

AVAILABLE SOON

EVALUATE IN YOUR LAB

Sickle Cell Disease added to SPOT-it™ panel

Simultaneous detection of

01

SCD

Sickle Cell Disease

NEW

02

SCID

Severe Combined
Immunodeficiency

03

SMA

Spinal Muscular
Atrophy

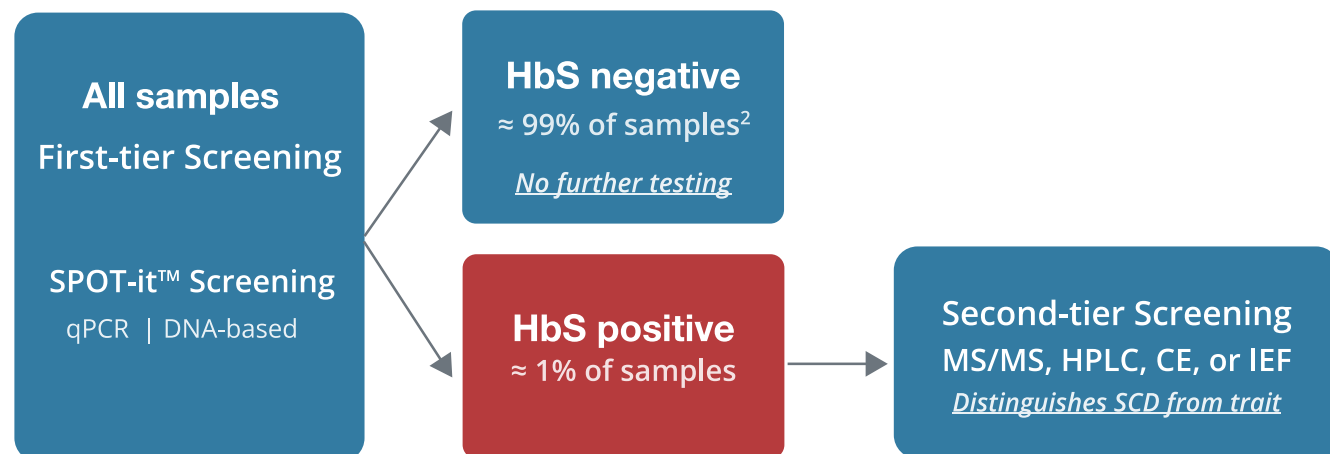
Designed for routine screening labs

- + No liquid-handling equipment or separate clean room required.
- + Pre-filled, ready-to-use reagent plates reduce hands-on time.
- + Lyophilized format for simplified storage and extended shelf life.

Screen for SCID, SMA and SCD simultaneously

The **SPOT-it™ TREC, SMN1 & HbS Lyo Screening Kit**¹ serves as a single-tier test for SCID and SMA and as a first-tier test for SCD. The assay advances only HbS-positive samples to second-tier testing to distinguish SCD from Sickle Cell Trait.

This approach significantly reduces sample volume requiring analysis through methods like MS/MS, HPLC, CE, or IEF, delivering rapid, cost-efficient results that are unaffected by prematurity or blood transfusions, reducing the need for repeat sampling.



¹ SPOT-it™ TREC, SMN1 & HbS Lyo Screening kit is currently not authorised for diagnostic use.

² Based on an assumed carrier frequency of 1%; this may vary significantly across populations and geographic regions.

Markers for SCID, SMA and SCD

- + TREC for SCID**
Severe Combined Immunodeficiency, or SCID, is a group of rare, inherited disorders caused by mutations in genes essential for the development of infection-fighting immune cells.
Early detection through newborn screening is based on the detection of **T-cell receptor excision circles** (TRECs)—circular DNA fragments formed during the T-cell maturation process.
- + SMN1 for SMA**
Spinal Muscular Atrophy, or SMA, is an autosomal recessive neuromuscular condition that affects motor neurons in the spinal cord, leading to progressive muscle weakness and atrophy.
In 95% of SMA patients, the disease is caused by a homozygous deletion of **exon 7 in the SMN1 gene**. This aberration is used as a marker for SMA in newborn screening.
- + HbS for SCD**
Sickle Cell Disease, or SCD, is an autosomal recessive disorder caused by a point mutation in the beta-globin gene, resulting in the production of abnormal hemoglobin, referred to as **hemoglobin S** (HbS).
HbS causes red blood cells to adopt a rigid, sickle shape, leading to vaso-occlusion, hemolytic anemia, and organ damage. The presence of **HbS point mutation** is used as the marker for SCD in newborn screening.

SPOT-it™ Kits — from DBS to result within 3 hours

01

Sample Distribution

A 3.2 mm diameter punch from each DBS sample is distributed into the SPOT-it™ Filter Plate.

02

Rinse Dried Blood Spots

SPOT-it™ Rinse Solution is added to each sample to remove hemoglobin and other impurities.

03

DNA Elution

Rinsed punches are transferred into the pre-filled SPOT-it™ Elution Plate using plate-stacking technology. The plate is heat-incubated to elute DNA.

04

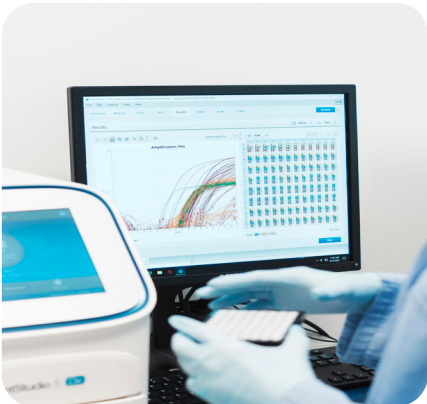
qPCR Amplification

Eluted DNA is transferred to the pre-filled SPOT-it™ qPCR Plate using plate-stacking technology, and the qPCR run is launched.

05

Result Interpretation

Control parameters are verified and results are visualized in the **SPOT-it™ Analysis Software**.



SPOT-it™ Newborn Screening

Product Name	Screening for		Samples per kit
SPOT-it™ TREC Screening Kit	SCID	CE IVD	328 or 984 samples
SPOT-it™ TREC & KREC Screening Kit	SCID & XLA	CE IVD	324 or 972 samples
SPOT-it™ TREC & SMN1 Screening Kit	SCID & SMA	CE IVD	328 or 984 samples
SPOT-it™ TREC, KREC & SMN1 Screening Kit	SCID, SMA & XLA	CE IVD	324 or 972 samples
SPOT-it™ TREC, SMN1 & HbS Lyo Screening Kit	SCID, SMA & SCD	Available soon	328 or 984 samples

*SPOT-it™ Screening kits are CE-marked (IVDD/IVDR) in vitro diagnostic medical devices for professional use. Products may not be authorised or available in all countries. Contact ImmunoIVD for availability and regulatory status in your region.

Early Detection for Life-Saving Interventions

>1M

Newborns screened with
SPOT-it™ kits each year

Since 2016

Trusted partner in Newborn
Screening Programs

Learn how SPOT-it™ assays
support efficient
SCID, SMA, and SCD screening

EVALUATE IN YOUR LAB

info@immunoivd.com



DOC-13913 ver.0