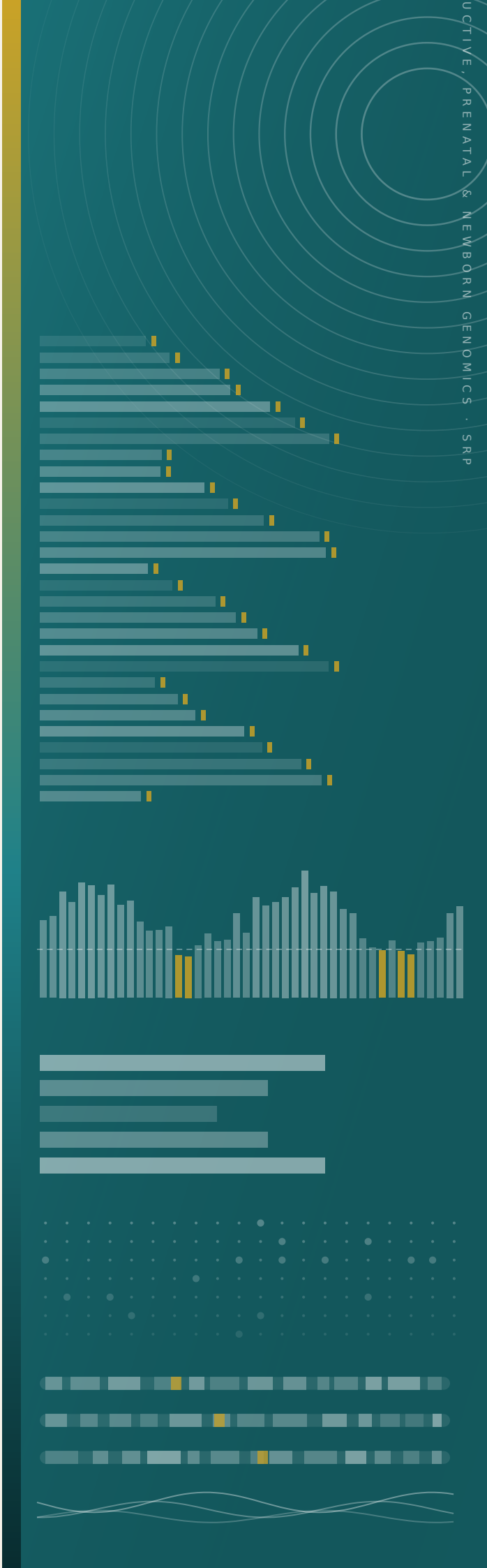
NTHRYS

OPC PVT LTD · HYDERABAD · INDIA

SECTOR CATALOGUE 03 · NGS DATA ANALYSIS

Reproductive, Prenatal & *Newborn Genomics*

Twenty-nine industrial training programmes, one for each distinct type of NGS data analysis performed in fertility clinics, prenatal diagnostic laboratories, preimplantation genetics units and newborn screening programmes. Every programme answers the questions a laboratory asks before it commits budget — why the analysis is done, when it is done, what it detects, what it does not detect, what makes it hard, who does this job and what a trainee can do afterwards — and is followed by its complete ordered workflow, step by step.



29	535	2598	443
ANALYSIS TYPES	STEPS	HOURS	DAYS

CATALOGUE SRP · 2026 EDITION

INDUSTRIAL TRAINING · REPRODUCTIVE GENOMICS PRACTICE

Reproductive, Prenatal & Newborn Genomics — *Analysis Types*

Twenty-nine distinct types of NGS data analysis performed across reproductive medicine, each an independent analysis with its own input, its own pipeline and its own report set. Each is listed under its formal title with the industry name and the shorthand a laboratory uses in practice. Every analysis type occupies a two-page entry: the first page answers why, when, what it detects, what it does not detect, who does the job and what follows; the second sets out the complete ordered workflow.

CODE	ANALYSIS TYPE · INDUSTRY NAME & SHORT NAME	STEPS	HOURS	DAYS	FEE (INR)
SRP-01	**Expanded Carrier Screening Analysis** Expanded carrier screening analysis · ECS analysis	18	88	15	2,05,000
SRP-02	**Couple-Based Carrier Risk Analysis** Combined couple reproductive risk analysis · Couple risk analysis	14	68	12	1,55,000
SRP-03	**Preconception Genomic Screening Analysis** Preconception genomic screening · Preconception screening	16	78	13	1,80,000
SRP-04	**Non-Invasive Prenatal Screening — Aneuploidy** NIPT aneuploidy analysis · NIPT · cfDNA screening	22	106	18	2,45,000
SRP-05	**NIPT Microdeletion & Subchromosomal Analysis** cfDNA microdeletion screening · Microdeletion NIPT	18	88	15	2,05,000
SRP-06	**Genome-Wide cfDNA Prenatal Analysis** Genome-wide cell-free DNA prenatal screening · Genome-wide NIPT	20	98	17	2,25,000
SRP-07	**Fetal Fraction Determination & Quality Analysis** Fetal fraction estimation and cfDNA quality control · Fetal fraction analysis	14	66	11	1,50,000
SRP-08	**Single-Gene Non-Invasive Prenatal Diagnosis** cfDNA single-gene prenatal diagnosis · NIPD · single-gene NIPT	21	102	17	2,35,000
SRP-09	**Fetal RHD & Blood Group Genotyping Analysis** Non-invasive fetal blood group genotyping · Fetal RHD typing	14	68	12	1,55,000
SRP-10	**Invasive Prenatal Diagnostic Analysis** CVS and amniocentesis sample analysis · Invasive prenatal diagnosis	19	92	16	2,10,000
SRP-11	**Prenatal Exome Analysis** Fetal exome sequencing analysis · Prenatal WES · fetal exome	24	116	20	2,65,000
SRP-12	**Prenatal Genome Analysis** Fetal whole genome sequencing analysis · Prenatal WGS · fetal genome	26	126	21	2,88,000
SRP-13	**Prenatal CNV & Digital Karyotyping Analysis** Prenatal copy number and chromosomal analysis · Prenatal CNV · digital karyotype	19	92	16	2,10,000
SRP-14	**Maternal Cell Contamination Analysis** MCC assessment in prenatal samples · MCC analysis	13	62	11	1,40,000
SRP-15	**Products of Conception & Pregnancy Loss Analysis** Miscarriage and fetal demise genomic analysis · POC analysis	17	82	14	1,90,000

Continued overleaf. Programme codes read SRP–01 and protocol references PRT/SRP/01.

Analysis Types — *16 to 29*

CODE	ANALYSIS TYPE · INDUSTRY NAME & SHORT NAME	STEPS	HOURS	DAYS	FEE (INR)
SRP-16	**Preimplantation Genetic Testing for Aneuploidy** PGT-A embryo aneuploidy analysis · PGT-A	22	108	18	2,45,000
SRP-17	**Preimplantation Genetic Testing for Monogenic Disease** PGT-M embryo single-gene analysis · PGT-M	24	118	20	2,70,000
SRP-18	**Preimplantation Testing for Structural Rearrangement** PGT-SR embryo rearrangement analysis · PGT-SR	20	98	17	2,25,000
SRP-19	**Embryo Mosaicism Analysis** Intermediate copy number and embryo mosaicism assessment · Mosaic embryo analysis	17	84	14	1,95,000
SRP-20	**Whole Genome Amplification Quality & Low-Input Analysis** Low-input amplification quality control · WGA QC · low-input analysis	15	72	12	1,65,000
SRP-21	**Sperm & Male Infertility Genomic Analysis** Male factor infertility genomic analysis · Male infertility panel	18	88	15	2,05,000
SRP-22	**Female Infertility & Ovarian Insufficiency Analysis** Female factor infertility genomic analysis · POI · female infertility panel	17	82	14	1,90,000
SRP-23	**Recurrent Pregnancy Loss Genomic Analysis** Recurrent miscarriage genomic investigation · RPL analysis	18	86	15	2,00,000
SRP-24	**Genomic Newborn Screening Analysis** Genomic newborn screening · gNBS · genomic newborn screening	23	112	19	2,55,000
SRP-25	**Rapid Neonatal Diagnostic Analysis** Rapid neonatal genomic diagnosis · Rapid neonatal analysis	25	122	21	2,80,000
SRP-26	**Newborn Pharmacogenomic Analysis** Neonatal and paediatric pharmacogenomic analysis · Newborn PGx	15	72	12	1,65,000
SRP-27	**Gamete Donor Screening & Matching Analysis** Donor genetic screening and recipient matching · Donor screening	16	78	13	1,80,000
SRP-28	**Mitochondrial Donation & Heteroplasmy Analysis** Mitochondrial heteroplasmy analysis in reproduction · Heteroplasmy analysis	17	84	14	1,95,000
SRP-29	**Fetal Sex Determination & X-Linked Risk Analysis** Non-invasive fetal sex determination · Fetal sexing	13	62	11	1,40,000
Complete Sector Catalogue 02 — all thirty-nine analysis types		**535**	**2598**	**443**	**59,68,000**

Each fee is derived independently from the step count, analytical complexity and reporting burden of that specific analysis type. There is no banded pricing. Days are calculated at six contact hours per working day and rounded up. Fees are per participant and exclusive of GST and the one-time registration charge. In-house delivery at the client laboratory and institutional rates are available on application.

SRP-01
SECTOR 01

Expanded Carrier Screening Analysis

(**Expanded carrier screening analysis** · ECS analysis)

WHY IT IS DONE

Most children born with a recessive condition have no family history of it, because both parents are unaffected carriers who had no reason to be tested. Screening healthy prospective parents identifies that risk before a pregnancy rather than after an affected birth.

WHAT IT ANSWERS

Is this individual a carrier for any condition on the screened gene set, and what reproductive risk does that carry?

WHEN IT IS DONE

Before conception where the decision window is widest, in early pregnancy, and before assisted reproduction. Increasingly offered routinely rather than only where family history suggests it.

WHAT TRIGGERS IT

Preconception counselling, an assisted reproduction pathway, known consanguinity, ancestry-associated founder risk, or a family history of recessive disease.

HOW IT IS DONE

The gene set version is authorised and coverage certified per gene, then sequence and copy number analysis run together with pseudogene-bearing loci routed to dedicated methods. Classification is deliberately conservative because the population is asymptomatic, and residual risk is calculated per gene against the reported ancestry.

WHAT IT PRODUCES

Per-gene carrier status, a per-gene coverage certificate, residual risk calculated by gene and ancestry, founder variant results, and a report supporting reproductive counselling.

WHAT IT DETECTS

Carrier status for sequence variants and copy number changes across the screened gene set, founder variants at population-appropriate sensitivity, and X-linked carrier status in female patients.

WHAT IT DOES NOT DETECT

Carrier status is never exhaustive, so residual risk always remains from variants outside the panel, uncertain variants and technically difficult genes. Several of the genes central to carrier screening carry pseudogene interference that defeats standard analysis, and severity or age of onset in an affected pregnancy cannot be predicted.

WHAT MAKES IT HARD

Residual risk is the number the couple actually needs and is the most frequently omitted deliverable. Carrier screening also concentrates on genes that are analytically awkward, so a panel-wide detection rate claim is unsupportable and gene-specific figures must be used instead.

WHO DOES THIS JOB

Variant scientist or clinical bioinformatician in reproductive genomics, mid level, working closely with genetic counselling.

WHAT YOU CAN DO AFTERWARDS

Operate a carrier screening pathway, manage pseudogene-bearing loci correctly, calculate and communicate per-gene residual risk, and support reproductive counselling with defensible figures.

WHO HIRES FOR IT

IVF and fertility chains, reproductive genetics laboratories, prenatal diagnostic services, and diagnostic chains offering preconception screening.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
18	**88 hours · 15 days**	Prior germline variant analysis experience	**2,05,000**

SRP-01
WORKFLOW

Expanded Carrier Screening Analysis

(**Expanded carrier screening analysis** · ECS analysis)

COMPLETE WORKFLOW — 18 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Gene set version authorisation**	Screening gene set version, condition list and severity criteria confirmed	Panel register	*Authorised gene set*
02	**Consent and scope confirmation**	Reproductive scope, partner testing and incidental finding policy confirmed	Consent record	*Analysis authorisation*
03	**Ancestry and family history capture**	Reported ancestry and family history captured for residual risk calculation	Structured capture	*Ancestry record*
04	**Data receipt and quality assessment**	Integrity and sequence quality verified against acceptance criteria	md5sum, FastQC, MultiQC	*QC acceptance record*
05	**Read preprocessing**	Adapters and low-quality tails removed with retention accounting	fastp	*Trimmed FASTQ*
06	**Alignment and processing**	Reads aligned, duplicate marked and recalibrated within panel intervals	bwa-mem2, Picard, GATK BQSR	*Analysis-ready BAM*
07	**Per-gene coverage certification**	Depth certified per gene and exon with shortfalls named individually	mosdepth, bedtools coverage	*Coverage certificate*
08	**Identity verification**	Fingerprint captured to confirm attribution before interpretation	somalier extract	*Identity clearance*
09	**Variant calling**	Variants called across the screened gene set with validated parameters	Validated caller	*Carrier VCF*
10	**Copy number calling**	Exon-level copy number called since deletions are a major carrier mechanism	gCNV, DECoN	*Carrier CNV calls*
11	**Pseudogene-aware locus handling**	Genes with pseudogene interference routed to dedicated methods	Specialist assay, long-read	*Specialist locus results*
12	**Founder variant screening**	Population-specific founder variants interrogated at required sensitivity	Founder variant panel	*Founder results*
13	**Repeat and structural allele assessment**	Repeat and structural alleles assessed where relevant to screened conditions	Repeat genotyping	*Structural allele results*
14	**Annotation and population filtering**	Variants annotated with carrier-appropriate ancestry-specific frequency thresholds	VEP, gnomAD by ancestry	*Annotated variants*
15	**Conservative classification**	Variants classified conservatively given an asymptomatic screened population	ACMG-AMP with gene specifications	*Classified variants*
16	**Per-gene residual risk calculation**	Post-test residual carrier risk calculated per gene against reported ancestry	Detection rate tables	*Residual risk figures*
17	**Orthogonal confirmation**	Reportable carrier findings confirmed by an independent method	Sanger, MLPA, ddPCR	*Confirmation record*
18	**Report generation and sign-out**	Carrier status, coverage and residual risk issued under dual signature	Validated report template	*Signed report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
18	**88 h · 15 d**	**SRP-01**	**2,05,000**

SRP-02
SECTOR 01

Couple-Based Carrier Risk Analysis

(**Combined couple reproductive risk analysis** · Couple risk analysis)

WHY IT IS DONE

A carrier result for one partner is not a reproductive risk on its own. Risk exists only where both partners carry variants in the same gene, so the analysis that matters clinically is the combined one, and it must account for the residual risk that testing leaves behind on both sides.

WHAT IT ANSWERS

What is this couple's combined probability of an affected pregnancy, for which conditions, and what reproductive options follow from it?

WHEN IT IS DONE

After both partners have been screened, in preconception counselling, before assisted reproduction, and when a new partner enters an existing reproductive plan.

WHAT TRIGGERS IT

Completion of screening in both partners, a positive result in one partner prompting testing of the other, or a preconception risk assessment request.

HOW IT IS DONE

Both partners' results are combined gene by gene with variant classification reviewed jointly, since a pairing of one pathogenic and one uncertain variant needs different handling from two pathogenic variants. Per-gene detection rates and ancestry-specific carrier frequencies are applied to derive combined and residual risk, and reproductive options are set out against each risk identified.

WHAT IT PRODUCES

The at-risk gene list with both partners' variants, combined risk per condition, residual risk after screening, phenotype information for identified conditions, and reproductive options documentation.

WHAT IT DETECTS

Genes in which both partners carry variants, the resulting autosomal recessive risk, X-linked risk from maternal carrier status, and the residual risk remaining after both screens.

WHAT IT DOES NOT DETECT

It cannot resolve risk arising from genes not screened in either partner, and it cannot predict phenotype severity where both partners carry different variants in the same gene. Risk figures depend on detection rates that vary by ancestry, so a couple of mixed or under-represented ancestry carries less certain figures.

WHAT MAKES IT HARD

Pairings involving a variant of uncertain significance are the recurring difficulty, because they cannot be dismissed and cannot be reported as a clear risk. Detection rates driving residual risk are ancestry-dependent and are frequently applied from a population that does not match the couple.

WHO DOES THIS JOB

Variant scientist or genetic counsellor with analytical training, mid level, in a reproductive genetics service.

WHAT YOU CAN DO AFTERWARDS

Combine partner results correctly, handle uncertain variant pairings defensibly, derive ancestry-appropriate residual risk, and present reproductive options accurately.

WHO HIRES FOR IT

IVF and fertility chains, reproductive genetics services, prenatal diagnostic laboratories, and genetic counselling services.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
14	**68 hours · 12 days**	Prior carrier screening analysis experience	**1,55,000**

SRP-02
WORKFLOW

Couple-Based Carrier Risk Analysis

(**Combined couple reproductive risk analysis** · Couple risk analysis)

COMPLETE WORKFLOW — 14 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Partner result verification**	Both partners' results confirmed complete with gene set versions recorded	Result review	*Verified partner results*
02	**Gene set concordance check**	Confirmation that both partners were screened on comparable gene sets	Version comparison	*Concordance record*
03	**Identity verification**	Both samples confirmed correctly attributed before combination	Fingerprint comparison	*Identity clearance*
04	**Ancestry recording**	Each partner's reported ancestry recorded for detection rate selection	Structured capture	*Ancestry records*
05	**Shared gene identification**	Genes in which both partners carry variants identified	Gene-level comparison	*Shared gene list*
06	**Variant classification review**	Variant pairings reviewed jointly since classification combinations differ in meaning	Joint classification review	*Reviewed pairings*
07	**Uncertain variant pairing handling**	Pairings involving uncertain variants handled under documented policy	Reporting policy	*Uncertain pairing decisions*
08	**Autosomal recessive risk calculation**	Combined risk calculated for each gene where both partners carry variants	Risk calculation	*Recessive risk figures*
09	**X-linked risk assessment**	Maternal X-linked carrier status assessed for risk to male offspring	X-linked risk logic	*X-linked risk*
10	**Per-gene detection rate application**	Ancestry-appropriate detection rates applied for each partner and gene	Detection rate tables	*Applied detection rates*
11	**Residual risk calculation**	Combined residual risk calculated after both partners' screening	Residual risk calculation	*Combined residual risk*
12	**Condition information assembly**	Phenotype and severity information assembled for each identified condition	Condition references	*Condition information*
13	**Reproductive options documentation**	Available options set out against each identified risk	Options framework	*Options documentation*
14	**Report generation and sign-out**	Combined risk, residual risk and options issued under dual signature	Validated report template	*Signed couple report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
14	**68 h · 12 d**	**SRP-02**	**1,55,000**

SRP-03
SECTOR 01

Preconception Genomic Screening Analysis

(**Preconception genomic screening** · Preconception screening)

WHY IT IS DONE

Screening before pregnancy gives the widest set of options — donor gametes, preimplantation testing, prenatal diagnosis or simply informed preparation — because no decision is yet constrained by an ongoing pregnancy and its timeline.

WHAT IT ANSWERS

What genomic risks relevant to reproduction does this individual or couple carry, before any pregnancy has begun?

WHEN IT IS DONE

During preconception planning, before assisted reproduction cycles, and in populations with recognised founder or consanguinity-related risk.

WHAT TRIGGERS IT

A preconception counselling appointment, planned assisted reproduction, a family history of genetic disease, or consanguinity.

HOW IT IS DONE

Scope and consent are established covering which finding categories will be returned, then carrier, structural and pharmacogenomic analyses are run against the authorised gene sets. Findings are classified conservatively for an asymptomatic population, residual risk is derived, and results are assembled to support a counselling conversation about options.

WHAT IT PRODUCES

Carrier and structural findings, pharmacogenomic results within scope, residual risk figures, a de novo limitation statement, and an options summary for counselling.

WHAT IT DETECTS

Recessive and X-linked carrier status, structural rearrangements relevant to reproductive outcome, pharmacogenomic variants relevant to pregnancy, and predisposition findings within the consented scope.

WHAT IT DOES NOT DETECT

It gives no information about the pregnancy itself, since none exists. De novo events, which cause a substantial share of severe childhood disease, are entirely unpredictable from parental screening, and this is the limitation prospective parents most often misunderstand.

WHAT MAKES IT HARD

Managing expectation is the central difficulty. Prospective parents frequently read a comprehensive negative as a guarantee, and the report must state plainly that the largest single cause of severe congenital disease is unpredictable de novo mutation.

WHO DOES THIS JOB

Variant scientist or clinical bioinformatician in reproductive genomics, mid level, with genetic counselling integral to delivery.

WHAT YOU CAN DO AFTERWARDS

Run preconception screening across multiple finding categories, derive residual risk, and produce reports that bound expectation honestly.

WHO HIRES FOR IT

Fertility and IVF chains, reproductive genetics services, preconception clinics, and consumer reproductive health providers.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
16	**78 hours · 13 days**	Prior germline variant analysis experience	**1,80,000**

SRP-03
WORKFLOW

Preconception Genomic Screening Analysis

(**Preconception genomic screening** · Preconception screening)

COMPLETE WORKFLOW — 16 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Scope and consent definition**	Finding categories to be returned defined and consented before analysis	Consent record	*Analysis authorisation*
02	**Gene set authorisation**	Carrier, structural and pharmacogenomic gene sets confirmed with versions	Gene set register	*Authorised gene sets*
03	**Ancestry and history capture**	Ancestry and reproductive history captured for risk interpretation	Structured capture	*Background record*
04	**Data receipt and quality assessment**	Integrity and sequence quality verified against acceptance criteria	md5sum, FastQC	*QC acceptance record*
05	**Read preprocessing and alignment**	Reads preprocessed and aligned with duplicate marking and recalibration	fastp, bwa-mem2, GATK BQSR	*Analysis-ready BAM*
06	**Per-gene coverage certification**	Depth certified across all authorised gene sets	mosdepth, bedtools coverage	*Coverage certificate*
07	**Identity verification**	Fingerprint captured to confirm attribution	somalier extract	*Identity clearance*
08	**Carrier variant calling**	Variants called across the carrier screening gene set	Validated caller	*Carrier variants*
09	**Copy number calling**	Copy number called across relevant genes	gCNV, DECoN	*Copy number findings*
10	**Structural rearrangement assessment**	Rearrangements with reproductive significance assessed	Structural analysis	*Structural findings*
11	**Pharmacogenomic analysis**	Pharmacogenes relevant to pregnancy analysed and diplotypes assigned	PharmCAT	*PGx results*
12	**Annotation and filtering**	Variants annotated with ancestry-appropriate frequency thresholds	VEP, gnomAD by ancestry	*Annotated variants*
13	**Conservative classification**	Variants classified conservatively for an asymptomatic population	ACMG-AMP with gene specifications	*Classified variants*
14	**Residual risk derivation**	Residual risk derived per gene against reported ancestry	Detection rate tables	*Residual risk*
15	**De novo limitation statement**	The unpredictability of de novo events stated explicitly in the report	Limitation template	*Limitation statement*
16	**Report generation and sign-out**	Findings, residual risk, limitations and options issued under dual signature	Validated report template	*Signed report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
16	**78 h · 13 d**	**SRP-03**	**1,80,000**

SRP-04
SECTOR 01

Non-Invasive Prenatal Screening — Aneuploidy

(**NIPT aneuploidy analysis** · NIPT · cfDNA screening)

WHY IT IS DONE

Screening for common trisomies from a maternal blood sample carries no procedural risk to the pregnancy, and its performance is far better than the serum and ultrasound screening it displaced. It has become the most widely performed cell-free DNA test in the world.

WHAT IT ANSWERS

Does this pregnancy show a cell-free DNA profile consistent with trisomy 21, 18 or 13, or a sex chromosome aneuploidy, and how reliable is that assessment given the fetal fraction achieved?

WHEN IT IS DONE

From around ten weeks of gestation, as a first-tier screen in many health systems and as a contingent screen after conventional risk assessment in others.

WHAT TRIGGERS IT

A routine antenatal screening pathway, advanced maternal age, an abnormal conventional screening result, or an ultrasound finding.

HOW IT IS DONE

Fetal fraction is determined first because it bounds everything that follows, then chromosomal representation is measured against a reference distribution with sequencing and GC bias corrected. Risk is computed against maternal age and gestational prior, results below the fetal fraction threshold are declared no-call rather than negative, and confounders are actively considered before reporting.

WHAT IT PRODUCES

Fetal fraction with its threshold assessment, per-chromosome risk scores with the statistical basis, no-call determination where applicable, sex chromosome findings within consented scope, and a screening report requiring confirmatory testing on any positive.

WHAT IT DETECTS

Chromosomal representation consistent with trisomy 21, 18 and 13, sex chromosome aneuploidies, and fetal sex where consented, with a risk score rather than a diagnosis.

WHAT IT DOES NOT DETECT

It is a screen, not a diagnosis, and every positive requires invasive confirmation. It does not detect most single-gene disorders, triploidy in many implementations, or structural abnormalities. Confined placental mosaicism, vanishing twin, maternal mosaicism and maternal malignancy all produce discordant results that are biologically real but not fetal.

WHAT MAKES IT HARD

Fetal fraction governs the entire result and a low-fraction sample must be reported as uninformative rather than low risk, which is the error with the gravest consequence. Confined placental mosaicism is the commonest cause of a true-positive test with a normal fetus, and reporting must convey that a positive is a screen result, not a finding.

WHO DOES THIS JOB

Clinical bioinformatician in prenatal genomics, mid to senior level, in a laboratory operating at high volume with strict quality control.

WHAT YOU CAN DO AFTERWARDS

Operate a cell-free DNA screening pipeline, determine and apply fetal fraction thresholds, compute risk correctly, and report screening results with appropriate confirmatory framing.

WHO HIRES FOR IT

Prenatal screening laboratories, IVF and maternity chains, national antenatal screening programmes, and diagnostic chains at scale.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
22	**106 hours · 18 days**	Prior cfDNA analysis experience	**2,45,000**

SRP-04
WORKFLOW

Non-Invasive Prenatal Screening — Aneuploidy

(**NIPT aneuploidy analysis** · NIPT · cfDNA screening)

COMPLETE WORKFLOW — 22 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Gestational age verification**	Gestational age confirmed against the assay's validated minimum	Clinical record	*Gestational age record*
02	**Consent and scope confirmation**	Screening scope including sex chromosome and fetal sex reporting confirmed	Consent record	*Analysis authorisation*
03	**Pre-analytical record capture**	Tube type, collection time and processing interval recorded	Pre-analytical log	*Pre-analytical record*
04	**Plasma separation quality assessment**	Separation quality assessed for maternal genomic DNA contamination	Fragment profile assessment	*Separation quality record*
05	**Cell-free DNA quantification**	Input quantity measured and assessed against assay requirements	Quantification	*Input record*
06	**Data receipt and quality assessment**	Integrity and sequence quality verified against acceptance criteria	md5sum, FastQC	*QC acceptance record*
07	**Alignment**	Reads aligned genome-wide with settings preserving fragment endpoints	bwa-mem2	*Aligned BAM*
08	**Duplicate and quality filtering**	Duplicates and low-quality alignments removed with counts recorded	Filtering	*Filtered BAM*
09	**GC bias correction**	Coverage corrected for GC bias which otherwise dominates representation	GC correction	*Corrected coverage*
10	**Mappability correction**	Coverage adjusted for mappability across chromosomes	Mappability correction	*Adjusted coverage*
11	**Fetal fraction determination**	Fetal fraction estimated by multiple independent methods	Fetal fraction estimation	*Fetal fraction*
12	**Fetal fraction threshold assessment**	Fraction assessed against the validated minimum for a reportable result	Threshold criteria	*Threshold decision*
13	**No-call determination**	Samples below threshold declared no-call rather than reported as low risk	No-call criteria	*No-call decision*
14	**Chromosomal representation measurement**	Representation measured per chromosome against reference distributions	Representation analysis	*Representation scores*
15	**Statistical risk computation**	Risk computed combining representation, fetal fraction and prior probability	Risk model	*Risk scores*
16	**Sex chromosome analysis**	Sex chromosome representation assessed within the consented scope	Sex chromosome analysis	*Sex chromosome findings*
17	**Confounder assessment**	Vanishing twin, maternal mosaicism and malignancy considered as explanations	Confounder review	*Confounder assessment*
18	**Quality control metric review**	Run and sample level metrics reviewed against acceptance criteria	QC review	*QC record*
19	**Positive predictive value statement**	Predictive value stated against maternal age and condition prevalence	PPV calculation	*PPV statement*
20	**Confirmatory framing**	Every positive framed as requiring invasive diagnostic confirmation	Reporting policy	*Confirmatory statement*
21	**Report generation**	Risk scores, fetal fraction and predictive value assembled	Validated screening report template	*Draft report*
22	**Clinical sign-out**	Report reviewed and authorised under dual signature	Sign-out procedure	*Signed screening report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
22	**106 h · 18 d**	**SRP-04**	**2,45,000**

SRP-05
SECTOR 01

NIPT Microdeletion & Subchromosomal Analysis

(**cfDNA microdeletion screening** · Microdeletion NIPT)

WHY IT IS DONE

Several microdeletion syndromes cause significant disability and are not associated with maternal age, so conventional risk-based screening never identifies them. Extending cell-free DNA analysis to subchromosomal regions offers detection, though at markedly lower positive predictive value than trisomy screening.

WHAT IT ANSWERS

Does this pregnancy show cell-free DNA evidence of a specific microdeletion or subchromosomal imbalance, and what is the predictive value of that result at this prevalence?

WHEN IT IS DONE

As an optional extension to routine cell-free DNA screening, and where an ultrasound finding suggests a specific microdeletion syndrome.

WHAT TRIGGERS IT

Patient election for extended screening, an ultrasound finding consistent with a microdeletion syndrome, or a family history of a specific deletion.

HOW IT IS DONE

Higher depth is applied than trisomy screening requires because the target regions are small, fetal fraction is assessed against a stricter threshold, and regional representation is measured with bias correction. Risk is computed with the condition prevalence made explicit so predictive value can be stated, and every positive is framed for confirmatory testing.

WHAT IT PRODUCES

Region-specific risk assessment, fetal fraction against the stricter threshold, explicitly stated positive predictive value at the relevant prevalence, and a report requiring confirmatory diagnosis.

WHAT IT DETECTS

Targeted microdeletion regions and, in genome-wide implementations, subchromosomal imbalances above a size threshold, reported as risk rather than diagnosis.

WHAT IT DOES NOT DETECT

Deletions below the size resolution, balanced rearrangements, and any region outside the design. Positive predictive value is low because these conditions are rare, so most positives are false, and this is the dominant clinical problem with the test rather than a technical footnote.

WHAT MAKES IT HARD

Predictive value is the whole difficulty. A test with excellent sensitivity and specificity still produces mostly false positives when prevalence is low, and reporting a risk score without the predictive value invites parents to act on a result that is more likely wrong than right.

WHO DOES THIS JOB

Clinical bioinformatician in prenatal genomics, mid to senior level, with statistical competence in predictive value reasoning.

WHAT YOU CAN DO AFTERWARDS

Extend cell-free DNA analysis to subchromosomal regions, apply stricter quality thresholds, and report risk with predictive value stated at the actual prevalence.

WHO HIRES FOR IT

Prenatal screening laboratories, maternity and IVF chains, and diagnostic providers offering extended screening menus.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
18	**88 hours · 15 days**	Prior NIPT analysis experience	**2,05,000**

SRP-05
WORKFLOW

NIPT Microdeletion & Subchromosomal Analysis

(**cfDNA microdeletion screening** · Microdeletion NIPT)

COMPLETE WORKFLOW — 18 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Region set authorisation**	The microdeletion regions covered and their size resolution confirmed	Assay register	*Authorised region set*
02	**Consent and scope confirmation**	Extended screening scope and its predictive value limitations consented	Consent record	*Analysis authorisation*
03	**Pre-analytical record capture**	Collection and processing variables recorded	Pre-analytical log	*Pre-analytical record*
04	**Cell-free DNA quantification**	Input quantity measured against the higher requirement for regional analysis	Quantification	*Input record*
05	**Data receipt and quality assessment**	Integrity and sequence quality verified against acceptance criteria	md5sum, FastQC	*QC acceptance record*
06	**Alignment and filtering**	Reads aligned and filtered with duplicate removal	bwa-mem2, filtering	*Filtered BAM*
07	**Bias correction**	GC and mappability bias corrected before regional analysis	Bias correction	*Corrected coverage*
08	**Fetal fraction determination**	Fetal fraction estimated by independent methods	Fetal fraction estimation	*Fetal fraction*
09	**Stricter threshold assessment**	Fraction assessed against the higher threshold regional analysis requires	Threshold criteria	*Threshold decision*
10	**Regional depth assessment**	Depth confirmed sufficient across each target region for detection	Regional coverage analysis	*Regional depth record*
11	**Regional representation measurement**	Representation measured across each microdeletion region	Regional analysis	*Regional scores*
12	**Size resolution statement**	The minimum detectable deletion size stated per region	Resolution calculation	*Resolution statement*
13	**Statistical risk computation**	Risk computed per region against reference distributions	Risk model	*Regional risk scores*
14	**Prevalence-based predictive value calculation**	Predictive value calculated using actual condition prevalence	PPV calculation	*PPV per region*
15	**Predictive value communication framing**	Results framed so a low predictive value is explicit rather than implied	Reporting policy	*Framing statement*
16	**Confounder assessment**	Maternal copy number variants considered as alternative explanations	Confounder review	*Confounder assessment*
17	**Confirmatory framing**	Every positive framed as requiring diagnostic confirmation	Reporting policy	*Confirmatory statement*
18	**Report generation and sign-out**	Regional risk, resolution and predictive value issued under dual signature	Validated report template	*Signed screening report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
18	**88 h · 15 d**	**SRP-05**	**2,05,000**

SRP-06
SECTOR 01

Genome-Wide cfDNA Prenatal Analysis

(**Genome-wide cell-free DNA prenatal screening** · Genome-wide NIPT)

WHY IT IS DONE

Restricting analysis to three chromosomes discards data that has already been generated. Genome-wide analysis detects rare autosomal trisomies and large subchromosomal imbalances that targeted screening cannot, some of which explain pregnancy complications that would otherwise remain unexplained.

WHAT IT ANSWERS

Does this pregnancy show any chromosome-wide or large subchromosomal imbalance anywhere in the genome, beyond the common trisomies?

WHEN IT IS DONE

Where the screening programme offers genome-wide analysis, in pregnancies with unexplained growth restriction or ultrasound findings, and in recurrent loss investigation.

WHAT TRIGGERS IT

Election of genome-wide screening, unexplained fetal growth restriction, ultrasound abnormality, or a history of pregnancy loss.

HOW IT IS DONE

Coverage is normalised genome-wide with GC and mappability bias corrected, fetal fraction is determined, and representation is assessed across all chromosomes and in sliding subchromosomal windows. Findings are scored against reference distributions, size resolution is stated, and results are interpreted against the strong prior that rare trisomies are placental.

WHAT IT PRODUCES

Genome-wide representation profiles, findings with size and location, the stated size resolution, fetal fraction assessment, placental origin interpretation, and confirmatory testing recommendations.

WHAT IT DETECTS

Rare autosomal trisomies, large subchromosomal deletions and duplications above the size resolution, and patterns suggestive of confined placental mosaicism.

WHAT IT DOES NOT DETECT

Events below the size resolution, balanced rearrangements, triploidy in most implementations, and single-gene disorders. The clinical significance of many genome-wide findings is unknown, and rare autosomal trisomies most often reflect placental rather than fetal chromosomes.

WHAT MAKES IT HARD

Most positive findings are placental rather than fetal, and communicating that without either alarming or falsely reassuring is genuinely difficult. Many findings have no established clinical meaning, so a policy on which are reported must exist before the first one appears.

WHO DOES THIS JOB

Clinical bioinformatician in prenatal genomics, mid to senior level, working with fetal medicine on reporting policy.

WHAT YOU CAN DO AFTERWARDS

Run genome-wide cell-free DNA analysis, state size resolution honestly, interpret findings against placental origin priors, and apply a defined reporting policy.

WHO HIRES FOR IT

Prenatal screening laboratories, fetal medicine services, maternity chains, and national screening programmes.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
20	**98 hours · 17 days**	Prior NIPT and copy number analysis experience	**2,25,000**

SRP-06
WORKFLOW

Genome-Wide cfDNA Prenatal Analysis

(**Genome-wide cell-free DNA prenatal screening** · Genome-wide NIPT)

COMPLETE WORKFLOW — 20 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Analysis scope authorisation**	Genome-wide scope, size resolution and reporting policy confirmed	Assay register	*Authorised scope*
02	**Consent and finding policy confirmation**	Which genome-wide finding categories will be reported confirmed with the patient	Consent record	*Analysis authorisation*
03	**Pre-analytical record capture**	Collection and processing variables recorded	Pre-analytical log	*Pre-analytical record*
04	**Cell-free DNA quantification**	Input quantity measured against assay requirements	Quantification	*Input record*
05	**Data receipt and quality assessment**	Integrity and sequence quality verified against criteria	md5sum, FastQC	*QC acceptance record*
06	**Alignment and filtering**	Reads aligned genome-wide and filtered with duplicates removed	bwa-mem2, filtering	*Filtered BAM*
07	**Genome-wide binning**	Coverage binned genome-wide at the resolution the design supports	Binning	*Binned coverage*
08	**GC and mappability correction**	Systematic coverage bias removed across all bins	Bias correction	*Corrected coverage*
09	**Fetal fraction determination**	Fetal fraction estimated by independent methods	Fetal fraction estimation	*Fetal fraction*
10	**Threshold assessment**	Fraction assessed against the validated minimum for genome-wide reporting	Threshold criteria	*Threshold decision*
11	**Whole chromosome representation analysis**	Representation assessed for every autosome, not only the common trisomies	Representation analysis	*Chromosome scores*
12	**Subchromosomal window analysis**	Sliding window analysis performed to detect large segmental imbalances	Window analysis	*Segmental findings*
13	**Size resolution determination**	The minimum detectable segmental size determined and stated	Resolution calculation	*Resolution statement*
14	**Statistical scoring**	Findings scored against reference distributions with confidence derived	Statistical model	*Scored findings*
15	**Rare trisomy interpretation**	Rare autosomal trisomies interpreted against the strong placental origin prior	Interpretation framework	*Placental interpretation*
16	**Confined placental mosaicism assessment**	Findings assessed for patterns consistent with placental rather than fetal origin	Pattern assessment	*CPM assessment*
17	**Clinical significance policy application**	Findings of unknown significance handled under the pre-agreed reporting policy	Reporting policy	*Policy application record*
18	**Growth restriction correlation**	Findings correlated against ultrasound growth findings where present	Clinical correlation	*Correlation record*
19	**Confirmatory recommendation**	Diagnostic confirmation recommended with the appropriate sample type specified	Recommendation criteria	*Confirmatory recommendation*
20	**Report generation and sign-out**	Findings, resolution and placental interpretation issued under dual signature	Validated report template	*Signed screening report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
20	**98 h · 17 d**	**SRP-06**	**2,25,000**

SRP-07
SECTOR 01

Fetal Fraction Determination & Quality Analysis

(**Fetal fraction estimation and cfDNA quality control** · Fetal fraction analysis)

WHY IT IS DONE

Every cell-free DNA prenatal result is bounded by how much of the circulating DNA is fetal. Below a threshold, a negative result carries no information at all, and reporting it as low risk is the single most consequential error available in prenatal screening.

WHAT IT ANSWERS

What proportion of the cell-free DNA in this sample is fetal, is that sufficient for a reliable result, and if not why?

WHEN IT IS DONE

Within every cell-free DNA prenatal analysis as a mandatory first step, and in investigating a no-call or a discordant result.

WHAT TRIGGERS IT

Any cell-free DNA prenatal test, a no-call result requiring explanation, or a discordant result under investigation.

HOW IT IS DONE

Fetal fraction is estimated by independent methods including fragment size distribution, Y chromosome representation in male pregnancies and polymorphic marker approaches, then cross-checked for agreement. The result is assessed against the assay's validated threshold, and low fractions are investigated against gestational age, maternal weight and processing quality.

WHAT IT PRODUCES

Fetal fraction estimates by each method with agreement assessment, the threshold determination, no-call decision where applicable, cause investigation for low fractions, and redraw recommendation with timing.

WHAT IT DETECTS

Fetal fraction with confidence bounds by one or more independent methods, its relationship to gestational age and maternal weight, and quality failures that suppress it.

WHAT IT DOES NOT DETECT

Fetal fraction does not indicate whether the fetus is chromosomally normal — it only bounds the reliability of the assessment. It also cannot distinguish a low fraction caused by early gestation from one caused by placental insufficiency, though the clinical implications differ substantially.

WHAT MAKES IT HARD

Single-method estimation can fail silently, so cross-method agreement is the practical safeguard. Distinguishing early gestation from placental insufficiency as the cause of a low fraction matters clinically, because one resolves with a redraw and the other is itself a finding.

WHO DOES THIS JOB

Clinical bioinformatician in prenatal genomics, mid level. A small analysis that governs the reliability of a very large volume of testing.

WHAT YOU CAN DO AFTERWARDS

Estimate fetal fraction by multiple methods, adjudicate disagreement, apply thresholds correctly, and investigate the cause of low fractions.

WHO HIRES FOR IT

Prenatal screening laboratories, maternity and IVF chains, and screening programme quality functions.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
14	**66 hours · 11 days**	Prior cfDNA analysis experience	**1,50,000**

SRP-07
WORKFLOW

Fetal Fraction Determination & Quality Analysis

(**Fetal fraction estimation and cfDNA quality control** · Fetal fraction analysis)

COMPLETE WORKFLOW — 14 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Method set specification**	The fetal fraction estimation methods to be applied specified and recorded	Assay specification	*Method set*
02	**Gestational age and maternal weight capture**	Gestational age and maternal weight recorded as determinants of fetal fraction	Clinical record	*Clinical variables*
03	**Pre-analytical record capture**	Collection and processing variables recorded as they suppress fetal fraction	Pre-analytical log	*Pre-analytical record*
04	**Plasma separation quality assessment**	Separation quality assessed for maternal genomic DNA release	Fragment profile assessment	*Separation quality*
05	**Fragment size distribution analysis**	Fragment lengths analysed since fetal fragments are systematically shorter	Fragment analysis	*Size distribution*
06	**Size-based fraction estimation**	Fetal fraction estimated from the fragment size distribution	Size-based estimation	*Size-based estimate*
07	**Y chromosome based estimation**	Fetal fraction estimated from Y representation in male pregnancies	Y-based estimation	*Y-based estimate*
08	**Polymorphic marker estimation**	Fetal fraction estimated from allelic imbalance at polymorphic sites	Marker-based estimation	*Marker-based estimate*
09	**Cross-method agreement assessment**	Estimates compared across methods with disagreement investigated	Agreement analysis	*Agreement record*
10	**Confidence bound derivation**	Confidence bounds derived for the adopted fetal fraction estimate	Confidence estimation	*Fraction bounds*
11	**Threshold assessment**	The estimate assessed against the validated minimum for the intended analysis	Threshold criteria	*Threshold decision*
12	**Low fraction cause investigation**	Low fractions investigated against gestational age, weight and processing quality	Cause analysis	*Cause assessment*
13	**Placental insufficiency consideration**	Placental insufficiency considered where low fraction is unexplained	Clinical correlation	*Insufficiency assessment*
14	**Redraw recommendation**	Redraw recommended with appropriate timing where fraction is insufficient	Recommendation criteria	*Redraw recommendation*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
14	**66 h · 11 d**	**SRP-07**	**1,50,000**

SRP-08
SECTOR 01

Single-Gene Non-Invasive Prenatal Diagnosis

(**cfDNA single-gene prenatal diagnosis** · NIPD · single-gene NIPT)

WHY IT IS DONE

Families at known risk of a specific monogenic condition previously faced a choice between an invasive procedure carrying miscarriage risk and not knowing. Determining fetal genotype from maternal plasma removes that choice entirely for a growing set of conditions.

WHAT IT ANSWERS

Has the fetus inherited the familial pathogenic variant or variants, determined from maternal plasma without an invasive procedure?

WHEN IT IS DONE

In pregnancies at known risk of a specific monogenic condition where the familial variants are already characterised, from around nine weeks of gestation.

WHAT TRIGGERS IT

A known familial pathogenic variant with an at-risk pregnancy, a previous affected child, or a carrier couple seeking definitive fetal genotyping.

HOW IT IS DONE

Familial variants are characterised and family samples used to construct parental haplotypes, then plasma is sequenced deeply at the relevant region. Paternal variants are detected directly by their absence from the maternal genome, and maternal inheritance is determined by relative haplotype dosage measuring the imbalance between maternal haplotypes in plasma.

WHAT IT PRODUCES

Fetal genotype for each familial variant, haplotype construction evidence, the statistical basis of dosage determination, fetal fraction and confidence assessment, and a report with confirmatory recommendations.

WHAT IT DETECTS

Paternally inherited variants directly, and maternally inherited variants through relative haplotype dosage where family reference material is available, with fetal genotype determined for the specific familial variants.

WHAT IT DOES NOT DETECT

Only the specific variants targeted, so it says nothing about any other condition. Maternal inheritance requires haplotype construction and therefore family samples, which are not always obtainable, and de novo variants in the fetus are not detectable by this approach.

WHAT MAKES IT HARD

Relative haplotype dosage is a small statistical signal riding on a large maternal background, so fetal fraction and depth requirements are severe. Constructing usable haplotypes needs the right family samples, and the analysis cannot proceed without them regardless of sequencing quality.

WHO DOES THIS JOB

Senior clinical bioinformatician in prenatal genomics with haplotype and statistical competence. A specialist and scarce skill.

WHAT YOU CAN DO AFTERWARDS

Construct parental haplotypes, apply relative haplotype dosage, determine fetal genotype from plasma, and state confidence defensibly.

WHO HIRES FOR IT

Specialist fetal medicine and prenatal diagnostic centres, national prenatal genomics programmes, and reference laboratories offering non-invasive diagnosis.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
21	**102 hours · 17 days**	Prior cfDNA and haplotype analysis experience	**2,35,000**

SRP-08
WORKFLOW

Single-Gene Non-Invasive Prenatal Diagnosis

(**cfDNA single-gene prenatal diagnosis** · NIPD · single-gene NIPT)

COMPLETE WORKFLOW — 21 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Familial variant characterisation**	The familial pathogenic variants fully characterised at sequence level	Prior genetic testing	*Variant characterisation*
02	**Family sample availability assessment**	Required family reference samples assessed for haplotype construction	Sample inventory	*Sample availability record*
03	**Feasibility determination**	Feasibility determined before the pregnancy proceeds to testing	Feasibility criteria	*Feasibility record*
04	**Informative marker selection**	Polymorphic markers flanking the locus selected for haplotype construction	Marker selection	*Informative marker set*
05	**Parental genotyping**	Parents and reference relatives genotyped at the marker set	Genotyping	*Parental genotypes*
06	**Haplotype construction**	Parental haplotypes constructed with the disease-carrying haplotype identified	Haplotype phasing	*Constructed haplotypes*
07	**Gestational age verification**	Gestational age confirmed against the assay minimum for adequate fetal fraction	Clinical record	*Gestational age record*
08	**Pre-analytical record capture**	Collection and processing variables recorded	Pre-analytical log	*Pre-analytical record*
09	**Cell-free DNA quantification**	Input quantity measured against the high requirement for dosage analysis	Quantification	*Input record*
10	**Deep targeted sequencing**	Plasma sequenced deeply across the locus and flanking marker region	Targeted deep sequencing	*Plasma data*
11	**Data quality assessment**	Depth and uniformity assessed across all marker positions	Coverage analysis	*Quality record*
12	**Fetal fraction determination**	Fetal fraction estimated and assessed against the dosage requirement	Fetal fraction estimation	*Fetal fraction*
13	**Paternal variant detection**	Paternally inherited variants detected by absence from the maternal genome	Direct detection	*Paternal variant status*
14	**Marker dosage measurement**	Relative dosage measured across maternal haplotype markers in plasma	Dosage measurement	*Dosage measurements*
15	**Relative haplotype dosage analysis**	Maternal inheritance determined from the statistical imbalance between haplotypes	RHDO analysis	*Maternal inheritance*
16	**Statistical confidence assessment**	Confidence derived from marker count, depth and fetal fraction	Statistical assessment	*Confidence record*
17	**Recombination assessment**	Recombination between markers assessed as a source of misassignment	Recombination analysis	*Recombination assessment*
18	**Fetal genotype determination**	Fetal genotype determined by combining paternal and maternal inheritance	Genotype logic	*Fetal genotype*
19	**Confirmatory recommendation**	Confirmation by invasive testing or at birth recommended per protocol	Recommendation criteria	*Confirmatory recommendation*
20	**Report generation**	Genotype, haplotype evidence and confidence assembled	Validated report template	*Draft report*
21	**Clinical sign-out**	Report reviewed and authorised under dual signature	Sign-out procedure	*Signed report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
21	**102 h · 17 d**	**SRP-08**	**2,35,000**

SRP-09
SECTOR 01

Fetal RHD & Blood Group Genotyping Analysis

(**Non-invasive fetal blood group genotyping** · Fetal RHD typing)

WHY IT IS DONE

Determining fetal blood group from maternal plasma allows anti-D prophylaxis to be given only to women carrying an antigen-positive fetus, avoiding unnecessary blood product administration in a large proportion of pregnancies and directing monitoring where alloimmunisation risk is real.

WHAT IT ANSWERS

Does this fetus carry the blood group antigen that the mother lacks, and is prophylaxis or enhanced monitoring therefore required?

WHEN IT IS DONE

In antigen-negative pregnant women for targeted prophylaxis, and in alloimmunised pregnancies to determine whether the fetus is at risk of haemolytic disease.

WHAT TRIGGERS IT

An antigen-negative pregnancy in a targeted prophylaxis programme, or maternal alloimmunisation requiring fetal risk assessment.

HOW IT IS DONE

Plasma is sequenced or genotyped at the relevant loci with multiple exons interrogated to distinguish gene deletion from variant alleles. Fetal DNA presence is confirmed independently by control markers, which is essential because a negative result and a failed test look identical without them, and population-specific variant alleles are actively considered.

WHAT IT PRODUCES

Fetal genotype with the exons interrogated, fetal DNA presence confirmation from control markers, variant allele assessment for the relevant population, and a report directing prophylaxis or monitoring.

WHAT IT DETECTS

Fetal blood group genotype at the relevant loci, distinguishing antigen-positive from antigen-negative fetuses, with control markers confirming fetal DNA presence.

WHAT IT DOES NOT DETECT

It cannot determine antigen expression level or predict the severity of haemolytic disease where the fetus is positive. Variant and silenced alleles common in some populations produce genotype-phenotype discordance, and a negative result without adequate positive control is uninterpretable rather than negative.

WHAT MAKES IT HARD

A negative fetal result and a technically failed test are indistinguishable unless fetal DNA presence is independently confirmed, which makes the control markers the critical component. Variant and silenced alleles differ by population and cause genotype-phenotype mismatch if not accounted for.

WHO DOES THIS JOB

Clinical bioinformatician in transfusion or prenatal genomics, mid level, working with transfusion medicine.

WHAT YOU CAN DO AFTERWARDS

Determine fetal blood group genotype from plasma, confirm fetal DNA presence independently, and handle population-specific variant alleles correctly.

WHO HIRES FOR IT

Transfusion services, blood transfusion reference laboratories, prenatal screening laboratories, and maternity services.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
14	**68 hours · 12 days**	Prior cfDNA analysis experience	**1,55,000**

SRP-09
WORKFLOW

Fetal RHD & Blood Group Genotyping Analysis

(**Non-invasive fetal blood group genotyping** · Fetal RHD typing)

COMPLETE WORKFLOW — 14 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Indication and antigen scope confirmation**	The maternal antigen status and target loci confirmed	Transfusion record	*Scope confirmation*
02	**Gestational age verification**	Gestational age confirmed against the assay's validated minimum	Clinical record	*Gestational age record*
03	**Pre-analytical record capture**	Collection and processing variables recorded	Pre-analytical log	*Pre-analytical record*
04	**Cell-free DNA quantification**	Input quantity measured against assay requirements	Quantification	*Input record*
05	**Maternal genotype confirmation**	Maternal genotype at the locus confirmed to interpret plasma correctly	Maternal genotyping	*Maternal genotype*
06	**Multi-exon target interrogation**	Multiple exons interrogated to distinguish gene deletion from variant alleles	Multi-target analysis	*Exon-level results*
07	**Control marker analysis**	Independent control markers analysed to confirm fetal DNA presence	Control marker analysis	*Fetal DNA confirmation*
08	**Fetal DNA presence determination**	Fetal DNA presence determined independently of the target result	Presence logic	*Presence determination*
09	**Variant allele assessment**	Population-specific variant and silenced alleles assessed	Variant allele analysis	*Variant allele findings*
10	**Genotype determination**	Fetal genotype determined across the interrogated exons	Genotype logic	*Fetal genotype*
11	**Discordance risk assessment**	Genotype-phenotype discordance risk assessed for the relevant population	Discordance analysis	*Discordance assessment*
12	**Inconclusive determination**	Results without adequate fetal DNA confirmation declared inconclusive	Inconclusive criteria	*Inconclusive decision*
13	**Clinical action derivation**	Prophylaxis or monitoring recommendation derived from the result	Clinical protocol	*Action recommendation*
14	**Report generation and sign-out**	Genotype, presence confirmation and action issued under dual signature	Validated report template	*Signed report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
14	**68 h · 12 d**	**SRP-09**	**1,55,000**

SRP-10
SECTOR 01

Invasive Prenatal Diagnostic Analysis

(**CVS and amniocentesis sample analysis** · Invasive prenatal diagnosis)

WHY IT IS DONE

Screening produces risk figures; only a diagnostic sample from the pregnancy itself produces an answer. Chorionic villus and amniotic fluid samples are the definitive material, and their analysis carries a decision timeline that no other diagnostic work does.

WHAT IT ANSWERS

What is the definitive genetic status of this pregnancy for the conditions being investigated?

WHEN IT IS DONE

After a positive screening result, following an abnormal ultrasound, in pregnancies at known monogenic risk, and where advanced maternal age or family history warrants diagnostic rather than screening testing.

WHAT TRIGGERS IT

A positive cell-free DNA or serum screening result, an ultrasound abnormality, a known familial variant, or parental carrier status requiring fetal diagnosis.

HOW IT IS DONE

Sample type and its specific limitations are recorded, maternal cell contamination is excluded before any result is interpreted, and the indicated analyses are run against a compressed clinical timeline. Findings are classified with fetal phenotype implications assessed, and results are communicated within the decision window the pregnancy allows.

WHAT IT PRODUCES

Maternal cell contamination assessment, the diagnostic result for the indication, mosaicism assessment where relevant, phenotype implications, and a report delivered within the clinical decision window.

WHAT IT DETECTS

Aneuploidy, copy number changes, and targeted or genome-wide sequence variants depending on the indication, with maternal cell contamination excluded.

WHAT IT DOES NOT DETECT

Anything outside the analysis requested, and it cannot resolve confined placental mosaicism from chorionic villus material without follow-up amniocentesis. Mosaicism within the sample itself may be missed depending on the level present and the method used.

WHAT MAKES IT HARD

Maternal cell contamination produces a normal-looking result on a maternal sample and is the most serious failure available here. Chorionic villus material reflects placenta rather than fetus, so a finding may be confined to the placenta and require amniocentesis to resolve.

WHO DOES THIS JOB

Clinical bioinformatician in prenatal genomics, mid to senior level, working with fetal medicine under time pressure.

WHAT YOU CAN DO AFTERWARDS

Analyse invasive prenatal samples, exclude maternal cell contamination rigorously, interpret sample-type-specific limitations, and deliver within a decision window.

WHO HIRES FOR IT

Fetal medicine centres, prenatal diagnostic laboratories, hospital genetics services, and reference laboratories.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
19	**92 hours · 16 days**	Prior germline variant and copy number analysis experience	**2,10,000**

SRP-10
WORKFLOW

Invasive Prenatal Diagnostic Analysis

(**CVS and amniocentesis sample analysis** · Invasive prenatal diagnosis)

COMPLETE WORKFLOW — 19 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Sample type and indication recording**	Sample type recorded with its specific limitations and the clinical indication	Requisition, sample record	*Sample record*
02	**Decision window recording**	The clinical decision window recorded as the operative delivery constraint	Clinical record	*Timeline record*
03	**Sample quality assessment**	Cellularity, contamination and material adequacy assessed on receipt	Sample assessment	*Adequacy record*
04	**Maternal sample requirement verification**	A maternal comparator sample confirmed available for contamination assessment	Sample inventory	*Comparator verification*
05	**Maternal cell contamination assessment**	Contamination assessed and quantified before any fetal result is interpreted	MCC analysis	*Contamination assessment*
06	**Contamination threshold decision**	Contamination assessed against the threshold appropriate to the requested analysis	Threshold criteria	*Threshold decision*
07	**Data receipt and quality assessment**	Integrity and sequence quality verified against acceptance criteria	md5sum, FastQC	*QC acceptance record*
08	**Alignment and processing**	Reads aligned, duplicate marked and recalibrated	bwa-mem2, Picard, GATK BQSR	*Analysis-ready BAM*
09	**Coverage certification**	Depth certified across the regions relevant to the indication	mosdepth	*Coverage certificate*
10	**Aneuploidy assessment**	Chromosomal aneuploidy assessed as a first-tier finding	Copy number analysis	*Aneuploidy results*
11	**Copy number analysis**	Genome-wide or targeted copy number called at the stated resolution	Copy number analysis	*Copy number findings*
12	**Targeted variant analysis**	Sequence variants analysed where the indication specifies a familial variant or gene set	Validated caller	*Variant results*
13	**Mosaicism assessment**	Mosaicism assessed and its detection floor stated for the sample type	Mosaicism analysis	*Mosaicism assessment*
14	**Placental confinement consideration**	Chorionic villus findings assessed for possible confinement to placenta	Interpretation framework	*Confinement assessment*
15	**Annotation and classification**	Findings annotated and classified with prenatal phenotype implications assessed	VEP, ACMG-AMP	*Classified findings*
16	**Follow-up sampling recommendation**	Amniocentesis recommended where placental confinement cannot be excluded	Recommendation criteria	*Follow-up recommendation*
17	**Orthogonal confirmation**	Reportable findings confirmed by an independent method within the timeline	Sanger, MLPA, ddPCR	*Confirmation record*
18	**Report generation**	Findings, contamination clearance and limitations assembled within the window	Validated report template	*Draft report*
19	**Clinical sign-out and communication**	Report authorised and communicated within the decision window	Sign-out procedure	*Signed diagnostic report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
19	**92 h · 16 d**	**SRP-10**	**2,10,000**

SRP-11
SECTOR 01

Prenatal Exome Analysis

(**Fetal exome sequencing analysis** · Prenatal WES · fetal exome)

WHY IT IS DONE

When ultrasound shows structural abnormalities but karyotype and microarray are normal, exome sequencing identifies a monogenic cause in a meaningful proportion of cases. That answer changes pregnancy management, delivery planning and recurrence counselling.

WHAT IT ANSWERS

Is there a monogenic explanation for the structural abnormalities seen on this fetus, and what does it mean for this pregnancy and for future ones?

WHEN IT IS DONE

After normal karyotype and microarray in a structurally abnormal fetus, in recurrent unexplained fetal abnormality, and in fetal demise with structural findings.

WHAT TRIGGERS IT

Structural abnormality on ultrasound with normal cytogenetic and microarray results, recurrence of an unexplained fetal phenotype, or unexplained fetal demise.

HOW IT IS DONE

Maternal cell contamination is excluded, then trio analysis is run where parents are available with de novo detection and inheritance filtering. Prioritisation uses the ultrasound phenotype expressed in structured terms, classification is conservative because fetal phenotype cannot be fully assessed, and results are delivered within the pregnancy decision window.

WHAT IT PRODUCES

Maternal cell contamination clearance, classified variants with fetal phenotype correlation, de novo findings with parental evidence, recurrence risk, and a report delivered inside the decision window.

WHAT IT DETECTS

Coding and splice variants explaining the fetal phenotype, with trio analysis where parental samples are available enabling de novo identification and inheritance filtering.

WHAT IT DOES NOT DETECT

Deep intronic, regulatory and structural causes, and conditions whose fetal presentation is not yet described. Fetal phenotype is incompletely observable by ultrasound, so phenotype-driven prioritisation operates on partial information, which is the defining analytical constraint.

WHAT MAKES IT HARD

The fetal phenotype is partial and evolving, so a variant that would be clearly causative in a described child may be uncertain in a fetus. Variants of uncertain significance are far harder to handle here because the decision that follows is irreversible and time-limited.

WHO DOES THIS JOB

Senior clinical bioinformatician or clinical scientist in prenatal genomics, working closely with fetal medicine and clinical genetics.

WHAT YOU CAN DO AFTERWARDS

Run prenatal trio exome analysis, prioritise against partial fetal phenotype, classify conservatively for prenatal context, and deliver within a decision window.

WHO HIRES FOR IT

Fetal medicine centres, national prenatal genomics programmes, hospital genetics laboratories, and specialist prenatal diagnostic services.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
24	**116 hours · 20 days**	Prior exome and trio analysis experience	**2,65,000**

SRP-11
WORKFLOW

Prenatal Exome Analysis

(**Fetal exome sequencing analysis** · Prenatal WES · fetal exome)

COMPLETE WORKFLOW — 24 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Indication and phenotype capture**	Ultrasound findings captured in structured terms as the prioritisation input	Structured HPO capture	*Structured fetal phenotype*
02	**Prior result verification**	Karyotype and microarray results verified as normal before exome proceeds	Prior result review	*Prior result record*
03	**Consent and finding scope confirmation**	Scope including incidental and parental findings confirmed with the family	Consent record	*Analysis authorisation*
04	**Decision window recording**	The pregnancy decision window recorded as the delivery constraint	Clinical record	*Timeline record*
05	**Sample adequacy assessment**	Fetal sample and parental samples assessed for adequacy	Sample assessment	*Adequacy record*
06	**Maternal cell contamination assessment**	Contamination excluded before any fetal result is interpreted	MCC analysis	*Contamination clearance*
07	**Data receipt and quality assessment**	Integrity and sequence quality verified for fetal and parental samples	md5sum, FastQC, MultiQC	*QC acceptance record*
08	**Read preprocessing**	Adapters and low-quality tails removed identically across all samples	fastp	*Trimmed FASTQ*
09	**Alignment and processing**	All samples aligned, duplicate marked and recalibrated identically	bwa-mem2, Picard, GATK BQSR	*Analysis-ready BAMs*
10	**Relatedness verification**	Parental relationships verified before inheritance filtering is applied	somalier relate	*Pedigree verification*
11	**Coverage and gap analysis**	Per-exon coverage certified with gaps declared for the fetal sample	mosdepth, bedtools coverage	*Coverage certificate*
12	**Variant calling**	Variants called with joint genotyping where parental samples are available	Validated caller	*Exome VCF*
13	**Exome copy number calling**	Exon-level copy number called against a matched reference pool	gCNV, DECoN	*Copy number findings*
14	**De novo variant identification**	De novo variants identified with stringent filtering where trio data exist	Trio logic	*De novo candidates*
15	**De novo read-level review**	Every de novo candidate reviewed at read level in all three samples	IGV review	*Reviewed de novo set*
16	**Inheritance model filtering**	All inheritance models applied in parallel against the fetal phenotype	PED-aware filtering	*Model-filtered candidates*
17	**Annotation and population filtering**	Variants annotated with model-specific frequency thresholds applied	VEP, gnomAD, ClinVar	*Annotated candidates*
18	**Phenotype-driven prioritisation**	Candidates ranked against the structured ultrasound phenotype	Exomiser, HPO terms	*Ranked candidates*
19	**Prenatal phenotype limitation assessment**	Prioritisation assessed against the incompleteness of observable fetal phenotype	Limitation review	*Phenotype limitation record*
20	**Conservative classification**	Variants classified conservatively given prenatal phenotype uncertainty	ACMG-AMP with gene specifications	*Classified variants*
21	**Multidisciplinary review**	Candidates reviewed with fetal medicine and clinical genetics	Scheduled review	*Review record*
22	**Orthogonal confirmation**	Reportable findings confirmed within the decision window	Sanger, MLPA	*Confirmation record*
23	**Report generation**	Findings, phenotype correlation, recurrence risk and limitations assembled	Validated report template	*Draft report*
24	**Clinical sign-out and communication**	Report authorised and communicated within the decision window	Sign-out procedure	*Signed clinical report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
24	**116 h · 20 d**	**SRP-11**	**2,65,000**

SRP-12
SECTOR 01

Prenatal Genome Analysis

(**Fetal whole genome sequencing analysis** · Prenatal WGS · fetal genome)

WHY IT IS DONE

Genome sequencing detects structural, repeat and non-coding causes that exome analysis misses, and does so from a single limited fetal sample rather than through sequential testing that the pregnancy timeline cannot accommodate.

WHAT IT ANSWERS

Is there any genomic explanation for this fetal presentation, across sequence, structural, copy number and repeat classes together?

WHEN IT IS DONE

In severe or complex fetal presentations, after a non-diagnostic exome, where sample material is too limited for sequential testing, and in programmes offering genome as the first-line diagnostic test.

WHAT TRIGGERS IT

A severe or multisystem fetal abnormality, a non-diagnostic prenatal exome, limited sample material, or programme protocol.

HOW IT IS DONE

Maternal cell contamination is excluded, then parallel analytical arms address every variant class with trio processing where parents are available. Candidates are prioritised against the structured ultrasound phenotype, classification is conservative given prenatal uncertainty, and delivery is compressed to fit the decision window.

WHAT IT PRODUCES

Contamination clearance, findings across all variant classes with inheritance where available, phenotype correlation, recurrence risk, and a report inside the decision window with prenatal uncertainty stated.

WHAT IT DETECTS

Sequence variants, copy number changes, structural rearrangements, repeat expansions and mitochondrial variants in one analysis, with trio inheritance where parents are available.

WHAT IT DOES NOT DETECT

Methylation and imprinting disorders, and the interpretation of non-coding findings remains extremely limited. The fetal phenotype constraint applies with even greater force, since a genome produces far more candidates against a phenotype that ultrasound describes only partially.

WHAT MAKES IT HARD

Genome breadth against partial fetal phenotype produces many plausible candidates and few certain ones. Non-coding findings are effectively uninterpretable prenatally, and the volume of uncertainty must be managed rather than transmitted to a family facing an irreversible decision.

WHO DOES THIS JOB

Senior clinical bioinformatician in prenatal genomics. Among the most demanding roles in this sector because analytical difficulty and decision pressure coincide.

WHAT YOU CAN DO AFTERWARDS

Run prenatal genome analysis across all variant classes, manage candidate volume against partial phenotype, and report prenatal uncertainty responsibly.

WHO HIRES FOR IT

National prenatal genome programmes, tertiary fetal medicine centres, and specialist prenatal diagnostic laboratories.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
26	**126 hours · 21 days**	Prior genome and prenatal exome analysis experience	**2,88,000**

SRP-12
WORKFLOW

Prenatal Genome Analysis

(**Fetal whole genome sequencing analysis** · Prenatal WGS · fetal genome)

COMPLETE WORKFLOW — 26 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Indication and phenotype capture**	Ultrasound findings captured in structured terms as prioritisation input	Structured HPO capture	*Structured fetal phenotype*
02	**Prior result verification**	Previous cytogenetic, microarray and exome results verified	Prior result review	*Prior result record*
03	**Consent and finding scope confirmation**	Scope including non-coding and incidental findings confirmed	Consent record	*Analysis authorisation*
04	**Decision window recording**	The pregnancy decision window recorded as the delivery constraint	Clinical record	*Timeline record*
05	**Sample adequacy assessment**	Fetal and parental sample adequacy assessed given limited material	Sample assessment	*Adequacy record*
06	**Maternal cell contamination assessment**	Contamination excluded before any fetal result is interpreted	MCC analysis	*Contamination clearance*
07	**Data receipt and quality assessment**	Integrity and sequence quality verified for all samples	md5sum, FastQC, MultiQC	*QC acceptance record*
08	**Read preprocessing**	Adapters and low-quality tails removed identically across samples	fastp	*Trimmed FASTQ*
09	**Alignment and processing**	All samples aligned and processed identically against a locked reference	bwa-mem2, Picard, GATK BQSR	*Analysis-ready BAMs*
10	**Relatedness verification**	Parental relationships verified before inheritance logic	somalier relate	*Pedigree verification*
11	**Coverage and callable region analysis**	Genome-wide coverage measured and callable footprint declared	mosdepth, CallableLoci	*Callable region BED*
12	**Small variant calling**	Variants called genome-wide with joint genotyping where trio data exist	Validated caller	*Genome VCF*
13	**Copy number calling**	Genome-wide copy number called with resolution stated	Copy number analysis	*Copy number findings*
14	**Structural variant calling**	Structural rearrangements called by ensemble with breakpoints resolved	Manta, Delly, SURVIVOR	*SV findings*
15	**Repeat expansion screening**	Pathogenic repeat loci genotyped against the catalogue	Repeat genotyping	*Repeat findings*
16	**Mitochondrial analysis**	Mitochondrial variants called with heteroplasmy quantified	Mitochondrial calling	*mtDNA findings*
17	**De novo variant identification**	De novo variants identified genome-wide with stringent filtering	Trio logic	*De novo candidates*
18	**De novo read-level review**	Every reportable de novo candidate reviewed at read level	IGV review	*Reviewed de novo set*
19	**Inheritance model filtering**	All inheritance models applied across coding and non-coding candidates	PED-aware filtering	*Model-filtered candidates*
20	**Annotation and population filtering**	All variant classes annotated with appropriate frequency thresholds	VEP, gnomAD, gnomAD-SV	*Annotated candidates*
21	**Phenotype-driven prioritisation**	Candidates ranked against the structured fetal phenotype	Exomiser, HPO terms	*Ranked candidates*
22	**Non-coding candidate restriction**	Non-coding findings restricted to phenotype-supported genes to bound uncertainty	Restriction policy	*Bounded non-coding set*
23	**Conservative classification**	Findings classified conservatively across all variant classes	ACMG-AMP, ACMG-ClinGen	*Classified findings*
24	**Multidisciplinary review**	Candidates reviewed with fetal medicine and clinical genetics	Scheduled review	*Review record*
25	**Orthogonal confirmation**	Reportable findings confirmed by class-appropriate methods within the window	Sanger, MLPA, ddPCR	*Confirmation record*
26	**Report generation and sign-out**	Findings, recurrence risk and prenatal uncertainty issued under dual signature	Validated report template	*Signed clinical report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
26	**126 h · 21 d**	**SRP-12**	**2,88,000**

SRP-13
SECTOR 01

Prenatal CNV & Digital Karyotyping Analysis

(**Prenatal copy number and chromosomal analysis** · Prenatal CNV · digital karyotype)

WHY IT IS DONE

Copy number analysis identifies the chromosomal and submicroscopic imbalances responsible for a substantial share of fetal abnormality, at resolution far beyond karyotype. It has become the standard first-tier diagnostic test on invasive prenatal samples.

WHAT IT ANSWERS

Does this fetus carry a chromosomal aneuploidy or a clinically significant copy number imbalance, and what is its expected phenotypic consequence?

WHEN IT IS DONE

As first-tier analysis on chorionic villus and amniotic fluid samples, after an abnormal ultrasound, following a positive screening result, and in pregnancy loss investigation.

WHAT TRIGGERS IT

An invasive prenatal sample, ultrasound abnormality, positive screening, or investigation of fetal demise.

HOW IT IS DONE

Maternal cell contamination is excluded first, then copy number is called genome-wide with resolution stated and mosaicism assessed. Findings are classified under copy number criteria with prenatal phenotype implications assessed, uncertain findings are handled under a policy agreed in advance, and parental testing is arranged where inheritance determines significance.

WHAT IT PRODUCES

Contamination clearance, copy number findings with resolution and mosaicism assessment, absence of heterozygosity results, classification with prenatal phenotype implications, and parental testing recommendations.

WHAT IT DETECTS

Aneuploidy, large and submicroscopic copy number imbalances, absence of heterozygosity indicating consanguinity or uniparental disomy, and mosaicism above the detectable threshold.

WHAT IT DOES NOT DETECT

Balanced rearrangements, low-level mosaicism below the detection floor, and single-gene disorders. Copy number changes of uncertain significance are common and their fetal consequence frequently cannot be predicted, which is the principal reporting difficulty.

WHAT MAKES IT HARD

Uncertain copy number findings are the recurring problem, because a family must make an irreversible decision on a variant whose consequence is unknown. Parental testing frequently resolves significance and must be arranged fast enough to matter within the decision window.

WHO DOES THIS JOB

Clinical bioinformatician in prenatal genomics, mid level, working with fetal medicine and genetic counselling on uncertain findings.

WHAT YOU CAN DO AFTERWARDS

Run prenatal copy number analysis with mosaicism assessment, classify under copy number criteria, and manage uncertain findings within a prenatal decision framework.

WHO HIRES FOR IT

Prenatal diagnostic laboratories, fetal medicine centres, cytogenetics services transitioning to sequencing, and hospital genetics laboratories.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
19	**92 hours · 16 days**	Prior copy number analysis experience	**2,10,000**

SRP-13
WORKFLOW

Prenatal CNV & Digital Karyotyping Analysis

(**Prenatal copy number and chromosomal analysis** · Prenatal CNV · digital karyotype)

COMPLETE WORKFLOW — 19 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Sample type and indication recording**	Sample type and clinical indication recorded with type-specific limitations	Requisition, sample record	*Sample record*
02	**Decision window recording**	The pregnancy decision window recorded as the delivery constraint	Clinical record	*Timeline record*
03	**Maternal comparator verification**	A maternal sample confirmed available for contamination assessment	Sample inventory	*Comparator verification*
04	**Maternal cell contamination assessment**	Contamination excluded before any fetal result is interpreted	MCC analysis	*Contamination clearance*
05	**Data receipt and quality assessment**	Integrity and data quality verified against acceptance criteria	md5sum, platform QC	*QC acceptance record*
06	**Coverage or signal extraction**	Depth or intensity extracted genome-wide at the design resolution	mosdepth, platform extraction	*Coverage data*
07	**Bias correction**	GC and mappability bias corrected before segmentation	Bias correction	*Corrected data*
08	**Segmentation**	Genome segmented into regions of consistent copy state	Segmentation algorithm	*Segments*
09	**Copy state assignment**	Integer copy states assigned with sex chromosome ploidy handled	Copy state assignment	*Copy number calls*
10	**Resolution determination**	Effective resolution determined and stated for the sample and design	Resolution calculation	*Resolution statement*
11	**Mosaicism assessment**	Mosaic copy number assessed with the detection floor stated	Mosaicism analysis	*Mosaicism findings*
12	**Absence of heterozygosity analysis**	Homozygous regions detected for consanguinity and uniparental disomy assessment	ROH detection	*AOH findings*
13	**Population and benign filtering**	Known benign copy number polymorphism removed	Population databases	*Filtered findings*
14	**ACMG-ClinGen classification**	Findings classified under copy number criteria with prenatal implications assessed	ACMG-ClinGen CNV framework	*Classified findings*
15	**Uncertain finding policy application**	Uncertain findings handled under the pre-agreed prenatal reporting policy	Reporting policy	*Policy application record*
16	**Parental testing arrangement**	Parental testing arranged where inheritance determines clinical significance	Parental testing protocol	*Parental testing record*
17	**Phenotype implication assessment**	Expected fetal phenotype assessed for each reportable finding	Clinical correlation	*Phenotype implications*
18	**Orthogonal confirmation**	Reportable findings confirmed by an independent method within the window	MLPA, qPCR, FISH	*Confirmation record*
19	**Report generation and sign-out**	Findings, resolution, mosaicism floor and implications issued under dual signature	Validated report template	*Signed clinical report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
19	**92 h · 16 d**	**SRP-13**	**2,10,000**

SRP-14
SECTOR 01

Maternal Cell Contamination Analysis

(**MCC assessment in prenatal samples** · MCC analysis)

WHY IT IS DONE

Every invasive prenatal sample can contain maternal cells, and a heavily contaminated sample produces a confidently normal result describing the mother rather than the fetus. It is the single most dangerous failure mode in prenatal diagnosis, and it is entirely preventable.

WHAT IT ANSWERS

Does this prenatal sample contain maternal cells, at what proportion, and is the fetal result therefore reliable?

WHEN IT IS DONE

On every chorionic villus, amniotic fluid and products of conception sample before any fetal result is interpreted or released.

WHAT TRIGGERS IT

Receipt of any invasive prenatal or pregnancy loss sample, or an unexpectedly normal result in a clinically abnormal pregnancy.

HOW IT IS DONE

Maternal and fetal samples are genotyped at a polymorphic marker panel and compared, with paternal alleles used where available to confirm fetal-specific material. Contamination proportion is estimated, assessed against the threshold appropriate to the intended analysis, and samples above threshold are rejected or reported with explicit qualification.

WHAT IT PRODUCES

Maternal and fetal genotype comparison, contamination proportion with bounds, fetal-specific allele confirmation, the analysis-specific threshold assessment, and an accept, qualify or reject determination.

WHAT IT DETECTS

Maternal cell presence with the proportion estimated, and where a paternal sample is available the fetal-specific alleles confirming genuinely fetal material.

WHAT IT DOES NOT DETECT

It cannot correct a contaminated result, only identify it, and in a female fetus with high maternal contamination the distinction becomes progressively harder. Very low contamination may be tolerable for some analyses and disqualifying for others, so the threshold is analysis-dependent rather than universal.

WHAT MAKES IT HARD

The threshold is not one number, because low-level contamination that is harmless for aneuploidy detection may be disqualifying for mosaicism or single-gene analysis. Female fetuses are the difficult case, and the marker panel must be informative enough to detect contamination at the level that matters.

WHO DOES THIS JOB

Variant scientist or clinical bioinformatician in prenatal genomics, entry to mid level. A small analysis with outsized consequence.

WHAT YOU CAN DO AFTERWARDS

Assess maternal cell contamination reliably, apply analysis-specific thresholds, confirm fetal-specific material, and make defensible accept or reject determinations.

WHO HIRES FOR IT

Prenatal diagnostic laboratories, fetal medicine services, cytogenetics laboratories, and reproductive genetics services.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
13	**62 hours · 11 days**	Prior genotype analysis experience	**1,40,000**

SRP-14
WORKFLOW

Maternal Cell Contamination Analysis

(**MCC assessment in prenatal samples** · MCC analysis)

COMPLETE WORKFLOW — 13 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Marker panel authorisation**	The polymorphic marker panel and its informativeness confirmed	Panel register	*Authorised marker panel*
02	**Analysis-specific threshold specification**	The contamination threshold appropriate to the intended analysis specified	Threshold criteria	*Threshold specification*
03	**Maternal sample verification**	A maternal comparator sample confirmed received and correctly attributed	Sample inventory	*Maternal sample verification*
04	**Paternal sample assessment**	Paternal sample availability assessed for fetal-specific allele confirmation	Sample inventory	*Paternal availability record*
05	**Fetal sample genotyping**	The prenatal sample genotyped across the marker panel	Genotyping	*Fetal genotypes*
06	**Maternal sample genotyping**	The maternal sample genotyped across the same marker panel	Genotyping	*Maternal genotypes*
07	**Informative marker identification**	Markers where maternal and fetal genotypes differ identified	Comparison analysis	*Informative markers*
08	**Informativeness adequacy assessment**	The number of informative markers assessed against detection requirements	Adequacy criteria	*Informativeness record*
09	**Contamination proportion estimation**	Maternal allele proportion estimated across informative markers	Proportion estimation	*Contamination estimate*
10	**Confidence bound derivation**	Confidence bounds derived for the contamination estimate	Confidence estimation	*Estimate bounds*
11	**Fetal-specific allele confirmation**	Paternal alleles confirmed present where a paternal sample is available	Allele confirmation	*Fetal material confirmation*
12	**Threshold assessment and determination**	The estimate assessed against the analysis-specific threshold	Threshold comparison	*Accept or reject determination*
13	**Reporting**	Contamination estimate, informativeness and determination assembled	Report template	*MCC report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
13	**62 h · 11 d**	**SRP-14**	**1,40,000**

SRP-15
SECTOR 01

Products of Conception & Pregnancy Loss Analysis

(**Miscarriage and fetal demise genomic analysis** · POC analysis)

WHY IT IS DONE

Around half of first-trimester losses are chromosomally abnormal, and establishing that gives a family an explanation and a recurrence risk. It also identifies the smaller group of losses caused by inherited rearrangements, where intervention can change future outcomes.

WHAT IT ANSWERS

Was there a chromosomal or genomic cause for this pregnancy loss, and what does it imply for recurrence?

WHEN IT IS DONE

After miscarriage, particularly recurrent loss, after stillbirth or fetal demise, and in couples undergoing recurrent loss investigation.

WHAT TRIGGERS IT

Pregnancy loss with tissue available, recurrent loss investigation, stillbirth, or termination for fetal abnormality.

HOW IT IS DONE

Tissue quality and degradation are assessed as the primary constraint, maternal cell contamination is excluded, and low-input analysis proceeds with methods tolerant of degraded material. Copy number and ploidy are determined, molar patterns and uniparental disomy are assessed, and recurrence implications are derived with parental karyotyping recommended where a rearrangement is suspected.

WHAT IT PRODUCES

Tissue quality and contamination assessment, ploidy and copy number findings, molar and uniparental disomy assessment, recurrence risk, and parental testing recommendations.

WHAT IT DETECTS

Aneuploidy, triploidy, copy number imbalances, molar pregnancy patterns, and uniparental disomy, with maternal cell contamination excluded.

WHAT IT DOES NOT DETECT

Balanced rearrangements, most single-gene causes unless specifically sought, and any cause in degraded tissue where DNA quality is insufficient. Losses with a non-genetic cause return normal results, which does not exclude a recurrent underlying problem.

WHAT MAKES IT HARD

Tissue is frequently degraded and heavily maternal, so a normal female result is the characteristic failure mode and must be actively excluded rather than accepted. Distinguishing genuine triploidy from technical artefact needs an allelic method rather than depth alone.

WHO DOES THIS JOB

Clinical bioinformatician in reproductive genomics, mid level, working with recurrent loss and fetal medicine services.

WHAT YOU CAN DO AFTERWARDS

Analyse degraded pregnancy loss material, exclude maternal contamination, determine ploidy reliably, and derive recurrence implications.

WHO HIRES FOR IT

Recurrent pregnancy loss services, fetal medicine centres, reproductive genetics laboratories, and IVF chains.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
17	**82 hours · 14 days**	Prior copy number and low-input analysis experience	**1,90,000**

SRP-15
WORKFLOW

Products of Conception & Pregnancy Loss Analysis

(**Miscarriage and fetal demise genomic analysis** · POC analysis)

COMPLETE WORKFLOW — 17 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Sample type and history recording**	Sample type, gestational age at loss and clinical history recorded	Requisition, clinical record	*Sample record*
02	**Tissue quality and degradation assessment**	Tissue degradation assessed as the primary constraint on analysis	Quality assessment	*Tissue quality record*
03	**Maternal comparator verification**	A maternal sample confirmed available for contamination assessment	Sample inventory	*Comparator verification*
04	**Maternal cell contamination assessment**	Contamination excluded, since maternal decidua frequently dominates the sample	MCC analysis	*Contamination assessment*
05	**Input quantification**	DNA yield measured and assessed against method requirements	Quantification	*Input record*
06	**Low-input method selection**	An analytical method tolerant of degraded low-yield material selected	Method selection	*Selected method*
07	**Data receipt and quality assessment**	Integrity and data quality verified against acceptance criteria	md5sum, platform QC	*QC acceptance record*
08	**Coverage extraction and correction**	Coverage extracted and corrected for bias in degraded material	Bias correction	*Corrected coverage*
09	**Copy number and ploidy analysis**	Copy number and overall ploidy determined across the genome	Copy number analysis	*Copy number findings*
10	**Allelic analysis**	Allele fractions analysed to detect triploidy and distinguish it from artefact	Allelic analysis	*Allelic findings*
11	**Triploidy determination**	Triploidy determined and its parental origin assessed where possible	Ploidy determination	*Triploidy assessment*
12	**Molar pregnancy pattern assessment**	Complete and partial molar patterns assessed from allelic and copy number data	Pattern analysis	*Molar assessment*
13	**Uniparental disomy assessment**	Uniparental disomy assessed from homozygosity and allelic patterns	UPD analysis	*UPD findings*
14	**Mosaicism assessment**	Mosaicism assessed with the detection floor stated for degraded material	Mosaicism analysis	*Mosaicism findings*
15	**Cross-loss pattern assessment**	Findings compared across previous losses where those results exist	Pattern comparison	*Cross-loss assessment*
16	**Recurrence implication derivation**	Recurrence risk derived with parental karyotyping recommended where indicated	Risk derivation	*Recurrence implications*
17	**Report generation and sign-out**	Findings, tissue quality, contamination and recurrence issued under dual signature	Validated report template	*Signed report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
17	**82 h · 14 d**	**SRP-15**	**1,90,000**

SRP-16
SECTOR 01

Preimplantation Genetic Testing for Aneuploidy

(**PGT-A embryo aneuploidy analysis** · PGT-A)

WHY IT IS DONE

Most embryos that fail to implant or miscarry are aneuploid. Testing a small biopsy before transfer allows euploid embryos to be prioritised, which reduces time to pregnancy and loss rate even though it does not change the number of viable embryos a patient has.

WHAT IT ANSWERS

Which embryos in this cohort are euploid, which are aneuploid, and which show mosaic or intermediate copy number profiles?

WHEN IT IS DONE

During an IVF cycle after trophectoderm biopsy at blastocyst stage, in advanced maternal age, recurrent implantation failure and recurrent loss.

WHAT TRIGGERS IT

An IVF cycle with embryo biopsy, advanced maternal age, recurrent implantation failure, or recurrent pregnancy loss.

HOW IT IS DONE

Whole genome amplification from a five to ten cell biopsy is quality controlled first because amplification failure dominates error, then copy number is called against a reference with amplification artefact modelled. Segmental resolution is stated, intermediate profiles are classified as mosaic against validated thresholds, and results are reported per embryo for transfer prioritisation.

WHAT IT PRODUCES

Per-embryo copy number profiles, euploid, aneuploid and mosaic classification, amplification quality metrics per biopsy, stated segmental resolution, and a transfer prioritisation report.

WHAT IT DETECTS

Whole chromosome aneuploidy, segmental imbalances above the resolution threshold, and intermediate copy number profiles reported as mosaic.

WHAT IT DOES NOT DETECT

Balanced rearrangements, single-gene disorders, polyploidy in many implementations, and any abnormality confined to cells not sampled by the biopsy. The biopsy is a handful of trophectoderm cells, so it represents the placenta rather than the embryo proper, which is the fundamental limitation.

WHAT MAKES IT HARD

Everything rests on amplification from a handful of cells, and amplification artefact mimics segmental imbalance convincingly. Mosaic classification is a threshold decision on a continuous signal, and embryos discarded on a mosaic call may have been viable, which makes threshold governance an ethical matter as much as a technical one.

WHO DOES THIS JOB

Clinical bioinformatician in reproductive genomics, mid to senior level, working within an IVF laboratory pathway.

WHAT YOU CAN DO AFTERWARDS

Analyse low-input embryo biopsies, model amplification artefact, classify mosaicism against validated thresholds, and support transfer prioritisation defensibly.

WHO HIRES FOR IT

IVF chains and fertility clinics, preimplantation genetics laboratories, reproductive genetics services, and embryology reference laboratories.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
22	**108 hours · 18 days**	Prior copy number and low-input analysis experience	**2,45,000**

SRP-16
WORKFLOW

Preimplantation Genetic Testing for Aneuploidy

(**PGT-A embryo aneuploidy analysis** · PGT-A)

COMPLETE WORKFLOW — 22 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Cycle and biopsy record capture**	Cycle identifiers, biopsy day and embryo identifiers recorded and linked	Embryology record	*Cycle record*
02	**Biopsy tracking verification**	Chain of custody from embryo to tube verified, since misattribution is unrecoverable	Tracking verification	*Custody record*
03	**Amplification performance assessment**	Whole genome amplification yield and profile assessed per biopsy	Amplification QC	*Amplification record*
04	**Amplification failure exclusion**	Biopsies with failed amplification excluded rather than analysed	Failure criteria	*Exclusion record*
05	**Data receipt and quality assessment**	Integrity and sequence quality verified per biopsy	md5sum, FastQC	*QC acceptance record*
06	**Alignment**	Reads aligned with settings appropriate to amplified low-input material	bwa-mem2	*Aligned BAM*
07	**Coverage uniformity assessment**	Uniformity assessed per biopsy since amplification bias mimics imbalance	Uniformity analysis	*Uniformity metrics*
08	**Reference set normalisation**	Coverage normalised against a reference set processed identically	Reference normalisation	*Normalised coverage*
09	**GC and amplification bias correction**	GC and amplification-specific bias corrected before segmentation	Bias correction	*Corrected coverage*
10	**Segmentation**	Genome segmented into regions of consistent copy state	Segmentation algorithm	*Segments*
11	**Whole chromosome copy state assignment**	Whole chromosome copy states assigned per embryo	Copy state assignment	*Chromosome calls*
12	**Segmental analysis**	Segmental imbalances assessed with the resolution limit stated	Segmental analysis	*Segmental findings*
13	**Resolution statement**	The minimum reliably detectable segment size stated for the assay	Resolution calculation	*Resolution statement*
14	**Intermediate value quantification**	Intermediate copy number levels quantified against validated thresholds	Level quantification	*Intermediate levels*
15	**Mosaic classification**	Intermediate profiles classified as mosaic under documented threshold policy	Classification policy	*Mosaic classification*
16	**Noise and artefact discrimination**	Amplification noise assessed as an alternative explanation for intermediate values	Noise assessment	*Discrimination record*
17	**Sex chromosome assessment**	Sex chromosome copy state assessed within the consented reporting scope	Sex chromosome analysis	*Sex chromosome findings*
18	**Per-embryo quality gating**	Each embryo result gated on its own amplification and uniformity metrics	Per-biopsy gating	*Quality gate record*
19	**No-result determination**	Embryos with inadequate data reported as no-result rather than as normal	No-result criteria	*No-result decisions*
20	**Transfer prioritisation**	Embryos ranked for transfer under the clinic's documented prioritisation policy	Prioritisation policy	*Transfer ranking*
21	**Report generation**	Per-embryo results, quality metrics and resolution assembled	Validated report template	*Draft report*
22	**Clinical sign-out**	Report reviewed and authorised under dual signature	Sign-out procedure	*Signed PGT report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
22	**108 h · 18 d**	**SRP-16**	**2,45,000**

SRP-17
SECTOR 01

Preimplantation Genetic Testing for Monogenic Disease

(**PGT-M embryo single-gene analysis** · PGT-M)

WHY IT IS DONE

Couples at known risk of a serious monogenic condition can avoid transmitting it entirely by transferring only unaffected embryos, without ever facing a decision about an established pregnancy. It is the intervention that removes the choice families most dread.

WHAT IT ANSWERS

Which embryos in this cohort have inherited the familial pathogenic variant or variants, and which are unaffected or carriers?

WHEN IT IS DONE

In IVF cycles for couples with a known familial monogenic condition, after a previous affected child, and in carrier couples choosing preimplantation testing over prenatal diagnosis.

WHAT TRIGGERS IT

A characterised familial pathogenic variant with an IVF pathway, a previous affected child, or carrier couple election.

HOW IT IS DONE

A pre-cycle workup characterises the familial variants and constructs informative marker haplotypes using family reference samples. Embryo biopsies are amplified with quality control, direct variant status and linked marker haplotypes are both determined, and concordance between the two is required before any embryo is called.

WHAT IT PRODUCES

The pre-cycle haplotype workup with informative marker set, per-embryo direct and haplotype-based results with concordance assessment, allele dropout assessment, concurrent aneuploidy results, and a transfer recommendation report.

WHAT IT DETECTS

Direct variant status per embryo where amplification permits, and haplotype-based inheritance using linked markers around the locus, with aneuploidy assessed concurrently in most workflows.

WHAT IT DOES NOT DETECT

Any condition other than the one tested, and it cannot exclude de novo events elsewhere in the embryo. Allele dropout during amplification can produce a false unaffected call, which is why direct variant detection alone is never sufficient.

WHAT MAKES IT HARD

Allele dropout is the defining risk and is why linked markers rather than direct detection alone must carry the call. The pre-cycle workup determines whether the test is possible at all, and families sometimes lack the reference samples needed to build informative haplotypes.

WHO DOES THIS JOB

Senior clinical bioinformatician or clinical scientist in preimplantation genetics, working with genetic counselling and embryology.

WHAT YOU CAN DO AFTERWARDS

Build informative haplotypes from family samples, analyse embryo biopsies with dropout control, require direct and haplotype concordance, and issue transfer recommendations.

WHO HIRES FOR IT

Preimplantation genetics laboratories, IVF chains, reproductive genetics services, and specialist fertility centres.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
24	**118 hours · 20 days**	Prior haplotype and low-input analysis experience	**2,70,000**

SRP-17
WORKFLOW

Preimplantation Genetic Testing for Monogenic Disease

(**PGT-M embryo single-gene analysis** · PGT-M)

COMPLETE WORKFLOW — 24 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Pre-cycle referral assessment**	The familial condition and variants assessed for testability before the cycle	Referral review	*Feasibility assessment*
02	**Familial variant characterisation**	Familial pathogenic variants characterised at sequence level from prior testing	Prior test results	*Variant characterisation*
03	**Family reference sample assessment**	Reference samples required for haplotype construction assessed for availability	Sample inventory	*Reference availability*
04	**Informative marker selection**	Polymorphic markers flanking the locus selected on both sides	Marker selection	*Informative marker set*
05	**Family genotyping**	Family reference samples genotyped across the marker set	Genotyping	*Family genotypes*
06	**Haplotype construction**	Disease and normal haplotypes constructed and phase established	Haplotype phasing	*Constructed haplotypes*
07	**Pre-cycle workup sign-off**	The workup confirmed sufficient before the clinical cycle commences	Workup criteria	*Workup sign-off*
08	**Cycle and biopsy record capture**	Cycle, biopsy day and embryo identifiers recorded and linked	Embryology record	*Cycle record*
09	**Biopsy tracking verification**	Chain of custody from embryo to tube verified	Tracking verification	*Custody record*
10	**Amplification performance assessment**	Amplification yield and profile assessed per biopsy	Amplification QC	*Amplification record*
11	**Data receipt and quality assessment**	Integrity and sequence quality verified per biopsy	md5sum, FastQC	*QC acceptance record*
12	**Direct variant genotyping**	The familial variant genotyped directly in each embryo biopsy	Targeted genotyping	*Direct variant results*
13	**Marker haplotype genotyping**	Linked markers genotyped in each biopsy to establish haplotype inheritance	Marker genotyping	*Marker results*
14	**Allele dropout assessment**	Dropout assessed at each marker and at the variant position	Dropout analysis	*Dropout assessment*
15	**Haplotype-based inheritance determination**	Inheritance determined from linked marker haplotypes independently of direct genotyping	Haplotype logic	*Haplotype-based genotype*
16	**Concordance requirement application**	Direct and haplotype-based results required to agree before any call	Concordance criteria	*Concordance record*
17	**Recombination assessment**	Recombination between flanking markers assessed as a misassignment risk	Recombination analysis	*Recombination assessment*
18	**Informative marker count assessment**	Sufficient informative markers on both sides confirmed per embryo	Adequacy criteria	*Marker adequacy record*
19	**Concurrent aneuploidy analysis**	Aneuploidy assessed concurrently in the same biopsy where the workflow supports it	Copy number analysis	*Aneuploidy results*
20	**Per-embryo quality gating**	Each embryo gated on amplification, dropout and marker adequacy	Per-biopsy gating	*Quality gate record*
21	**No-result determination**	Embryos with discordant or inadequate data reported as no-result	No-result criteria	*No-result decisions*
22	**Transfer recommendation**	Unaffected embryos identified and ranked for transfer	Recommendation policy	*Transfer recommendation*
23	**Report generation**	Per-embryo genotype, haplotype evidence and dropout assessment assembled	Validated report template	*Draft report*
24	**Clinical sign-out**	Report reviewed and authorised under dual signature	Sign-out procedure	*Signed PGT report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
24	**118 h · 20 d**	**SRP-17**	**2,70,000**

SRP-18
SECTOR 01

Preimplantation Testing for Structural Rearrangement

(**PGT-SR embryo rearrangement analysis** · PGT-SR)

WHY IT IS DONE

Balanced translocation and inversion carriers are healthy but produce a high proportion of unbalanced gametes, causing repeated loss and affected pregnancies. Selecting embryos without the unbalanced derivative addresses the cause directly.

WHAT IT ANSWERS

Which embryos carry an unbalanced product of the parental rearrangement, and which are balanced or normal?

WHEN IT IS DONE

In IVF cycles for carriers of a balanced translocation, inversion or other structural rearrangement, typically after recurrent loss or an affected pregnancy.

WHAT TRIGGERS IT

A known parental balanced rearrangement with an IVF pathway, recurrent pregnancy loss in a carrier couple, or a previous unbalanced pregnancy.

HOW IT IS DONE

The parental rearrangement is characterised with breakpoints defined as precisely as available methods permit, then embryo biopsies are amplified and copy number called with attention to the specific segments involved. Segments near breakpoints are assessed carefully because resolution limits detection there, and carrier status is determined where the method supports it.

WHAT IT PRODUCES

The parental rearrangement characterisation, per-embryo segmental copy number with the derivative segments assessed, aneuploidy results, carrier status where determinable, and a transfer recommendation report.

WHAT IT DETECTS

Unbalanced products of the parental rearrangement as segmental gains and losses, concurrent whole chromosome aneuploidy, and with breakpoint-informative methods the distinction between balanced carrier and genuinely normal embryos.

WHAT IT DOES NOT DETECT

Standard copy number methods cannot distinguish a balanced carrier embryo from a normal one, since both have normal copy number. Distinguishing them requires breakpoint-spanning or haplotype-based methods that are not universally available.

WHAT MAKES IT HARD

Small derivative segments near the resolution limit are exactly the ones that matter, so segmental resolution around the specific breakpoints must be established rather than assumed from a global claim. Carrier discrimination requires methods many laboratories do not run, and its absence should be stated rather than implied.

WHO DOES THIS JOB

Clinical bioinformatician in preimplantation genetics, mid to senior level, working with cytogenetics on rearrangement characterisation.

WHAT YOU CAN DO AFTERWARDS

Characterise parental rearrangements, assess derivative segments at appropriate resolution, determine carrier status where possible, and support transfer decisions.

WHO HIRES FOR IT

Preimplantation genetics laboratories, IVF chains, cytogenetics services, and reproductive genetics centres.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
20	**98 hours · 17 days**	Prior copy number and structural variant experience	**2,25,000**

SRP-18
WORKFLOW

Preimplantation Testing for Structural Rearrangement

(**PGT-SR embryo rearrangement analysis** · PGT-SR)

COMPLETE WORKFLOW — 20 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Parental rearrangement characterisation**	The parental rearrangement characterised with breakpoints defined as precisely as possible	Cytogenetic and molecular records	*Rearrangement characterisation*
02	**Derivative segment definition**	The segments gained or lost in unbalanced products defined explicitly	Segment analysis	*Derivative segment definitions*
03	**Breakpoint resolution assessment**	Assay resolution around the specific breakpoints assessed before the cycle	Resolution analysis	*Breakpoint resolution record*
04	**Feasibility determination**	Testability confirmed given the segment sizes involved	Feasibility criteria	*Feasibility record*
05	**Cycle and biopsy record capture**	Cycle, biopsy day and embryo identifiers recorded and linked	Embryology record	*Cycle record*
06	**Biopsy tracking verification**	Chain of custody from embryo to tube verified	Tracking verification	*Custody record*
07	**Amplification performance assessment**	Amplification yield and profile assessed per biopsy	Amplification QC	*Amplification record*
08	**Data receipt and quality assessment**	Integrity and sequence quality verified per biopsy	md5sum, FastQC	*QC acceptance record*
09	**Alignment and normalisation**	Reads aligned and coverage normalised against a matched reference set	bwa-mem2, normalisation	*Normalised coverage*
10	**Bias correction**	GC and amplification bias corrected before segmentation	Bias correction	*Corrected coverage*
11	**Targeted segment analysis**	The specific derivative segments analysed at maximum available resolution	Targeted segmental analysis	*Segment copy states*
12	**Genome-wide copy number analysis**	Whole chromosome aneuploidy assessed concurrently across the genome	Copy number analysis	*Aneuploidy results*
13	**Unbalanced product determination**	Embryos carrying unbalanced derivative products identified	Determination logic	*Unbalanced determinations*
14	**Carrier discrimination assessment**	Balanced carrier status assessed where the method supports it	Breakpoint or haplotype method	*Carrier status*
15	**Carrier discrimination limitation statement**	Inability to distinguish carrier from normal stated where applicable	Limitation template	*Limitation statement*
16	**Per-embryo quality gating**	Each embryo gated on amplification and segmental resolution achieved	Per-biopsy gating	*Quality gate record*
17	**No-result determination**	Embryos with inadequate resolution at critical segments reported as no-result	No-result criteria	*No-result decisions*
18	**Transfer recommendation**	Balanced or normal embryos identified and ranked for transfer	Recommendation policy	*Transfer recommendation*
19	**Report generation**	Per-embryo segment results, resolution and carrier status assembled	Validated report template	*Draft report*
20	**Clinical sign-out**	Report reviewed and authorised under dual signature	Sign-out procedure	*Signed PGT report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
20	**98 h · 17 d**	**SRP-18**	**2,25,000**

SRP-19
SECTOR 01

Embryo Mosaicism Analysis

(**Intermediate copy number and embryo mosaicism assessment** · Mosaic embryo analysis)

WHY IT IS DONE

A substantial proportion of embryo biopsies return intermediate copy number values that are neither clearly euploid nor clearly aneuploid. How these are classified determines whether embryos are transferred or discarded, and mosaic-classified embryos do produce healthy births.

WHAT IT ANSWERS

Does this intermediate copy number profile represent genuine mosaicism, technical noise, or a true whole-cell abnormality, and what transfer priority follows?

WHEN IT IS DONE

Within every PGT-A workflow where intermediate values arise, and in reviewing classification thresholds across a laboratory's embryo cohort.

WHAT TRIGGERS IT

An intermediate copy number result on embryo biopsy, or a laboratory review of mosaic classification policy.

HOW IT IS DONE

Amplification quality is assessed first because noise and mosaicism are confounded, then intermediate levels are quantified against validated thresholds derived from control material. Whole chromosome and segmental profiles are distinguished, the chromosome involved is assessed for viability implications, and classification is applied under a documented laboratory policy.

WHAT IT PRODUCES

Quantified intermediate levels with amplification quality context, whole chromosome against segmental classification, chromosome-specific viability assessment, threshold policy application, and a transfer prioritisation recommendation.

WHAT IT DETECTS

Intermediate copy number levels with the proportion estimated, the chromosome and segment involved, and the distinction between whole chromosome and segmental intermediate profiles.

WHAT IT DOES NOT DETECT

It cannot determine whether the rest of the embryo shares the finding, because only the biopsied cells were measured. Technical noise from amplification produces intermediate values indistinguishable from biological mosaicism in a single biopsy, and no method resolves this definitively.

WHAT MAKES IT HARD

Technical noise and true mosaicism are genuinely confounded in a single small biopsy, so thresholds must be derived from control material rather than chosen by convention. The consequences are asymmetric and irreversible, since a discarded embryo cannot be recovered while a transferred mosaic embryo frequently produces a healthy child.

WHO DOES THIS JOB

Senior clinical bioinformatician in preimplantation genetics, working with the laboratory director on classification policy.

WHAT YOU CAN DO AFTERWARDS

Distinguish amplification noise from biological mosaicism, derive thresholds from control material, and apply classification policy consistently and defensibly.

WHO HIRES FOR IT

Preimplantation genetics laboratories, IVF chains, reproductive genetics services, and fertility research programmes.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
17	**84 hours · 14 days**	Prior PGT-A analysis experience	**1,95,000**

SRP-19
WORKFLOW

Embryo Mosaicism Analysis

(**Intermediate copy number and embryo mosaicism assessment** · Mosaic embryo analysis)

COMPLETE WORKFLOW — 17 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Classification policy verification**	The laboratory's mosaic classification thresholds and policy confirmed	Policy register	*Authorised policy*
02	**Threshold derivation evidence review**	Evidence deriving thresholds from control material reviewed rather than assumed	Validation records	*Threshold evidence*
03	**Amplification quality assessment**	Per-biopsy amplification quality assessed as the primary confounder	Amplification QC	*Amplification record*
04	**Noise floor determination**	The technical noise floor determined from control material for this assay	Noise analysis	*Noise floor*
05	**Coverage uniformity assessment**	Uniformity assessed per biopsy since non-uniformity produces intermediate values	Uniformity analysis	*Uniformity metrics*
06	**Intermediate value identification**	Copy number values falling between euploid and aneuploid identified	Value analysis	*Intermediate values*
07	**Level quantification**	The intermediate level quantified with confidence bounds	Quantification	*Quantified levels*
08	**Whole chromosome and segmental discrimination**	Whole chromosome and segmental intermediate profiles distinguished	Profile analysis	*Profile classification*
09	**Noise discrimination**	Each intermediate value assessed against the noise floor for this biopsy	Noise comparison	*Discrimination record*
10	**Chromosome-specific viability assessment**	The chromosome involved assessed for viability and phenotype implications	Chromosome-specific criteria	*Viability assessment*
11	**Level banding**	Intermediate levels banded under the documented classification policy	Banding policy	*Level bands*
12	**Classification assignment**	Mosaic classification assigned with the band and evidence recorded	Classification logic	*Mosaic classification*
13	**Sampling limitation statement**	The limitation that only biopsied cells were measured stated explicitly	Limitation template	*Limitation statement*
14	**Transfer priority derivation**	Transfer priority derived under policy considering level, chromosome and cohort	Prioritisation policy	*Transfer priority*
15	**Counselling information assembly**	Information on mosaic embryo outcomes assembled for counselling	Outcome references	*Counselling information*
16	**Cohort context assessment**	The embryo assessed in the context of the full cohort available for transfer	Cohort review	*Cohort context*
17	**Reporting**	Levels, classification, limitations and priority assembled	Report template	*Mosaicism report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
17	**84 h · 14 d**	**SRP-19**	**1,95,000**

SRP-20
SECTOR 01

Whole Genome Amplification Quality & Low-Input Analysis

(**Low-input amplification quality control** · WGA QC · low-input analysis)

WHY IT IS DONE

Every preimplantation and single-cell reproductive analysis begins with amplifying a few picograms of DNA by several orders of magnitude. The artefacts that process introduces are the dominant source of error downstream, and detecting them is what separates a reliable embryo result from a plausible-looking one.

WHAT IT ANSWERS

Did amplification of this low-input sample succeed, what artefacts did it introduce, and is the resulting data fit for the analysis intended?

WHEN IT IS DONE

Within every preimplantation genetic testing workflow, in single-cell and polar body analysis, and in degraded or minimal-input reproductive samples.

WHAT TRIGGERS IT

Any analysis beginning from single-cell or few-cell input, an anomalous downstream result requiring explanation, or amplification method validation.

HOW IT IS DONE

Amplification yield and fragment profile are assessed, then coverage uniformity is measured genome-wide with dropout regions mapped. Allele dropout rate is estimated at heterozygous control positions, amplification bias and chimeric artefacts are quantified, and the data are assessed against the specific requirements of the intended downstream analysis.

WHAT IT PRODUCES

Amplification yield and profile assessment, coverage uniformity metrics with dropout maps, allele dropout rate estimate, bias and artefact quantification, contamination screening, and a fitness-for-purpose determination.

WHAT IT DETECTS

Amplification success or failure, coverage uniformity and dropout regions, allele dropout, preferential amplification bias, chimeric artefacts, and contamination introduced during amplification.

WHAT IT DOES NOT DETECT

It cannot recover information lost to allele dropout, only identify that dropout occurred. It also cannot distinguish an amplification artefact from a genuine biological finding in every case, which is why downstream calls near thresholds remain uncertain.

WHAT MAKES IT HARD

Fitness for purpose is analysis-specific, since data adequate for whole chromosome aneuploidy may be entirely inadequate for segmental calling or single-gene analysis. Allele dropout is invisible without heterozygous control positions, so the assessment must be designed in rather than inferred afterwards.

WHO DOES THIS JOB

Clinical bioinformatician in preimplantation or single-cell genomics, mid level.

WHAT YOU CAN DO AFTERWARDS

Assess amplification quality quantitatively, map dropout, estimate allele dropout rate, and make analysis-specific fitness determinations.

WHO HIRES FOR IT

Preimplantation genetics laboratories, IVF chains, single-cell technology providers, and reproductive genetics services.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
15	**72 hours · 12 days**	Prior low-input sequencing analysis experience	**1,65,000**

SRP-20
WORKFLOW

Whole Genome Amplification Quality & Low-Input Analysis

(**Low-input amplification quality control** · WGA QC · low-input analysis)

COMPLETE WORKFLOW — 15 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Input material recording**	Cell number, source and collection method recorded as the analytical starting point	Sample record	*Input record*
02	**Amplification method recording**	The amplification chemistry and protocol version recorded	Protocol record	*Method record*
03	**Yield measurement**	Amplification yield measured against the expected range for the input	Quantification	*Yield record*
04	**Fragment profile assessment**	Amplified product fragment profile assessed for characteristic failure patterns	Fragment analysis	*Fragment profile*
05	**Negative control assessment**	No-template controls assessed for contamination introduced during amplification	Control analysis	*Contamination assessment*
06	**Data receipt and quality assessment**	Sequence quality verified against acceptance criteria	FastQC, MultiQC	*QC acceptance record*
07	**Alignment**	Reads aligned with settings appropriate to amplified material	bwa-mem2	*Aligned BAM*
08	**Coverage uniformity measurement**	Genome-wide coverage uniformity measured against uniformity benchmarks	Uniformity analysis	*Uniformity metrics*
09	**Dropout region mapping**	Regions with no coverage mapped as amplification dropout	Dropout analysis	*Dropout map*
10	**Allele dropout estimation**	Allele dropout rate estimated at heterozygous control positions	Dropout estimation	*Allele dropout rate*
11	**Preferential amplification assessment**	Systematic allelic bias assessed at heterozygous positions	Bias analysis	*Amplification bias*
12	**Chimeric artefact quantification**	Chimeric molecules produced during amplification identified and quantified	Chimera analysis	*Chimera rate*
13	**Contamination screening**	Foreign human and non-human DNA screened for in the amplified product	Contamination screening	*Contamination result*
14	**Analysis-specific requirement comparison**	Metrics compared against the requirements of the intended downstream analysis	Requirement criteria	*Requirement comparison*
15	**Fitness-for-purpose determination**	A documented determination made on whether the data support the intended analysis	Determination criteria	*Fitness determination*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
15	**72 h · 12 d**	**SRP-20**	**1,65,000**

SRP-21

SECTOR 01

Sperm & Male Infertility Genomic Analysis

(**Male factor infertility genomic analysis** · Male infertility panel)

WHY IT IS DONE

Male factor contributes to around half of infertility, and a meaningful proportion has an identifiable genetic cause. Establishing that cause changes treatment selection, predicts surgical sperm retrieval success, and identifies risks transmissible to offspring.

WHAT IT ANSWERS

Is there a genetic cause for this man's impaired spermatogenesis or obstruction, and what does it mean for treatment and for any children conceived?

WHEN IT IS DONE

In azoospermia and severe oligozoospermia, before surgical sperm retrieval, in congenital absence of the vas deferens, and in unexplained male infertility.

WHAT TRIGGERS IT

Azoospermia or severe oligozoospermia on semen analysis, planned surgical sperm retrieval, congenital vas deferens absence, or recurrent IVF failure with male factor.

HOW IT IS DONE

Y chromosome deletion regions are interrogated with the required marker set including partial deletion assessment, karyotype-level imbalances are assessed, and cystic fibrosis gene analysis includes intronic and polymorphic repeat alleles that standard panels miss. Monogenic spermatogenic failure genes are analysed and findings interpreted for retrieval prognosis and transmission risk.

WHAT IT PRODUCES

Y chromosome deletion findings with region specificity, chromosomal findings, cystic fibrosis gene results including repeat alleles, monogenic findings, retrieval prognosis interpretation, and transmission risk to offspring.

WHAT IT DETECTS

Y chromosome microdeletions in the azoospermia factor regions, karyotype-level abnormalities, cystic fibrosis gene variants including intronic and repeat alleles, and monogenic spermatogenic failure variants.

WHAT IT DOES NOT DETECT

Acquired and environmental causes, epigenetic causes of spermatogenic failure, and sperm DNA fragmentation, which requires a functional rather than genomic assay. Many cases remain unexplained after complete genomic workup, and this should be stated rather than implied as a normal result.

WHAT MAKES IT HARD

Y deletion region specificity determines prognosis, since complete deletion of one region predicts unsuccessful retrieval while another does not, so reporting a generic deletion is clinically inadequate. Cystic fibrosis gene analysis in this context requires intronic and repeat allele assessment that routine panels omit.

WHO DOES THIS JOB

Clinical bioinformatician in reproductive genomics, mid level, working with andrology and fertility services.

WHAT YOU CAN DO AFTERWARDS

Analyse Y chromosome deletions with region specificity, perform extended cystic fibrosis gene analysis, and interpret findings for retrieval prognosis and transmission.

WHO HIRES FOR IT

IVF and andrology services, fertility chains, reproductive genetics laboratories, and urology genetics services.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
18	**88 hours · 15 days**	Prior germline variant and copy number analysis experience	**2,05,000**

SRP-21
WORKFLOW

Sperm & Male Infertility Genomic Analysis

(**Male factor infertility genomic analysis** · Male infertility panel)

COMPLETE WORKFLOW — 18 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Indication and semen parameter capture**	Semen analysis findings and clinical indication captured as interpretive context	Andrology report	*Clinical record*
02	**Consent and scope confirmation**	Scope including transmission risk findings confirmed with the patient	Consent record	*Analysis authorisation*
03	**Data receipt and quality assessment**	Integrity and sequence quality verified against acceptance criteria	md5sum, FastQC	*QC acceptance record*
04	**Alignment and processing**	Reads aligned, duplicate marked and recalibrated	bwa-mem2, Picard, GATK BQSR	*Analysis-ready BAM*
05	**Coverage certification**	Depth certified across all target regions including Y chromosome loci	mosdepth	*Coverage certificate*
06	**Identity and sex verification**	Fingerprint captured and chromosomal sex verified	somalier, sex determination	*Identity clearance*
07	**Y chromosome region interrogation**	Azoospermia factor regions interrogated with the required marker set	Region-specific analysis	*Y region results*
08	**Partial deletion assessment**	Partial deletions within regions assessed, since they carry different prognosis	Partial deletion analysis	*Partial deletion findings*
09	**Region specificity determination**	The specific regions deleted determined rather than reporting a generic deletion	Specificity analysis	*Region specificity*
10	**Y chromosome copy number assessment**	Copy number variation across the Y chromosome assessed	Copy number analysis	*Y copy number findings*
11	**Chromosomal abnormality assessment**	Karyotype-level imbalances assessed from sequencing data	Copy number analysis	*Chromosomal findings*
12	**Cystic fibrosis gene analysis**	Sequence variants called across the gene with full exon coverage	Validated caller	*CFTR variants*
13	**Intronic and repeat allele analysis**	Intronic variants and polymorphic repeat alleles analysed with dedicated methods	Repeat and intronic analysis	*Repeat allele results*
14	**Monogenic gene analysis**	Spermatogenic failure genes analysed and variants classified	Validated caller, ACMG-AMP	*Monogenic findings*
15	**Retrieval prognosis interpretation**	Findings interpreted for surgical sperm retrieval prognosis	Clinical interpretation	*Prognosis interpretation*
16	**Transmission risk assessment**	Risk of transmission to offspring assessed for each finding	Risk assessment	*Transmission risk*
17	**Unexplained case statement**	Cases remaining unexplained stated as such rather than reported as normal	Reporting policy	*Unexplained statement*
18	**Report generation and sign-out**	Findings, prognosis and transmission risk issued under dual signature	Validated report template	*Signed report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
18	**88 h · 15 d**	**SRP-21**	**2,05,000**

SRP-22
SECTOR 01

Female Infertility & Ovarian Insufficiency Analysis

(**Female factor infertility genomic analysis** · POI · female infertility panel)

WHY IT IS DONE

Premature ovarian insufficiency and unexplained female infertility have identifiable genetic causes in a significant minority of cases. Identifying them alters fertility preservation timing, predicts response to stimulation, and detects the premutation carriers whose families face wider risk.

WHAT IT ANSWERS

Is there a genetic explanation for this woman's ovarian insufficiency or infertility, and what does it imply for fertility preservation and for her family?

WHEN IT IS DONE

In premature ovarian insufficiency, diminished ovarian reserve at a young age, poor response to stimulation, and unexplained infertility with a suggestive family history.

WHAT TRIGGERS IT

Premature ovarian insufficiency, unexpectedly diminished reserve, poor stimulation response, or a family history of early menopause.

HOW IT IS DONE

FMR1 repeat status is assessed with sizing limitations recognised and orthogonal confirmation arranged for expanded results, X chromosome and autosomal imbalances are assessed, and monogenic insufficiency genes are analysed. Findings are interpreted for fertility preservation timing and for family implications, particularly where a premutation is identified.

WHAT IT PRODUCES

FMR1 repeat status with sizing method and limitations, chromosomal findings, monogenic variant classification, fertility preservation implications, and family cascade recommendations for premutation carriers.

WHAT IT DETECTS

FMR1 premutation range repeats, karyotype-level abnormalities including X chromosome imbalances, monogenic ovarian insufficiency variants, and DNA repair gene variants associated with reduced reserve.

WHAT IT DOES NOT DETECT

Autoimmune, iatrogenic and environmental causes, and the majority of cases remain genetically unexplained. Repeat sizing from short reads is limited, so premutation range determination requires an orthogonal method for accurate sizing.

WHAT MAKES IT HARD

FMR1 premutation carries implications far beyond fertility, affecting the woman's own later neurological risk and her relatives' risk of fragile X syndrome, so it requires a counselling pathway rather than a laboratory report alone. Accurate repeat sizing needs a method short reads cannot provide.

WHO DOES THIS JOB

Clinical bioinformatician in reproductive genomics, mid level, with genetic counselling integral to premutation reporting.

WHAT YOU CAN DO AFTERWARDS

Assess FMR1 status with appropriate sizing methods, analyse ovarian insufficiency genes, and manage the family implications of premutation findings.

WHO HIRES FOR IT

Fertility and IVF chains, reproductive endocrinology services, reproductive genetics laboratories, and genetic counselling services.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
17	**82 hours · 14 days**	Prior germline variant and repeat analysis experience	**1,90,000**

SRP-22
WORKFLOW

Female Infertility & Ovarian Insufficiency Analysis

(**Female factor infertility genomic analysis** · POI · female infertility panel)

COMPLETE WORKFLOW — 17 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Indication and reserve parameter capture**	Ovarian reserve markers and clinical indication captured as context	Clinical record	*Clinical record*
02	**Consent and scope confirmation**	Scope including premutation and family implications confirmed	Consent record	*Analysis authorisation*
03	**Family history capture**	Family history of early menopause and fragile X captured	Structured capture	*Family history record*
04	**Data receipt and quality assessment**	Integrity and sequence quality verified against acceptance criteria	md5sum, FastQC	*QC acceptance record*
05	**Alignment and processing**	Reads aligned, duplicate marked and recalibrated	bwa-mem2, Picard, GATK BQSR	*Analysis-ready BAM*
06	**Coverage certification**	Depth certified across the gene set and repeat regions	mosdepth	*Coverage certificate*
07	**Identity verification**	Fingerprint captured to confirm attribution	somalier extract	*Identity clearance*
08	**FMR1 repeat genotyping**	The repeat locus genotyped with sizing limitations recognised	Repeat genotyping	*Repeat genotype*
09	**Repeat sizing limitation statement**	Short-read sizing limitations stated with orthogonal confirmation arranged	Limitation template	*Sizing limitation*
10	**Orthogonal repeat sizing**	Expanded results routed to a method capable of accurate sizing	Repeat-primed PCR, Southern blot	*Confirmed sizing*
11	**X chromosome copy number analysis**	X chromosome imbalances and mosaic monosomy assessed	Copy number analysis	*X chromosome findings*
12	**Autosomal chromosomal assessment**	Karyotype-level autosomal imbalances assessed	Copy number analysis	*Chromosomal findings*
13	**Monogenic gene analysis**	Ovarian insufficiency genes analysed and variants classified	Validated caller, ACMG-AMP	*Monogenic findings*
14	**DNA repair gene assessment**	Repair gene variants associated with reduced reserve assessed	Gene analysis	*Repair gene findings*
15	**Fertility preservation implication derivation**	Findings interpreted for fertility preservation timing and approach	Clinical interpretation	*Preservation implications*
16	**Family cascade assessment**	Family implications assessed with cascade recommendations for premutation carriers	Cascade protocol	*Cascade recommendations*
17	**Report generation and sign-out**	Findings, sizing method, implications and cascade issued under dual signature	Validated report template	*Signed report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
17	**82 h · 14 d**	**SRP-22**	**1,90,000**

SRP-23
SECTOR 01

Recurrent Pregnancy Loss Genomic Analysis

(**Recurrent miscarriage genomic investigation** · RPL analysis)

WHY IT IS DONE

Repeated loss is one of the most distressing experiences in reproductive medicine and frequently goes unexplained. A genomic investigation identifies the parental rearrangements and recurrent embryonic causes that can be acted on, and gives a recurrence figure where it cannot.

WHAT IT ANSWERS

Is there a genetic cause for this couple's recurrent losses, in the parents or in the pregnancies themselves, and what intervention follows?

WHEN IT IS DONE

After two or more pregnancy losses, in couples with a previous chromosomally abnormal pregnancy, and where loss has followed assisted reproduction repeatedly.

WHAT TRIGGERS IT

Recurrent pregnancy loss meeting the service threshold, a previously identified abnormal pregnancy, or repeated implantation failure.

HOW IT IS DONE

Both partners are analysed for balanced rearrangements and reproductively significant copy number variants, and any available loss tissue is analysed with maternal contamination excluded. Patterns across multiple losses are assessed, recurrence risk is derived from the findings, and preimplantation testing options are set out where a parental rearrangement is identified.

WHAT IT PRODUCES

Parental rearrangement and copy number findings, loss tissue results where available, cross-loss pattern assessment, derived recurrence risk, and preimplantation testing recommendations where applicable.

WHAT IT DETECTS

Parental balanced rearrangements, parental copy number variants of reproductive significance, recurrent aneuploidy patterns across losses where tissue was analysed, and uniparental disomy or imprinting findings.

WHAT IT DOES NOT DETECT

Thrombophilic, immunological, anatomical and endocrine causes, which fall outside genomic testing entirely. A normal genomic investigation does not exclude a recurrent cause, and most recurrent loss remains unexplained after complete workup.

WHAT MAKES IT HARD

Most investigations return normal, and delivering that without implying nothing is wrong requires care. Where a parental rearrangement is found, translating it into a realistic recurrence figure and a proportionate recommendation is harder than detecting it.

WHO DOES THIS JOB

Clinical bioinformatician in reproductive genomics, mid level, working with recurrent loss clinics and genetic counselling.

WHAT YOU CAN DO AFTERWARDS

Investigate recurrent loss across parental and conceptus material, derive realistic recurrence figures, and set out proportionate intervention options.

WHO HIRES FOR IT

Recurrent loss clinics, IVF and fertility chains, reproductive genetics laboratories, and maternal-fetal medicine services.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
18	**86 hours · 15 days**	Prior copy number and structural variant experience	**2,00,000**

SRP-23
WORKFLOW

Recurrent Pregnancy Loss Genomic Analysis

(**Recurrent miscarriage genomic investigation** · RPL analysis)

COMPLETE WORKFLOW — 18 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Loss history capture**	Number, timing and investigation status of previous losses captured	Clinical record	*Loss history record*
02	**Consent and scope confirmation**	Scope for both partners and any available loss tissue confirmed	Consent record	*Analysis authorisation*
03	**Sample inventory assessment**	Available parental and conceptus material inventoried	Sample inventory	*Sample inventory record*
04	**Data receipt and quality assessment**	Integrity and quality verified for all samples	md5sum, FastQC	*QC acceptance record*
05	**Parental alignment and processing**	Both partners' data aligned and processed identically	bwa-mem2, Picard, GATK BQSR	*Analysis-ready BAMs*
06	**Parental identity verification**	Both samples confirmed correctly attributed	somalier extract	*Identity clearance*
07	**Parental copy number analysis**	Copy number analysed in both partners for reproductively significant variants	Copy number analysis	*Parental CNV findings*
08	**Parental structural rearrangement analysis**	Balanced rearrangements sought in both partners	Structural analysis	*Parental rearrangement findings*
09	**Breakpoint characterisation**	Any rearrangement breakpoints characterised for downstream testing design	Breakpoint analysis	*Breakpoint characterisation*
10	**Loss tissue contamination assessment**	Maternal contamination excluded in any available loss tissue	MCC analysis	*Contamination assessment*
11	**Loss tissue ploidy analysis**	Ploidy and copy number determined in available loss tissue	Copy number and allelic analysis	*Loss tissue findings*
12	**Cross-loss pattern assessment**	Findings across multiple losses assessed for a recurrent pattern	Pattern analysis	*Cross-loss assessment*
13	**Uniparental disomy and imprinting assessment**	Uniparental disomy and imprinting findings assessed where indicated	UPD analysis	*UPD findings*
14	**Non-genetic cause acknowledgement**	Non-genetic causes outside scope acknowledged explicitly in the report	Reporting policy	*Scope statement*
15	**Recurrence risk derivation**	Recurrence risk derived from the findings and the loss history	Risk derivation	*Recurrence risk*
16	**Preimplantation testing assessment**	Preimplantation testing options assessed where a rearrangement is identified	Options assessment	*PGT recommendation*
17	**Counselling information assembly**	Information assembled to support a counselling conversation on options	Counselling references	*Counselling information*
18	**Report generation and sign-out**	Findings, recurrence risk and options issued under dual signature	Validated report template	*Signed report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
18	**86 h · 15 d**	**SRP-23**	**2,00,000**

SRP-24
SECTOR 01

Genomic Newborn Screening Analysis

(**Genomic newborn screening** · gNBS · genomic newborn screening)

WHY IT IS DONE

Biochemical newborn screening detects a few dozen conditions; genomic screening can detect hundreds of treatable childhood-onset disorders before symptoms appear. Where treatment exists and early intervention changes outcome, detection at birth is the difference between a managed condition and irreversible harm.

WHAT IT ANSWERS

Does this newborn carry a pathogenic variant in any gene on the screening set for a treatable childhood-onset condition?

WHEN IT IS DONE

At birth within a genomic newborn screening programme, alongside or following conventional biochemical screening.

WHAT TRIGGERS IT

Programme enrolment at birth, an abnormal biochemical screening result requiring genomic follow-up, or parental election in an offered programme.

HOW IT IS DONE

The curated gene set is authorised with actionability and penetrance criteria applied to inclusion, coverage is certified per gene, and variants are called and classified against a deliberately restrictive reporting policy that excludes uncertain findings. Positive results are confirmed orthogonally and routed to a defined clinical follow-up pathway before any family communication.

WHAT IT PRODUCES

Per-gene coverage certification, classified findings under the restrictive reporting policy, orthogonal confirmation of every positive, the follow-up pathway referral, and a report bounded explicitly by the screening gene set.

WHAT IT DETECTS

Pathogenic and likely pathogenic variants in the curated screening gene set, copy number changes where the design supports them, and carrier status only where programme policy includes it.

WHAT IT DOES NOT DETECT

Conditions outside the curated set, variants of uncertain significance which programme policy generally excludes from reporting, and conditions with adult onset or no available intervention. Penetrance in an asymptomatic newborn is far less certain than in a symptomatic child, which is the central interpretive difficulty.

WHAT MAKES IT HARD

Penetrance is the problem. A variant classified pathogenic from symptomatic patients may never cause disease in a screened asymptomatic newborn, so reporting it can medicalise a healthy child. The reporting policy must be restrictive by design, and the false positive consequences here are lifelong rather than transient.

WHO DOES THIS JOB

Clinical bioinformatician or curation scientist in newborn screening, mid to senior level, in a programme with strong governance.

WHAT YOU CAN DO AFTERWARDS

Curate and govern a newborn screening gene set, apply restrictive reporting policy, confirm positives orthogonally, and operate within a follow-up pathway.

WHO HIRES FOR IT

National newborn screening programmes, public health genomics services, children's hospital laboratories, and research programmes in genomic screening.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
23	**112 hours · 19 days**	Prior germline analysis and curation experience	**2,55,000**

SRP-24
WORKFLOW

Genomic Newborn Screening Analysis

(**Genomic newborn screening** · gNBS · genomic newborn screening)

COMPLETE WORKFLOW — 23 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Gene set curation review**	Gene set inclusion criteria covering actionability and penetrance reviewed	Curation framework	*Curation review record*
02	**Gene set version authorisation**	The screening gene set version confirmed and recorded	Gene set register	*Authorised gene set*
03	**Reporting policy confirmation**	The restrictive reporting policy excluding uncertain findings confirmed	Reporting policy	*Policy confirmation*
04	**Consent and enrolment verification**	Parental consent and programme enrolment verified before analysis	Consent record	*Analysis authorisation*
05	**Specimen and identity verification**	Specimen adequacy and newborn identity attribution verified	Sample verification	*Identity clearance*
06	**Data receipt and quality assessment**	Integrity and sequence quality verified against acceptance criteria	md5sum, FastQC, MultiQC	*QC acceptance record*
07	**Read preprocessing**	Adapters and low-quality tails removed with retention accounting	fastp	*Trimmed FASTQ*
08	**Alignment and processing**	Reads aligned, duplicate marked and recalibrated	bwa-mem2, Picard, GATK BQSR	*Analysis-ready BAM*
09	**Per-gene coverage certification**	Depth certified per gene on the screening set with shortfalls named	mosdepth, bedtools coverage	*Coverage certificate*
10	**Variant calling**	Variants called across the screening gene set with validated parameters	Validated caller	*Screening VCF*
11	**Copy number calling**	Copy number called where the design and gene set require it	gCNV, DECoN	*Copy number findings*
12	**Annotation and population filtering**	Variants annotated with stringent frequency filters for a screened population	VEP, gnomAD, ClinVar	*Annotated variants*
13	**Classification**	Variants classified with only pathogenic and likely pathogenic reportable	ACMG-AMP with gene specifications	*Classified variants*
14	**Uncertain variant exclusion**	Uncertain variants excluded from reporting under the restrictive policy	Reporting policy	*Exclusion record*
15	**Penetrance assessment**	Reportable findings assessed for penetrance in an asymptomatic newborn	Penetrance evidence	*Penetrance assessment*
16	**Actionability confirmation**	Each finding confirmed to have an available intervention before reporting	Actionability criteria	*Actionability confirmation*
17	**Carrier status policy application**	Carrier findings handled strictly under programme policy	Programme policy	*Carrier policy record*
18	**Orthogonal confirmation**	Every positive confirmed by an independent method before communication	Sanger, ddPCR	*Confirmation record*
19	**Biochemical correlation**	Findings correlated against conventional biochemical screening where available	Correlation review	*Correlation record*
20	**Follow-up pathway referral**	Confirmed positives routed into the defined clinical follow-up pathway	Referral protocol	*Referral record*
21	**Family communication preparation**	Communication prepared with counselling support before family contact	Communication protocol	*Communication preparation*
22	**Report generation**	Findings bounded by the gene set with penetrance caveats assembled	Validated report template	*Draft report*
23	**Clinical sign-out**	Report reviewed and authorised under dual signature	Sign-out procedure	*Signed screening report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
23	**112 h · 19 d**	**SRP-24**	**2,55,000**

SRP-25
SECTOR 01

Rapid Neonatal Diagnostic Analysis

(**Rapid neonatal genomic diagnosis** · Rapid neonatal analysis)

WHY IT IS DONE

A critically unwell newborn cannot wait weeks for a diagnosis. Rapid genomic analysis identifies treatable metabolic and other conditions within days, and where no treatment exists it allows care decisions to be made with knowledge rather than uncertainty.

WHAT IT ANSWERS

Is there a genetic diagnosis explaining this newborn's critical presentation, and does it change management in the next few days?

WHEN IT IS DONE

In neonatal intensive care for critically unwell infants with a suspected genetic cause, in unexplained metabolic crisis, and in severe congenital abnormality.

WHAT TRIGGERS IT

A critically unwell newborn with suspected genetic cause, unexplained metabolic decompensation, or severe congenital anomalies requiring management decisions.

HOW IT IS DONE

The pathway is activated with committed turnaround and compute reserved, phenotype is captured from the neonatal team before data arrive, and treatable condition gene sets are screened first regardless of prior probability. Trio analysis is used where parental samples are available, findings are communicated verbally when actionable, and management implications are stated rather than classifications alone.

WHAT IT PRODUCES

Treatable condition screen results, prioritised findings with phenotype correlation, verbal communication records with timing, the signed report with management implications, and a turnaround audit.

WHAT IT DETECTS

Sequence variants, copy number changes and where genome-based structural and repeat causes, prioritised so that treatable conditions are examined first.

WHAT IT DOES NOT DETECT

The same limitations as the underlying assay, compounded by compressed analysis. Methylation and imprinting conditions are missed, and a rapid negative is weaker than an unhurried one, which must be communicated rather than lost in the urgency.

WHAT MAKES IT HARD

Neonatal phenotypes are non-specific and evolving, so prioritisation operates on weak signal. Consent is taken from parents in acute distress and must follow a protocol designed for that situation, and results must be expressed as management changes rather than as genetic classifications.

WHO DOES THIS JOB

Senior clinical bioinformatician on a rapid rota in neonatal or paediatric genomics, working with neonatal intensive care.

WHAT YOU CAN DO AFTERWARDS

Operate a rapid neonatal pathway, screen treatable conditions first, manage acute-setting consent and verbal reporting, and express findings as management implications.

WHO HIRES FOR IT

Neonatal intensive care genomics services, children's hospitals, national rapid genome programmes, and paediatric reference laboratories.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
25	**122 hours · 21 days**	Prior rapid genome or exome analysis experience	**2,80,000**

SRP-25
WORKFLOW

Rapid Neonatal Diagnostic Analysis

(**Rapid neonatal genomic diagnosis** · Rapid neonatal analysis)

COMPLETE WORKFLOW — 25 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Rapid pathway activation**	Case accepted with clock started and turnaround committed	Escalation protocol	*Activated case*
02	**Joint clinical framing**	The clinical question framed as the management decision awaiting genomic input	Joint protocol	*Framed question*
03	**Acute setting consent**	Consent taken from parents under a protocol designed for acute distress	Acute consent protocol	*Documented consent*
04	**Neonatal phenotype capture**	Presentation captured in structured terms from the neonatal team	Structured HPO capture	*Structured phenotype*
05	**Parental sample assessment**	Parental samples assessed for availability to enable trio analysis	Sample inventory	*Trio configuration*
06	**Compute pre-provisioning**	Dedicated capacity reserved with no queueing permitted	Reserved capacity	*Provisioned compute*
07	**Streaming quality assessment**	Quality assessed during sequencing with early accept or abort decision	Real-time metrics	*Early acceptance decision*
08	**Accelerated alignment**	Reads aligned using a validated accelerated implementation	Accelerated aligner	*Aligned BAM*
09	**Accelerated variant calling**	Variants called using the validated accelerated configuration	Accelerated caller	*Variant call set*
10	**Concurrent identity verification**	Identity and contamination verified in parallel to protect the clock	somalier, VerifyBamID2	*Identity clearance*
11	**Transfusion interference assessment**	Recent transfusion assessed as a source of chimerism and identity artefact	Transfusion record review	*Interference assessment*
12	**Coverage sufficiency assessment**	Coverage assessed across curated treatable condition gene sets	mosdepth on gene sets	*Coverage record*
13	**Treatable condition first-pass screen**	Curated treatable condition genes screened first regardless of prior probability	Treatable gene sets	*Treatable condition result*
14	**Metabolic gene screen**	Inborn errors of metabolism screened for immediate management relevance	Metabolic gene sets	*Metabolic result*
15	**Concurrent copy number calling**	Copy number called in parallel using a pre-built reference pool	gCNV	*Copy number findings*
16	**Trio analysis**	De novo and inheritance analysis applied where parental samples exist	Trio logic	*Trio findings*
17	**Phenotype-driven prioritisation**	Candidates ranked against the evolving neonatal phenotype	Exomiser, HPO terms	*Ranked candidates*
18	**Genome-wide expansion**	Analysis widened beyond treatable sets while priority review proceeds	Genome-wide filtering	*Expanded candidates*
19	**Rapid multidisciplinary review**	Candidates reviewed with the neonatal and genetics teams on a committed timeline	Scheduled review	*Review record*
20	**Classification**	Candidates classified with reasoning documented under time pressure	ACMG-AMP with gene specifications	*Classified variants*
21	**Management-linked verbal communication**	Findings communicated as the management change indicated, with timing recorded	Verbal protocol	*Communication record*
22	**Accelerated orthogonal confirmation**	Confirmation initiated on an accelerated pathway in parallel	Rapid Sanger, ddPCR	*Confirmation record*
23	**Report generation**	Findings, management implications and limitations assembled	Validated rapid template	*Draft report*
24	**Clinical sign-out**	Report authorised under dual signature superseding verbal communication	Sign-out procedure	*Signed clinical report*
25	**Turnaround and outcome audit**	Stage timestamps and the management change actually made recorded	Audit procedure	*Audit record*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
25	**122 h · 21 d**	**SRP-25**	**2,80,000**

SRP-26

SECTOR 01

Newborn Pharmacogenomic Analysis

(**Neonatal and paediatric pharmacogenomic analysis** · Newborn PGx)

WHY IT IS DONE

Newborns and infants receive drugs with well-characterised pharmacogenomic risks, and a result generated at birth remains valid for life. Establishing it early prevents avoidable adverse reactions across an entire childhood of prescribing.

WHAT IT ANSWERS

What pharmacogenomic phenotypes does this newborn carry, and what prescribing guidance follows for the agents likely to be used in childhood?

WHEN IT IS DONE

At birth within a screening or precision medicine programme, before neonatal intensive care drug exposure, and after an unexpected drug reaction in infancy.

WHAT TRIGGERS IT

Programme enrolment, planned neonatal drug exposure with known pharmacogenomic risk, or an adverse drug reaction requiring investigation.

HOW IT IS DONE

Coverage is confirmed at allele-defining positions, variants including structural and repeat alleles are called, and diplotypes are assigned against a versioned definition table. Phenotypes are translated and mapped to current guidance, with paediatric-specific considerations noted, and results are formatted for durable storage in the child's lifelong record.

WHAT IT PRODUCES

Diplotype calls with definition version, phenotype translation, guideline-mapped prescribing recommendations with paediatric considerations, ambiguity statement, and a durable lifelong record.

WHAT IT DETECTS

Star-allele diplotypes across the relevant pharmacogenes, translated metabolic phenotypes, and guideline-mapped prescribing recommendations relevant to paediatric practice.

WHAT IT DOES NOT DETECT

Developmental changes in drug metabolism, which vary substantially through infancy and are not genetically determined. Rare uncatalogued alleles default to normal function, and the analysis addresses only the genetic component of drug response.

WHAT MAKES IT HARD

Genotype does not translate directly to phenotype in early infancy because enzyme systems are still maturing, so recommendations must carry a developmental caveat. The record's value depends entirely on it remaining findable and interpretable years later, which is a data governance problem as much as an analytical one.

WHO DOES THIS JOB

Clinical bioinformatician or pharmacogenomics scientist, mid level, working with paediatric pharmacy.

WHAT YOU CAN DO AFTERWARDS

Call pharmacogene diplotypes in newborns, apply paediatric-appropriate guidance with developmental caveats, and produce durable lifelong records.

WHO HIRES FOR IT

Children's hospitals, newborn screening programmes, paediatric pharmacy services, and precision medicine programmes.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
15	**72 hours · 12 days**	Prior pharmacogenomic analysis experience	**1,65,000**

SRP-26
WORKFLOW

Newborn Pharmacogenomic Analysis

(**Neonatal and paediatric pharmacogenomic analysis** · Newborn PGx)

COMPLETE WORKFLOW — 15 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Gene and agent scope confirmation**	The pharmacogenes and paediatric agents in scope confirmed	Assay register	*Scope confirmation*
02	**Allele definition version lock**	Star-allele definition tables confirmed with versions recorded	Definition register	*Definition version*
03	**Consent and record retention confirmation**	Consent and lifelong record retention arrangements confirmed	Consent record	*Analysis authorisation*
04	**Coverage verification at defining positions**	Depth confirmed adequate at every allele-defining position	mosdepth	*Coverage certificate*
05	**Identity verification**	Fingerprint captured to confirm attribution to the newborn	somalier extract	*Identity clearance*
06	**Variant calling**	Variants called at defining positions including small insertions and deletions	Validated caller	*Variant set*
07	**Structural and repeat allele analysis**	Gene deletions, duplications and repeat alleles assessed with dedicated methods	Copy number and repeat analysis	*Structural findings*
08	**Phasing**	Variants phased into haplotypes where data permit	Phasing method	*Phased haplotypes*
09	**Diplotype assignment**	Diplotypes assigned against the versioned definition table	PharmCAT	*Diplotype calls*
10	**Ambiguity documentation**	Unresolvable diplotypes and uncatalogued alleles documented	Ambiguity register	*Ambiguity statement*
11	**Phenotype translation**	Diplotypes translated to metabolic phenotype using current activity scores	Activity score tables	*Metabolic phenotype*
12	**Developmental caveat application**	Developmental variation in enzyme maturity noted against each recommendation	Paediatric guidance	*Developmental caveat*
13	**Guideline mapping**	Phenotypes mapped to current prescribing guidance with version recorded	CPIC, DPWG guidelines	*Prescribing recommendations*
14	**Durable record preparation**	Results formatted for retrieval and interpretation years later	Structured record format	*Durable record*
15	**Report generation and sign-out**	Diplotypes, phenotypes and paediatric guidance issued under dual signature	Validated report template	*Signed report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
15	**72 h · 12 d**	**SRP-26**	**1,65,000**

SRP-27
SECTOR 01

Gamete Donor Screening & Matching Analysis

(**Donor genetic screening and recipient matching** · Donor screening)

WHY IT IS DONE

A single gamete donor may contribute to many families, so an undetected carrier state propagates far beyond one pregnancy. Screening donors and matching them against recipients on carrier status prevents recessive disease at the point where prevention is simplest.

WHAT IT ANSWERS

What carrier states does this donor hold, and which recipients can be matched to them without creating an at-risk pairing?

WHEN IT IS DONE

At donor recruitment before any allocation, on periodic rescreening as gene sets expand, and at each recipient matching decision.

WHAT TRIGGERS IT

Donor recruitment into a programme, an expanded screening gene set requiring rescreening, or a recipient matching request.

HOW IT IS DONE

Donors are screened against the authorised gene set with per-gene coverage certified and results retained in a structured, queryable form. Chromosomal analysis is performed, and matching is executed by comparing donor and recipient carrier profiles gene by gene, with residual risk calculated for each proposed pairing.

WHAT IT PRODUCES

The donor carrier profile in structured form, chromosomal findings, per-pairing matching assessment, residual risk for each proposed pairing, and rescreening recommendations as gene sets expand.

WHAT IT DETECTS

Carrier status across the screening gene set, chromosomal rearrangements relevant to reproduction, and matching incompatibility against a specific recipient's carrier profile.

WHAT IT DOES NOT DETECT

Conditions outside the screened set, de novo events in resulting children, and any risk arising from the recipient's untested partner. Matching reduces risk substantially but cannot eliminate it, and the residual figure must be stated to recipients.

WHAT MAKES IT HARD

Donor results must remain queryable and comparable for years as recipients present and gene sets expand, which is a data architecture problem rather than an analytical one. Retrospective notification when an expanded panel reveals a carrier state in an already-used donor is the situation programmes handle worst.

WHO DOES THIS JOB

Variant scientist or clinical bioinformatician in reproductive genomics, mid level, working with donor programme coordination.

WHAT YOU CAN DO AFTERWARDS

Screen and structure donor carrier profiles, execute recipient matching with residual risk, and manage rescreening and retrospective notification.

WHO HIRES FOR IT

Gamete banks and donor programmes, IVF chains, reproductive genetics laboratories, and fertility clinic networks.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
16	**78 hours · 13 days**	Prior carrier screening analysis experience	**1,80,000**

SRP-27
WORKFLOW

Gamete Donor Screening & Matching Analysis

(**Donor genetic screening and recipient matching** · Donor screening)

COMPLETE WORKFLOW — 16 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Screening gene set authorisation**	The donor screening gene set and version confirmed and recorded	Gene set register	*Authorised gene set*
02	**Donor consent and use scope confirmation**	Consent covering screening, storage and recipient matching confirmed	Consent record	*Analysis authorisation*
03	**Donor identity verification**	Donor identity verified and linked to the programme record	Identity verification	*Identity clearance*
04	**Data receipt and quality assessment**	Integrity and sequence quality verified against acceptance criteria	md5sum, FastQC	*QC acceptance record*
05	**Alignment and processing**	Reads aligned, duplicate marked and recalibrated	bwa-mem2, Picard, GATK BQSR	*Analysis-ready BAM*
06	**Per-gene coverage certification**	Depth certified per gene with shortfalls named individually	mosdepth, bedtools coverage	*Coverage certificate*
07	**Variant calling**	Variants called across the screening gene set	Validated caller	*Donor VCF*
08	**Copy number calling**	Copy number called across genes where deletions are a carrier mechanism	gCNV, DECoN	*Copy number findings*
09	**Pseudogene-aware locus handling**	Genes with pseudogene interference routed to dedicated methods	Specialist assay	*Specialist results*
10	**Chromosomal rearrangement assessment**	Balanced rearrangements and reproductively significant imbalances assessed	Structural analysis	*Chromosomal findings*
11	**Annotation and classification**	Variants annotated and classified conservatively for a healthy donor	VEP, ACMG-AMP	*Classified variants*
12	**Structured profile generation**	The carrier profile stored in a structured, queryable form for future matching	Structured storage	*Donor carrier profile*
13	**Recipient matching execution**	Donor and recipient carrier profiles compared gene by gene	Matching logic	*Matching assessment*
14	**Residual risk calculation**	Residual risk calculated for each proposed pairing with ancestry applied	Risk calculation	*Pairing residual risk*
15	**Rescreening assessment**	Need for rescreening against expanded gene sets assessed and scheduled	Rescreening policy	*Rescreening record*
16	**Report generation and sign-out**	Donor profile, matching outcome and residual risk issued under dual signature	Validated report template	*Signed report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
16	**78 h · 13 d**	**SRP-27**	**1,80,000**

SRP-28
SECTOR 01

Mitochondrial Donation & Heteroplasmy Analysis

(**Mitochondrial heteroplasmy analysis in reproduction** · Heteroplasmy analysis)

WHY IT IS DONE

Mitochondrial disease transmission depends on heteroplasmy level rather than presence, and that level shifts unpredictably between generations. Measuring it in oocytes, embryos and after mitochondrial donation is what allows reproductive risk to be quantified at all.

WHAT IT ANSWERS

What is the mitochondrial heteroplasmy level in this material, and what transmission or recurrence risk does it imply?

WHEN IT IS DONE

In reproductive counselling for women carrying pathogenic mitochondrial variants, in preimplantation testing for mitochondrial disease, and in monitoring carryover after mitochondrial donation.

WHAT TRIGGERS IT

A maternal pathogenic mitochondrial variant with reproductive plans, preimplantation testing for mitochondrial disease, or a mitochondrial donation procedure.

HOW IT IS DONE

Deep mitochondrial sequencing is performed with nuclear mitochondrial sequence excluded, and heteroplasmy is quantified against an empirical error model rather than by simple allele fraction. Low-level detection limits are established, variation across a cohort of embryos or oocytes is characterised, and results are interpreted against known bottleneck behaviour.

WHAT IT PRODUCES

Heteroplasmy levels with confidence bounds, the detection limit achieved, cohort variation characterisation, carryover measurement where applicable, and transmission risk interpretation.

WHAT IT DETECTS

Heteroplasmy level with confidence bounds in the material tested, variation between embryos or oocytes in a cohort, and residual maternal mitochondrial carryover following donation.

WHAT IT DOES NOT DETECT

It cannot predict the heteroplasmy level a resulting child will have in affected tissues, because the genetic bottleneck and tissue distribution are not predictable from a single measurement. Low-level carryover may expand unpredictably over time, which no single timepoint measurement can anticipate.

WHAT MAKES IT HARD

Quantification at low heteroplasmy sits near the assay's error floor, so an empirical error model is essential rather than optional. The genetic bottleneck means a measured embryo level does not predict the level in a resulting child's affected tissues, and counselling must reflect that honestly.

WHO DOES THIS JOB

Senior clinical bioinformatician in mitochondrial or reproductive genomics. A small and highly specialised field.

WHAT YOU CAN DO AFTERWARDS

Quantify heteroplasmy near the error floor, establish detection limits, measure carryover, and interpret transmission risk against bottleneck behaviour.

WHO HIRES FOR IT

Mitochondrial disease specialist centres, mitochondrial donation programmes, reproductive genetics services, and research programmes.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
17	**84 hours · 14 days**	Prior mitochondrial and low-fraction analysis experience	**1,95,000**

SRP-28
WORKFLOW

Mitochondrial Donation & Heteroplasmy Analysis

(**Mitochondrial heteroplasmy analysis in reproduction** · Heteroplasmy analysis)

COMPLETE WORKFLOW — 17 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Indication and variant confirmation**	The maternal pathogenic mitochondrial variant confirmed and characterised	Prior test results	*Variant characterisation*
02	**Material type recording**	The material being tested recorded, since heteroplasmy differs across tissues	Sample record	*Material record*
03	**Input quantification**	Input material quantified against the requirements of deep sequencing	Quantification	*Input record*
04	**Data receipt and quality assessment**	Integrity and sequence quality verified against acceptance criteria	md5sum, FastQC	*QC acceptance record*
05	**Mitochondrial read extraction**	Mitochondrial reads extracted with circularity handled correctly	samtools extraction	*Mitochondrial reads*
06	**Nuclear mitochondrial sequence exclusion**	Nuclear mitochondrial sequence excluded before quantification	NUMT-aware realignment	*Filtered reads*
07	**Deep alignment**	Reads aligned to the mitochondrial reference with shifted-origin handling	Mitochondrial alignment	*Mitochondrial BAM*
08	**Coverage assessment**	Depth and uniformity assessed across the mitochondrial genome	mosdepth	*Coverage record*
09	**Background error model construction**	Error rates measured empirically at known-negative positions	Error profiling	*Empirical error model*
10	**Heteroplasmy quantification**	Heteroplasmy quantified against the error model with confidence bounds	Quantification	*Heteroplasmy estimates*
11	**Detection limit determination**	The minimum reliably detectable heteroplasmy level determined	Sensitivity calculation	*Detection limit*
12	**Artefact filtering**	Strand bias and alignment artefacts removed before interpretation	Bias metrics	*Filtered results*
13	**Cohort variation characterisation**	Variation across embryos or oocytes in a cohort characterised	Cohort analysis	*Cohort variation*
14	**Carryover measurement**	Residual maternal mitochondrial carryover measured after donation	Carryover analysis	*Carryover measurement*
15	**Bottleneck limitation statement**	The unpredictability of transmission through the genetic bottleneck stated	Limitation template	*Limitation statement*
16	**Transmission risk interpretation**	Findings interpreted for reproductive transmission risk with caveats	Clinical interpretation	*Risk interpretation*
17	**Report generation and sign-out**	Levels, detection limit, cohort variation and caveats issued under dual signature	Validated report template	*Signed report*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
17	**84 h · 14 d**	**SRP-28**	**1,95,000**

SRP-29

SECTOR 01

Fetal Sex Determination & X-Linked Risk Analysis

(**Non-invasive fetal sex determination** · Fetal sexing)

WHY IT IS DONE

In pregnancies at risk of a serious X-linked condition, determining fetal sex from maternal plasma spares female pregnancies from invasive testing entirely. It is also required to guide management in congenital adrenal hyperplasia, where treatment timing depends on it.

WHAT IT ANSWERS

Is this fetus male or female, determined from maternal plasma, and does that determination change the management of an X-linked or sex-dependent condition risk?

WHEN IT IS DONE

In pregnancies at risk of X-linked disease from around seven to nine weeks, in congenital adrenal hyperplasia risk, and in specific disorders of sex development pathways.

WHAT TRIGGERS IT

A maternal carrier state for a serious X-linked condition, risk of congenital adrenal hyperplasia, or a suspected disorder of sex development.

HOW IT IS DONE

Multiple Y chromosome targets are interrogated rather than one, and fetal DNA presence is confirmed independently so that absence of Y signal can be distinguished from assay failure. Gestational age is assessed against the assay's validated minimum, and results are reported with confidence and confirmatory recommendations.

WHAT IT PRODUCES

Fetal sex assignment with the Y targets interrogated, independent fetal DNA presence confirmation, gestational age adequacy assessment, confidence statement, and downstream management recommendations.

WHAT IT DETECTS

Presence or absence of Y chromosome sequence in maternal plasma, with fetal DNA presence independently confirmed, giving fetal sex assignment with confidence bounds.

WHAT IT DOES NOT DETECT

It cannot determine whether a male fetus is affected by the X-linked condition, only that risk applies. Vanishing twin, previous male pregnancy contamination and maternal sex chromosome abnormalities all produce misleading results, and disorders of sex development mean genotype and phenotype may not align.

WHAT MAKES IT HARD

A female result and a failed test are indistinguishable without independent fetal DNA confirmation, which makes that control the critical component of the entire analysis. Testing too early gives insufficient fetal fraction, and the result is used to make invasive testing decisions, so a wrong call has direct procedural consequences.

WHO DOES THIS JOB

Clinical bioinformatician in prenatal genomics, entry to mid level.

WHAT YOU CAN DO AFTERWARDS

Determine fetal sex from plasma with multiple targets, confirm fetal DNA presence independently, and interpret results within an X-linked management pathway.

WHO HIRES FOR IT

Prenatal diagnostic laboratories, fetal medicine services, clinical genetics services, and maternity chains.

COMMERCIALS

STEPS IN WORKFLOW	TRAINING DURATION	PREREQUISITE	TRAINING FEE (INR)
13	**62 hours · 11 days**	Prior cfDNA analysis experience	**1,40,000**

SRP-29
WORKFLOW

Fetal Sex Determination & X-Linked Risk Analysis

(**Non-invasive fetal sex determination** · Fetal sexing)

COMPLETE WORKFLOW — 13 ORDERED STEPS

#	STEP	WHAT HAPPENS	TOOL / METHOD	OUTPUT
01	**Indication and risk confirmation**	The X-linked or sex-dependent condition risk confirmed and recorded	Clinical record	*Indication record*
02	**Gestational age verification**	Gestational age confirmed against the assay's validated minimum	Clinical record	*Gestational age record*
03	**Consent and disclosure scope confirmation**	Consent including fetal sex disclosure scope confirmed	Consent record	*Analysis authorisation*
04	**Pre-analytical record capture**	Collection and processing variables recorded	Pre-analytical log	*Pre-analytical record*
05	**Cell-free DNA quantification**	Input quantity measured against assay requirements	Quantification	*Input record*
06	**Data receipt and quality assessment**	Integrity and sequence quality verified against acceptance criteria	md5sum, FastQC	*QC acceptance record*
07	**Multi-target Y interrogation**	Multiple Y chromosome targets interrogated rather than a single locus	Multi-target analysis	*Y target results*
08	**Control marker analysis**	Independent control markers analysed to confirm fetal DNA presence	Control analysis	*Fetal DNA confirmation*
09	**Fetal DNA presence determination**	Fetal DNA presence determined independently of the Y result	Presence logic	*Presence determination*
10	**Confounder assessment**	Vanishing twin, prior male pregnancy and maternal abnormality considered	Confounder review	*Confounder assessment*
11	**Sex assignment**	Fetal sex assigned with confidence bounds from the target results	Assignment logic	*Sex assignment*
12	**Inconclusive determination**	Results without fetal DNA confirmation declared inconclusive rather than female	Inconclusive criteria	*Inconclusive decision*
13	**Management pathway derivation**	Downstream management derived for the specific X-linked or sex-dependent risk	Clinical protocol	*Management recommendation*

STEPS IN WORKFLOW	TRAINING DURATION	PROGRAMME CODE	TRAINING FEE (INR)
13	**62 h · 11 d**	**SRP-29**	**1,40,000**

Build and staff a *reproductive genomics service*

Each of the twenty-nine programmes in this catalogue trains one complete type of NGS data analysis as a reproductive genomics laboratory performs it — not a tool, not a step, but the whole analysis from the data that arrives to the report that is signed. Trainees run the pipeline on live clinical data, break it, diagnose the failure, and produce the deliverable report set a laboratory would place in a case file and defend to an assessor. Programmes are named the way laboratories name their own work, so a trainee returns to the bench able to run the analysis on the requisition rather than a generic version of it. Individual programmes may be taken alone, combined into a track, or delivered in-house at the client laboratory against that laboratory's own assays and gene lists.

CATALOGUE

SRP · Sector 03 of 16

SCOPE

29 analysis types · 535 steps

DURATION

2598 contact hours · 443 working days

COMPLETE CATALOGUE FEE

INR 38,79,000 (list 59,68,000)

15%	22%	28%	35%
ANY THREE PROGRAMMES	CORE TRACK	PROFESSIONAL TRACK	COMPLETE CATALOGUE