



## TaqMan SCID/SMA Plus Assay

Multiplex real-time PCR assay for SCID and SMA  
newborn screening research

# TaqMan SCID/SMA Plus Assay

For newborn screening research



The Applied Biosystems™ TaqMan® SCID/SMA Plus Assay is a multiplex real-time PCR assay for simultaneous screening of severe combined immunodeficiency (SCID) and spinal muscular atrophy (SMA) directly from dried blood spot (DBS) samples.

## Key features

- Optimized assay for SCID and SMA
- Direct extraction from DBS results in less hands-on time
- Compatible with 96- or 384-reaction protocols
- High-quality results for 96- or 384-well format with less than 45 min qPCR run time using fast cycling protocol
- Minimal liquid-dispensing steps for easy automation

## Background

Intensive research in early detection of SCID and SMA is essential for prevention of permanent disabilities or death of infants.

SCID is a primary immunodeficiency disorder characterized by lack of functional T and B cells, resulting in reduced cellular and humoral immunity. Several genes may be involved, so analysis is based on measurement of episomal DNA products resulting from immune cell development.

SMA is an autosomal recessive disorder characterized by degeneration of motor neurons in the spinal cord and brain, leading to progressive muscular weakness. The disease is caused by defects in the *SMN1* gene, which codes for a protein necessary for normal functioning of motor neurons.

## Complete research solution from DBS to result

The TaqMan SCID/SMA Plus Assay detects *SMN1*, *TREC*, and *KREC*, as well as the RNase P gene as an internal genomic control. The assay solution contains a mixture of primers and probes for each target plus an *SMN2* blocker. With only 4 liquid-dispensing steps, the DBS sample preparation workflow requires less than 40 min of hands-on time for desiccated DBS samples, or less than 20 min when using freshly prepared, routine DBS samples. Protocols for both 96- and 384-well plates are available, making this a scalable assay adaptable to high-throughput environments. A real-time PCR instrument with at least 5 filters is required to detect the reporter dyes plus the passive reference dye.

## TaqMan SCID/SMA Plus Assay

Consistent and robust-performing multiplex assay for SCID/SMA. TaqMan® SCID/SMA Plus Assay has been optimized for high specificity to detect homozygous deletion of *SMN1*.

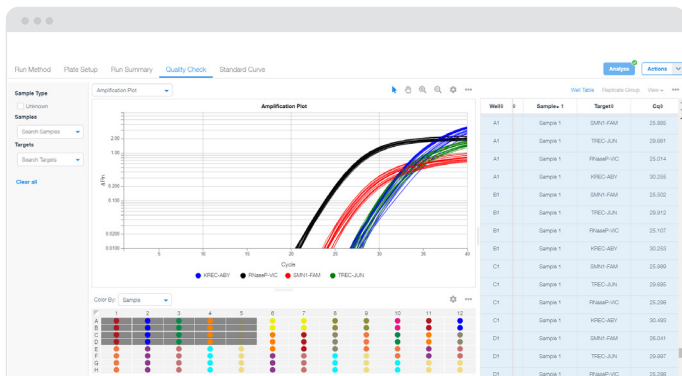
## QuantStudio real-time PCR systems

The TaqMan SCID/SMA Plus Assay is compatible across multiple Applied Biosystems™ QuantStudio™ real-time PCR systems. The assay requires a real-time PCR system with at least 5 filters. Recommended QuantStudio™ real-time PCR systems (96 and 384 blocks):

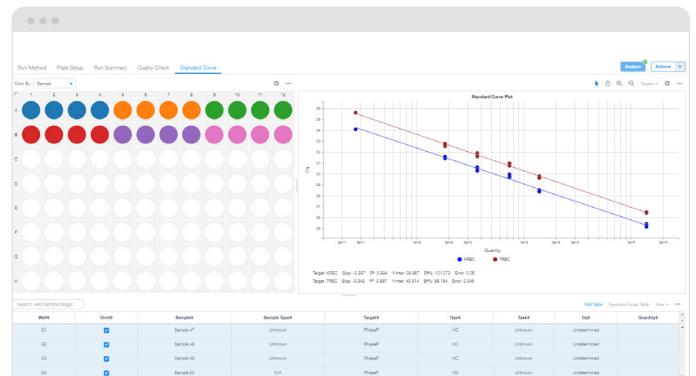
- QuantStudio 5 system
- QuantStudio 6 Flex system
- QuantStudio 7 Flex system
- QuantStudio Dx system (in RUO mode)
- QuantStudio 12K Flex system

## Software: Design and Analysis

Easy-to-use software with integrated analysis suite.



Design and analysis results: Target amplification



Design and analysis results: Standard curve

## A simple, automatable workflow

### TaqMan SCID/SMA Plus Assay: 3-step workflow from sample to results



Sample Preparation



Amplification



Results

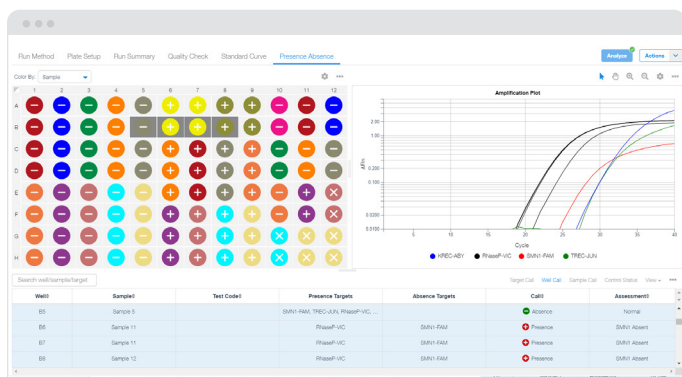
A direct DNA extraction from Dried Blood Spot (DBS), with minimal hands on time.

A rapid amplification method, with a 30\* minute data acquisition time.

Robust multiplex assay with consistent performance and high target specificity.

\*Note: Using a QuantStudio 5 96W 0.1 mL instrument

## Kit performance



Design and analysis result: Absence presence calling

Since the implementation of a single test for both SMA and SCID is currently sought after globally, Thermo Fisher Scientific has developed a real-time SMA/SCID multiplex assay that permits concomitant measurement of *SMN1*, *TREC*, *KREC* and *RNaSP* reference. We have designed the SMA assay to target exon 7 of the *SMN1* gene and effectively eliminated non-specific detection of the highly similar *SMN2* gene by competitive inhibition. This high *SMN1* target specificity of SMA assay limits both ambiguous calls and requirement for retesting. We also confirmed *TREC* and *KREC* copy number detection capability of as low as 10 copies per reaction. Additionally, we were able to substantially improve DBS sample preparation method and reduce the number of steps to a minimum. In conclusion, Thermo Fisher Scientific has developed a **highly specific, sensitive, and robust multiplex assay for SMA and SCID testing with a rapid and streamlined turnaround workflow to aid further research efforts.**

## Why choose the TaqMan SCID/SMA Plus Assay for your newborn screening research



### Quality

- Consistent and robustly performing multiplex assay for SCID/SMA
- Optimized for high specificity to detect homozygous deletion of *SMN1*



### Simplicity

- No DNA purification needed after DBS extraction, resulting in less hands-on-time
- Only 4 liquid-dispensing steps, and a qPCR Fast cycling protocol of <20 - 40 min\*



### Reliability

- Consistent quality\*\*
- Complete workflow in one kit
- Global support network including on-site customer training



### Cost Savings

- Workflow adaptable to standard laboratory equipment and compatible with QuantStudio platforms
- With minimal liquid-dispensing steps, the assay can be easily automated

\*<20 minutes of hands-on time for sample preparation when using freshly prepared, routine DBS samples as opposed to desiccated DBS specimens.

\*\*verified design and manufacturing process for reagents

### Ordering information

Product	Quantity	Cat. No.
TaqMan SCID/SMA Plus Assay	100 reactions	A52275
	1,000 reactions	A48566
	4,000 reactions	A48567
	8,000 reactions	A48568
	20,000 reactions	A48569
QuantStudio Real-Time PCR System	1 instrument	Various*
On-site training	1 day	A48619

\*Assay is compatible with QuantStudio 5, 6 Flex, 7 Flex, 12K Flex, and Dx (in RUO mode) systems.

Find out more at [thermofisher.com/nbs](https://thermofisher.com/nbs)

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