



BLOW UP YOUR GENOMICS WORKFLOW.

Automate nucleic acid QC and get on with your life sciences.

If sample QC takes you more than two minutes, it's too manual. Fragment Analyzer™ takes the job off your hands—streamlining lab operations and wiping out errors. Just pipette once and it delivers truly reliable results via automated capillary electrophoresis.

No chips. No tapes. No compromises.

- Setup in seconds
- Get resolution down to 2 base pairs
- Detection starts at 5 pg/μL

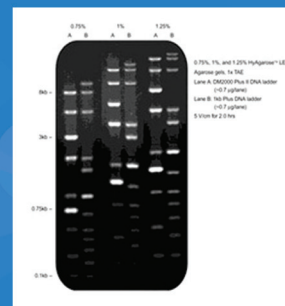
DITCH YOUR TIRED OLD WORKFLOW AT AATI-US.COM.



Agarose for Molecular Biology

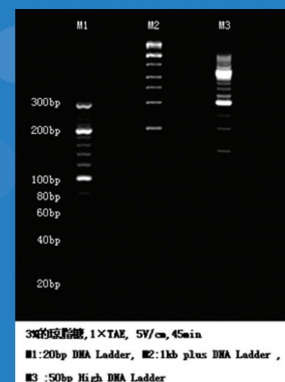
Besta™ LE Agarose, Multi-purpose

Besta™ LE Agarose is a low EEO, multi-purpose, standard melting point agarose that yields high resolution sharp DNA bands with high clarity and low background. Its optimized gel strength enhances ease of gel processing and handling.



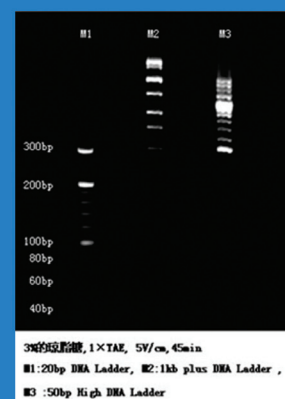
Besta™ LM Agarose

Besta™ LM Agarose is a Low Melting and gelling point agarose producing gels with great sieving properties and higher clarity when compared with standard agarose. Besta™ LM Agarose is ideal for in-gel manipulations which can be performed without prior extraction of the DNA from the gel slice.



Besta™ HR Agarose

Besta™ HR Agarose is a PCR grade, intermediate melting point agarose that efficiently separates small DNA fragments between 20 and 800 bp in length and yields ultra-high resolution with high clarity and low background. It is suitable for the analysis of AFLP's (Amplified Fragment Length Polymorphisms), STR's (Short Tandem Repeats) and tetra-nucleotide repeats.



Success is contagious

In 2011, Ion AmpliSeq™ technology enabled targeted sequencing from 10 ng of DNA or less. And the world responded.

In just four years we've designed over 25,000 custom panels for researchers around the world. With 10 ng of DNA or less, you can easily target sets of genes, and make sequencing a success for even old or degraded FFPE samples. Now that's an idea worth spreading.



AmpliSeq it. Because every sample matters.

Get more from your samples at thermofisher.com/ampliseq

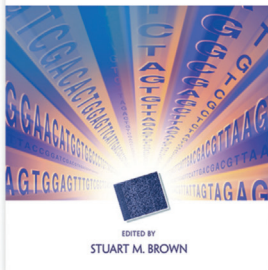
ThermoFisher
SCIENTIFIC



NEXT-GENERATION DNA SEQUENCING INFORMATICS

SECOND EDITION

NEXT-GENERATION
DNA SEQUENCING
INFORMATICS
SECOND EDITION



Edited by Stuart M. Brown, *New York University School of Medicine*

Next-generation DNA sequencing (NGS) technology has revolutionized biomedical research, making genome and RNA sequencing an affordable and frequently used tool for a wide variety of research applications including variant (mutation) discovery, gene expression, transcription factor analysis, metagenomics, and epigenetics. Bioinformatics methods to support DNA sequencing have become and remain a critical bottleneck for many researchers and organizations wishing to make use of NGS technology. This new edition provides a thorough, plain-language introduction to the necessary informatics methods and tools for analyzing NGS data and provides detailed descriptions of algorithms, strengths and weaknesses of specific tools, pitfalls, and alternative methods. Four new chapters cover experimental design, sample preparation, and quality assessment of NGS data; public databases for DNA sequencing data; de novo transcript assembly; proteogenomics; and emerging sequencing technologies. The remaining chapters from the first edition have been updated with the latest information. This book also provides extensive reference to best-practice bioinformatics methods for NGS applications and tutorials for common workflows. This edition addresses the informatics needs of students, laboratory scientists, and computing specialists who wish to take advantage of the explosion of research opportunities offered by new DNA sequencing technologies.

2015, 402 pages, illustrated (81 4C, 20 B&W), index
Hardcover \$61

ISBN 978-1-621821-23-6

CONTENTS

- | | | |
|---|--|--|
| Preface | 8 De Novo Assembly of Bacterial Genomes from Short Sequence Reads
<i>Silvia Argimón and Stuart M. Brown</i> | 14 Metagenomics
<i>Guillermo I. Perez-Perez, Miroslav Blumenberg, and Alexander V. Alekseyenko</i> |
| Acknowledgments | 9 De Novo Transcriptome Assembly
<i>Lisa Cohen, Steven Shen, and Efstratios Efstathiadis</i> | 15 Proteogenomics
<i>Kelly V. Ruggles and David Fenyo</i> |
| About the Authors | 10 Genome Annotation
<i>Steven Shen and Stuart M. Brown</i> | 16 Emerging DNA Sequencing Technologies and Applications
<i>Gerald A. Higgins and Brian D. Athey</i> |
| 1 Introduction to DNA Sequencing
<i>Stuart M. Brown</i> | 11 Using Next-Generation Sequencing to Detect Sequence Variants
<i>Jinhua Wang, Zuojuan Tang, and Stuart M. Brown</i> | 17 Cloud-Based Next-Generation Sequencing Informatics
<i>Konstantinos Krampis, Efstratios Efstathiadis, and Stuart M. Brown</i> |
| 2 Quality Control and Data Preprocessing
<i>Stuart M. Brown</i> | 12 ChIP-seq
<i>Stuart M. Brown, Zuojuan Tang, Christina Schweikert, and D. Frank Hsu</i> | Glossary |
| 3 History of Sequencing Informatics
<i>Stuart M. Brown</i> | 13 RNA Sequencing with Next-Generation Sequencing
<i>Stuart M. Brown and Jeremy Goecks</i> | Index |
| 4 Public Sequence Databases
<i>Stuart M. Brown</i> | | |
| 5 Visualization of Next-Generation Sequencing Data
<i>Phillip Ross Smith, Kranti Konganti, and Stuart M. Brown</i> | | |
| 6 DNA Sequence Alignment
<i>Efstratios Efstathiadis</i> | | |
| 7 Genome Assembly Using Generalized de Bruijn Digraphs
<i>D. Frank Hsu</i> | | |



www.cshlpress.org



Looking for A Genomic Research Partner?

Next-Generation Sequencing(NGS)

Whole genome Sequencing (Hiseq X Ten)
Exome Sequencing
Targeted Sequencing
Long Read Sequencing
Transcriptome Analysis, small RNA
Epigenomics

Bio Informatics

Assembly / Mapping
Variant (SNP / Indel) calling
CNV & Breakpoints
Expression Profiles
DEGs / miRNA
Enrichment Profiles
Gene Annotation

Capillary Sequencing

Microarray

Oligonucleotide Synthesis

Genetically Engineered Mouse

Next-Gen Sequencing

Getting the most out of your single-cell RNA-seq data?

TRANSCRIPTOME ANALYSIS WITH NEXT-GEN SEQUENCING

Be confident using tools with the highest sensitivity and reproducibility

SMART-Seq™ v4 Ultra™ Low Input RNA Kit for Sequencing



Accurate, sensitive, single-cell mRNA-seq

The SMART-Seq v4 Ultra Low Input RNA Kit for Sequencing, built on a combination of our experience and new LNA technology, is our most sensitive single-cell mRNA-seq kit. With direct cDNA synthesis from 1–1,000 intact cells (or 10 pg–10 ng of total RNA), mRNA-seq libraries generated from this kit outperform previously published protocols. We've used this kit to explore the differences and similarities between individual cells, showing the high-quality, consistent data that can be expected with the newest ultra-low input kit. Our continuous drive to improve our already best-in-class tools helps you push the frontiers of ultra-low input RNA-seq.

See more by visiting
www.clontech.com/single-cell-v4
or call 1.800.662.2566



Scan to find out more

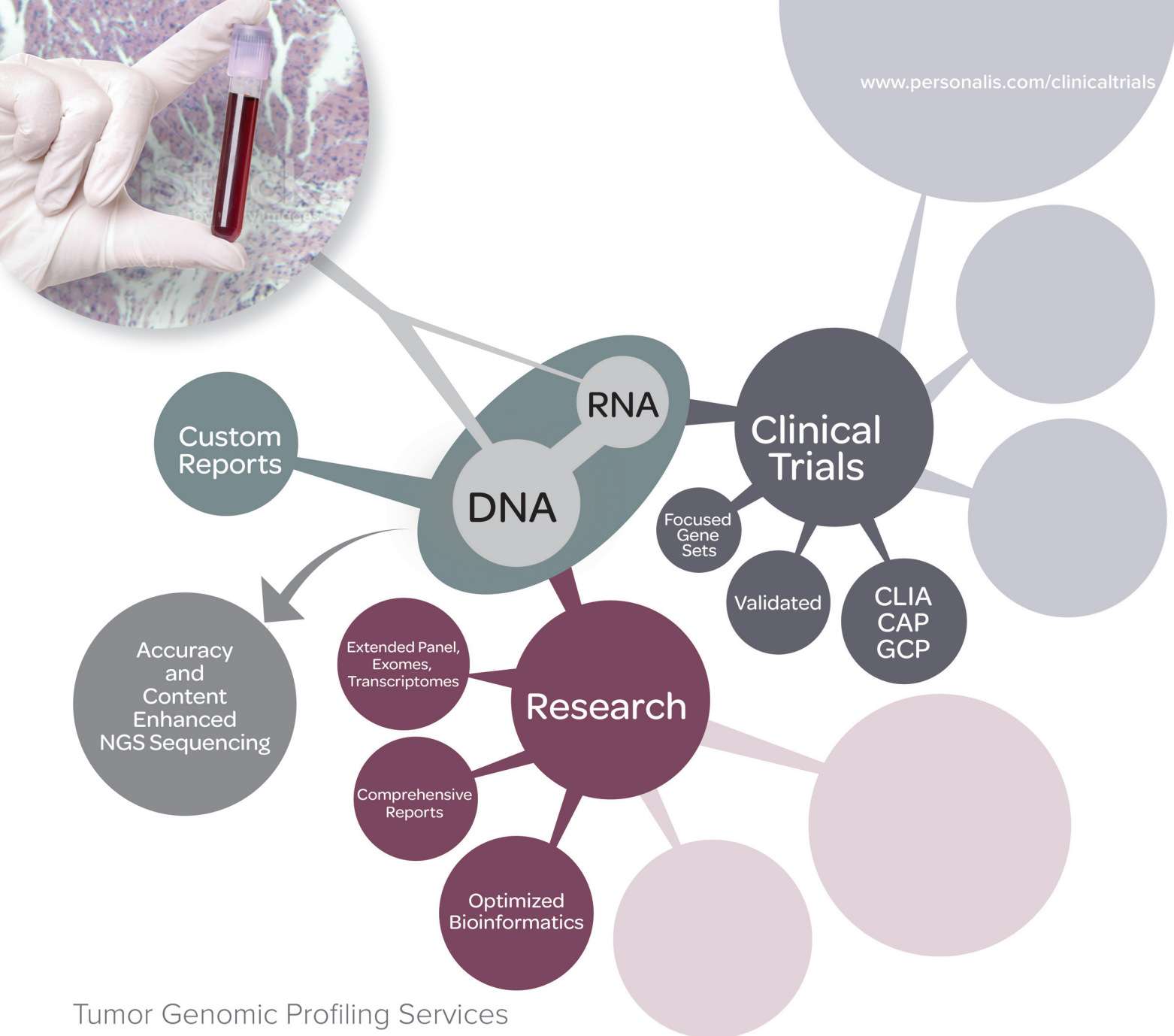


Takara  **Clontech**

Clontech Laboratories, Inc. • A Takara Bio Company

United States/Canada: +1.800.662.2566 • Asia Pacific: +1.650.919.7300 • Europe: +33.(0)1.3904.6880 • Japan: +81.(0)77.543.7247
For Research Use Only. Not for use in diagnostic or therapeutic procedures. Not for resale. Takara and the Takara logo are trademarks of TAKARA HOLDINGS, Kyoto, Japan. Clontech, the Clontech logo, SMART-Seq, that's GOOD science!, and Ultra are trademarks of Clontech Laboratories, Inc. All other marks are the property of their respective owners. Certain trademarks may not be registered in all jurisdictions.
© 2015 Clontech Laboratories, Inc.

www.clontech.com 06.15 US (633648)



Tumor Genomic Profiling Services for Clinical Trials

Comprehensive Coverage, Tailored Results

Based on the Accuracy and Content Enhanced (ACE) platform, our advanced tumor profiling services provide the most complete genomic data from any tumor sample type. Our cancer panel includes coverage of over 1,300 genes, including clinically actionable genes, known driver mutations, immuno-oncology and key cancer pathway genes. Our service is tailored to address the needs of your clinical trial enrollment requirements while simultaneously making a broader research dataset available. As a result, we maximize the return on every tumor sample.

- ✓ DNA and RNA analysis from a single sample for maximum insight
- ✓ More coverage of key cancer pathway genes
- ✓ Alterations reported include SNVs, CNVs, gene fusions and low-level variant expression
- ✓ Comprehensive analysis with flexible reporting of the complete panel or a focused set of genes, depending on your requirements

www.personalis.com | info@personalis.com

+1 855-GENOME4 (436-6634)

+1 650-752-1300 (outside U.S.)



Personalis[®]
Pioneering Genome-Guided Medicine

Discover 1% Somatic Mutation Detection from 10 ng DNA in 2 Hours

Accel-Amplicon™ Panels

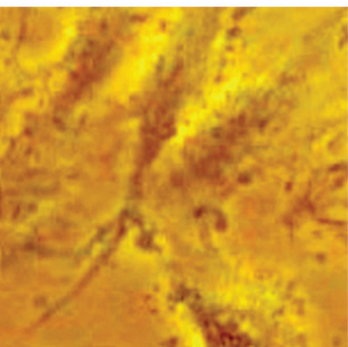
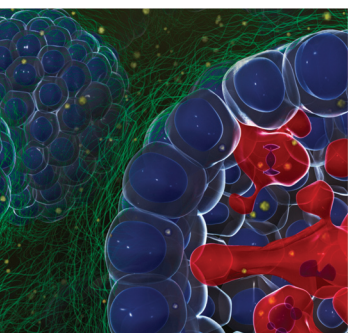
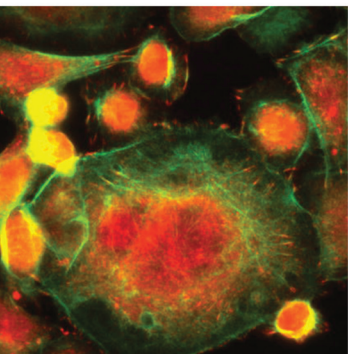
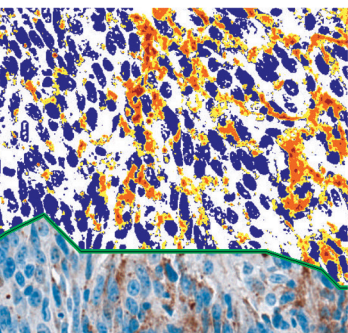
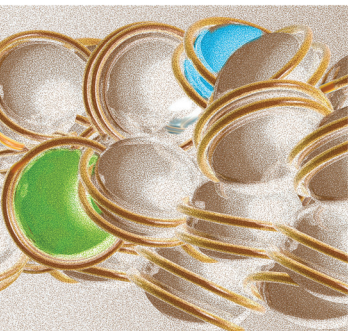
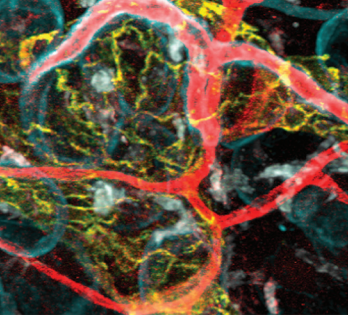
Single-tube Multiplex Assays for Illumina® Platforms

- 2 Hour Workflow Sample to Sequencer
- 1% Mutant Allele Frequency Detection
- 10 ng Sample Input
- Validated for Circulating Cell-free DNA, FFPE, Fresh-frozen Samples

www.swiftbiosci.com



© 2015, Swift Biosciences, Inc. The Swift logo and Accel-Amplicon are trademarks of Swift Biosciences. Illumina is a trademark of Illumina, Inc. 15-0299, 05/15



2015-2016 SCIENTIFIC CONFERENCES

Presenting the most significant research on cancer etiology, prevention, diagnosis, and treatment

CRI-CIMT-EATI-AACR The Inaugural International Cancer Immunotherapy Conference: Translating Science into Survival
September 16-19, 2015 • New York, NY

Chromatin and Epigenetics in Cancer
Co-Chairpersons: Peter A. Jones, Sharon Y. R. Dent, and Charles W. M. Roberts
September 24-27, 2015 • Atlanta, GA

Advances in Breast Cancer Research
Co-Chairpersons: Matthew J. Ellis, Charles M. Perou, and Jane E. Visvader
October 17-20, 2015 • Bellevue, WA

Advances in Ovarian Cancer Research: Exploiting Vulnerabilities
Co-Chairpersons: Kathleen R. Cho, Douglas A. Levine, and Benjamin G. Neel
October 17-20, 2015 • Orlando, FL

Fourth AACR International Conference on Frontiers in Basic Cancer Research
Chairperson: M. Celeste Simon
Co-Chairpersons: James P. Allison, John E. Dick, Nathanael S. Gray, and Victor E. Velculescu
October 23-26, 2015 • Philadelphia, PA

The Basic Science of Sarcomas
Co-Chairpersons: Robert G. Maki, Angelo Paolo Dei Tos, Jonathan A. Fletcher, Lee J. Helman, and Brian A. Van Tine
November 3-4, 2015 • Salt Lake City, UT

AACR-NCI-EORTC International Conference on Molecular Targets and Cancer Therapeutics
Scientific Committee Co-Chairpersons: Levi A. Garraway, Lee J. Helman, and Jean-Charles Soria
November 5-9, 2015 • Boston, MA

Advances in Pediatric Cancer Research: From Mechanisms and Models to Treatment and Survivorship
Co-Chairpersons: Scott A. Armstrong, Charles G. Mullighan, Kevin M. Shannon, and Kimberly Stegmaier
November 9-12, 2015 • Fort Lauderdale, FL

New Horizons in Cancer Research: Bringing Cancer Discoveries to Patients Shanghai 2015
Co-Chairpersons: Lewis C. Cantley and Carlos L. Arteaga
November 12-15, 2015 • Shanghai, China

Eighth AACR Conference on the Science of Cancer Health Disparities in Racial/Ethnic Minorities and the Medically Underserved
Co-Chairpersons: John M. Carethers, Marcia R. Cruz-Correa, Mary Jackson Scroggins, Edith A. Perez, Beti Thompson, and Cheryl L. Willman
November 13-16, 2015 • Atlanta, GA

Developmental Biology and Cancer
Co-Chairpersons: Hans Clevers, Stuart Orkin, and Suzanne Baker
November 30-December 3, 2015 • Boston, MA

Tumor Metastasis
Co-Chairpersons: Bruce R. Zetter, Melody A. Swartz, and Jeffrey W. Pollard
November 30-December 3, 2015 • Austin, TX

CSHA/AACR Joint Meeting: Big Data, Computation, and Systems Biology in Cancer
Conference Organizers: Andrea Califano, William C. Hahn, Satoru Miyano, and Xuegong Zhang
December 1-5, 2015 • Suzhou, China

EORTC-NCI-EMA-AACR International Conference on Innovation and Biomarkers in Cancer Drug Development
Co-Chairpersons: Denis A. Lacombe and John W. Martens
December 3-4, 2015 • Brussels, Belgium

Noncoding RNAs and Cancer
Co-Chairpersons: Howard Y. Chang, Jeannie T. Lee, and Joshua Mendell
December 4-7, 2015 • Boston, MA

San Antonio Breast Cancer Symposium
Co-Directors: Carlos L. Arteaga, Virginia Kaklamani, and C. Kent Osborne
December 8-12, 2015 • San Antonio, TX

AACR-IASLC Joint Conference on Lung Cancer
Co-Chairpersons: Alice T. Shaw and Karen L. Kelly
January 4-7, 2016 • San Diego, CA

The Function of Tumor Microenvironment in Cancer Progression
Co-Chairpersons: Raghu Kalluri, Robert A. Weinberg, Douglas Hanahan, and Morag Park
January 7-10, 2016 • San Diego, CA

Patient-Derived Cancer Models: Present and Future Applications from Basic Science to the Clinic
Co-Chairpersons: Manuel Hidalgo, Hans Clevers, S. Gail Eckhardt, and Joan Seoane
February 11-14, 2016 • New Orleans, LA

10th AACR-JCA Joint Conference Breakthroughs in Cancer Research: From Biology to Therapeutics
Co-Chairpersons: Frank McCormick and Tetsuo Noda
February 16-20, 2016 • Maui, HI

The Cancer Cell Cycle: Tumor Progression and Therapeutic Response
Co-Chairpersons: Julien Sage, J. Alan Diehl, and Karen E. Knudsen
February 27-March 2, 2016 • Orlando, FL

AACR Annual Meeting 2016
Program Committee Chairperson: Scott Armstrong
April 16-20, 2016 • New Orleans, LA

Learn more and register at
www.AACR.org/Calendar

AACR American Association
for Cancer Research

FINDING CURES TOGETHERSM



Orphan

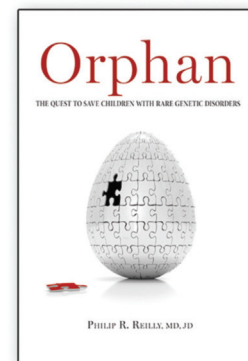


THE QUEST TO SAVE CHILDREN WITH RARE GENETIC DISORDERS

By Philip R. Reilly, MD, JD

Orphan is about the struggle to save the lives of children who, because of an unlucky roll of the genetic dice, are born with any one of several thousand rare genetic disorders. Many are burdened with diseases that carry mysterious names, some of which you can read about for the first time in this book, along with compelling stories about the physicians, scientists, and parents who have taken them on. The diseases include phenylketonuria, sickle cell anemia, dystrophic epidermolysis bullosa, X-linked hypohidrotic ectodermal dysplasia, and Friedreich's ataxia—just a few of the more than 1000 genetic disorders that are well-described and many more that are not. Many manifest in infancy. Some show up in mid-childhood, others later in childhood, and still others among adults.

They touch almost every extended family. *Orphan* is more than a book about disease and research—it gives voice to thousands of people who, all too often, have endured terrible illnesses, bravely faced arduous clinical trials, and, sometimes, have gained victories, almost always in silence. This book recounts extraordinary breakthroughs and hopes for the future. Many of the disorders that will end our lives are in some part genetically influenced. We really are all orphans, and this book is for all of us.



2015, 408 pages, illustrated (12 page insert of B&W images), index

Hardcover \$29

ISBN 978-1-621821-37-3

Contents

Preface

Acknowledgments

Introduction

1. Diet

2. The Rise of Medical Genetics

3. Blood

4. Genetic Testing: Avoiding Disease

5. Stem Cells: Creating Human Mosaics

6. Enzyme Replacement Therapy: Genetically Engineered Drugs

7. Gene Therapy: Using Viruses to Deliver Normal Genes

8. Overcoming Mutations

9. Butterfly Children: Rebuilding the Skin

10. Ligands: Turning Genes On

11. Mending Broken Proteins

12. What Is Next: Emerging Therapies

13. We Are All Orphans: Lessons for Common Diseases

Bibliography

Index

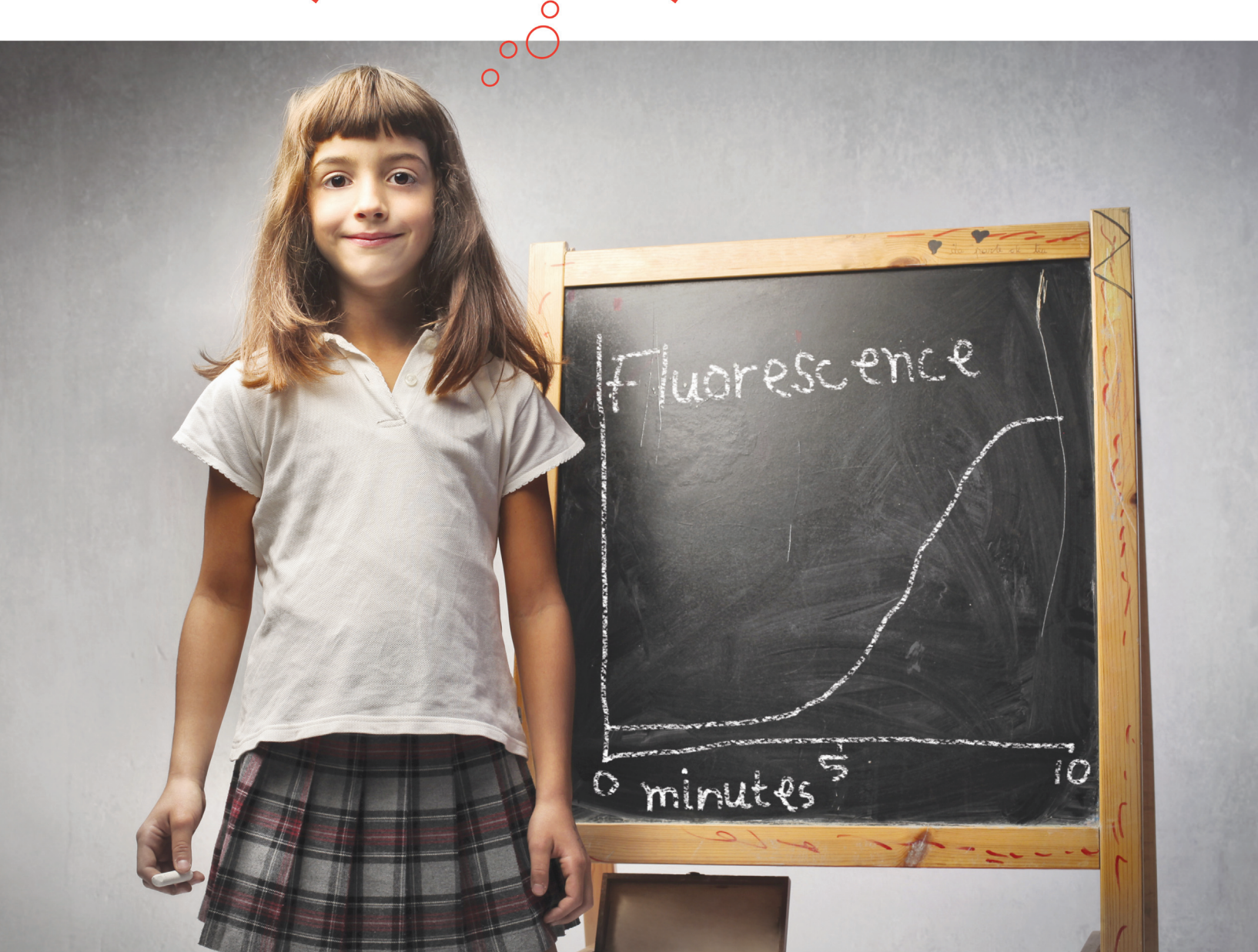
About the author: Philip R. Reilly earned his undergraduate degree at Cornell University, studied human genetics at the University of Texas Graduate School of Biomedical Sciences, and graduated from Yale Medical School in 1981. He did his medical residency at Boston City Hospital. He earned board certification in internal medicine and clinical genetics, and a law degree at Columbia University. He has served on the Board of Directors of the American Society of Human Genetics, and he is a Founding Fellow of the American College of Medical Genetics. He twice served as President of the American Society of Law, Medicine, and Ethics. During the 1990s, Reilly was the Executive Director of the Eunice Kennedy Shriver Center for

Mental Retardation in Waltham, Massachusetts, a nonprofit that worked on understanding childhood and adult neurological disorders. Dr. Reilly has held faculty positions at Harvard Medical School and Brandeis University. Since 2009 he has worked as a venture partner at Third Rock Ventures in Boston where he focuses on helping to start companies to develop innovative therapies for orphan genetic diseases. Over the years he has published six books and many articles about the impact of advances in genetics. Reilly frequently works with patient groups who are concerned with rare genetic disorders.



Learn more, read a free chapter, and order your copy today! Visit cshlpress.org/orphan

Goodbye thermocycler.



Enjoy more playtime with **Recombinase Polymerase Amplification** (RPA) the isothermal amplification that really works. RPA uses a recombinase-based process instead of thermocycling to amplify DNA, meaning **real-time detection within 15 minutes**.

Read over 50 RPA publications @
twistdx.co.uk/publications

RPA. It really works.
twistdx.co.uk | +44 (0)1223 496700

Break free from qPCR

Find gene targets faster



Validating gene targets with qPCR is often slow and costly due to lengthy primer design, experimental assay optimization, and sample processing time.

QuantiGene® Plex Assays validate gene targets faster:

- No RNA purification or reverse transcription is required
- Multiplex panels of up to 80 genes are functionally validated
- One QuantiGene Plex® plate provides as much information as 20 qPCR plates



Download **"The 7 best reasons to break free from qPCR"**
www.ebioscience.com/breakfree-genres

Biology for a better world.

NORTH AMERICA: 888-999-1371 ■ EUROPE: +43 1 796 40 40-305 ■ JAPAN: +81 (0)3 6430 4020 ■ INQUIRIES: info@ebioscience.com

©2015 Affymetrix, Inc. All rights reserved. For Research Use Only. Not for use in diagnostic or therapeutic procedures.