CENTOGENE
GUIDING PRECISION MEDICINE

PHARMA SERVICES

Real-world data for rare & genetic disease — accelerating drug discovery & clinical development



Translational research, patient identification, biomarkers / real-world data — and clinical trial support.

For Translational Research teams | July 2026 | CENTOGENE Japan K.K.

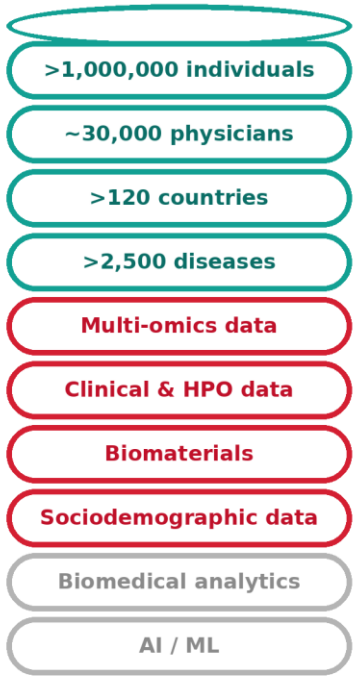
COMPANY OVERVIEW

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Accelerating drug discovery with real-world data in rare & genetic disease

CENTOGENE is a global genetic diagnostics and data company focused on rare, genetic and neurological diseases. Built on one of the world's largest Biodatabanks, we accelerate and de-risk the journey from drug discovery to patient access.

CENTOGENE Biodatabank



Real-world data resource for rare & neurodegenerative disease

One of the world's largest data resources

An integrated, multi-omic real-world repository in rare & neurodegenerative disease — continuously expanded through both diagnostics and pharma partnerships.

Broadest testing portfolio

From WES/WGS and multi-omics to NGS panels — comprehensive genetic testing for rare disease that drives high diagnostic yield.

End-to-end support

From target assessment, biomarkers and patient identification to market access — accelerating and de-risking discovery, development and commercialization.

DISEASE FOCUS

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Priority disease areas & flagship programs

Anchored in rare, genetic and neurological disease, with data accrued through real global programs — covering ~2,500 diseases.

Neurology & neurodegeneration

- **Parkinson's disease** ROPAD study (~15,000 pts / GBA · LRRK2)
- **Frontotemporal dementia (FTD)** EFRONT study (>3,000 pts)
- **Hereditary ataxia** Friedreich's ataxia, and more

Lysosomal & metabolic disorders

- **Gaucher disease** LysoProof natural-history study (>250 pts)
- **Niemann-Pick type C (NPC)**
- **Mucopolysaccharidosis (MPS) / ASMD**

Neuromuscular & bone metabolism

- **Duchenne muscular dystrophy (DMD)**
- **Hypophosphatasia (HPP)** enzyme replacement therapy target

Amyloidosis & hematology

- **Hereditary ATTR amyloidosis (hATTR)** long-term study, 5,000 enrolled
- **Pyruvate kinase deficiency (PKD)** Phase II/III · 20 countries

These are representative examples. CENTOGENE covers ~2,500 rare & genetic diseases and accesses patient data through a ~30,000-physician network across 120 countries and one of the world's largest Biodatabanks.

* Disease names, programs and figures are illustrative, based on the v05 proposal and CENTOGENE-provided materials (not warranted as confirmed information).

VALUE FOR TRANSLATIONAL RESEARCH

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Accelerating translational research with genetically defined disease data

Target safety, biomarkers, patient identification — CENTOGENE answers TR's common challenges with deep data in rare / genetic / neurological disease.

1

Target safety (human genetics)

From ~200,000 exomes/genomes, assess early target safety (Therapeutic Risk) using phenotypes of human-KO / loss-of-function (LoF) carriers.

2

Biomarker discovery & validation

Identify and validate biomarkers/assays using patient-derived cell models and multi-omics (transcriptome / metabolome).

3

Patient ID & stratification

Identify rare and undiagnosed patients across a ~30,000-physician network in 120 countries and a 2,500-disease Biodatabank, then stratify by genotype/phenotype.

* Figures are based on CENTOGENE-provided materials. Specific applications are discussed case by case for the target disease/gene.

CLINICAL TRIAL SUPPORT

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Running trials and testing for patient enrollment

From identifying eligible patients to protocol-aligned genetic testing, enrollment and registry build-out — end-to-end clinical development support across countries.

1

Patient ID & stratification

Identify and stratify eligible patients via Biodatabank retrospective search and candidate cohorts.

→

2

Enrollment testing

Provide protocol-aligned genetic testing (NGS / WES / WGS / MLPA).

→

3

Site selection & enrollment

Support enrollment via a ~30,000-physician network across 120 countries and patient groups.

→

4

Trial conduct & registry

Accelerate development with ongoing molecular diagnostics and disease-registry build-out.

Clinical development & registry track record (examples)

Pyruvate kinase deficiency
Phase II/III: causal-variant identification & genetic testing · 20 countries

Hereditary ATTR amyloidosis
Long-term molecular diagnostics via NGS + MLPA · 5,000 enrolled

Parkinson's disease (ROPAD)
Genetic testing incl. LRRK2 · ~15,000 pts / positives are trial candidates

Frontotemporal dementia (EFRONT)
Enrollment & genetic testing of >3,000 pts · 7 countries

* Cases and figures are illustrative, based on the v05 proposal and CENTOGENE-provided materials (not warranted as confirmed information).

REAL WORLD INSIGHTS REPORTS

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Insights reports for decision-ready evidence

We analyze CENTOGENE's in-house clinical and genetic data and deliver purpose-built reports — from target assessment to patient selection and regulatory-grade evidence.

Therapeutic Risk

Assess early safety risk from phenotypes of human-KO / LoF carriers of a target gene

Variant Classification

ACMG-based classification for patient selection, stratification and regulatory evidence

Prevalence

Model global and regional prevalence from in-house variant observations

Patient Cohort

Describe and analyze diagnosed / candidate cohorts to assess trial feasibility

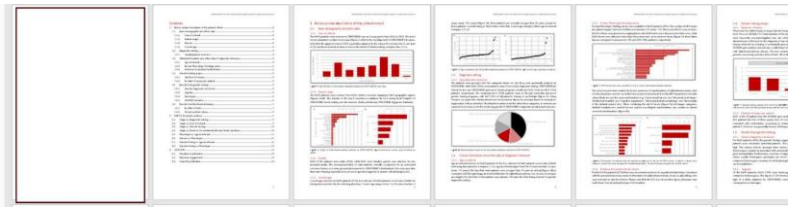
Mutational Spectrum

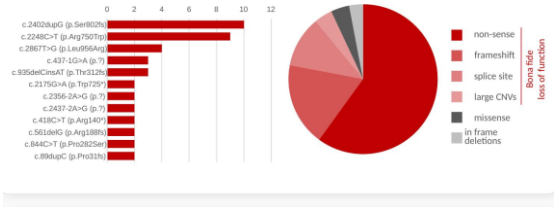
Present the spectrum of disease-associated genetic variants

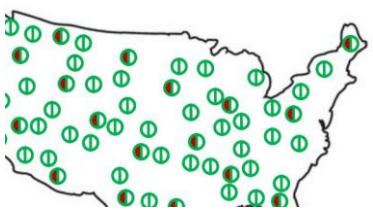
Data Licensing

Provide anonymized datasets and patient cohorts on third-party platforms

Sample report output
(illustrative data)







* Illustrative report formats based on example data ([gene]/[disease]). Actual outputs are produced per your request.

Contact: CENTOGENE Japan K.K.

Thank you.

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Guiding Precision Medicine in Clinical Diagnostics

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