Seraseq® NIPT Reference Materials

Assay validation and daily-run QC material for Non-Invasive Prenatal Testing (NIPT)

INTRODUCTION

Non-invasive Prenatal Testing (NIPT) of circulating cell-free DNA (cfDNA) present in the blood of pregnant women assesses the likelihood that the fetus will be born with a genetic abnormality such as Down Syndrome (trisomy 21). Given the clinical implications of the test result, it is critical that NIPT assays deliver highly accurate and reliable detection of aneuploidies.

LGC Clinical Diagnostics matched maternal-fetal reference materials are biomimetic: they are derived from pregnant maternal plasma and thus maintain the characteristics of a clinical sample (patient-like). This enables accurate assessment of a broad range of NIPT assays, including those based on whole-genome sequencing or targeted approaches including single nucleotide polymorphism (SNP)-based assays, microarrays, or those using bioinformatic pipelines based on differences in maternal and fetal cfDNA representations.

The portfolio encompasses the most prevalent chromosomal aneuploidies (trisomy 21, 18, and 13), sex chromosome aneuploidies (SCAs) such as Turner, Klinefelter, Jacobs, and XXX syndromes, as well as 22q11.2 microdeletions (DiGeorge syndrome) and monogenic disorder Spinal Muscular Atrophy (SMA). Additionally, reference materials for twin pregnancies have been introduced to expand the offering.

Supplied in our SeraCon Matribase (simulated plasma matrix derived from human), they are designed to be used in the same way as clinical samples and enable monitoring of the full NIPT process - from extraction to reporting of the NIPT results.

FEATURES

- Collection of common aneuploidies for singleton and twin pregnancies: trisomy 21, 18, 13, XO, XXY, XYY, XXX; microdeletion 22q11, SMA as well as euploid reference materials
- Proprietary method maintains native cfDNA size profile of ~170 bp size distribution, natural maternal-fetal size difference and SNP content
- Long shelf-life of 4 years allowing for repeated use of the same lot for training, validation, or assay performance assessment
- Scalable technology allows for easy customization of fetal fraction, concentration, and material formulation (either in plasma or fragmented DNA in buffer format)
- A new line of postpartum cfDNA in buffer and matching antepartum cfDNA drawn from the same patient is also available, enabling limit of detection testing at critically low fetal fraction by diluting the apparent antepartum fetal fraction

RELIABLE, CONSISTENT REFERENCE MATERIAL

As USA-manufactured reference materials, developed under cGMP compliance in ISO 13485 certified facilities, the Seraseq NIPT portfolio ensure a reliable, sustainable and lot-to lot consistent supply. This 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.

Not for In Vitro Diagnostic Use. Research Use Only.

HIGHLIGHTS

Patient-like: derived from pregnant patient samples and containing maternal and fetal cfDNA.

High-quality third party QC material to develop, validate, monitor, and troubleshoot your NIPT assay.

Convenient full process control: single vial format of plasma-like material to save time, cost, and increase QC consistency.

NGS technology agnostic: compatible with a broad range of NIPT assays.

Saves time and cost procuring remnant samples or producing homebrew reagents.

ORDERING INFORMATION

Patient-like and full process matched NIPT Reference Materials

Condition	Material #	Product	Fill Size	Intended Use
Euploid	0720-0169	Seraseq® Euploid Male - Matched Reference Material	1 vial x 1 mL	Full-process reference materials meant to be used alongside clinical samples to assess the entire workflow. DNA extraction is required.
	0720-0170	Seraseq® Euploid Female - Matched Reference Material		
T21	0720-0167	Seraseq® Trisomy 21 Male - Matched Reference Material		
	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		
	0720-0780	Seraseq® Trisomy 13 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) - Matched Reference Material		
	0720-0953	Seraseq® Klinefelter Syndrome (XXY) - Matched Reference Material		
	0720-0954	Seraseq® Jacobs Syndrome (XYY) - Matched Reference Material		
	0720-1069	Seraseq® Triple X Syndrome (XXX) - Matched Reference Material		

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
T21	0720-1100	Seraseq® Antepartum T21 Male - Matched cfDNA	1 vial x 25uL (1 mM Tris, 0.1 mM EDTA, 10 mM KCl)	Purified cfDNA in 0.1xTE buffer to be added during the library preparation. No purification or DNA extraction is required.
	0720-1101	Seraseq® Postpartum T21 Male - Matched cfDNA		
22q11	0720-1102	Seraseq® Antepartum 22q11 Male - Matched cfDNA		
	0720-1103	Seraseq® Postpartum 22q11 Male - Matched cfDNA		
Spinal Muscular Atrophy (SMA)	0720-1115	Seraseq® Antepartum SMA Male - Matched cfDNA		
	0720-1129	Seraseq® Postpartum SMA Male - Matched cfDNA		

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

Condition	Material #	Product	Fill Size	Intended Use
Euploid Twins	0720-1117	Seraseq® Euploid Male Twins - Matched cfDNA	1 vial x 25uL (1 mM Tris, 0.1 mM EDTA, 10 mM KCl)	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		

Custom NIPT Materials

The above products can be customized with regard to fetal fraction level, concentration, volume and format (DNA mix or plasma). Other genetic conditions available. Please contact us for details.	Custom-made to suit your unique project requirements

PRODUCT DESIGN

Our 2nd generation products are derived from rare clinical samples rather than from cell lines, thus avoiding the inherent limitations of artificially fragmented genomic DNA. They were developed using an innovative and proprietary technology (patent-pending) which utilizes source plasma material obtained from pregnant patients carrying a fetus with a known condition, through a IRB-approved collaboration with Stanford University. The DNA is amplified, encapsulated, and formulated in plasma matrix which enables the material to be processed similarly to a patient specimen, as well as increases the product stability.

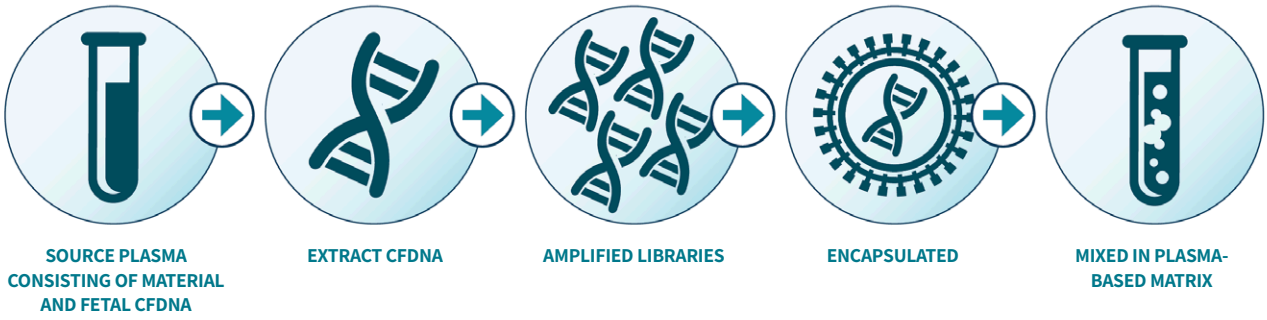

FIGURE 1: Matched NIPT reference material preparation from full-process reference materials in maternal plasma.

REAL-WORLD APPLICATION OF THE NIPT REFERENCE MATERIALS

The Seraseq NIPT reference materials have been successfully used in numerous labs across the world for a variety of applications ranging from new assay development to validation, proficiency assessment and routine run QC.

Assay Development

- Enable sensitivity, specificity, limit of detection (LOD) studies and more.
- Expedite new assay development and protocol optimization.
- Test range validity thanks to our customizable technology which allows for specific volume, concentration and fetal fraction.

Analytical Validation

- Confirm analytical performance and ability to correctly identify all the aneuploidies/microdeletions.
- Perform External Quality Assessment (EQA) and proficiency testing to ensure accuracy and concordance of clinical assays.
- Conduct new assay installation and training.

Routine Run QC

- Comply with good laboratory practices, ISO guidelines and recommendations.
- Monitor daily run performance.
- Provide peace of mind that the clinical assay provides reliable results.
- Detect, monitor, and mitigate or troubleshoot issues at each step of the workflow.

ABOUT US

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 includes high quality ground-truth RNA, ctDNA and genomic DNA-based reference materials that are NGS platform agnostic for tumor profiling, immuno-oncology, liquid biopsy, NIPT, germline cancer and pharmacogenomics assay workflows. **For more information visit seracare.com**

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