

Three tests on one knowledgebase calibrated on Indian population data, every one ordered by a doctor. This brochure sets out when to use each, what to write on the prescription, what the report contains, and the evidence behind it.

The tests and when to use each

Genavli Screen

With your doctor's prescription

For a healthy person: the clinical genes read at high depth, at lower cost than a whole genome.

- 775 core carrier genes · 600 conditions, reported per person
- Medically actionable findings (84 genes) and pharmacogenomics (96 gene–drug pairs)
- Ancestry and traits, imputed from the low-pass data
- Spinal muscular atrophy, thalassaemia and fragile X by bundled assays

Genavli Wellness Genome

With your doctor's prescription

For a healthy person, no symptoms needed: the whole genome, re-readable for life.

- 1,141 carrier genes · 947 conditions, plus novel variants
- Actionable findings and pharmacogenomics, including full CYP2D6
- Ancestry and traits observed, not imputed
- Polygenic scores, research-grade and labelled, calibrated against an Indian reference

Genavli Diagnostic Genome

Doctors and laboratories only

For a patient with a suspected rare or genetic disorder.

- Every variant ranked against the patient's clinical features (HPO terms)
- Structural variants, copy number, repeat expansions and hard genes such as SMN1 and CYP21A2
- Actionable findings unless the patient opts out
- Optional carrier screening (1,141 genes · 947 conditions) and pharmacogenomics

Couple option (Screen or Wellness Genome)

- Both partners take the same test: Genavli Screen for both, or the Wellness Genome for both, never mixed.
- Each partner has their own doctor's prescription, blood sample and consent, and agrees to share carrier results for the joint report: conditions where both carry a disease-causing variant in the same recessive gene, and X-linked conditions the female partner carries, with the chance for each pregnancy and the residual risk after a negative result.
- Pre-conception only. No prenatal testing.

Newborn screening

- Ordered by the baby's doctor, a paediatrician or neonatologist, with the parents' consent, from a blood sample.
- Looks for conditions that begin in infancy or childhood and can be treated or managed early, from a newborn-specific gene and variant list with its own report; conditions that begin only in adulthood are not reported for a child.
- Done on the screening backbone (a low-pass genome with high-depth enrichment of the clinical genes) or on the whole genome (30×).

Every figure, with its counting rule

600–947

conditions screened

Distinct conditions, deduplicated by disease identifier; ClinGen Definitive or Strong validity plus 14 society-recommended genes. Genavli Screen to the whole-genome tests.

775–1,141

carrier genes on the panel

Autosomal-recessive and X-linked genes on the reported panel. Core set on Genavli Screen; core plus expanded set on the Wellness and Diagnostic Genomes.

84

medically actionable genes

ACMG SF v3.3, reported at individual level under separate consent. Not part of the carrier count.

96

pharmacogenomic gene–drug pairs

CPIC Level A. Expected diplotype resolution modelled on South Asian allele frequencies, not measured sensitivity.

What to write on the prescription

For Genavli Screen and the Wellness Genome a person can register and order themselves, but only with your written prescription. On your official prescription pad, write:

1. The patient's name
 2. The test name, for example "Genavli Wellness Genome" or "Genavli Screen" (for the couple option, each partner needs their own prescription)
 3. The indication, if there is one
 4. Your name, your medical council and registration number
 5. Your signature and the date

What happens next

- The person uploads a photo or scan of the prescription at registration, with your name, registration number, address, contact number and email.
- Genavli staff check your registration in the medical council register and phone you to confirm before the order is accepted.
- You receive the results too; the person is responsible for discussing them with you.
- The Diagnostic Genome and newborn screening are ordered by you or your laboratory directly.

Counselling and follow-up

A session is included at no extra charge when the report has a medically actionable finding, a carrier result relevant to having children, or a positive Diagnostic Genome result; for any other result a session can be booked on demand for an additional fee.

What you are expected to discuss

- The result itself: you receive the report, and the person is responsible for discussing it with you.
- The next step each finding leads to, which the report names per area (below).
- That a screening result is not a diagnosis: a negative result reduces but does not eliminate risk, and residual risk is stated per condition.
- For a carrier result relevant to having children: the partner's testing, through the couple option.

Each area of the report and where a finding leads

Reproductive and carrier risk	Partner testing, pre-conception counselling, prenatal diagnosis through a licensed centre.
Heart and circulation	Cardiology referral, lipid management, cascade testing of relatives.
Metabolic and kidney	Endocrinology or nephrology referral, treatment choice in MODY, family screening.
Cancer predisposition	Oncogenetics referral, surveillance schedule, risk-reducing options, cascade testing.
Medicines and dosing	Prescribing guidance to the treating physician; pharmacogenomic card for the patient.
Neurological and neuromuscular	Neurology referral, orthogonal repeat sizing (RP-PCR or Southern blot), family counselling.
Ancestry and traits	Lifestyle discussion; no clinical action is implied.

What the report contains

Report modules

Rare-disease diagnosis. On the Diagnostic Genome: every variant ranked against the patient's clinical features (HPO terms) for a diagnosis. Newborn screening, ordered by the baby's doctor, runs on the whole genome or on the screening backbone.

Medically actionable findings. The 84 ACMG SF v3.3 genes, reported at individual level under separate consent, with a stated management pathway.

Consanguinity and residual risk. Runs of homozygosity measured from the assay, not assumed from a pedigree, feeding consanguinity-adjusted risk.

Reanalysis and existing data. Whole genomes are stored and can be reinterpreted when the knowledgebase changes, without a new sample. Existing data (VCF, BAM, CRAM or FASTQ) can be brought for interpretation: wellness-level results when an individual registers it with their own doctor's prescription, diagnostic-level when a doctor or laboratory orders it.

Detection status per variant class

Stated in every report: observed, imputed, covered by a bundled assay, partial or not assessed.

Variant class	Screen	Wellness Genome	Diagnostic Genome
Rare pathogenic carrier variants	Observed	Observed	Observed
Novel / previously unreported variants	Not assessed	Observed	Observed
Ancestry and traits	Imputed	Observed	Not offered
Polygenic scores	Not offered	Research-grade, Indian calibration	Not offered
Spinal muscular atrophy (SMN1)	Bundled assay	Paralogue-aware caller	Paralogue-aware caller
α- and β-thalassaemia	Bundled assay	Sequencing + targeted caller	Sequencing + targeted caller
Fragile X (FMR1 CGG repeat)	Bundled assay	Bundled assay	Bundled assay
CYP2D6 full diplotype	Partial	Full, incl. deletions, duplications, hybrids	Full, when pharmacogenomics is added
Structural variants, inversions, translocations	Not assessed	Observed	Observed
Reanalysis without a new sample	Limited to captured content	Full genome reinterpretable	Full genome reinterpretable

Where a test cannot see something, we say so

Every report carries per-section limitation statements naming what was not assessed and how it can be covered. No screening test detects everything; a report implying otherwise is the more dangerous product.

Carrier screening. Per-person carrier status for autosomal-recessive and X-linked conditions, with residual risk after a negative result, and a joint report when both partners of a couple are tested.

Pharmacogenomics. 96 CPIC Level A gene–drug pairs with the diplotype, the phenotype and the prescribing guidance that follows from it.

Ancestry, traits and polygenic scores. Wellness report, separate from the clinical report: ancestry composition and research-grade traits with Indian reference percentiles. On the Wellness Genome only, polygenic scores calibrated against an Indian reference, labelled research-grade.

No polygenic scores on clinical reports. Never on a Diagnostic Genome report. Scores derived from European cohorts do not transfer reliably; on the Wellness Genome they are calibrated against an Indian reference and labelled research-grade.

Pre-conception and postnatal use only. No prenatal diagnosis, no embryo testing, no pre-implantation testing; fetal sex is never determined.

Screening backbone or whole genome

Genavli Screen runs on the screening backbone; the Wellness and Diagnostic Genomes run on the whole genome. Couple and newborn screening can use either, and the choice decides what the report can see.

Screening backbone

Low-pass genome plus high-depth targeted enrichment of the clinical genes — Genavli Screen

Advantages

- Costs less than a whole genome.
- Reads the clinical genes at high depth, so rare carrier and actionable variants in them are observed directly, never inferred.
- Bundled confirmatory assays cover spinal muscular atrophy, thalassaemia and fragile X.
- Ancestry and traits come from the low-pass data, without a second test.

Limitations

- Only the targeted genes are read at clinical depth. Variants outside the targets (other genes, most non-coding and deep intronic changes, many structural variants and repeat expansions) are not assessed.
- Novel or previously unreported variants are not assessed; ancestry and traits are imputed, not observed.
- CYP2D6 is only partly resolved.
- Later reanalysis is limited to what was captured.

Whole genome

30× whole-genome sequencing — Wellness and Diagnostic Genomes

Advantages

- Reads all genes and the regions between them, so novel variants are observed and nothing has to be anticipated in advance.
- Finds structural variants, exon-level copy number, repeat expansions and the mitochondrial genome.
- Specialised callers resolve SMN1, thalassaemia, CYP21A2, GBA1 and full CYP2D6 without add-on assays.
- Can be reanalysed for new genes as knowledge grows, without a new sample.

Limitations

- Costs more and generates more data.
- Produces more findings that need careful interpretation.
- Fragile X still needs a bundled assay; repeat expansions longer than a read are reported as unconfirmed until orthogonally tested.

Evidence

- ACMG/AMP classification with ClinGen specifications: expert-panel criteria where they exist; every call carries its evidence.
- Indian population frequencies: GenomeIndia allele frequencies with gnomAD South Asian data, thresholds recalibrated.
- NABL-accredited, ISO 15189 partner laboratory: Genavli interprets; an accredited laboratory sequences and confirms.
- Analytical validation published at genavli.in/validation: variant calling and imputation on GIAB and South Asian reference genomes, by depth and allele frequency.
- Counting rules published at genavli.in/tests: every figure says how it was counted.

Contact

Questions about a patient, a prescription or a laboratory partnership: write to us.

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