

The simple and elegant way to unlock genetic information

Extracta DBS rapidly recovers PCR-ready genomic DNA from challenging dried blood spot samples in 30 minutes



Mei Baker, MD, FACMG
Co-Director of Newborn Screening, Wisconsin State Laboratory of Hygiene, Professor of Pediatrics, University of Wisconsin School of Medicine and School of Public Health

"We make our technology choices based on several criteria, such as ease of use, consistent performance, and applicability to challenging samples. I have tried several alternatives and the combination of Quantabio Extracta DBS and ToughMix® reagent provided the best overall assay performance when we performed real-time PCR assays to measure T cell receptor excision circles (TRECs) used for SCID screening."

There are currently tens of millions of DNA samples processed around the world each year. Sample types range from fresh-frozen to FFPE to plasma to even dried blood spots (DBS). Unfortunately, these samples are often limited in quantity and quality, resulting in lower yields of DNA. Lower yields often negatively impact the assay performance for downstream translational research applications that use PCR, real-time PCR or even Next-Generation Sequencing (NGS) technologies to screen and analyze for genetic mutations associated with inherited and infectious diseases.

Over the past decade, there have been significant advancements in the way researchers extract and recover high-quality DNA. One of the early pioneers in public health genetics, Dr. Mei Baker at the Wisconsin State Laboratory of Hygiene, has developed a higher throughput and more efficient DNA extraction chemistry. Dr. Baker is best known for establishing the world's first Newborn Screening (NBS) program for Severe Combined Immunodeficiency (SCID), commonly known as "bubble boy disease".

We recently caught up with Dr. Baker to discuss her latest research projects, including her DNA extraction technique, which was patented by the Wisconsin Alumni Research Foundation

(WARF) and is now licensed and sold as part of Quantabio's Extracta DBS. She has been able to demonstrate more efficient and reproducible target quantification by combining Extracta DBS with the Quantabio qPCR ToughMix* reagents.

Personal commitment to improving NBS protocols

During my career, I have gained a deep appreciation for both the clinical and research side of public health genetics and genomics. After graduating from the Anhui Medical University, I practiced medicine for several years in China. I received molecular genetics and clinical biochemical training in the United States. The combination of professional training, job opportunity, and scientific passion led me to public health genetics and genomics. I have focused my career on applying emerging technologies to implement new and improve ongoing screening tests for disorders such as, SCID, cystic fibrosis, fragile X, Pompe Disease, and spinal muscular atrophy.

*Custom qPCR ToughMix



With more than 10 years of working experience in the public health field I have learned that for these tests to be routine, we need more practical and simple approaches. I call it the need for "simple elegance." Simple elegance does not mean cheap, it means robust, easy-to-use and dependable technology. A simple and elegant sample prep solution leads to better downstream application performance.

Challenges with sample quality and quantity

We have 30 people in our lab, handling everything from data entry to testing, reporting, and follow-up. In terms of sample volume, we process more than 90,000 samples per year, and our daily sample load can be as high as 700.

With DBS samples, you are dealing with significant quality and quantity challenges. The collection card captures a very limited and variable amount of DNA due to individual variability and used portions of dried blood specimens. A typical punch disk is 3.2 mm, which is equivalent to 3.2 µl of blood. In addition, amplifying small quantities of DNA could potentially lead to unwanted preferential amplification in PCR, and result in bias in subsequent data.

Since millions of samples are processed in public health labs every year, we need a high-throughput solution capable of handling this demand. People usually think this means an automated liquid handling platform, but these instruments

often face some difficulties associated with the physical nature of the dried blood spots. The robustness of Extracta DBS should help reduce or eliminate the need for these expensive robots.

Reproducible Quantification of TRECs for SCID

We make our technology choices based on several criteria, such as ease of use, consistent performance, and applicability to challenging samples. I have tried several alternatives and the combination of Quantabio Extracta DBS and ToughMix reagent provided the best overall assay performance when we performed real-time PCR assays to measure T cell receptor excision circles (TRECs) used for SCID screening.

Extracta DBS offers significant throughput and efficient improvements over available alternatives since it was designed for the higher throughput public health laboratory requirements. This simple 30-minute single step solution for crude extraction of genomic DNA does not require a purification step. The extracted DNA has been successfully used for Sanger and next generation sequencing assays.

References

1. Baker et al. 2016. Improving newborn screening for cystic fibrosis using next-generation sequencing technology: a technical feasibility study. *Genet Med.* 18(3):231-238.
2. Baker et al. 2010. Implementing Routine Testing for Severe Combined Immunodeficiency within Wisconsin's Newborn Screening Program. *Public Health Rep.* 125 Suppl 2, 88-95.
3. Verbsky et al. 2012. Newborn Screening for Severe Combined Immunodeficiency, The Wisconsin Experience (2008-2011). *J. Clin. Immunol.* 32, 82-88.

Extracta DBS is intended for molecular biology applications. This product is not intended for the diagnosis, prevention or treatment of a disease.

Extracta DBS is licensed under US Patent 9,206,468 and foreign equivalents for non-commercial research purposes only.

Trademarks: PerfeCta®, and ToughMix® are registered Trademarks of Quantabio, LLC.

Quantabio products are intended for molecular biology applications. The products are not intended for the diagnosis, prevention or treatment of a disease.
MK-CS-0001 REV 02 Mei Baker 0121