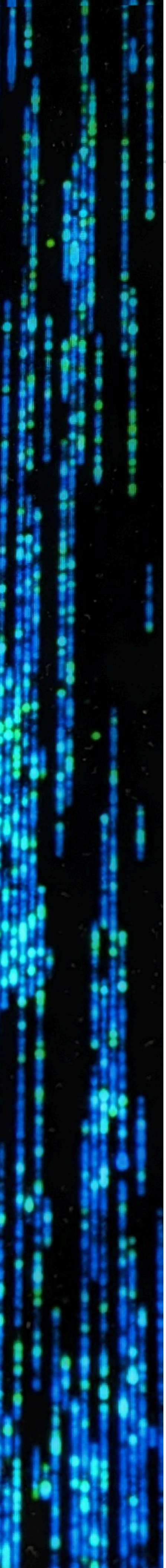

PRAXIS GENOMICS

Upgradeable Testing Architecture

If a simpler test does not yield a definitive diagnosis, any test can be upgraded to the next tier without requesting a new sample. The patient pays only the difference in test price at the time of upgrade, saving both time and the inconvenience of repeat sample collection. This upgradeable framework applies across constitutional diagnostic, carrier, and somatic tests. **The flowchart below uses blood as the example sample type**; some sample types (saliva, buccal swab, FFPE, etc.) support a narrower range of upgrades, and OGM and Transcriptome have additional sample requirements (see notes below).

> **Note — Transcriptome (SRT) sample requirements:** *When blood is the starting sample, the* **only accepted source for Transcriptome is a Tempus™ tube**. *Other sample types (frozen or FFPE tissue from a tissue where the gene of interest is expressed) are also acceptable. If a transcriptome upgrade may be needed, please submit a Tempus™ tube alongside the primary blood draw — Praxis will store it at no charge pending an upgrade request. Contact us at* **support@praxisgenomics.com** *if you are unsure what acceptable material would be for your gene(s) of interest.*

Microarray Equivalent / NIPT Confirmation Testing (SRG801)

- Aneuploidy detection and CNV calling with 10 kb resolution

↓ *Upgradeable to Basic Exome, Expanded Exome, WGS, or any combined PRX test*

Basic Exome (SRG501–504)

- All known pathogenic variants anywhere in the genome (as classified in ClinVar and other curated databases)
- SNV and small indels in coding regions +/− 100 bp upstream and downstream of genes and +/- 20 bp upstream and downstream of exons
- Single exon resolution CNV calling irrespective of exon size
- Mitochondrial genome variants (SNVs, deletions)

↓ *Upgradeable to Expanded Exome, WGS, or any combined PRX test*

Expanded Exome (SRG507–510)

- All the above plus repeat expansion assessment for all known repeat expansion associated conditions

↓ *Upgradeable to WGS or any combined PRX test*

Whole Genome Sequencing — short read (SRG001–004; SRG005–006)

- All the above plus SNV, CNV, and SV calling genome-wide
- Mitochondrial genome copy number assessment (depletion/amplification)

↓ *Upgradeable to any combined PRX test — pay only the difference; Transcriptome requires Tempus™ tube at original collection*

Long Read Whole Genome Sequencing (LRS001)

- Simultaneous small variant and structural variant from a single dataset
- Precise repeat-expansion sizing and confirmation of structural variant breakpoints
- Variant phasing on the same chromosome

↓

Long Read Methylome Sequencing (LRS101)

- Detection of methylation and imprinting defects genome-wide
- Direct methylated base detection without bisulfite conversion

↓

Optical Genome Mapping (OGM001–004; OGM201–204; OGM005–006)

- Detection of balanced translocations and inversions
- Repeat expansion sizing and macrosatellite repeat contraction detection (e.g. D4Z4, FSHD)
- Complex structural variant characterization

↓ *Upgradeable to include Transcriptome via combined PRX testing*

Transcriptome (SRT401–404)

- Functional assessment of SNVs, SVs, CNVs; detection of expression level changes, fusion genes, and aberrant splicing
- Requires RNA stabilization in a Tempus™ tube collected at the time of original sample submission or frozen/fixed tissue (FFPE)
- Can be combined with any tier above — contact us at the time of original ordering to ensure proper gene specific tissue type selection

Somatic-specific test codes: SRG601–602 (Somatic WGS), SRT701–702 (Somatic Transcriptome), OGM601–602 (Somatic OGM), PRX601–602 (Somatic combined OGM + WGS). The same upgrade architecture applies.