



# SOLUTIONS AND TOOLS FOR SMA AND PID SCREENING

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**PerkinElmer 2022**

**Terhi Helenius**

Together we are making

# A DIFFERENCE FOR THE BETTER

Our newborn screening technologies have aided in screening over **750 MILLION BABIES WORLDWIDE** for a variety of life-threatening diseases, helping them get a healthier start.

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We help save more than **70 BABIES DAILY**

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Dedicated to develop **molecular NBS tests since 2014**



**50M**

SCREENED BABIES  
ANNUALLY



**100+**

CUSTOMERS  
WORLDWIDE



HELP SAVE

**70 BABIES**

DAILY USING  
PERKINELMER  
TECHNOLOGIES



**>30Y**

OF EXPERIENCE  
IN SCREENING

# PERKINELMER MOLECULAR PLATFORM



## EnLite Neonatal TREC

- SCID
- CE-IVD/FDA
- 2014



## Eonis

- SCID, XLA, SMA
- CE-IVD
- 2020

# EONIS™ SYSTEM OVERVIEW

SAMPLE  
COLLECTION

SAMPLE  
PREPARATION

ANALYSIS /  
MEASUREMENT

REAGENTS

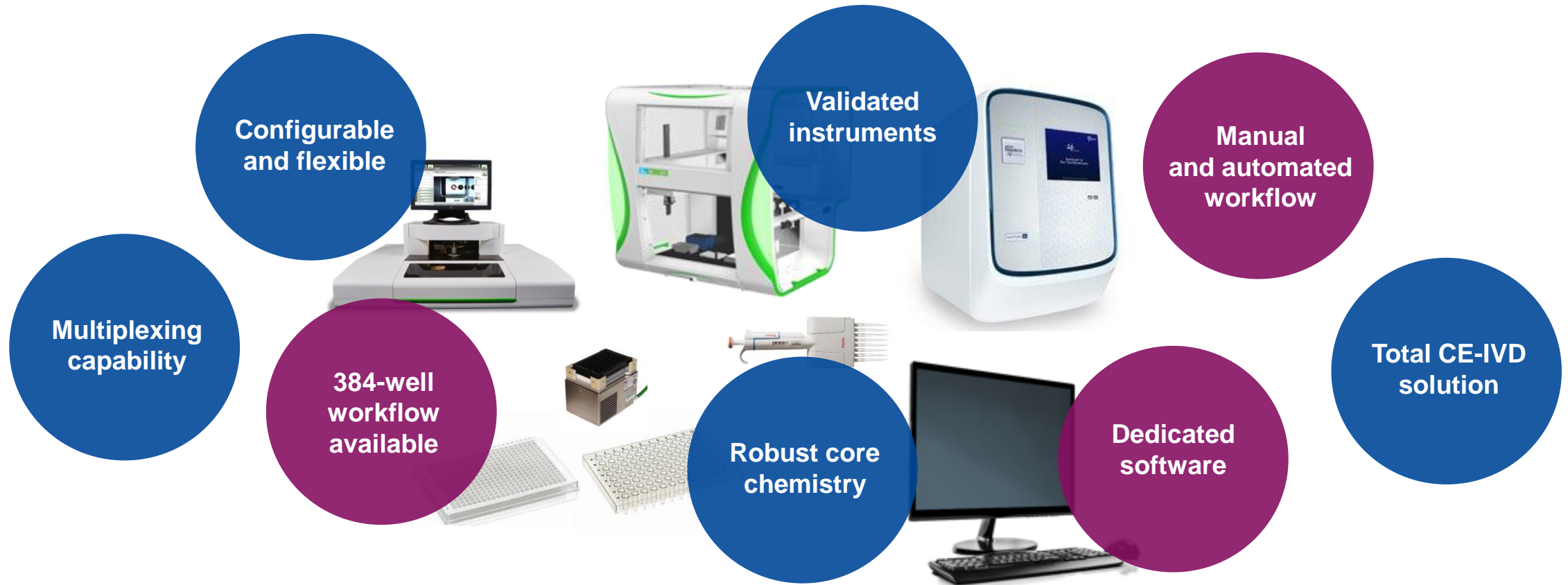
SOFTWARE



**MULTIPLEXED DETECTION OF SMA, SCID AND XLA FROM ONE DBS SPOT  
TOTAL CE-IVD SOLUTION**

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# EONIS™ SYSTEM KEY FEATURES



**CONFIGURABLE TO MEET YOUR LABORATORY'S THROUGHPUT NEEDS  
ROBUST CHEMISTRY**

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# PERKINELMER HAS GLOBAL PRESENCE

## UNITED STATES

> 2 M BABIES  
SCREENED FOR  
SCID SINCE 2014

## EUROPE

UK, GERMANY,  
ITALY, SPAIN,  
DENMARK

## AUSTRALIA

> 250 000 BABIES  
SCREENED FOR  
SMA SINCE 2018

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# CUSTOMER CASE: SMA SCREENING IN AUSTRALIA

**22 SCREEN POSITIVE**

MEDIAN AGE AT DIAGNOSIS **15 DAYS**

MEDIAN AGE AT TREATMENT **25 DAYS**

INCIDENCE APPROX. **1 IN 10 000**

**252 081**  
**BABIES**  
**SCREENED**

**→ RERUN RATE**  
**LESS THAN**  
**1%**

*"One has to remember this is  
a screening test."*



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- 2020









## Eonis Q

- SCID, XLA, SMA
- CE-IVD
- Coming soon



# EONIS Q – DESIGNATED BY FEEDBACK

-  Robust chemistry
-  TAT less than 2 hours
-  Accessible to all
-  Easy to train
-  Easy to implement
-  Easy to store and ship

*Product is under development*

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# NEW EONIS Q DRY CHEMISTRY SYSTEM WORKFLOW



**MOLECULAR TESTING MADE EASY AND ACCESSIBLE  
TO ALL LABORATORIES**

*Product is under development*

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# EONIS Q DRY CHEMISTRY SYSTEM

## KEY FEATURES



*Product is under development.*

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**THANK YOU!**

Questions?

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