SeraCare
Clinical Diagnostics

SERASEQ®

Enabling the Advancement of Prenatal Genetic Testing

Seraseq® Reference Materials for Reproductive Health

LGC Diagnostics & Genomics

Complex NGS NIPT assays require sophisticated, groundtruth reference materials

Non-invasive prenatal test (NIPT) based on cell-free DNA in the maternal blood circulation continues to be widely adopted by clinical laboratories around the world. Thanks to continuous technology advancement, they allow screening for not only the most common fetal aneuploidies such as trisomy 21 (Down Syndrome) but also other autosomal and sex chromosome aneuploidies as well as some microdeletions, large copy number variants, and monogenic disorders.

Due to low incidence of some chromosome aberrations (1:50,000 for rare microdeletions), it is often difficult for laboratories to find positive patient samples to robustly evaluate their test performance characteristics. Inadequately validated NIPT assays may result in false results, which can lead to anxiety, invasive diagnostic tests, and difficult decisions for future parents. Moreover, the existence of various testing platforms (chromosome counting, SNP frequencies, microarray etc.), different bioinformatic pipelines and sensitivity of the technology make the development, validation, and comparison of NIPT assays even more challenging.

A critical component of a robust molecular assay is the reference material, which ensures the test is performing as expected and delivers confidence in results used to guide pregnancy outcomes. Not all reference materials are created equal, and selection of the provider and quality standards is a key consideration when a new assay is developed, validated, and monitored.

Reference materials from LGC Clinical Diagnostics are the industry gold standard

As the leading provider of off-the-shelf and custom reference materials, we support NIPT assay development, validation and routine monitoring; leading to increased confidence in the clinical deployment of these assays and ensuring the highest quality and safety for patients.

Expert-Designed Reference Materials to Support the Entire Assay Lifecycle

Development

Designing clinical assays that are highly specific, sensitive and robust is crucial. Seraseq® reference materials support the development and validation of new assays by providing a patient-like solution to hard-to-source, low volume and rare clinical samples. Assay variability is reduced thanks to lot-to-lot consistency, long-term stability and the sustainable source of our reference materials.

Analytical Validation

When first implemented in a clinical setting, an assay must be validated to ensure that the test is suitable for its intended purpose. It is essential for providing a safe and useful service to clinicians and patients. With the extremely low availability and stability of clinical samples; Seraseq® reference materials are the perfect surrogate solution.

Performance Monitoring

To ensure a method is working consistently, internal quality controls (IQC) should be implemented. Not only can they improve the consistency of assay performance; QC gives the laboratory confidence in results – in other words, decide whether a clinical test result can be trusted and is reliable before being reported to patients. In general, manufacturers' controls are optimized for use with the manufacturer's test system, and thus are less likely to identify assay performance issues.

Troubleshooting

Strong quality control measures are required in each step of the workflow to identify, monitor, and mitigate errors. NGS workflows have many complex steps that need to be tracked and controlled carefully to achieve consistency. Appropriate reference materials formulated to mimic patient samples support quality assurance programs. They can highlight suboptimal performance and help in identifying and correcting the root cause.

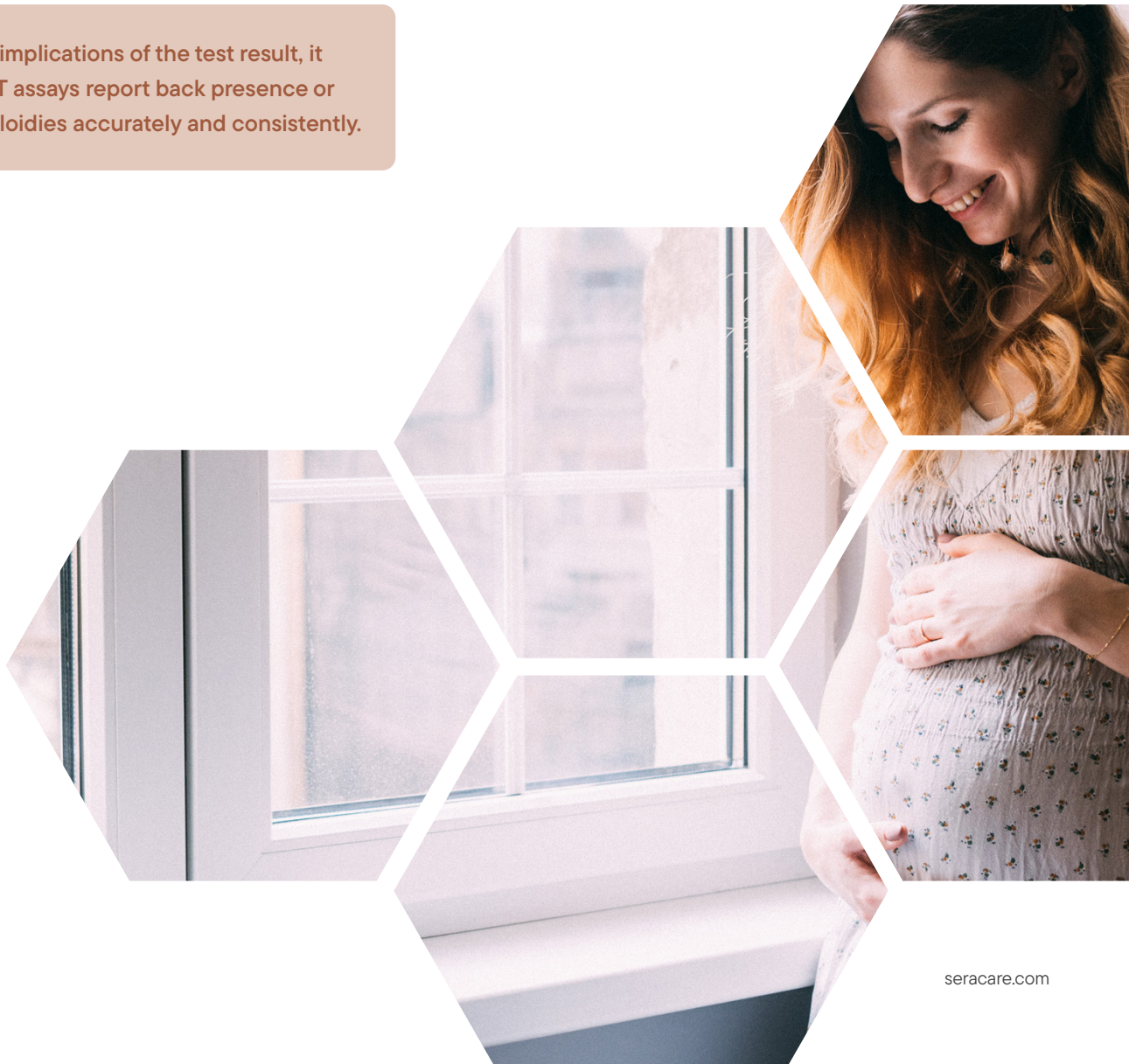
Personnel Training

As per regulatory requirements, all procedures must be performed exclusively by trained and skilled employees. Personnel on-the-job training should not only be offered frequently, but also accurately and in real-case scenarios. It is therefore essential to have quality reference materials to ensure adequate training.

Compliance

Establishments that comply with CLIA, CAP and certain ISO standards require participation in external quality assessment (EQA) or proficiency testing (PT) which represent an external comparison of clinical, analytical, and interpretation performance between laboratories and against international standards. They are essential to ensure result concordance and accuracy. It is especially important as NIPT is delivered using a variety of methodologies. EQA performance can be utilized as a tangible measure of the quality of a laboratory's performance. Seraseq® reference materials are used by international leaders of EQA schemes and proficiency testing exercises.

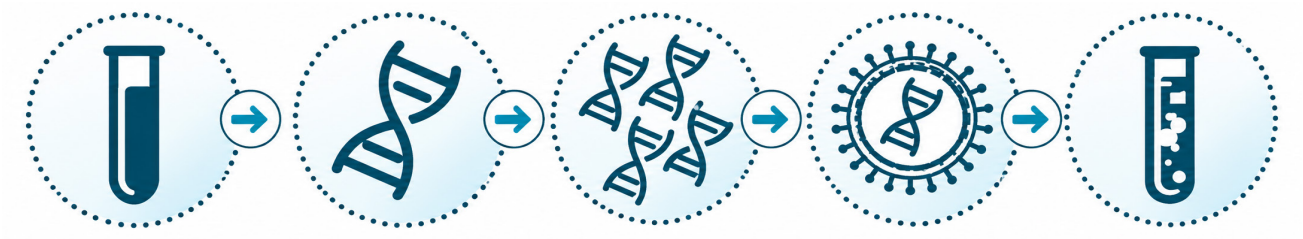
Given the clinical implications of the test result, it is critical that NIPT assays report back presence or absence of aneuploidies accurately and consistently.



Seraseq® Innovative Design Process

Seraseq® NIPT reference materials are manufactured from cfDNA extracted from plasma of pregnant patients carrying a known and confirmed aneuploidy. First, the cfDNA is isolated and amplified via a proprietary patent-pending method. This manufacturing process maintains natural cfDNA size profiles of both fetal and maternal cfDNA of approximately 170bp on average, the fetal fraction of the original sample, the single-nucleotide polymorphism (SNP) content of mother and fetus, as well as the size difference of the cfDNA.

This DNA is then innovatively stabilized via encapsulation and introduced into a dilution of simulated plasma matrix. This makes our materials look and perform like the real patient samples they were derived from.



Source plasma consisting of material and fetal cfDNA

Extract cfDNA

Amplified libraries

Encapsulated

Mixed in plasma based matrix

It is important that quality-control materials be as similar to patient specimens in order to monitor the quality of all analytical steps of the testing process.

LGC Diagnostics & Genomics

As close to a patient sample as technologically feasible

Seraseq® NIPT reference materials are the only patient-derived and patient-matched reference materials available on the market. They are derived from actual clinical samples rather than cell-lines thus avoiding the inherent limitations of artificially made genomic DNA.

Mechanisms responsible for the characteristics of circulating nucleic acids are still poorly understood. However, it is known that mechanical shearing of DNA produces fragments which differ in size distribution from those found in maternal serum, with less small DNA fragments present. This is due to the lack of nucleases which are involved in the fragmentation of plasma cfDNA.

This is extremely important as some NIPT bioinformatics pipeline are based on different maternal and fetal cfDNA representations (multivariate machine learning methods, SeqFF for fetal fraction estimation, singlenucleotide polymorphism (SNP)-based methods, etc.).

NIPT Platform Agnostic

Compatible with all NGS NIPT on the market, including SNP-based fetal fraction estimation methods assays.

High-Quality

Produced in ISO 13485 certified, cGMP compliant, US-FDA audited facilities, Seraseq® reference materials are traceable from sourcing through processing and delivery to provide you with the highest level of confidence. They provide lot-to-lot consistency and long-term stability of up to 4 years. Further quality checks are established by an external NIPT laboratory to ensure accuracy.

Full-process

Quality control in NIPT should evaluate every stage of the procedure. Supplied in our in-house artificial plasma, SeraCon Matribase, Seraseq® mimic clinical samples and allow monitoring of an entire NIPT workflow: from DNA extraction to reporting of test results including sample registration.

Patient-derived Reference Materials

Compatible with all NGS platforms including SNP-based fetal fraction estimation methods assays which determine the chromosomal copy number by looking for specific patterns in allelic measurements/targeted MPS methods analyze enriched regions on specific chromosome selectively.

Patient-like and full process matched NIPT Reference Materials

Condition	Material #	Product	Fill Size	Intended Use, Conc. & Fetal Fraction
T21	0720-0167	Seraseq® Trisomy 21 Male – Matched Reference Material	1 vial x 1 mL 20-35 ng/mL (Synthetic Plasma)	Full-process reference materials meant to be used alongside clinical samples to assess the entire workflow. DNA extraction is required. Please refer to the Technical Product Report for more information.
	0720-0168	Seraseq® Trisomy 21 Female – Matched Reference Material		
T18	0720-0171	Seraseq® Trisomy 18 Male – Matched Reference Material		
	0720-0172	Seraseq® Trisomy 18 Female – Matched Reference Material		
T13	0720-0779	Seraseq® Trisomy 13 Male – Matched Reference Material		
	0720-0180	Seraseq® Trisomy 13 Female – Matched Reference Material		
	0720-0170	Seraseq® Euploid Female – Matched Reference Material		
Euploid	0720-0169	Seraseq® Euploid Male – Matched Reference Material		
	0720-0170	Seraseq® Euploid Female – Matched Reference Material		
22q11	0720-0173	Seraseq® 22q11 Male – Matched Reference Material		
	0720-0174	Seraseq® 22q11 Female – Matched Reference Material		
Sex Chromosome Aneuploidies	0720-0952	Seraseq® Turner Syndrome (XO) Reference Material		
	0720-0953	Seraseq® Klinefelter Syndrome (XXY) Reference Material		
	0720-0954	Seraseq® Jacobs Syndrome (XYY) Reference Material		
	0720-1069	Seraseq® Triple X Syndrome (XXX) Reference Material		
Lyophilise	0720-1136	Seraseq® Euploid Female-Matched Reference Material (Lyophilized)	1 vial lyophilized originally at 1 mL volume (Lyophilized TE buffer)	Reconstitute in 1 mL molecular grade water and extract cfDNA before performing molecular analysis.
	0720-1137	Seraseq® Trisomy 21 Male-Matched Reference Material (Lyophilized)		
	0720-1138	Seraseq® Trisomy 18 Female-Matched Reference Material (Lyophilized)		

Cell-free DNA derived from patient's blood identified with twin gestations:

Condition	Material #	Product	Fill Size	Intended Use, Conc. & Fetal Fraction
Euploid Twins	0720-1117	Seraseq® Euploid Male Twins - Matched cfDNA	1 vial x 25uL Supplied in low TE buffer	Purified cfDNA in 0.1xTE buffer to be added during the library preparation. No purification or DNA extraction is required
22q11 Twins	0720-1116	Seraseq® 22q11 Male Twins - Matched cfDNA		

Cell-free DNA derived from patients' antepartum and postpartum blood, for dilution of fetal fraction and assay development applications:

Condition	Material #	Product	Fill Size	Intended Use, Conc. & Fetal Fraction
T21	0720-1100	Seraseq® Antepartum T21 Male cfDNA	1 vial x 25uL Supplied in low TE buffer >10 ng/ µL (250ng cfDNA)	Purified cfDNA in 0.1xTE buffer to be added during the library preparation. No purification or DNA extraction is required Please refer to the Technical Product Report for more information.
	0720-1101	Seraseq® Postpartum T21 Male cfDNA		
22q11	0720-1102	Seraseq® Antepartum 22q11 Male cfDNA		
	0720-1103	Seraseq® Postpartum 22q11 Male cfDNA		
Spinal Muscular Atrophy (SMA)	0720-1115	Seraseq® Antepartum SMA Male cfDNA		
	0720-1129	Seraseq® Postpartum SMA Male cfDNA		

Note: For rolling-amplification assays, such as the Vanadis® NIPT platform (Revvity) we offer cell-line based reference materials. Please contact us for more information.

Newborn Screening

Traditionally, newborn screening relied on biochemical markers (e.g., enzyme activity or metabolite levels) via dried blood spots. However, the field is rapidly shifting toward Genomic Newborn Screening.

By using NGS, labs can screen for a much broader array of conditions with higher specificity, often identifying disorders before clinical symptoms appear. Despite rapid adoption of genomic newborn screening, laboratories often face limited access to well-characterised reference materials that accurately reflect real clinical samples.

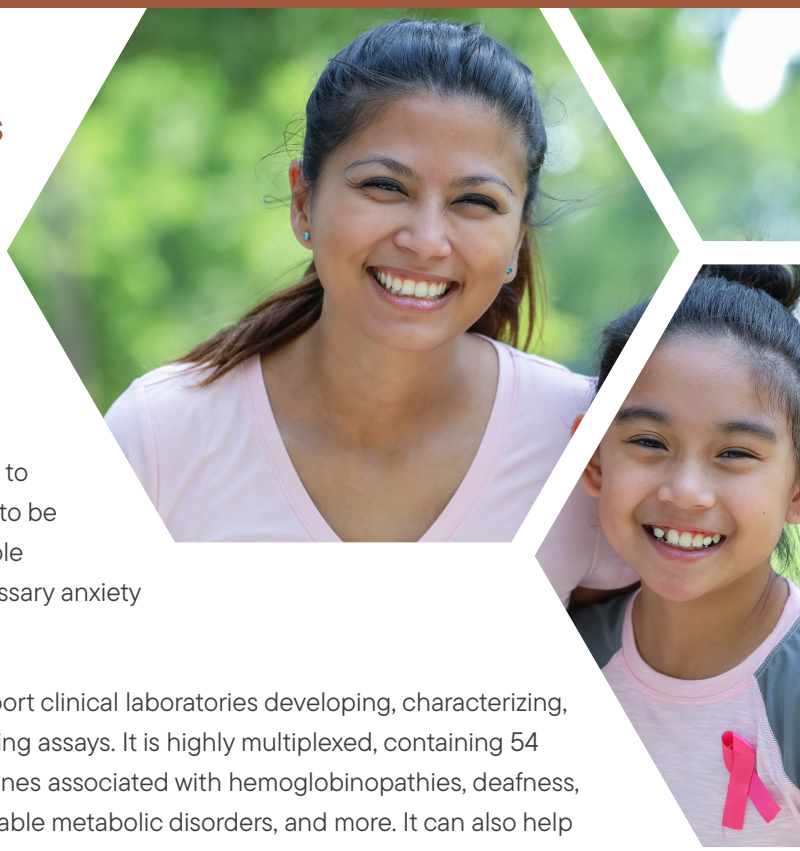
Seraseq Newborn DNA Mix SMA reference materials address this gap by providing clinically relevant, inheritanceready genomic controls specifically designed for newborn screening applications. They consist of genomic DNA isolated from immortalized maternal and fetal cell lines collected from either a Hemophilia B carrier pregnant with a non-affected fetus or from a healthy pregnant patient carrying a fetus with Spinal Muscular Atrophy (SMA).

Features and Benefits

- Patient-derived: genomic DNA extracted from proprietary cell lines isolated from cord blood or maternal blood. Inheritance analysis-ready design: materials available as matched maternal + offspring duo to support evaluation of inheritance & variant transmission.
- Clinically relevant content: includes clinically significant variants in genes relevant to Hemophilia B (F9) and Spinal Muscular Atrophy (SMN1).
- Supported by long-read sequencing data, available on demand.
- Sustainable, scalable supply: cell line-based approach supports lot consistency and ongoing availability for repeated validation and longitudinal QC.

Material Number	Product	Concentration	Fill in Size	Total Mass
0720-1173	Seraseq® Newborn DNA Mix SMA - Child	15 ng/µL	1 vial x 25µL	>375ng
0720-1174	Seraseq® Newborn DNA Mix SMA - Mother	15 ng/µL	1 vial x 25µL	>375ng
0720-1175	Seraseq® Newborn DNA Mix Hemo - Child	15 ng/µL	1 vial x 25µL	>375ng
0720-1176	Seraseq® Newborn DNA Mix Hemo - Mother	15 ng/µL	1 vial x 25µL	>375ng

Genetic Testing For Inherited Disorders



Carrier screening is a genetic test that identifies whether a person carries a gene variant for an inherited disorder. It focuses on autosomal recessive and X-linked conditions, where carriers often have no symptoms but may pass a condition on to their children. Results can help individuals and couples make more informed family-planning and healthcare decisions.

Expanded carrier screening assesses a broader range of disorders to provide a more comprehensive view of genetic risk. For screening to be clinically useful, assay accuracy and precision are essential - reliable performance reduces the risk of false positives that cause unnecessary anxiety and false negatives that provide false reassurance.

Seraseq® Carrier Screening reference material is designed to support clinical laboratories developing, characterizing, validating, and routinely monitoring expanded NGS carrier screening assays. It is highly multiplexed, containing 54 clinically important SNVs, deletions, and duplications across 48 genes associated with hemoglobinopathies, deafness, heart disease, immunodeficiency, neuromuscular conditions, treatable metabolic disorders, and more. It can also help laboratories detect systematic errors and sources of variability (reagents, instrumentation, or human factors) and support compliance with quality standards such as CAP and CLIA.

Features and Benefits

- Develop and validate carrier screening-based NGS assays with confidence using a highly multiplexed reference material containing important monogenic disorder biomarkers.
- Contains 54 clinically important variants from 48 genes associated with the most common monogenic disorders: Cystic Fibrosis, Spinal Muscular Atrophy (SMA), a- and ß-thalassemia, congenital deafness, fragile X syndrome, sickle cell disease, Tay–Sachs disease, Gaucher disease, Phenylketonuria, Cardiomyopathy, Smith–Lemli–Opitz syndrome and many more.
- Variants are present within a single well-characterized genomic background (GM24385) at clinically relevant allele frequencies, precisely quantitated by digital PCR and orthogonally verified by NGS.
- Manufacturing of large batches with long shelf-life allows for repeated use of the same lot for validation, or assay performance assessment over time.

Material Number	Product	Concentration	Fill in Size	Total Mass
0730-0569	Seraseq® Carrier Screening DNA Mix	30 ng/µL	1 vial x 20µL	600ng

Our Custom Offering

We design and produce sophisticated, high quality reference materials tailored to your specifications

With our custom services, you have a high degree of flexibility, access to our technical and scientific experts, and a rapid turnaround time for your unique requirements and complex biomarkers.

For example, LGC Clinical Diagnostics can offer custom PGT-A reference materials for the most common trisomies 21, 18 and 13 along with euploid material which can be used as a negative reference.

All our NIPT and PGT-A products can be customized with regard to concentration, fetal fraction ratio, format, volume or blending to create mosaicism samples.

We have a biobank of rare antepartum, postpartum and cord blood cfDNA samples from singleton and twin pregnancies affected with genetic conditions. Examples include: Cystic Fibrosis, SMA, Sjögren's Syndrome, Noonan Syndrome, Maple Syrup Urine Disease, alpha & beta thalassemias, rare CNVs and many more.

Preimplantation Genetic Testing for Aneuploidies (Pgt-A) - Custom Made

Assay Validation and Daily-Run QC Material

Pre-implantation Genetic Testing (PGT) is a genetic test performed on embryos produced by in vitro fertilization (IVF). The aim of the testing is to screen embryos for genetic anomalies and improve the odds of a successful pregnancy.

Patient-derived Reference Materials

Product
Seraseq® PGT-A Trisomy 21 Reference Material
Seraseq® PGT-A Trisomy 18 Reference Material
Seraseq® PGT-A Trisomy 13 Reference Material
Seraseq® PGT-A Euploid Reference Material

These products can be customized with regard to concentration, volume or blending to create mosaicism samples.

Reliable, Consistent Reference Material

Seraseq® PGT-A Reference Materials are produced from gDNA extracted from a fetal source sample with confirmed Trisomy (or Euploid status) and provided in a 1 mM Tris / 0.1 mM EDTA pH 8.0 buffer.

Developed under cGMP compliance in ISO 13485 certified facilities, our products provide a consistent source of reference material for your PGT assay. This not only ensures a reliable supply which is consistent from lot-to-lot; it also eliminates the need to obtain, characterize, blend, and document your own mixes of cell lines, saving you time and resources in your assay development and validation efforts.

- Ideal for introduction to the PGT-A workflow at the WGA (amplification) step
- Compatible with NGS as well as array-based PGT-A assays
- Eliminate the need to find, source and maintain remnant samples
- Can be used for PGT-A assay development, validation or routine QC
- Conveniently formatted for ease of use
- Save time, cost and increase QC consistency

LGC Diagnostics & Genomics

SeraCare
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SeraCare offers a comprehensive portfolio of reference materials for oncology and reproductive health, designed and manufactured to meet the precision demanded by NGS assays. The portfolio comprises high-quality, ground-truth RNA, ctDNA, and genomic DNA-based reference materials that are NGS platform-agnostic for tumor profiling, immuno-oncology, liquid biopsy, non-invasive prenatal testing (NIPT), pharmacogenomics, and germline cancer assay workflows.

SeraCare is now part of LGC Clinical Diagnostics, Inc. LGC Clinical Diagnostics develops and manufactures over 3,650 catalog and custom-developed diagnostic-quality solutions and component materials for the global life sciences industry. We partner with IVD assay developers, and pharmaceutical, CRO, and academic institutions in commercialization activities across the entire diagnostic pipeline - from concept and early-stage research, through expedited product development and onwards into routine clinical use.

Laboratory and diagnostic professionals across various disciplines, including clinical chemistry, immunochemistry, serology, molecular diagnostics, and clinical genomics, rely on LGC Clinical Diagnostics' products to support accurate and reliable diagnostic results.

How to Order

For more information about products listed in this catalog or to speak with your sales representative, contact us at:

Online: **www.seracare.com**
Phone: **+1-508-244-6400** (International)
800-676-1881 (Toll free US)
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