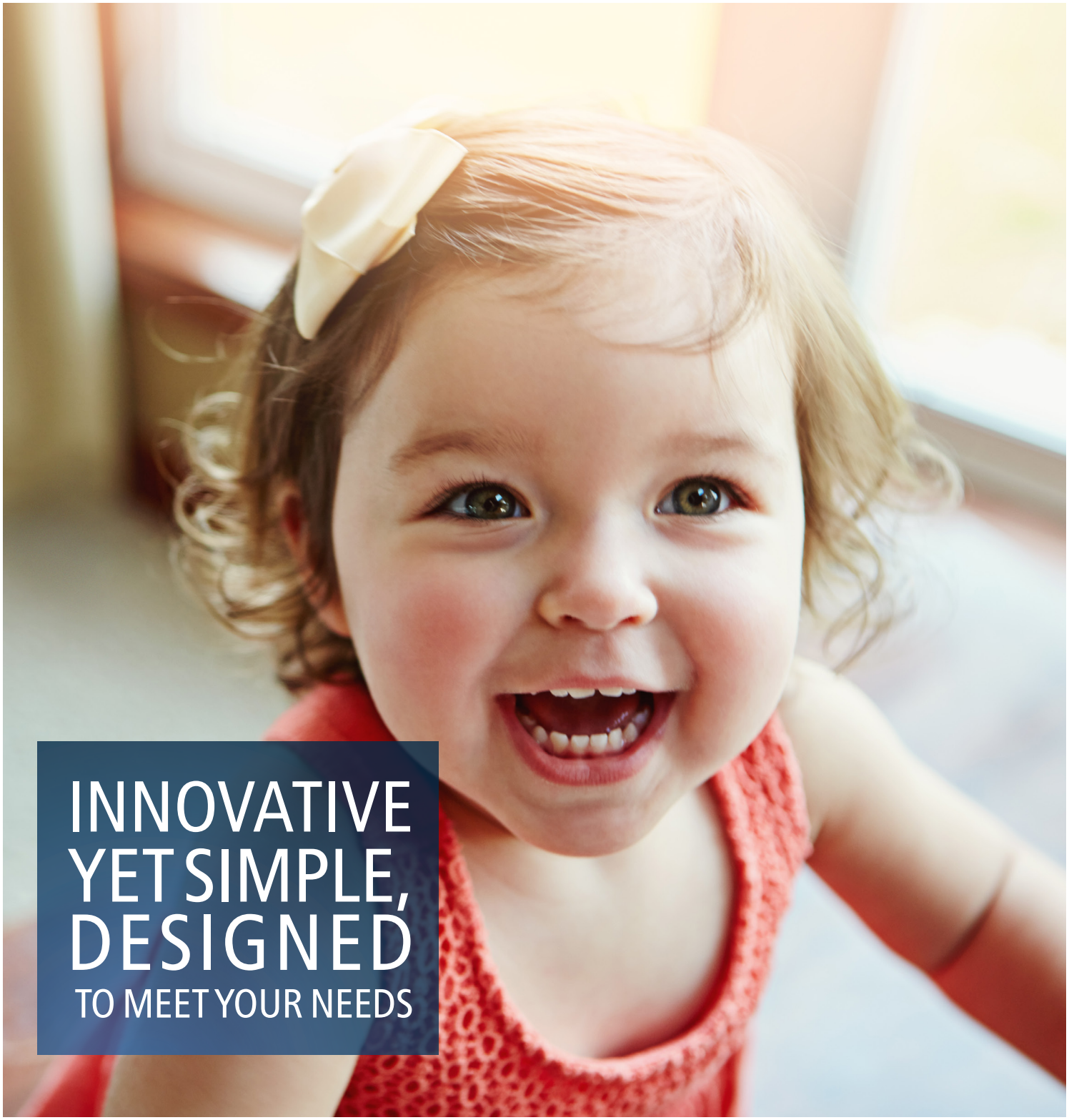
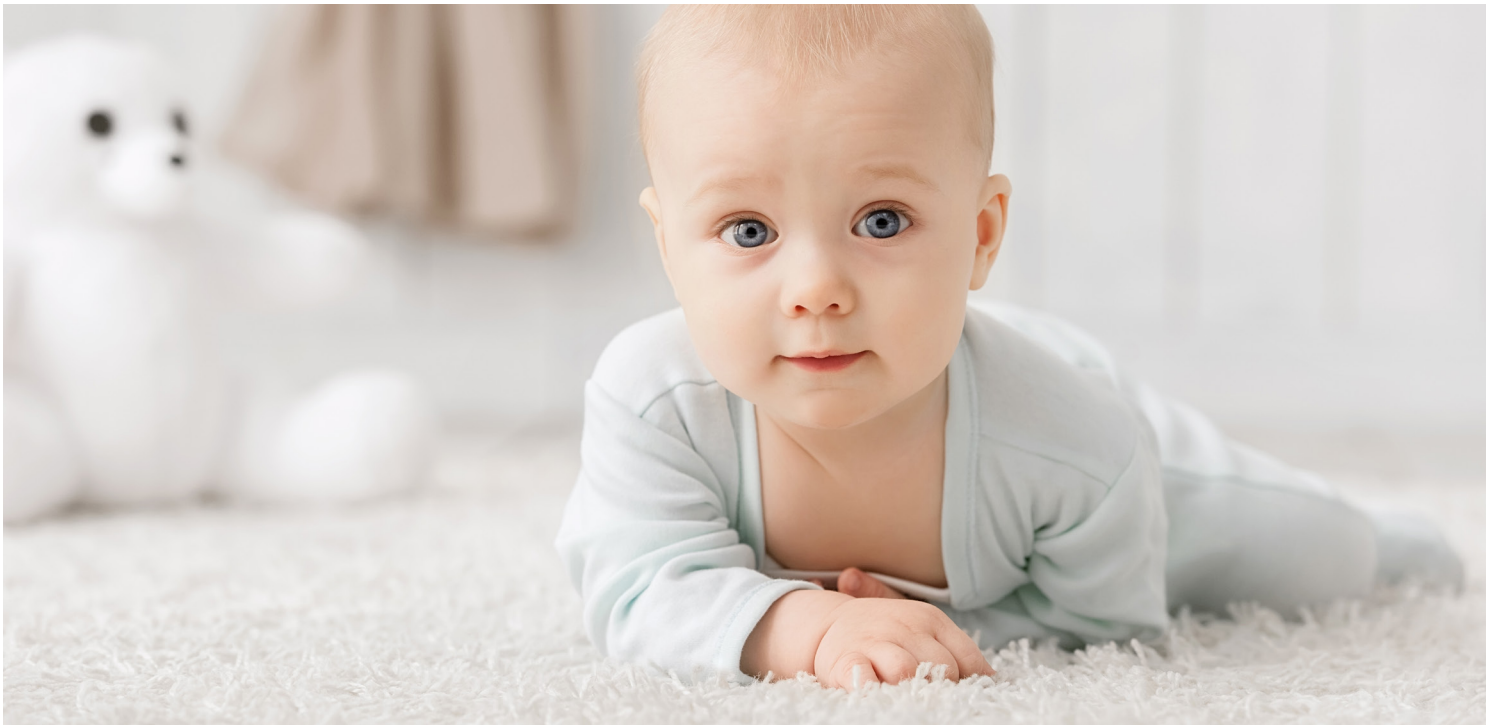


EONIS™ Platform

PerkinElmer®
For the Better



EONIS™ platform - Simultaneous screening of SMA, SCID and XLA from a single DBS punch

Multiplexing of SMA, SCID and XLA screening can now be done in one assay utilizing Real-Time PCR technology, without increasing daily hands-on work load and complexity. This will provide a future-proof solution for cost-efficiently obtaining additional disorder information.

Combination of DNA extraction, multiplexing and automation allows maximum efficiency in workflow. Analytes are quantified using innovative analysis method without calibrators, saving more room for your precious samples. For maximum flexibility and throughput, as well as for minimum hands-on time, liquid handling can be used both for DNA Extraction and for PCR setup in the EONIS™ platform.

SCALABLE.
ROBUST.
FUTURE-PROOF.

KEY BENEFITS OF THE EONIS™ PLATFORM

- ☑ Excellent screening performance
- ☑ Ability to multiplex analytes
- ☑ Innovative analysis method without calibrators
- ☑ Robust streamlined workflow with possibility for automation
- ☑ Flexibility in throughput
- ☑ Dedicated Analysis Software

SMA

Why screen for SMA?

Spinal muscular atrophy (SMA) is leading genetic cause of infant death. It is a neuromuscular disease inherited in an autosomal recessive manner.

SMA is characterized by muscle weakness and atrophy resulting from progressive degeneration and loss of the lower motor neurons in the spinal cord and the brain stem nuclei. The onset of weakness ranges from before birth to adolescence or young adulthood and life span varies from less than 6 months to normal.

SMA patients often require comprehensive medical care involving multiple disciplines but the field has recently seen major advances in clinical therapies approved by FDA and EMA. [2b]

What is SMN1?

SMA is caused by mutations in the survival motor neuron gene 1 (SMN1). The survival motor neuron gene (SMN) is composed of 9 exons, with a stop codon present near the end of exon 7 and it has been shown to be the primary spinal muscular atrophy–determining gene [2b]. The molecular diagnosis of SMA consists of the detection of the absence of exon 7 of SMN1. [2.f]

SCID

Why screen for SCID?

Severe Combined Immunodeficiency (SCID) is a group of disorders characterized by a severe defect in T cell production and function.

Typically, infants with SCID will die due to infection by one year of age unless the infant's immune system is restored through treatment [1a].

The preferred treatment is bone marrow/stem cell transplantation. Evidence from large case series indicates that children receiving early stem-cell transplant for SCID have improved outcomes compared with children who are treated later [1a].

What are TRECs?

T-cell Receptor Excision circles (TRECs) are circular DNA fragments generated during T-cell receptor rearrangement. In healthy neonates, TRECs are made in large numbers, while in infants with SCID, they are barely detectable.

XLA

Why screen for XLA?

X-linked agammaglobulinemia (XLA) is a condition occurring almost exclusively in males and characterized by lack of B-cells which helps protect the body against infection.

Children with XLA are usually healthy for the first 1 or 2 months of life. After this time children develops recurrent infections that can develop into severe, life-threatening bacterial infections. With treatment to replace antibodies, infections can usually be prevented, improving the quality of life for people with XLA. [3a],[3b]

What are KRECs?

Kappa-deleting element recombination circle (KREC) are extra-chromosomal circular DNA segments generated in B-cells during their maturation in the bone marrow. The quantification of KRECs provides novel information in the management of B-cell immunity-related diseases and is considered as an effective marker to screen for XLA in newborns [3c],[3d].

4 EASY STEPS OF EONIS™ ASSAY

EONIS™ assay consists of four easy steps; punching, extraction, amplification and data analysis.

Dedicated analysis software enables quantification of TREC and KREC, while SMN1 results are reported qualitatively. SMA carrier status will not be detected or reported. RPP30 is used as an internal amplification control as well as basis for the quantification. Software analysis includes automated run acceptance criteria from kit controls to ensure that the quality of measured data is not compromised.

Punching of DBS samples and controls

Extraction with manual or automated workflow

Amplification of TREC, KREC, SMN1 & RPP30

Result interpretation in the EONIS™ analysis software

Amplification curves

PUNCHING → **EXTRACTION** → **AMPLIFICATION** → **DATA ANALYSIS**

CONFIGURABLE TO YOUR THROUGHPUT NEEDS

Panthera Puncher™ 9 Instrument
DBS Puncher

EONIS™ DNA Extraction kit
EONIS™ SCID-SMA kit

JANUS® Extraction Instrument
JANUS® Mini Extraction instrument
JANUS® PCR mastermix instrument

QuantStudio™ Dx Real-Time PCR instrument

EONIS™ analysis software

The design of the assay and system enables automation to further streamline the workflow of high-throughput laboratories without compromising the sample traceability from punch to result.

EONIS™ ORDER GUIDE

REAGENTS

- 3240-0010/-001U EONIS™ DNA Extraction Kit (1152 reactions)
- 3241-0010/-001U EONIS™ SCID-SMA kit (384 reactions)
- 3242-0010/-001U EONIS™ SCID-SMA kit (3 x 384 reactions)

SOFTWARE

- 5020-1000 EONIS™ analysis software
- 1003-0580 Personal Computer

ASSAY CONSUMABLES

- 4174-0010 96-well PCR plate
- 4158-0010 384-well PCR plate
- 4157-0010 DNA Extraction Plate
- 4159-0010 Optical PCR Seal
- 4156-0010 Adhesive Foil Seal

EONIS™ INSTRUMENTS

- 2031-0020 QuantStudio™ Dx 96 well, fast (CE-IVD)
- 2031-0010 QuantStudio™ Dx 384 well, fast (CE-IVD)
- 2031-1010 QuantStudio™ Dx Extended Warranty
- CLS150056 JANUS Extraction Instrument
- CLS153416 JANUS Mini Extraction Instrument
- CLS150057 JANUS PCR Mastermix Instrument

CALIBRATION CONSUMABLES

- 3243-0010 96-well Calibration Plates Kit
- 3248-0010 96-well Calibration Plate, Background
- 4172-0010 96-well Spectral Calibration Plate with Cy5 Dye
- 4173-0010 96-well Spectral Calibration Plate with Cy5.5 Dye
- 3225-0010 384-well Calibration Plates Kit
- 3226-0010 384-well Calibration Plate, Background
- 4178-0010 384-well Spectral Calibration Plate with Cy5 Dye
- 4179-0010 384-well Spectral Calibration Plate with Cy5.5 Dye

1. Background publications for SMA

a. ACOG Committee Opinion, Spinal Muscular Atrophy, 2009, reaffirmed 2014

b. Prior and Finanger, Spinal Muscular Atrophy, GeneReviews (https://www.ncbi.nlm.nih.gov/books/NBK1352/), accessed on Oct, 2018

c. Boardman, Young, Griffiths, Newborn screening for spinal muscular atrophy: the views of affected families and adults, Am J Med Genet, 2016

d. Wang et al., Consensus Statement for Standard of Care in Spinal Muscular Atrophy, Journal of Child Neurology, 2007

e. Lefebvre S, Burglen L, Reboullet S, et al. Identification and characterization of a spinal muscular atrophy-determining gene, Cell, 1995

f. Prior TW, Spinal Muscular Atrophy Diagnostics, Journal of Child Neurology, 2007

g. Wang et al., Consensus Statement for Standard of Care in Spinal Muscular Atrophy, Journal of Child Neurology, 2007

2. Background publications for SCID

a. American College of Medical Genetics, Newborn Screening ACT Sheet, Severe Combined Immunodeficiency (SCID) and Conditions Associated with T Cell Lymphopenia, https://www.acmg.net/PDFLibrary/SCID.pdf

b. Buckley, Molecular defects in human severe combined immunodeficiency and approaches to immune reconstitution, Annu Rev Immunol, 2004

c. Kwan and Puck, History and Current Status of Newborn Screening for Severe Combined Immunodeficiency, Semin Perinatol, 2015

d. van der Spek et al., TREC Based Newborn Screening for Severe Combined Immunodeficiency Disease: A Systematic Review, J Clin Immunol, 2015

e. Puck et al., Newborn Screening for Severe Combined Immunodeficiency in 11 Screening Programs in the United States, JAMA ,2014

f. Serana et al, Use of V(D)J recombination exicision circles to identify T- and B-cell defects and to monitor the treatment in primary and acquired immunodeficiencies, Journal of Translational Medicine, 2013

3. Background publications for XLA:

a. Smith, E. and Berglöf, A. X-Linked Agammaglobulinemia in GeneReviews®, Adam MP, Ardinger HH, Pagon RA, et al., editors. Seattle (WA): University of Washington, Seattle; 1993-2019.

b. Shillitoe, B and Gennery, A. (2017): X-Linked Agammaglobulinaemia: Outcomes in the modern era. Clinical Immunology 183, 54-62.

c. Nakagawa, N. et al. (2011): Quantification of kappa-deleting recombination excision circles in Guthrie cards for the identification of early B-cell maturation. J. Allergy Clin Immunol. 128(1), 223-235 e2.

d. Barbaro, M. et al. (2017): Newborn Screening for Severe Primary Immunodeficiency Diseases in Sweden – a 2-year pilot TREC and KREC screening study. J. Clin Immunol. 37(1), 51-60.

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