Neurology
Service Specification Sheet

GenomeScan® Neuro-*GBA1*Screen

Transform your neuro research with precision-driven *GBA1* sequencing

Neurodegenerative diseases demand answers and the *GBA1* gene is at the center of some of the field's most transformative breakthroughs. As the strongest genetic contributor to Parkinson's disease (PD), *GBA1* plays a major role in pathways involving lysosomal dysfunction, α-synuclein accumulation, and cellular stress responses. At the same time, pathogenic *GBA1* variants are the established cause of Gaucher disease, making this gene a critical biomarker across both neurodegeneration and lysosomal storage disorders.

The *GBA1* gene has a highly homologous pseudogene, *GBAP1*, located ~16 kb downstream from the functional gene. This high sequence similarity, reaching up to 98% across coding regions, makes *GBA1* analysis intrinsically challenging. *GBAP1* can interfere with read mapping and variant detection, and recombination events between the two loci can generate hybrid alleles that standard sequencing workflows often miss. At GenomeScan, complexity becomes our competitive edge. We have engineered a next-generation *GBA1* workflow that cuts through pseudogene interference with precision, combining optimized laboratory protocols with advanced bioinformatic filtering to deliver clear, confident variant calls.

In this rapidly evolving landscape, Next Generation Sequencing based *GBA1* sequencing has become indispensable, powering diagnostic workflows, risk prediction models, and next generation therapeutic research. From precision medicine initiatives to clinical trial enrolment, scientists and clinicians rely on high-confidence variant detection to uncover disease mechanisms, stratify patient groups, and accelerate translational breakthroughs.

GenomeScan brings extensive expertise in *GBA1* sequencing, delivering the accuracy and depth your research demands. Our dedicated scientists provide reliable, high-resolution and meaningful results you can trust.

Whether you're advancing PD research, exploring lysosomal biology, validating biomarkers, or designing therapeutic studies, our *GBA1* sequencing services equip you with the data to move confidently from discovery to impact.

Input material

Samples

- Saliva (minimum of 1 mL required)
- Whole Blood (minimum of 1 mL required)

Isolated gDNA requirements

- Optimal total gDNA input: 100 ng of gDNA
- Minimal volume: 25 µL
- Minimal quality: OD260/280 ~1.8-2.0 and OD260/230 ~2.0-2.2

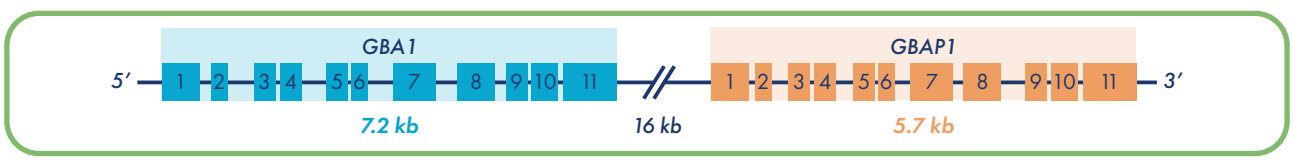
Raw data output

Sequencing on Illumina NovaSeq

- Standard sequencing depth 0.4 M paired-end reads/sample

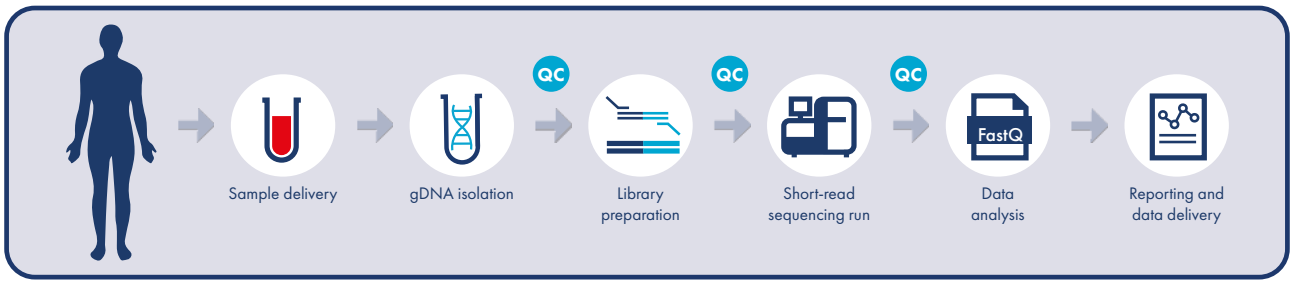
Deliverables

- FASTQ files
- Quality Score Q30 > 80%
- Optional data analysis with comprehensive report



GBA1 and *GBAP1* gene homology influence detection of SNVs, indels and recombinant alleles

GenomeScan Neuro-*GBA1*Screen Workflow



Our workflow incorporates library preparation, quality control steps, and short-read sequencing using Illumina platforms to enable accurate and reliable *GBA1* variant detection.

Biological Insights

GenomeScan Neuro-*GBA1*Screen provides deep biological insight into:

- **Lysosomal Function:** Identifies variants that reduce GCase activity and disrupt lysosomal homeostasis.
- **Neurodegeneration Pathways:** Reveals mutations linked to α-synuclein accumulation and increased PD risk.
- **Gaucher Disease Biology:** Defines pathogenic variants driving sphingolipid accumulation and disease severity.
- **Therapeutic Development:** Supports gene-therapy strategies aimed at restoring GCase function and reducing α-synuclein aggregation.
- **Clinical Trial Stratification:** Helps stratify patients based on severity of *GBA1* variants, guiding recruitment for targeted clinical trials in *GBA1*-associated PD.
- **Gene Therapy Trials:** Sequencing is essential to select and monitor participants in ongoing *GBA1* targeted gene therapy trials.

One Step Solution

GenomeScan may contribute during the whole process, from gDNA isolation to secondary data analysis. Our team will guide you through every step and advise on the best strategy for your project.

Get in touch with us to discuss your study design, request a quote, or explore how our Neuro-*GBA1*Screen solutions can support your research.

Data analysis

Our BioIT team has developed a data analysis workflow that provides high-quality, reproducible insights through a standardized and validated bioinformatics pipeline. Including:

- Alignment of sequencing reads to a *GBA1*-optimised reference genome.
- Variants reported in accordance with field-standard conventions, including exclusion of the 39 residue signal sequence.
- Variant detection and characterization across the *GBA1* gene, including identification of single nucleotide variants (SNVs), insertions and deletions.
- Comprehensive batch reporting, generated through our Mutation Analysis pipeline, supplemented with indepth annotation of all variants detected in the *GBA1* gene.

About GenomeScan

As an ISO-accredited leading Dutch Next Generation Sequencing service provider, GenomeScan develops customizable NGS solutions for pharmaceutical and biotech companies, healthcare providers and academic institutions. By providing state-of-the-art tools to analyze genetic disorders fast, affordably, and effectively, GenomeScan fosters innovation through partnership with medical centers and research laboratories.

Our services are designed to accelerate therapeutic innovation by delivering precise, reproducible, and regulatory-compliant genomic data. Whether supporting early-stage research or clinical development, GenomeScan empowers pharma partners with the tools and insights needed to advance personalized medicine and targeted therapies.

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