SeraCare
Clinical Diagnostics

SERASEQ®

Reference Materials for Germline Genetic Testing

Industry Gold Standard Reference Materials

Supporting the development, validation, and routine performance monitoring of high-complexity molecular assays.

LGC Diagnostics & Genomics

Reference Materials from SeraCare Clinical Diagnostics are the Industry Gold Standard

Germline testing identifies variants that increase a person's risk of developing certain conditions, such as cancers or cardiovascular diseases. Identifying these inherited risks early enables proactive monitoring, preventive measures, and timely interventions, which can significantly enhance patient outcomes.

Understanding a patient's germline genetic makeup allows providers to personalize treatment strategies, such as the use of targeted therapies or more aggressive treatments. For example, pharmacogenomics uses genetic information to predict how a patient will respond to specific medications, allowing doctors to select the most effective drugs and dosages while minimizing adverse effects.

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

Contents

Expert-Designed Reference Materials for the Entire Assay Lifecycle

Assay development & optimization

Support highly specific, sensitive, and robust clinical assays

Seraseq reference materials support development and validation by providing patient-like solutions for rare, hard-to-source clinical samples, with lot-to-lot consistency, long-term stability, and a sustainable source.

Analytical validation

Ensure assays are fit for their intended purpose

Seraseq reference materials offer an ideal surrogate solution, addressing the limited availability and stability of clinical samples, and provide a safe and effective service to clinicians and patients.

Performance Monitoring

Trust assay performance

Being confident that you consistently produce reliable test results is critical. Manufacturer-specific controls are optimized for use with their own system, making them less likely to identify performance issues, while Seraseq third-party reference materials are unbiased.

Compliance

Reduce the stress of meeting quality measures

CLIA, CAP, and ISO standards require participation in external quality assessment (EQA) or proficiency testing (PT) programs. Seraseq reference materials are widely used to ensure accuracy and concordance of results across laboratories and methodologies.

Personnel Training

Rely on high-quality materials for effective skill development

Regulatory requirements mandate that procedures must be performed by trained staff. Meeting that standard depends on realistic, regular training. Using trustworthy reference materials is crucial for the best training experience.

Troubleshooting

Scrutinize quality control throughout the workflow

Patient-like reference materials uphold quality assurance at every step by detecting, monitoring, and addressing errors. Robust quality control identifies suboptimal performance, facilitating the identification and correction of root causes.

Seraseq® for Inherited Cancer Testing

Inherited cancer syndrome refers to a genetic predisposition to certain cancers, which have a specific associated gene and inheritance pattern. Genetic testing identifies pathogenic variants that may increase the risk of cancer, providing invaluable information that informs prevention, detection, and treatment choices. Common inherited cancer syndromes, include: BRCA mutations (BRCA1 and BRCA2), Cowden syndrome (multiple hamartoma syndrome), familial adenomatous polyposis (FAP), Li-Fraumeni syndrome (LFS), and Lynch Syndrome.

Hereditary cancers pose significant risks to individuals and their families. Detecting these mutations accurately is crucial for early intervention and personalized treatment strategies. It allows for informed decisions about ways to reduce cancer risks, such as someone who's found to have BRCA1 or BRCA2 gene mutations may opt to start breast cancer screening early, have surgery, or begin taking medications to lower their risk.

Treatment for breast cancer is highly targeted with medication. PARP inhibitor cancer therapy blocks the PARP protein, preventing cancer cells from repairing damaged DNA. This therapy is particularly effective for those with BRCA1 or BRCA2 gene mutations.

Comprehensive mutation coverage

Our reference materials are designed to cover a wide range of mutations associated with inherited cancers, ensuring that your assays can detect the most challenging and clinically relevant variants. Screen confidently for up to 61 clinically relevant variants across 56 cancer-associated genes.

Unmatched complexity

Difficult-to-detect mutations, such as the MSH2 Boland inversion, PMS2 pseudogene homologous variant, and EPCAM 3' large deletion, are incorporated, ensuring robust testing capabilities.

Precision and reliability

You can validate your assays with confidence, knowing that our synthetic DNA materials offer unparalleled accuracy and reproducibility.

Streamlined workflow integration

The Seraseq® portfolio is compatible with a variety of testing platforms, allowing seamless integration into existing workflows.

Single-vial convenience

Simplify workflows with a highly multiplexed reference material packaged in a single vial, reducing costs and saving time while boosting QC consistency.

Inherited Cancers / Parp Inhibitor Product List

The Inherited Disease reference materials consist of the following products in purified DNA format to mimic heterozygous patient samples.

Product Description	Material #	Genes	# Variants	Format
Seraseq® Inherited Cancer DNA Mix v2	0730-0069	55 Pan-Cancer genes (breast, ovarian, colorectal, endocrine, renal, dermatologic, and multi-organ cancer predisposition syndromes): APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, DICER1, EPCAM, FH, FLCN, HOXB13, KIT, MAX, MEN1, MET, MITF, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PDGFRA, PMS2, POLD1, POLE, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, STK11, TMEM127, TP53, TSC1, TSC2, VHL BRCA2,CDKN2A, MLH1, MSH2, MSH6, PMS2	61	Purified DNA 1 vial x 20µL >500ng
Seraseq® gDNA BRCA1/2 LGR Inherited Mutation Mix	0730-0568	2 genes: BRCA1, BRCA2	20	Purified DNA 1 vial x 25µL >375ng
Seraseq® BRCA1/2 Exon Deletions DNA Mix	0730-0570	2 genes: BRCA1, BRCA2	2	Purified DNA 1 vial x 25µL >375 ng

Complementary Somatic Oncology Products

Product Description	Material #	Genes	# Variants	Format
Seraseq® ctDNA Prostate Mix WT	0710-3327	40 genes: APC, AR, ATM, ATR, BARD1, BRAF, BRCA1, BRCA2, BRIP1, CCND1, CDK12, CDKN2A, CHEK1, CHEK2, ERG, ETV1, FANCA, FANCL, KIT, KRAS, MAP4K3, MLH1, MRE11, MSH2, MSH6, MYC, NBN, PALB2, PIK3CA, PIK3R1, PMS2, PTEN, RAD51B, RAD51C, RAD54L, RB1, SLC7A8, SPOP, TP53, ZNF2	54	Purified DNA 1 vial x 25µL >250ng
Seraseq® ctDNA Prostate Mix AF1%	0710-3328			Purified DNA 1 vial x 25µL >250ng
Seraseq® ctDNA Prostate Mix AF0.5%	0710-3329			Purified DNA 1 vial x 25µL >250ng
Seraseq® ctDNA ESR1 Mix WT	0710-3564	2 genes: ESR1, PIK3CA	22	Purified DNA 1 vial x 25µL >250ng
Seraseq® ctDNA ESR1 Mutation Mix AF1%	0710-3565			Purified DNA 1 vial x 25µL >250ng

Seraseq® for Inherited Cardiovascular Diseases

Inherited cardiovascular diseases (ICVDs) are genetic disorders affecting the heart and blood vessels. These diseases often run in families and can lead to life-threatening conditions like sudden cardiac arrest, heart failure, or stroke.

Genetic testing for cardiovascular disease recognizes inherited mutations that put people at risk of heart conditions such as cardiomyopathies, arrhythmic disorders, thoracic aortic aneurysms, and aortic dissections. Testing can help confirm diagnoses, clarify conditions, provide insights, and facilitate the development of gene-based management strategies.

Early detection is crucial for prevention and informed patient care management. Testing for germline variants is a first step for those likely to have an ICVD or a family history of unexpected cardiac events.

Accurate genetic testing for inherited cardiovascular conditions relies on the development, validation, and routine performance assessment of assays. SeraCare's Seraseq Reference Materials play a critical role in ensuring high-quality and reliable genetic testing for ICVDs.

Significant & rare mutations

Seraseq reference materials are biosynthetic, DNA-based materials that mimic real patient samples. Labs can validate new genetic testing panels for ICVDs, detecting all relevant germline variants, and confirm assay sensitivity and specificity across a range of conditions, including rare and difficult-to-detect mutations.

Routine performance monitoring

These materials allow laboratories to regularly assess the accuracy and reproducibility of their genetic tests, ensuring consistent results over time. They provide a standardized approach to identifying and resolving technical issues in workflows.

Support for emerging technologies

Seraseq materials facilitate the adoption of new technologies such as NGS and whole-genome sequencing by providing a robust tool for assay optimization and troubleshooting.

Comprehensive variant coverage

ICVD testing often involves analyzing multiple genes simultaneously. Our Seraseq materials cover a broad spectrum of clinically relevant germline variants, making them ideal for complex panels targeting ICVD-related genes.

Validated assays supported by Seraseq reference materials ensure that laboratories can accurately detect pathogenic variants, including insertions, deletions, single-nucleotide variants (SNVs), and structural rearrangements. They provide reliable data to inform clinical decisions, such as cascade testing for at-risk family members.

Product Description	Material #	Genes	# Variants	Format
Seraseq® Inherited Cardiovascular DNA Mix	0740-0132	56 genes: ABCC9, ACTC1, ACTN2, ALPK3, BAG3, CACNA1C, CALM1, CALM2, CASQ2, CDH2, CTNNA3, CRYAB, DES, DMD, DSC2, DSG2, DSP, EMD, FKTN, FLNC, GAA, GATA6, GLA, HCN4, JPH2, JUP, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYPN, NEXN, NKX2-5, PKP2, PLN, PPA2, PRDM16, PRKAG2, PTPN11, RAF1, RBM20, RYR2, SCN5A, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTR, TTN, SHOC2	61	Purified DNA 1 vial x 25µL >250 ng

Pharmacogenomics

Pharmacogenomics is revolutionizing the way medications are prescribed, enabling the development of personalized treatment plans tailored to an individual's genetic makeup. Reference materials are critical in pharmacogenomics, as they provide standardized benchmarks for validating and ensuring the accuracy of genetic tests.

These materials, such as those offered by SeraCare, help laboratories and researchers verify that their assays can accurately detect genetic variations that affect drug metabolism and response. Without reliable reference materials, the risk of inaccurate test results increases, potentially leading to improper treatment decisions. This is why high-quality reference materials are essential for advancing personalized medicine and ensuring patient safety in clinical settings.

Product Description	Material #	Genes	# Variants	Format
Seraseq® Pharmacogenomics DNA Mix	0750-9503	13 genes: CYP2C9, CYP2C19, CYP2D6, CYP3A4, CYP3A5, CYP4F2, DPYD, G6PD, NUDT15, SLCO1B1, TPMT, UGT1A1, VKORC	79	Purified DNA 1 vial x 20µL 30ng/µL >600ng

Identifying mutations that influence drug metabolism and response relies on consistently accurate genetic testing.

First multiplex pharmacogenomics reference material on the market. Includes a strategic mix of 33x Tier 1/ Core (high clinical impact) and 35x hard-to-source Tier 2 AMP-recommended variants to assess assay performance in a single vial.

DPYD Testing

One example of pharmacogenomics that helps patients avoid a life-threatening situation is genotyping the DPYD gene variants. People with dihydropyrimidine dehydrogenase (DPD) enzyme deficiency can have severe toxic reactions to fluoropyrimidine-based (FU) chemotherapy. Accurate DPYD genetic testing allows clinicians to create personalized treatments and therapies and proactively prevent toxicities.

This test analyzes the DPYD gene, which encodes the enzyme dihydropyrimidine dehydrogenase (DPD). The enzyme plays a crucial role in the metabolism of fluoropyrimidines, a class of chemotherapeutic agents that includes 5-fluorouracil (5-FU) and capecitabine, which have been used over the last 50 years to treat various types of cancers. However, approximately 2-8% of patients have variations in the DPYD gene that lead to reduced or absent DPD enzyme activity. These genetic variations can result in severe, and sometimes fatal, toxicity when standard doses of fluoropyrimidines are administered.

Assay validation and daily-run QC material

SeraCare Seraseq® DPYD DNA Mutation Mix is a highly multiplexed reference material containing 39 DPYD variants, carefully selected based on clinical guidelines and recommendations (AMP, EMA, CPIC, DPWG), national public health programs, and commercial assay panels. The mutation mix includes variants known to be clinically non-functional or variants with unknown function that are found at higher frequencies in diverse ethnic groups, creating the first pan-ethnic DPYD reference material.

Product Description	Material #	Concentration	Fill in Size	Total Mass
Seraseq® DPYD DNA Mutation Mix	0750-9502	30 ng/µL	1 vial x 20µL	600ng

Advantages of Seraseq® DPYD DNA Mutation Mix

- Clinically relevant variants conferring no or decreased DPYD gene function
- Contains 4 times more variants than alternative products
- 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

Expanded Carrier Screening

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.

Advantages of Seaseq® Carrier Screening DNA Mix

- 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 a long shelf life allows for the repeated use of the same lot for validation or assay performance assessment over time.

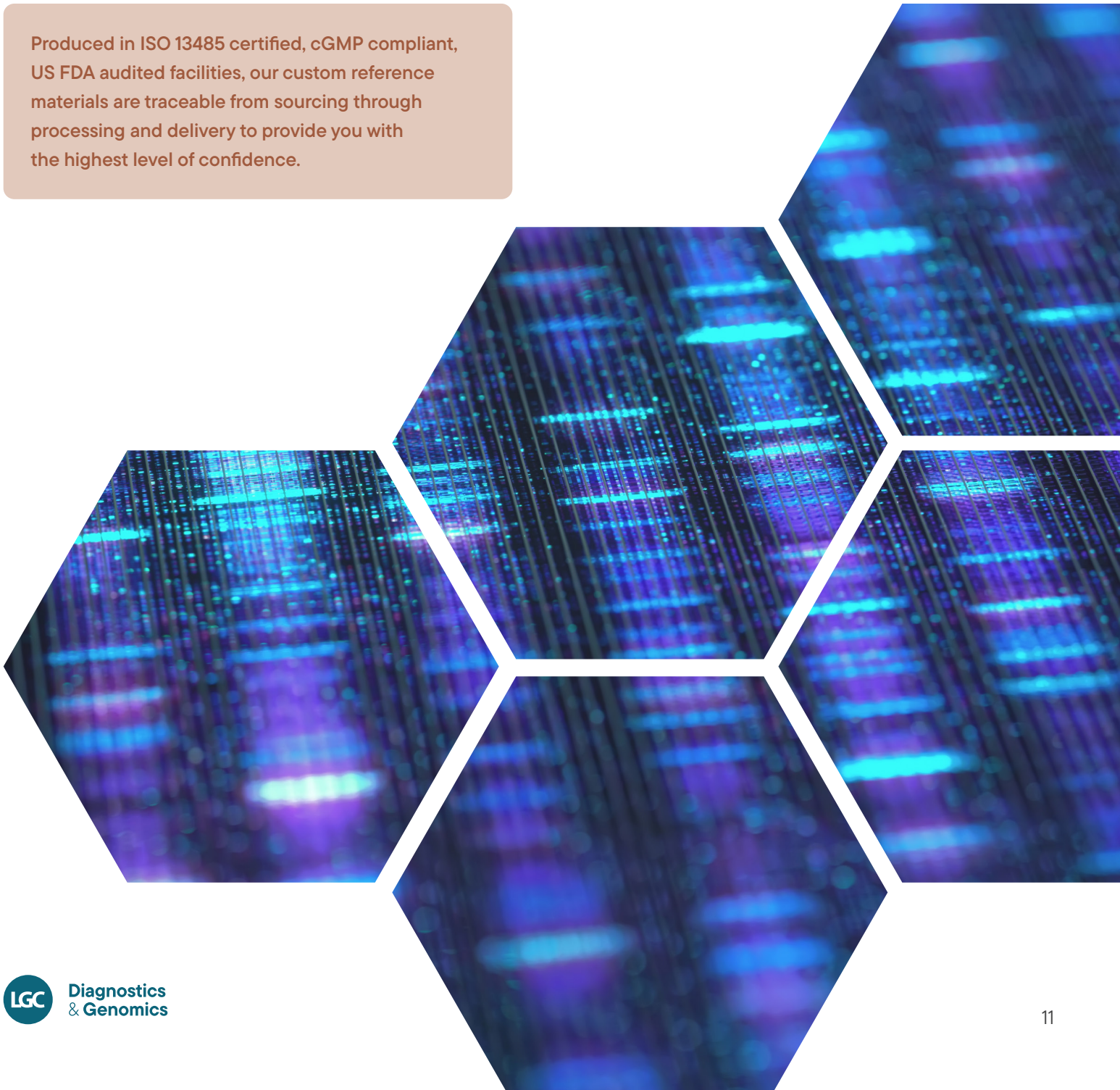
Product Description	Material #	Concentration	Fill in Size	Total Mass
Seraseq® Carrier Screening DNA Mix	0730-0569	30 ng/µL	1 vial x 20µL	600ng

Our Custom Offering

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

With our custom services, you enjoy a high degree of flexibility, access to our technical and scientific experts, and a rapid turnaround time tailored to your unique requirements and complex biomarkers. Complementing our portfolio of ready-to-use reference materials for NGS- and PCR-based assays, SeraCare develops custom, highly multiplexed solutions with any genomic variants relevant to your needs. Custom reference materials gives you unparalleled flexibility and scalability for validating your NGS workflow. Our biosynthetic spike-in technology let us provide a rapid turnaround time for your custom materials – typically 8-12 weeks.

Produced in ISO 13485 certified, cGMP compliant, US FDA audited facilities, our custom reference materials are traceable from sourcing through processing and delivery to provide you with the highest level of confidence.



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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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