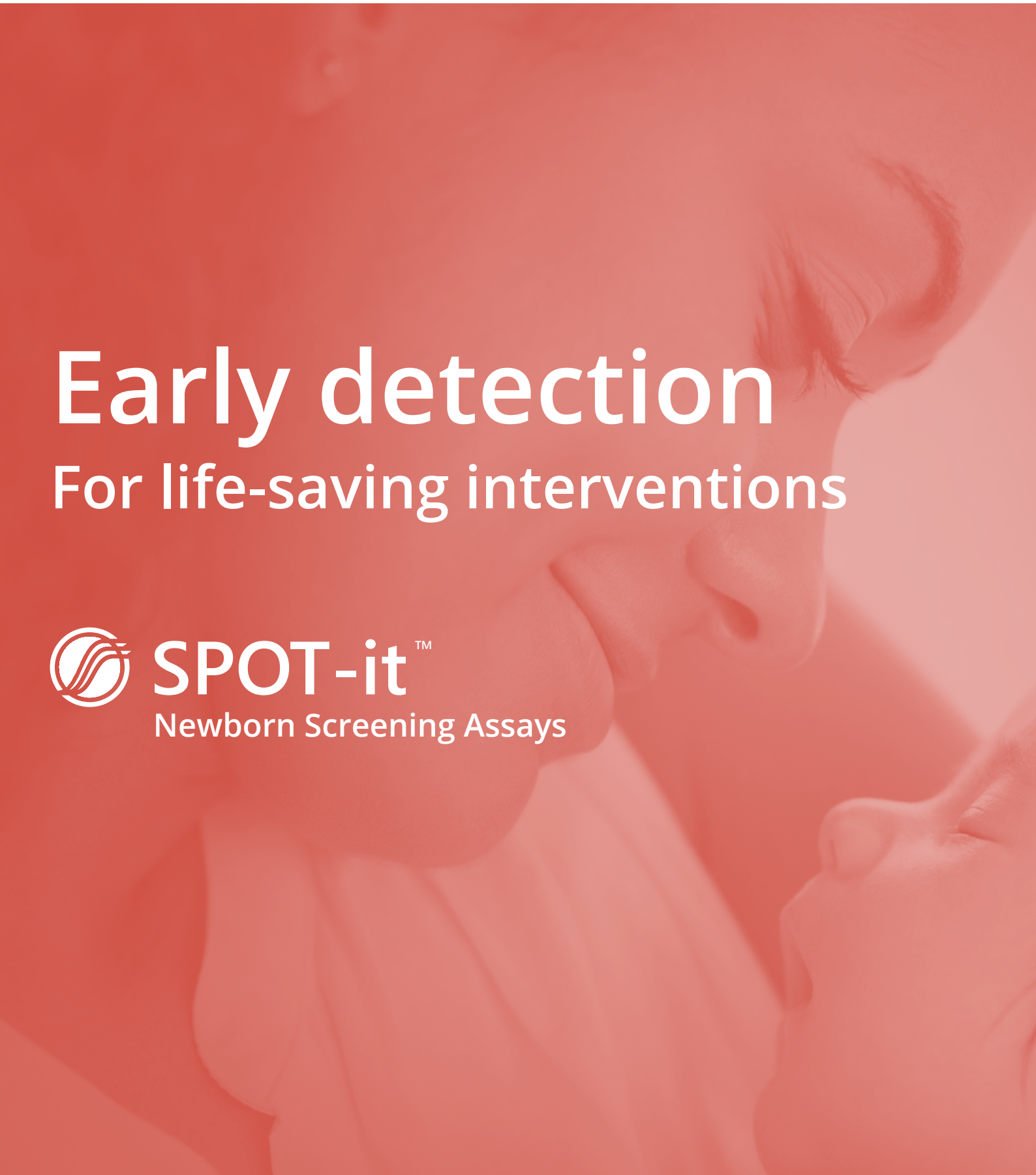
Immuno IVD

Early detection

For life-saving interventions



SPOT-it™

Newborn Screening Assays

Immuno IVD

SCID Screening

Severe Combined Immunodeficiency (SCID) is a genetic condition where children do not produce functional T-cells and, in some cases, B-cells; both of which are essential for a healthy immune system. Newborns with SCID suffer from a reduced immune response to infections which, without early detection and treatment, can be fatal.

TRECs are produced during the maturation of T-cells. Lack of TRECs can indicate low or no production of T-cells, and newborns identified with low or no TRECs may suffer from SCID and should be referred for confirmatory testing.

Newborn screening programs for SCID can detect a lack of T-cell production within a few days after birth, identifying children who may have the disease before they fall ill. This technology allows rapid intervention and effective treatment before the child develops severe infections.

SMA Screening

Spinal Muscular Atrophy (SMA) is a genetic neuromuscular condition where patients gradually lose muscle strength, affecting their ability to sit up, walk and, in severe cases, breathe and swallow. The disease is autosomal recessive and caused by genetic variants in the survival motor neuron 1 (*SMN1*) gene, leading to very low levels of the SMN protein which is crucial for functional motor neurons. In 95% of SMA patients the disease is caused by a homozygous deletion of exon 7 of *SMN1*.

Newborn screening for SMA can detect the genetic variant in the *SMN1* gene before symptoms appear and allow for the earliest possible treatment with the opportunity to delay muscular degeneration. Due to this fast-moving field, there are few long-term studies on early-onset treatment prognosis, but it is expected to offer a higher quality of life for patients for as long as possible.

XLA Screening

X-linked agammaglobulinemia (XLA) is a genetic disorder where patients are not able to produce enough functional B-cells. Newborns with XLA suffer from a reduced immune response to infections, and the condition may be life-threatening. XLA is an X-linked recessive disease which almost exclusively affects males.

KRECs are produced during the process of recombination of B-cells. Lack of KRECs can indicate low or no production of functional B-cells, and newborns identified with low or no KRECs may suffer from XLA and should be referred for confirmatory testing.

Newborn screening for XLA can detect the lack of KRECs within a few days after birth, facilitating an earlier diagnosis for patients with XLA and earlier initiation of treatment.

Immuno IVD

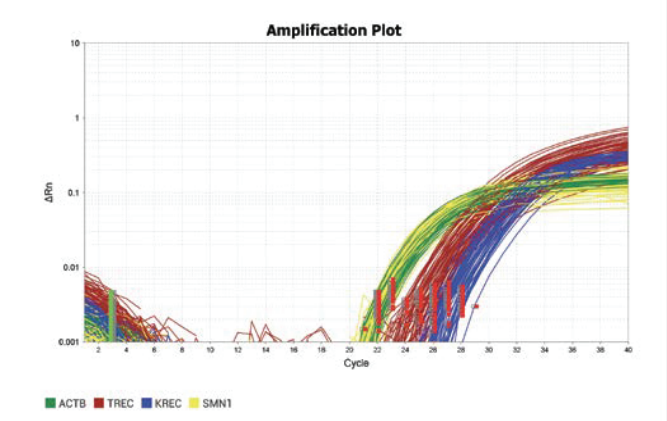
SPOT-it™ Screening Assay

- Complete solution for SCID, XLA and SMA screening – including DNA extraction
- From Dried Blood Spot to screening result within three hours
- Pre-filled and ready-to-use plates including standard curve, control samples and ACTB reference
- Just two pipetting steps for quick and easy handling

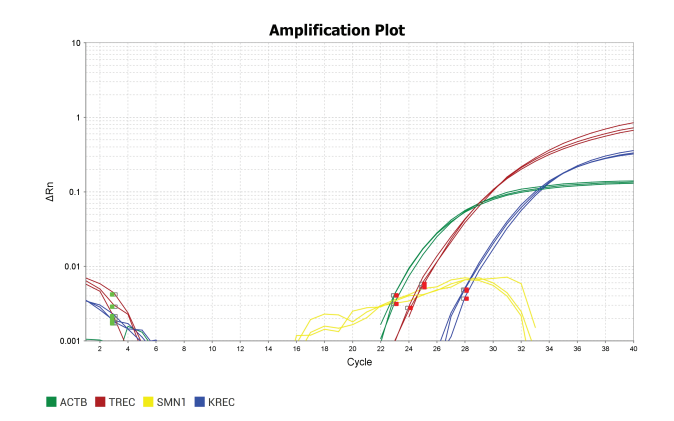
4-step Process

1. **Sample Distribution**
   Use a 3.2 mm diameter punch to distribute samples from Dried Blood Spots
2. **Rinse DBS**
   Rinse and rehydrate the Dried Blood Spots for 20 minutes on a plate shaker
3. **DNA Elution**
   Elute DNA by heating samples to 95 °C for 30 minutes
4. **qPCR**
   Amplify and quantify targets using qPCR

Amplification Plots from SPOT-it™ TREC, KREC & SMN1 Screening Kit



Amplification plot from normal newborn samples



Amplification plot from SMA control samples

References

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SPOT-it™ Analysis Software

The SPOT-it™ Analysis Software is intended for assisting laboratory technicians in the qualitative visualization of data obtained from the qPCR instrument software when using any of the SPOT- it™ Screening Kit products. The software checks all quality parameters of a plate run (e. g. the standard curve acceptance criteria, the quality controls and the internal control gene), and assigns the labels “Within Range”, “Out of Range” or “Inconclusive” to each sample. Plates that fail on quality control parameters are automatically flagged, and samples are colour-coded for easy identification.

Product Information

Product code	Product name	Screening for: SCID	XLA	SMA	
12-2015-T	SPOT-it™ TREC Screening Kit	✓			CE IVD
12-2015-TK	SPOT-it™ TREC & KREC Screening Kit	✓	✓		CE IVD
12-2020-TS	SPOT-it™ TREC & SMN1 Screening Kit	✓		✓	CE IVD
12-2020-TKS	SPOT-it™ TREC, KREC & SMN1 Screening Kit	✓	✓	✓	CE IVD
10.180	SPOT-it™ Analysis Software	✓	✓	✓	CE IVD

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