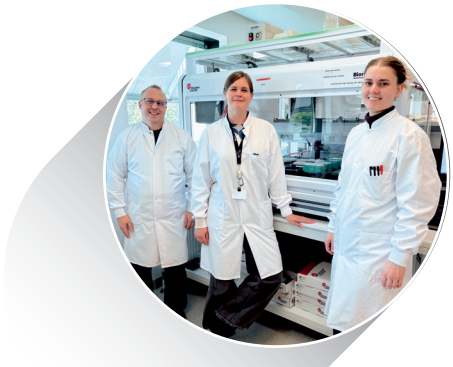
In Denmark, Newborn Screening Gets a Boost from Streamlined DNA Purification

The sparQ™ Lysis Kit delivers consistent, reliable results, even from very low sample volumes



From left: **Jacob Sønderby Pedersen**, Lab Automation Technician, **Caroline Mentzel, Ph.D.**, and **Amalie Kjær Hartman Jacobsen, MSc.** at SSI, using the sparQ Lysis Kit within the high-capacity automated process.

There are any number of traits that make babies unique, but in Denmark, Greenland, and the Faroe Islands, there's one thing they have in common: their newborn screening is performed by highly trained scientific experts at the Statens Serum Institut (SSI). Part of the Danish Ministry of Health, SSI has a broad remit to improve health through disease control and research. When it comes to newborns, scientists screen for 25 diseases, all chosen because they can be prevented or treated with early detection.

Screening just about every baby born in Denmark, Greenland, and the Faroe Islands is no trivial task. SSI typically screens 55,000 to 60,000 babies every year – the equivalent of 160 or so babies each day! Fortunately, the diseases included in the newborn screening process are quite rare. About 70 babies each year test positive for one of the 25 diseases.

Caroline Mentzel, Ph.D., NGS scientist in the institute's Department of Congenital Disorders, is a key member of SSI's newborn genetics screening laboratory. The team quickly process the newborn samples, which are collected two to three days after birth, and perform follow-up confirmatory testing and disease-focused gene sequencing for the babies who test positive.

Newborn screening is a high-stakes operation. False positive results could subject a baby to invasive follow-up testing and the family to unnecessary anxiety; false negatives mean that preventable or easily treatable diseases could progress to much more serious stages before being detected. Mentzel and her team at SSI know that results have to be accurate – which means that every component in the screening workflow has to be scrutinized for reliability.

High-Throughput Screening

Recently, SSI began investigating the possibility of incorporating nanopore sequencing technology to resolve a genetic region that couldn't be investigated with traditional short-read sequencing. Doing so would require more DNA volume than her lab was getting from its regular DNA purification kit.

But the team didn't just need more DNA; they also needed a solution that worked well with their highly automated laboratory setup. To screen as many babies as they do, they use a high-capacity automated liquid handling workstation, running the screening assay on all available samples once a week. The assay itself involves many cumbersome steps. Even with the automation assist, "it's a lot of work," Mentzel says.

In the face of all of these challenges, Mentzel's team evaluated the new sparQ Lysis Kit from Quantabio. The kit is the latest addition to Quantabio's comprehensive newborn screening workflow for sequencing dried blood spot samples. Extracting genomic DNA from the dried blood spot samples collected from newborns can be challenging due to limited sample volume, potential DNA degradation, and the presence of inhibitors from

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the blood matrix and filter paper. This can reduce the yield and quality of DNA, compromising the accuracy and reliability of downstream genetic analyses. The new sparQ Lysis Kit uses advanced chemistry that enables rapid and efficient extraction of double-stranded genomic DNA from just three standard blood spot punches in less than 50 minutes. The kit was designed to work well with automated and high-throughput workflows.

In the SSI evaluation, the sparQ Lysis Kit got high marks for ease of use, particularly with the bead-based extraction process used in the lab. "Some of the other kits involve more processes. My student tried this kit and liked it better because it's so straightforward," Mentzel says. "I go down to the lab and hear from the technicians that this is an easy process, and they appreciate it."

Production Mode

After a successful evaluation in the lab, the sparQ Lysis Kit is set to become a regular part of SSI's newborn screening workflow. "This kit runs very well with our liquid handling workstation," Mentzel says. "It definitely solves a problem we had with automation and low yield." Even with very small sample volumes, her team saw consistently reliable results when using the new kit.

In addition to standard newborn screening, Mentzel also sees a role for this kit in a research project that will involve batch sequencing using pooled DNA samples. During the analysis process, sequence results have to be deconvoluted and assigned back to their sample of origin – something that can only happen with clean, reliable results generated from high DNA input volumes.

Based on the SSI experience, Mentzel would recommend the sparQ Lysis Kit to other scientists running high-throughput workflows. "If you need a lot of DNA and it's a high-throughput project, this is a kit that's easy to automate," she says. "It wins over other bead based kits and over column purification in projects with a lot of samples."

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For more information about the sparQ Lysis Kit, visit **https://www.quantabio.com/product/sparq-lysis-kit**.