

Your one answer

Diagnosis to Treatment for Rare Disease Patients

3billion

© 2025 3billion, Inc.

Global Innovator in Rare Disease Diagnosis

Founded in

2016

Countries

70+

Physicians

1600+

Cumulative Examination Patients

75,000+

Institutions

700+

Diagnostic Rate

32%

Mission

Diagnosis to Treatment for Rare Disease Patients

3billion entered the genetic testing market in 2016 to provide answers for patients with rare disease.

We are committed to helping patients and their families on their journey, starting with genetic testing and diagnosis.

We strive towards a world where rare disease patients are not neglected in diagnosis and treatment.



Company Timeline

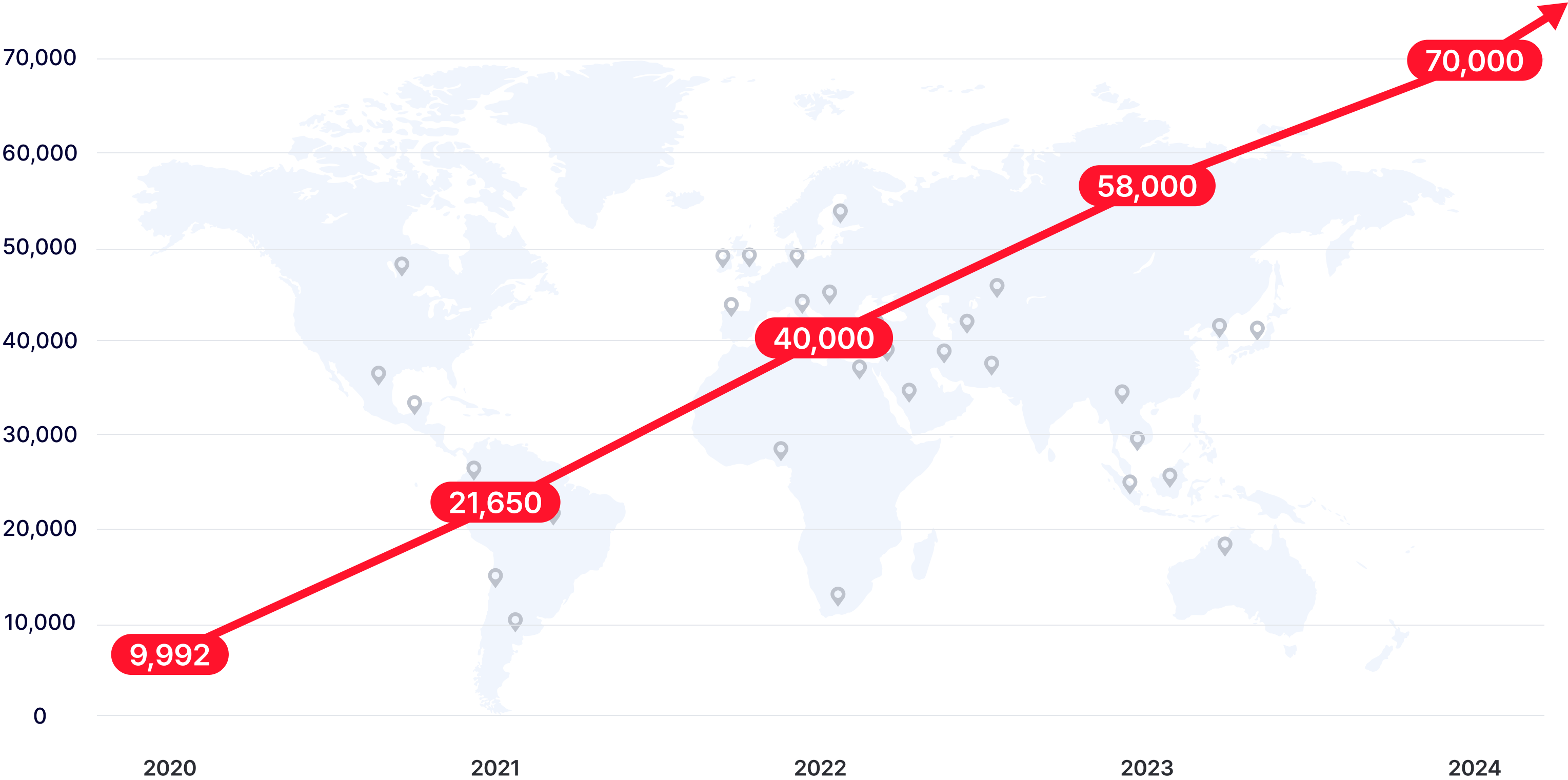
Continuous growth in the last 9 years

2016	Oct.	Founded 3billion			
2017	Mar.	Launched the beta version of Direct-to-Consumer (DTC) rare disease diagnostic service			
2018	Apr.	Participated in national project (NIPA) to establish an AI-driven diagnostic system for pediatric rare diseases			
	Jun.	Raised Series A funding			
2019	Feb.	Launched <3B-EXOME>, a WES-based rare disease diagnostic test			
	Oct.	Raised Series B funding			
2020	Feb.	Established facilities for next-generation sequencing analysis			
2021	Mar.	Raised Series C funding			
	Apr.	Participated in the national Bio Big Data project			
	Dec.	Obtained CAP certification			
2022	Apr.	Raised Pre-IPO funding			
	Jun.	Launched <3B-GENOME>, a WGS-based rare disease diagnostic test			
	Dec.	Obtained CLIA certification			
2023	Apr.	Obtained CDPH Laboratory License			
	Dec.	Data-Based Genetic Testing <3B-INTERPRETER> Launched			
2024	Jul.	Participation in non-metropolitan genetic diagnosis support project (KR-RDSP)			
	Nov.	Listed on KOSDAQ			

Rapid Growth of Genomic Database

Since 2020, we have accumulated over 70,000 genomic data entries

Cumulative number of data



National Project

Contributing to the public by participating in national projects



Development of intelligent S/W that connects and integrates medical data on 8 major disease areas

- Development of web-based variant diagnostic S/W for patients with hereditary hearing loss and developmental delay
- Selected as Top 12 out of 70,000 national projects in 2020



AI solution for the diagnosis, treatment, and management of pediatric rare disease patients

- Selected as the developer for AI S/W solution for pediatric rare disease patients
- Currently in the development and certification process for diagnostic S/W



Establishing a National Genomic Database to predict and diagnose population-specific diseases

- Analysis of 10,000 patients' genomic data (WGS) to produce diagnostic reference reports



The Korea Centers for Disease Control and Prevention's project to enhance rare disease prognosis through early diagnosis and timely treatment in non-metropolitan areas

- Conduct full-genome tests for suspected rare disease patients in non-metropolitan areas to aid early diagnosis.

Advancing knowledge of rare disease through continuous research

111 +

38 +



5,800 +



NIM_024666_S(AAGAG)-c.481C>T (p.Arg161Ter)		Cite this record	Cite this record	Cite this record		
Interpretation:	Pathogenic					
Review status:	☆ ☆ ☆ ☆ criteria provided, single submitter					
Submissions:	5					
First in ClinVar:	Sep 29, 2014					
Most recent Submission:	Jan 15, 2022					
Last evaluated:	Jan 15, 2022					
Accession:	NV000009732.4					
Variants ID:	29732					
Description:	single nucleotide variant					
Variant details						
Conditions	NM_024666.5(AAGAG)(c.481C>T (p.Arg161Ter))					
Disease(s)	Ablau ID: Variant type: Variant length: Oncogenic location: Genomic location:	48331 single nucleotide variant 1 bp 15q23 15: 47,237,868 (GRCh38) GRCh38 UCSC 15: 47,524,204 (GRCh37) GRCh37 UCSC	Molecular consequences: non-sense non-sense non-sense	HE HE		
HGVG:	Nucleotide Protein	NC_024666.5.g.481C>T NP_079942.3.p.Arg161Ter	HE HE	HE HE		
Protein change:	R161P, R62*	NP_021268.1.p.L164V NP_021268.1.p.L164V NP_021268.1.p.L164V				
Other names:	NG_000015.10:g.7231987.G.A					
Clinical significance:	-					
Functional consequence:	-					
Global minor allele frequency (gnAF):	-					
Allele frequency:	Terra-Genics for Precision Medicine (TPMPM) 0.00000 Exome Aggregation Consortium (EXAC) 0.00001 Terra-Genics for Precision Medicine (TPMPM) 0.00001 The Genome Aggregation Database (gnAD), exomes 0.00001					
Links:	ClinGen: CA762708 OMIM: 614868.001 dbSNP: c.74803412 VUSome					
Comment on variant:	ClinGen panel contributed the HVG's expression for this variant.					
Submitted interpretations and evidence						
Interpretation (Last updated):	Review status (Criteria provided):	Condition (MIM#):	More information	HE HE HE		
Pathogenic (Jan 15, 2022)	criteria provided, single submitter	Panethelium leucodermatide, psoriasis	Boltun Kowalenko SC000000039.1 From Simon, Jan 15, 2022 (last updated: Jan 15, 2022)	Publications PMID:33007148 Step-gated conversion predicted to increase or decrease protein function through non-sense mediated decay (NSD) or protein truncation. (allele pathogenic variants are -)	allele: predicted to increase or normal high non-sense NSD or protein pathogenic variants are -	allele: predicted to increase or normal high non-sense NSD or protein pathogenic variants are -

Awards and Recognition

Establishing diagnostic capabilities through global competitions

2017

Nov.
Selected as finalist for Fx2017 Startup Award

Jul.
Selected as finalist for Roche's Future X Healthcare 2017 Startup Award
Won the Korean representative selection for the 1776 Challenge Cup global startup competition

Jan.
Won first place at IPMC Precision Medicine Startup Competition

2020

Sep.
Won the Next Unicorn award at ASEAN-Korea Scale Up Competition



2021

Dec.
Selected as tech innovator by Novartis' 2nd Health X-Challenge Seoul



2022

Nov.
Selected as Korean Society of Medical Genetics and Genomics Autumn Conference (Individual) Best Research Award: Excellent Poster Award

Oct.
ASHG Poster selected for Reviewers' Choice

Aug.
Selected as a Top Tech Company of 2022 by Global Data Analysis Company Tracxn (2 consecutive years)

May.
Won in CAGI6, a global AI genomics analysis competition

2024

Nov.
Korea Wins 'Excellent Award' for Venture-Startup Patent



Global Partnerships

Working with over 700 institutions in over 70 countries worldwide



Business Area

We collaborate throughout the entire journey, from the diagnosis of rare diseases to the development of drugs.

3billion provides NGS-based diagnostic services and provides custom diagnostics to pharmaceutical companies to increase patients' access to treatment.

3billion also develops and provides an AI platform for the development of new rare disease therapies.

Diagnostic
Testing Services



Genetic Testing
Support Programs
with Pharma



AI-Driven
Drug Discovery



Diagnostic Testing Services

Identify the causative variants of a patient's symptoms with 3billion, using next generation sequencing (NGS)

3billion continues to improve the accuracy of diagnosis by leveraging its diagnostic and AI technology.

3billion also provides lifetime reanalysis at no additional costs to reflect the latest research.



Services

Various coverage options, all for rare disease diagnoses

3B-GENOME

Diagnosis based on **Whole genome sequencing**

Search for disease-causing variants in the entire human genome

3B-EXOME

Diagnosis based on **Whole exome sequencing**

Identify disease-causing variants in exon regions of over 20,000 genes and adjacent regions

3B-VARIANT

Diagnosis based on **Sanger sequencing**

Confirm specific variants found in 3B-EXOME / 3B-GENOME testing and for family testing

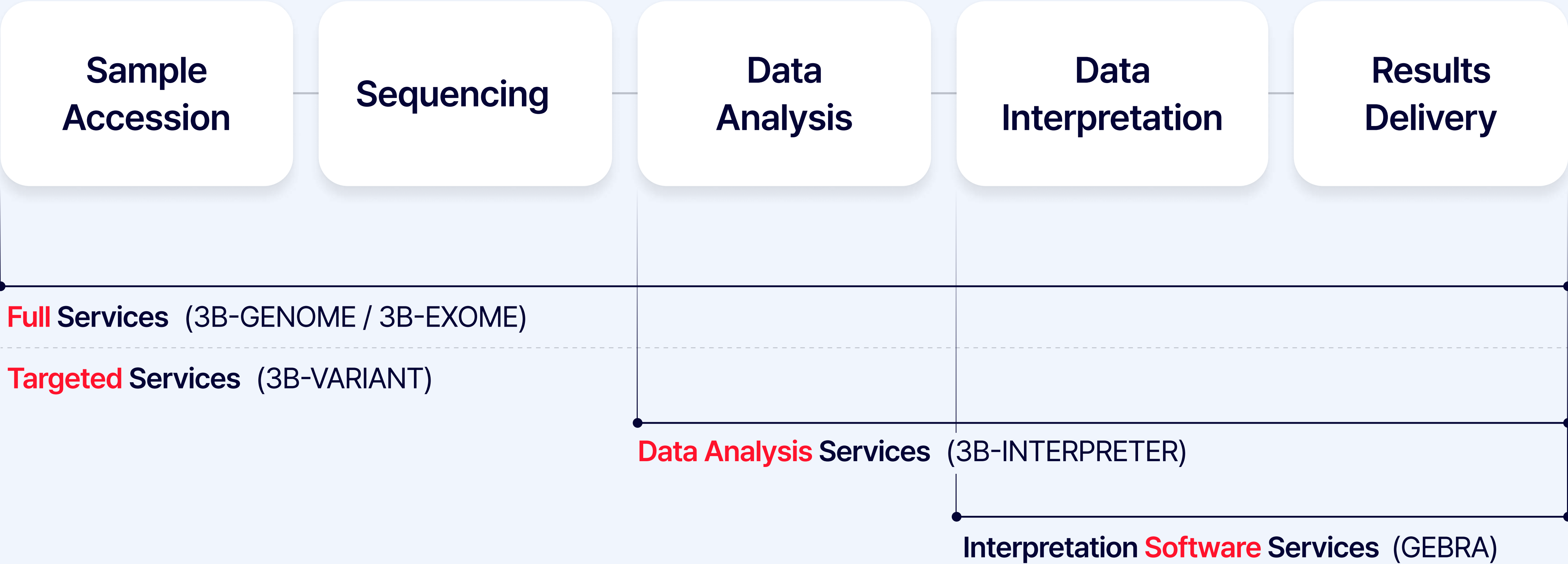
3B-INTERPRETER

Diagnosis based on **FASTQ, VCF Data**

Genomic data analysis and interpretation service

A Wide Range of Services

End-to-end services for rare disease diagnosis



Advanced Diagnostic Technology

Proprietary data analysis technology for diagnosis using AI and big data

EVIDENCE

Examines vast amounts of variant data to prioritize variants of interest

EVIDENCE reduces potential diagnostic biases and the time needed for analysis.

- Daily update of variant database
- Interpretation and classification of 100,000 variants within 1 minute
- Utilization of proprietary genomic database

3Cnet

An AI-based tool for predicting variant pathogenicity

3Cnet predicts the pathogenicity of detected variants by learning from various types of data.

- High sensitivity with a 99.99% accuracy in predicting pathogenicity of variants
- Algorithm minimizes bias through learning from multiple data sets

3ASC

Assists clinical geneticists in diagnosis

Using the information extracted by EVIDENCE, clinical geneticists select final candidates for causative variants associated with symptoms.

- Determines artifacts (data errors due to equipment)
- Includes causative variants within the top 5 results with a 96% success rate

Reliable Laboratory

Internationally certified laboratory infrastructure, protocols in line with global standards



Accreditation by College of American Pathologists
CAP License # 8750906,
AU-ID# 2052626



Certification by Clinical Laboratory Improvement
Amendments
CLIA ID # 99D2274041



International Information Security Management
System (ISMS) Standard Certification
ISO 27001:2022



ISO/IEC 27001:2022



CDPH(California Department of Public Health)
Clinical and Public Health Laboratory License



Genetic Testing Support Programs with Pharma

**Identify the most relevant rare disease
patient population with 3billion**

Since orphan drugs have a small patient population who have difficulties with diagnosis, it can be challenging to recruit patients.

Find the right patients for the right treatment with 3billion.



Service

Helping you find patients who will benefit most from treatment



Collaboration with Global Pharma

Offering customized genetic testing for certain rare diseases

Support Program for Lysosomal Storage Disorders

3billion is providing accessing to genetic testing to patients suspected with Lysosomal Storage Disorders in collaboration with a global pharmaceutical company.



Support Program for Inherited Retinal Disorders

3billion is providing a joint genetic testing program for patients with inherited retinal disorder with a global pharmaceutical company.



Support Program for Atypical Cerebral Palsy

3billion is providing access to genetic testing for patients with suspected Atypical Cerebral Palsy in collaboration with a global pharmaceutical company.



Support Program for Dysplasia & Seizure

3billion is providing access to genetic testing for patients with suspected Skeletal Dysplasia and Seizure in collaboration with a global pharmaceutical company.



Partner Benefits

A cooperative structure that benefits each stakeholder group

Pharmaceutical Company

Expand access to treatment



Healthcare Provider

Propose genetic testing to patients at
no additional cost



Patient

Receive financial support for genetic
testing and quickly identify the
cause of disease



AI-Driven Drug Discovery

Support pharma in developing new orphan drugs using MIN-T, an AI-based drug development technology

3billion's drug discovery technology meets various demands of pharmaceutical companies, such as target identification, target validation, hit-to-lead, and experimental validation, aiding in new drug development.

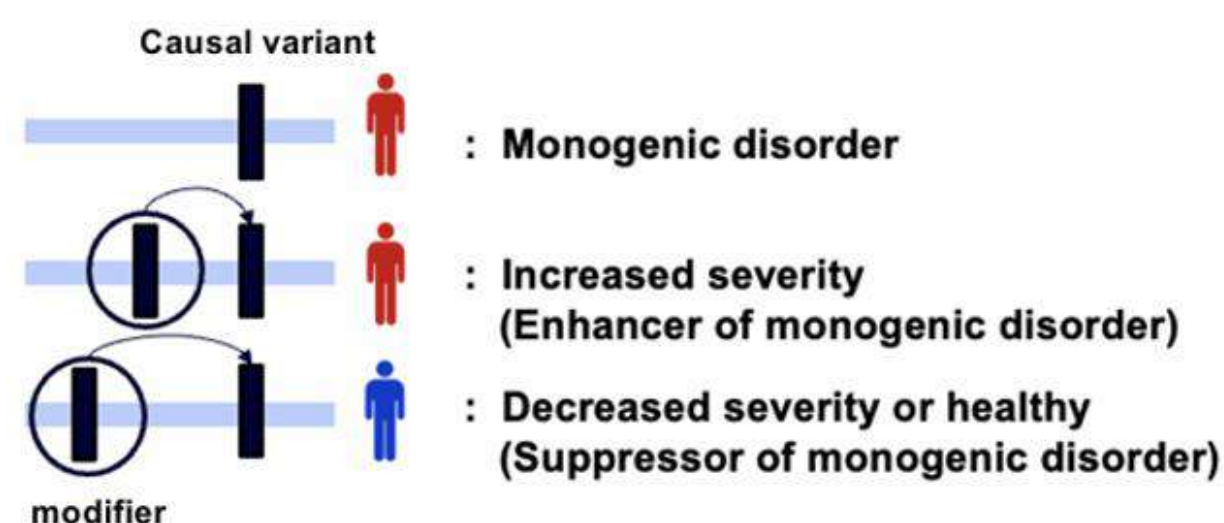


Service

Utilizing patient genomic data to discover novel targets and pockets for small molecules so that we can treat the diseases known to be undruggable target

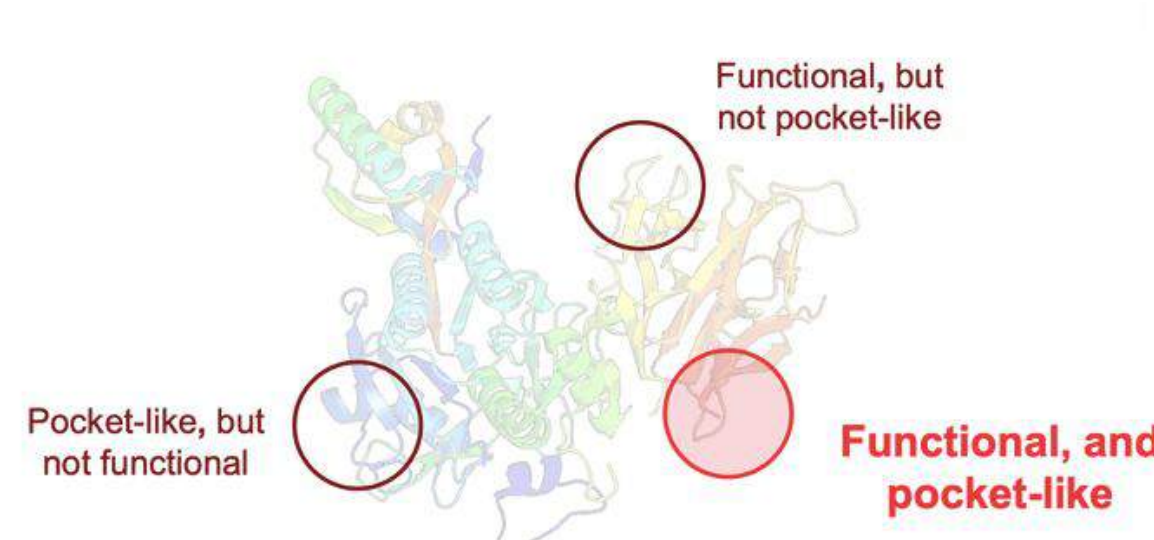
Target Discovery

- Finding modifier gene for the disease using WES/WGS patient data
- Finding unknown GoF mechanism diseases which could be promising targets for SM



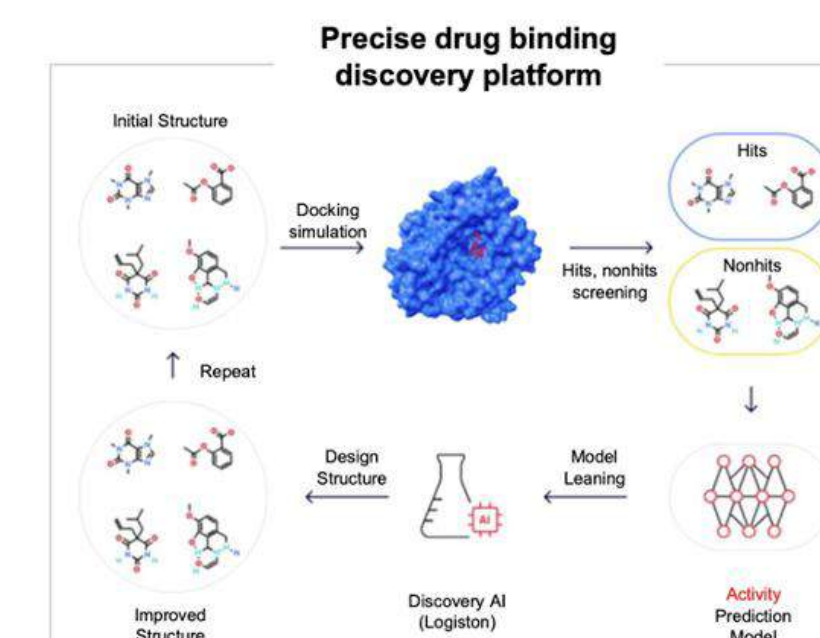
Pocket Discovery

- Find effective pockets even without references based on the variant interpretation
- The active site indicates the site of the protein target which is both functional and pocket-like.



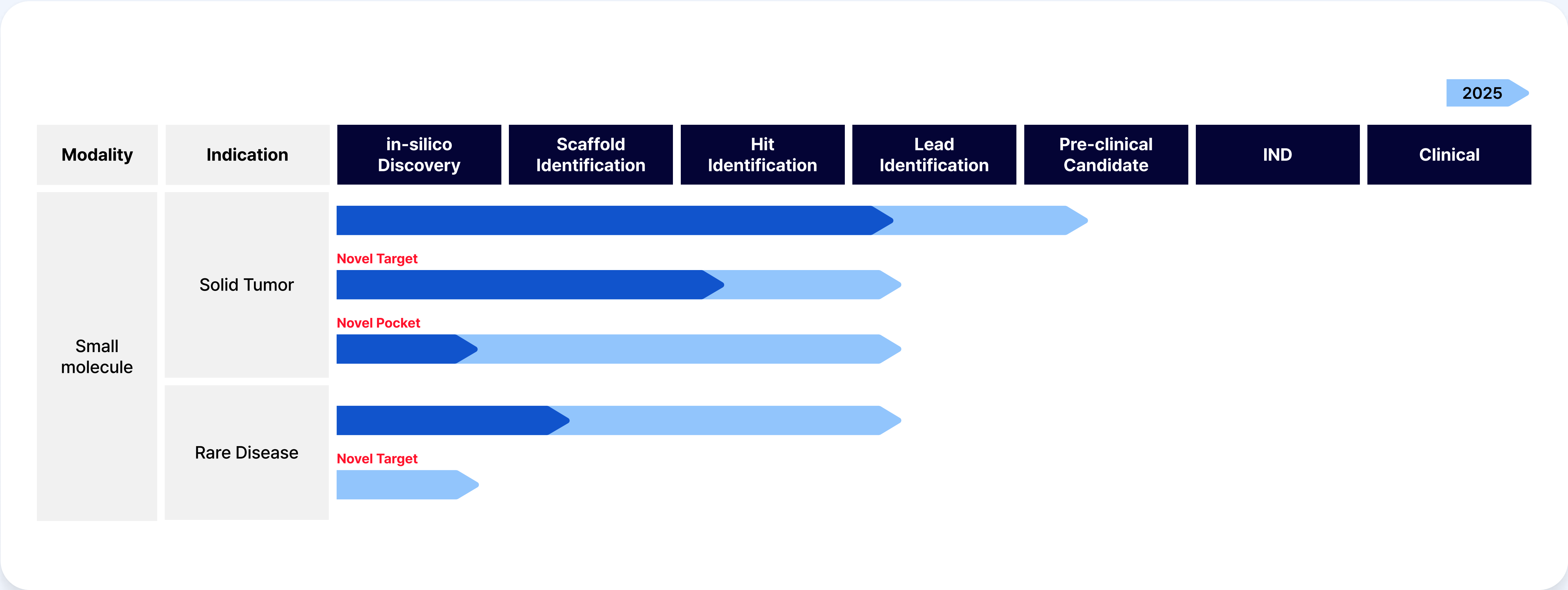
Active Compound Design

- Reaction based novel compound synthesis and active compound design methods
- Structurally novel but synthesizable compounds generated by AI



Drug Development Platform Pipeline

We are utilizing AI to discover leads, currently in the validation stage
In 2025, we aim to expand into lead validation for rare disease drug development



Partner Benefits

A drug discovery structure that benefits all participating groups

Pharmaceutical Company

Significantly reduce time to
discovery and costs for drug
development



Healthcare Provider

Increase the probability of finding
optimal treatment for patients



Patient

Improve chances of accessing
treatment more quickly through
shorter drug development cycles



We strive to provide end-to-end solution through our Genomic data platform for patients through diagnosis, treatment, prevention

3billion is changing the rare disease diagnostic ecosystem through technology and our passion.

We hope to change the world by applying AI to genomic data to provide the best-in-class diagnostic technology and development of rare disease treatment.

We will create value throughout the patient's entire journey, including diagnosis, treatment, and prevention, with our genomic platform.



Web. 3billion.io

Order. portal.3billion.io

Email. support@3billion.io

Address. 8th, 415 Teheran-ro, Gangnam-gu, Seoul, South Korea



Last Updated 2025.01.

© 2025 3billion, Inc.