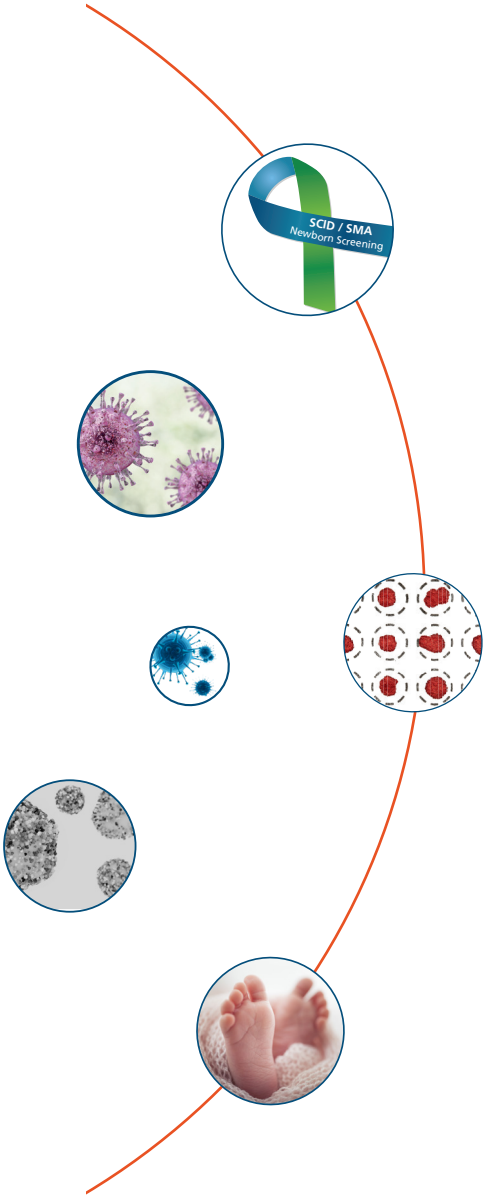
Stronger Science for Newborn Screening

As newborn screening expands to include a broader range of genetic and infectious conditions, the need for sensitive, reliable molecular tools continues to grow. Quantabio's PCR, qPCR, and NGS solutions enable accurate detection of SNPs, gene deletions and duplications, and pathogens. These capabilities support critical applications with precision and speed to help laboratories meet rising demands with stronger, faster science.

Overcoming Molecular Testing Challenges

Genetic tests and assays are often plagued by difficult to amplify genomic regions: repeat regions, AT- and GC-rich regions along with PCR inhibitors. Quantabio's PCR, qPCR and NGS products are designed to amplify through these difficult regions ensuring high assay performance.



SNP DETECTION

Rapidly detect SNPs in genomic DNA via singleplex or multiplex real-time qPCR assays enabling variant detection in genes causing SCID and SMA.

PATHOGEN DETECTION

Optimized, molecular workflows using qPCR enable early detection of critical pathogens such as Cytomegalovirus (CMV).

WHOLE GENOME SEQUENCING

Enables rapid detection of multiple SNPs and small structural variants in critically ill newborns, establishing a stronger, more scalable standard for modern screening.

Learn More

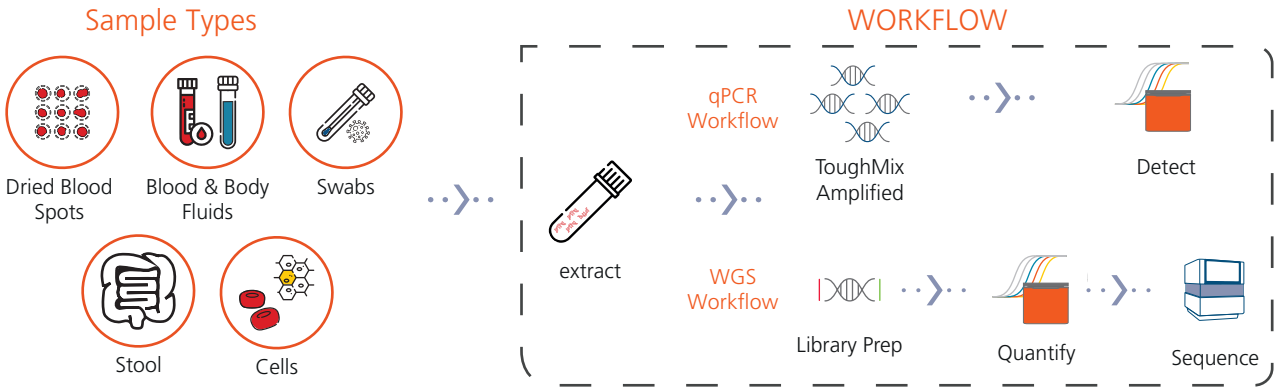
ToughMix. Experience the Amplification Difference!

ToughMix chemistry is the trade secret difference to our mastermix formulations. For more than 20 years, it has helped scientists amplify and analyze challenging samples by overcoming common PCR inhibitors. ToughMix formulations can be used directly from crude lysates as well as from purified, extracted samples.

What can ToughMix do?

- Work directly with crude lysates
- Avoid expensive and time-consuming purification steps
- Compatible with a wide range of probe designs and detection chemistries
- High quality *Taq* polymerase – free of residual host DNA
- Neutralize problem causing inhibitors present in crude samples

Inhibitor	Common sources	Reagent performance	
		Competitor	ToughMix
Hematin	Blood Dried blood spots	–	✓
Hemoglobin	Blood	✓	✓
Polysaccharides	Feces Plant tissues	–	✓
Melanin	Hair Skin	–	✓



Discover the Right Product by Application

	Product	Pathogen Detection	SNP Detection (qPCR)	Amplicon Seq	Whole Genome
Extraction	Extracta DBS	X	X	X	
	sparQ Lysis		X	X	X
qPCR	PerfeCTa qPCR ToughMix	X	X		
	PerfeCTa qPCR Multiplex ToughMix	X	X		
NGS	sparQ DNA Frag and Library Prep				X
	sparQ DNA Library Prep				X
	sparQ PureMag Beads			X	X
	sparQ Universal Library Quant			X	X

ToughMix.
Designed for Diverse Newborn Screening qPCR Applications.

SNP Detection - Germline Mutations

Starting from just a few drops of blood spotted onto storage cards, various qPCR assays are run to detect commonly known SNPs associated with disease status. Rapid testing is essential to quickly identify Single Nucleotide Polymorphisms (SNPs) or other mutations, report findings to the family and begin treatment. Many public health labs use Extracta DBS for fast extraction of DNA from dried blood spots (DBS). This crude, dirty extraction method is then well-suited for use with ToughMix formulations for qPCR analysis.

Taylor *et al*. (2015) aimed to expand the existing, widely used assay for severe combined immunodeficiency (SCID) by adding a target for the detection of spinal muscular atrophy (SMA). By utilizing PerfeCTa qPCR ToughMix, they generated clear, distinct amplification curves for each condition by qPCR (Figure 1).

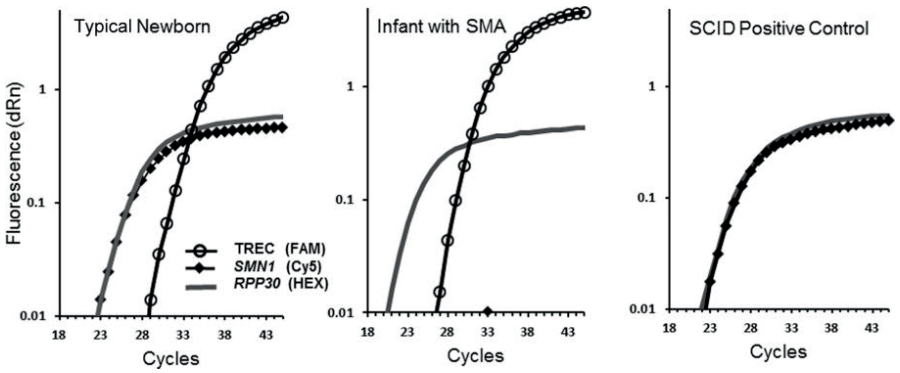


Figure 1 **Triplex TREC–SMN1–RPP30 DBS real-time PCR amplification curves.** DNA targets were amplified directly from dried blood spot (DBS) punches. Amplification of RPP30 was observed in all samples, confirming sample integrity. SMN1 was absent in the spinal muscular atrophy (SMA) patient sample, while TREC was undetectable in the severe combined immunodeficiency (SCID)-positive control. Figure from Taylor *et al*. Newborn blood spot screening test using multiplexed real-time PCR to simultaneously screen for Spinal Muscular Atrophy and Severe Combined Immunodeficiency, 2015.

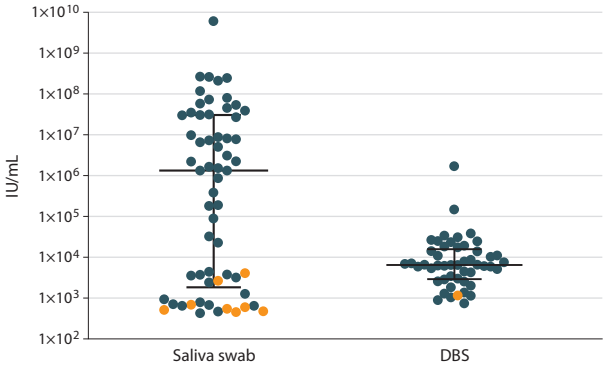


Figure 2 **Viral load distribution of cytomegalovirus in saliva and dried blood spots.** Figure from Dollard et al. Sensitivity of dried blood spot testing for detection of congenital cytomegalovirus infection, 2021.

Pathogen Detection

Viral pathogenic infections at birth also pose a serious public health problem. For example, congenital cytomegalovirus (CMV) infection is one of the most common infections leading to developmental problems and birth defects. Dollard *et al*. (2021) utilized Extracta DBS for extraction from dried saliva swabs and dried blood spots for rapid crude extraction. Additionally, the associated CDC laboratory utilized PerfeCTa FastMix II for CMV detection (Figure 2).

sparQ.
Designed for Newborn Screening Sequencing Applications.

WHOLE GENOME SEQUENCING

As sequencing costs continue to decrease, whole genome sequencing (WGS) can provide comprehensive, reliable, and rapid genetic information regarding genetic disorders present in newborns. Quantabio has developed the sparQ DBS-seq Workflow to provide a complete end-to-end solution for extraction and library prep for rapid WGS for newborn screening (Figure 3). This workflow features the sparQ Lysis Kit to facilitate rapid extraction of high-molecular-weight genomic DNA from DBS samples. The extracted DNA can then be used as input directly into the sparQ DNA Frag & Library Prep Kit for a robust and scalable solution for PCR-free or amplified whole genome library preparation.

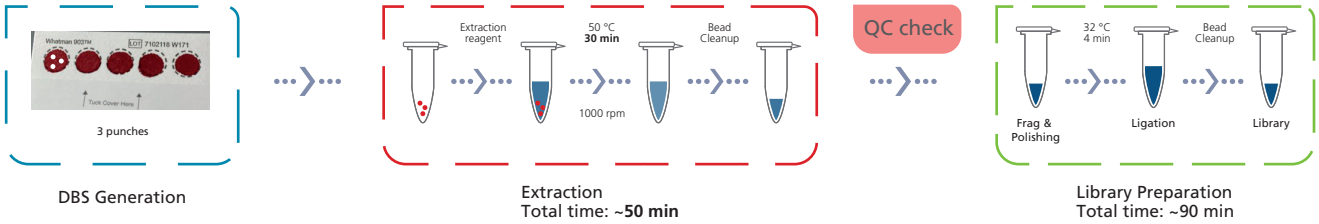


Figure 3 sparQ DBS-seq Workflow pairing the sparQ Lysis Kit for extraction with the sparQ DNA Frag & Library Preparation kit for library preparation. The sparQ Lysis Kit allows for extraction of high-quality DNA from DBS punches in only 50 minutes. After a QC check of the resulting DNA, library preparation can be performed in approximately 90 minutes.

The sparQ Lysis Kit features fast, high-quality extraction. Following the protocol of the sparQ Lysis Kit, high molecular weight (>50 kb) double-stranded gDNA is isolated from three 3.2 mm DBS punches in 50 minutes, yielding an average of 210 ng from newborn blood and 300 ng from cord blood (Figure 4A). Sequencing of PCR-free libraries generated by the sparQ DNA Frag & Library Prep Kit on NovaSeq® 6000 Xp (Illumina) with a target of 160 Gb per sample resulted in a uniform >30X genome coverage, low duplication rates (<8%), and expected variant calling (Figure 4B).

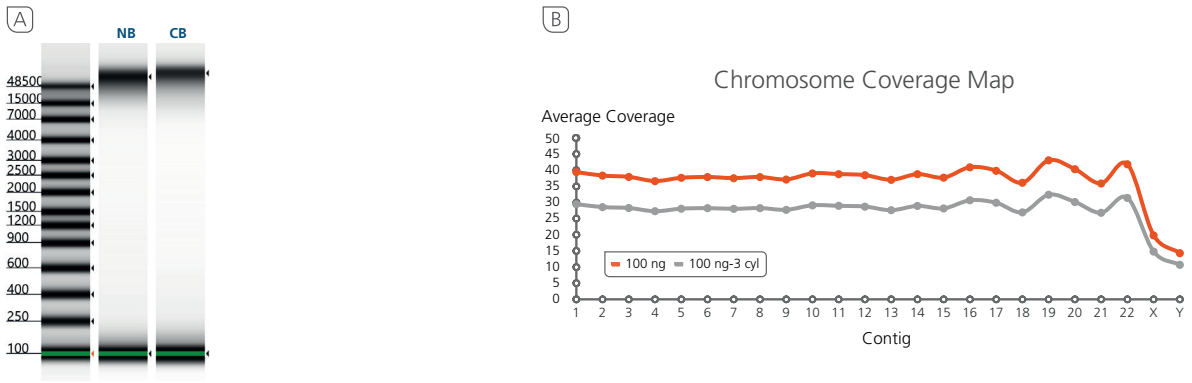


Figure 4 DNA recovered from dried blood spots on Whatman W903 cards. **A** DNA sizes >48 kb were generated using the sparQ Lysis kit from three DBS punches generated using newborn blood (NB) or cord blood (CB), as seen by TapeStation gDNA ScreenTape. **B** Sequencing results showed genome-wide coverage was greater than 30X across all chromosomes, which is essential for determining most germline mutations with high confidence.

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