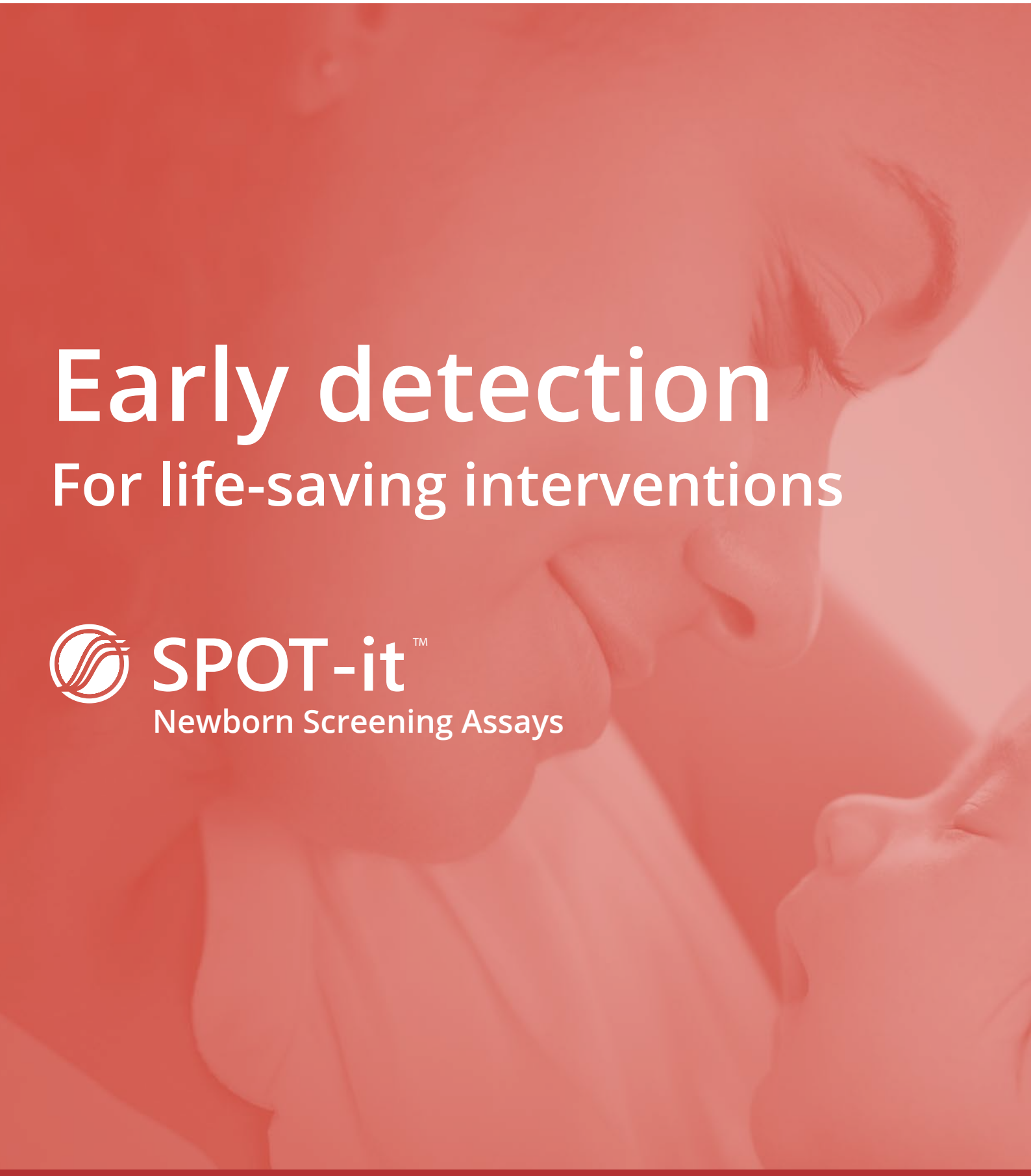
Immuno IVD

Early detection

For life-saving interventions

SPOT-it™

Newborn Screening Assays



Immuno IVD

SCID Screening

Severe Combined Immunodeficiency, or 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 lead to severe complications and is 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, or 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.

References

· Blom, M. *et al*. Introducing Newborn Screening for Severe Combined Immunodeficiency (SCID) in the Dutch Neonatal Screening Program. Int. J. Neonatal Screen. (2018), 4,40.doi.org/10.3390/ijns4040040

· Blom, M. *et al*. Parents' Perspectives and Societal Acceptance of Implementation of Newborn Screening for SCID in the Netherlands. J Clin Immunol (2020). doi.org/10.1007/s10875-020-00886-4

· Cossu, F. Genetics of SCID. Ital J Pediatr, 76 (2010). doi.org/10.1186/1824-7288-36-76

· D'Amico, A. *et al*. Spinal muscular atrophy. Orphanet J Rare Dis (2011). 6,71. doi.org/10.1186/1750-1172-6-71

· Glascock, J. *et al*.Treatment Algorithm for Infants Diagnosed with Spinal Muscular Atrophy through Newborn Screening. J Neuromuscul Dis (2018). 5(2):145-158. doi.org/10.3233/-JND-180304

· Heimall, J. *et al*. Immune reconstitution and survival of 100 SCID patients post–hematopoietic cell transplant: a PIDTC natural history study. Blood (2017). doi.org/10.1182/blood-2017-05-781849

· Puck, J. Neonatal Screening for Severe Combined Immunodeficiency (SCID). Curr Opin Pediatr. (2011). doi: 10.1097/MOP.0b013e32834cb9b0

· SMA Europe. SMA Newborn Screening Alliance website(www.sma-screening-alliance.org). Accessed 2021.

Immuno IVD

SPOT-it™ Screening Assay

· Complete solution for SCID & SMA screening - including DNA extraction
· From Dried Blood Spot to screening result within 3 hours
· Pre-filled and ready-to-use plates including samples for standard curve generation
· Just two pipetting steps for quick and easy handling
· Uses conventional 3.2 mm Dried Blood Spots as input
· CE-IVD marked assay available for TREC, TREC & KREC kits

4-step Process

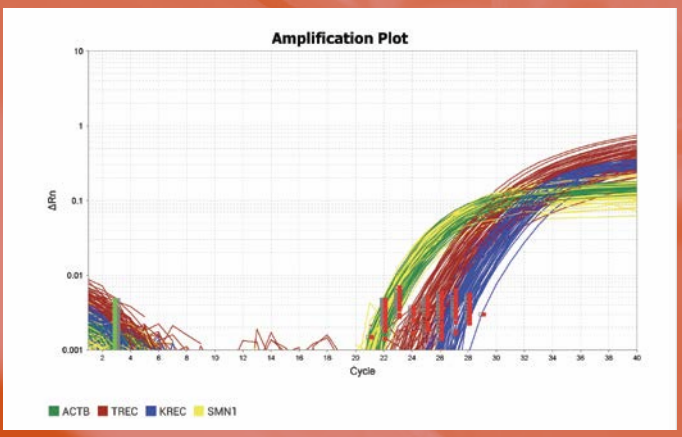
1. **Sample Distribution**
Use a 3.2 mm diameter punch to distribute samples from Dried Blood Spots.

2. **Rinse DBS**
Rinse and rehydrate the Dried Blood Spots for 20 minutes on a plate shaker

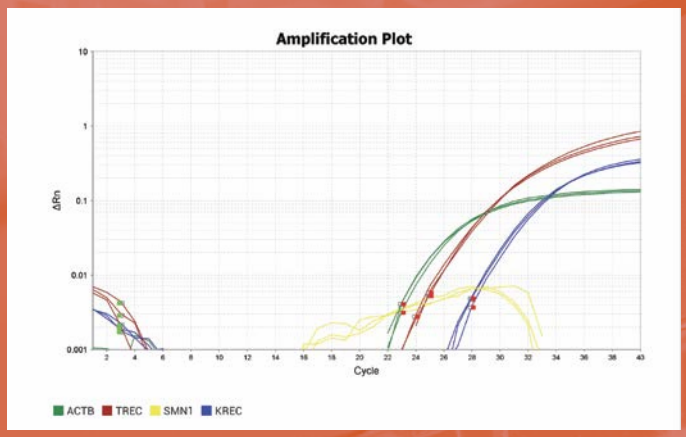
3. **DNA Elution**
Elute DNA by heating samples to 95 °C for 30 minutes

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

*Preliminary data from kit under development

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	✓	✓	✓	Coming soon!

About ImmunoIVD

ImmunoIVD is a Swedish company founded in 2015 by researchers who wanted to develop a sensitive SCID assay, suitable for usage in routine newborn screening laboratories. We are proud of our status as a specialized, research-intensive company keeping the focus on developing rapid and effective assays for newborn screening to facilitate early detection, prompt treatment and more successful outcomes for children affected by primary immunodeficiencies and other rare diseases.

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