinical testing.
[3] Only acceptable for 3B-EXOME and 3B-VARIANT.

## Results

Results are categorized into Positive, Inconclusive, and Negative.
A variant in a known disease gene that would fit the patient's phenotype is reported.

View Sample Report

### Positive




- **AD / XL**: 1 heterozygous or hemizygous P/LP variant
- **AR**: 1 homozygous or 2(compound heterozygous) P/LP variants

### Inconclusive

Automatic Reanalysis




- **AD / XL**: 1 VUS
- **AR**: ≥2 heterozygous or 1 homozygous VUS
- **AR**: 1 heterozygous P/LP variant
- **Gene of Uncertain Significance**: P/LP variant in a gene with emerging disease evidence

### Negative

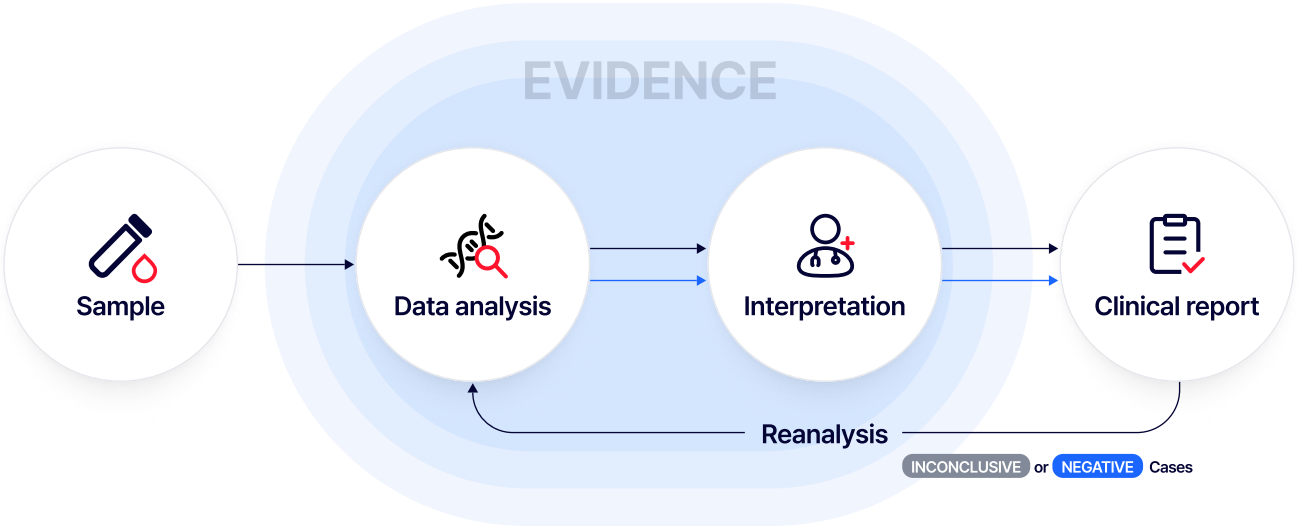
Automatic Reanalysis




- No clinically significant variant consistent with the phenotype identified

# One-stop Service for Rare Disease Diagnosis

Experience our all-inclusive rare disease diagnostic service – a single platform connecting sample, sequencing data analysis, interpretation, and clinical report. Interpretation of genomic data becomes faster and more accurate with 3billion's AI technology.




## How can AI improve diagnostic testing for rare diseases?

### Highly automated

- Streamlined variant calling workflow from FASTQ to annotated VCF
- Variant interpretation based on ACMG guidelines
- Daily DB update
- Similarity scoring between the symptoms of the patient and the disease.
- Variant pathogenicity prediction (3Cnet)
- Variant prioritization (3ASC)

**Improved efficiency**
with minimized resources

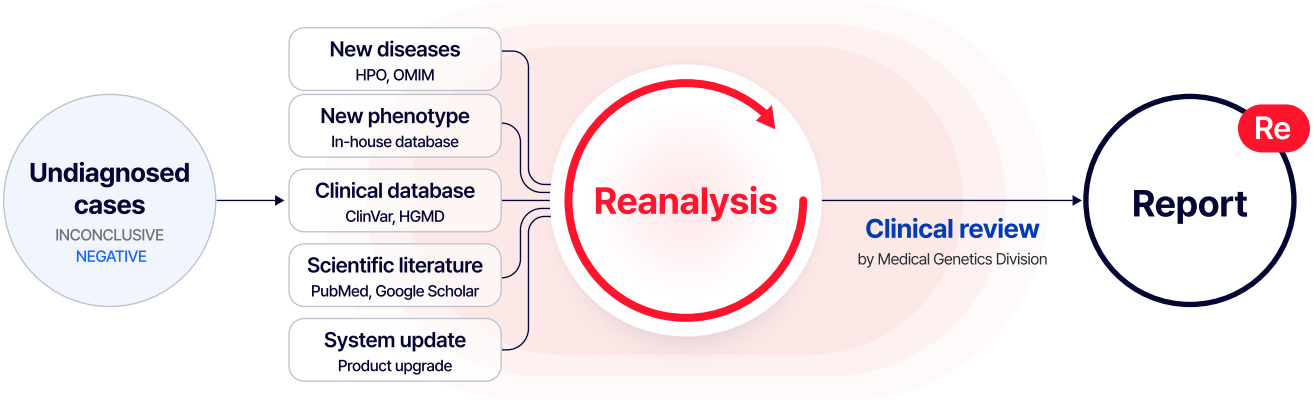
**Increased reliability**
with minimized human error

**Competitive price**

**Faster turnaround**

**Accurate diagnosis**

# Automated Reanalysis Until Diagnosis

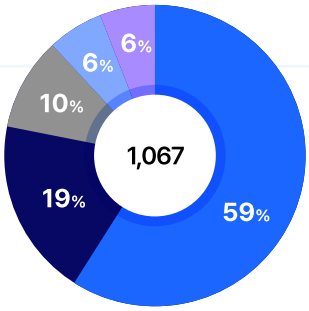



For undiagnosed patients, 3billion reanalyzes the sequencing data with updated database information. When a new molecular diagnosis is identified, an updated report is provided at no additional cost.

## 3billion's Reanalysis Insight

**With 1,067 successful reanalyses conducted between 2024 and 2025, 3billion's evolving system continues to bring more families closer to the answers they need.**

- Variant DB Update (59%)
- Additional Phenotype (10%)
- Pipeline Update (6%)
- Disease DB Update (19%)
- Segregation Analysis (6%)




## Real case of reanalysis

**Disease DB Update Case**

NEGATIVE → (2 Weeks) → INCON-CLUSIVE → (7 Weeks) → POSITIVE

Feb, 2021
No clinically significant variants identified

Mar, 2021
Article published
Am J Hum Genet. 2021;108(3):502-516

May, 2021
New disease updated
Radio-Tartaglia syndrome (OMIM 619312)

# Consult our Medical Genetics Division for Your Inquiries

*response within 48 hours

48hr

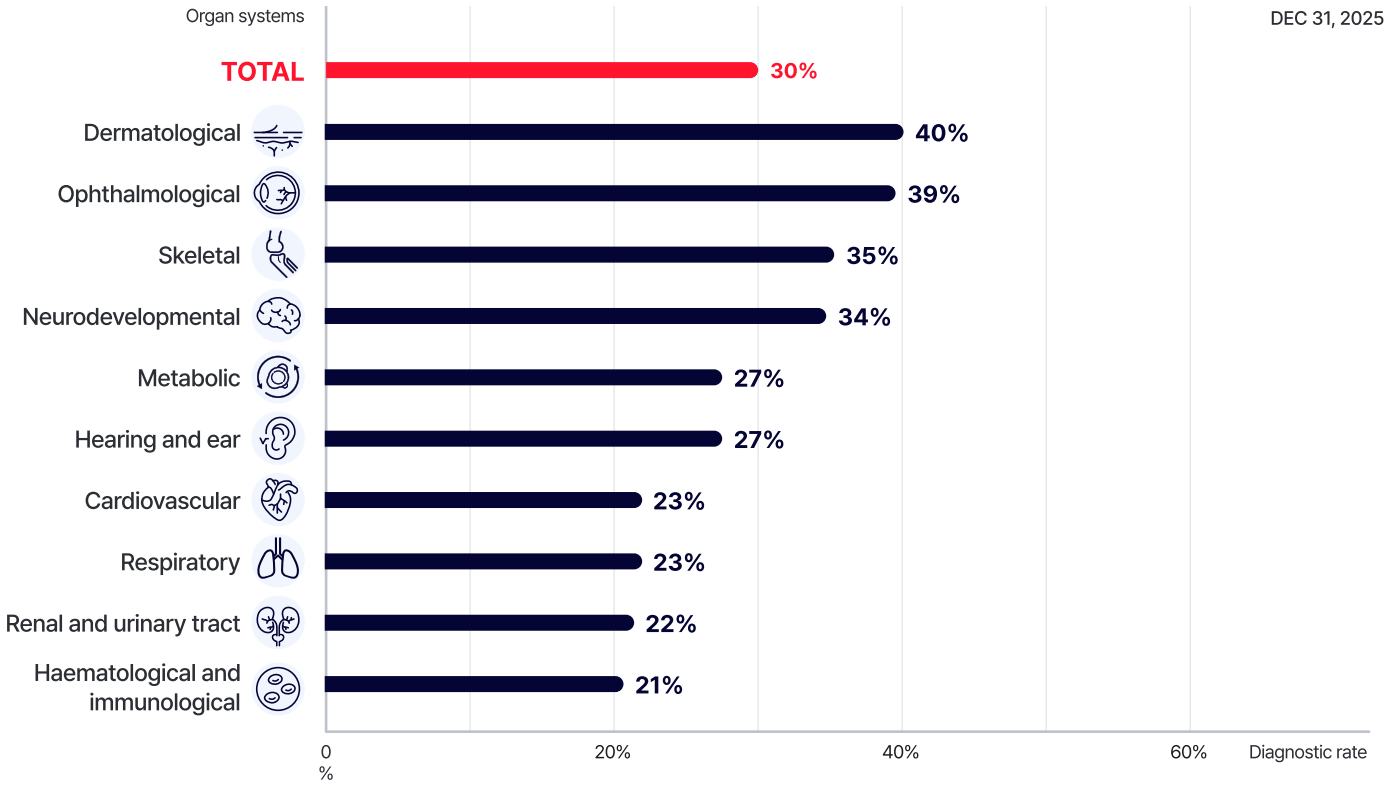
- Clinical consultation regarding the test results
- Case-based academic research collaboration
- National-level collaboration on rare diseases
- Collaboration with pharmaceutical companies

**Request a clinical consultation at** genetics@3billion.io

# Diagnostic Rate

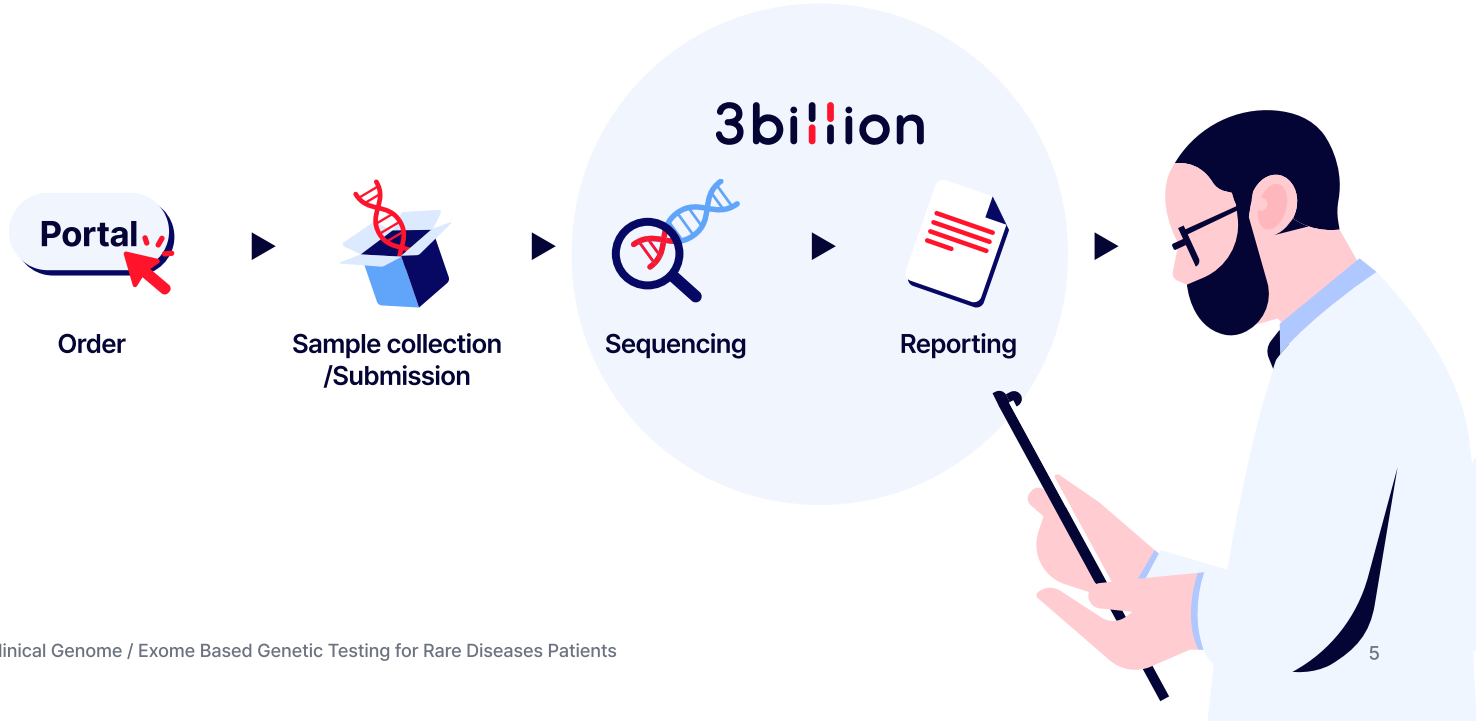
As of Dec. 2025, 30% of the patients have completed the diagnostic journey with us by receiving a clear molecular diagnosis. These patients were diagnosed with a wide spectrum of diseases.

The diagnostic rate for different organ systems are as follows:




# Genetic Testing in Practice

Physicians are placing orders and receiving results through our user-friendly online portal. On the portal, you can place an order, check the status of the test, and view the results. You can also share the results with your colleagues who have an account on our portal.



3billion

HEALTHCARE PROFESSIONALS
**2,200+**

INSTITUTIONS
**790+**

COUNTRIES
**70+**

# 1 MISSION

3billion is here to help physicians to diagnose and treat rare diseases through genetic testing.

Updated. Oct, 2026


**Web.** 3billion.io
**Order.** portal.3billion.io
**Email.** support@3billion.io
**Address.** 8th, 415 Teheran-ro, Gangnam-gu, Seoul, South Korea




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