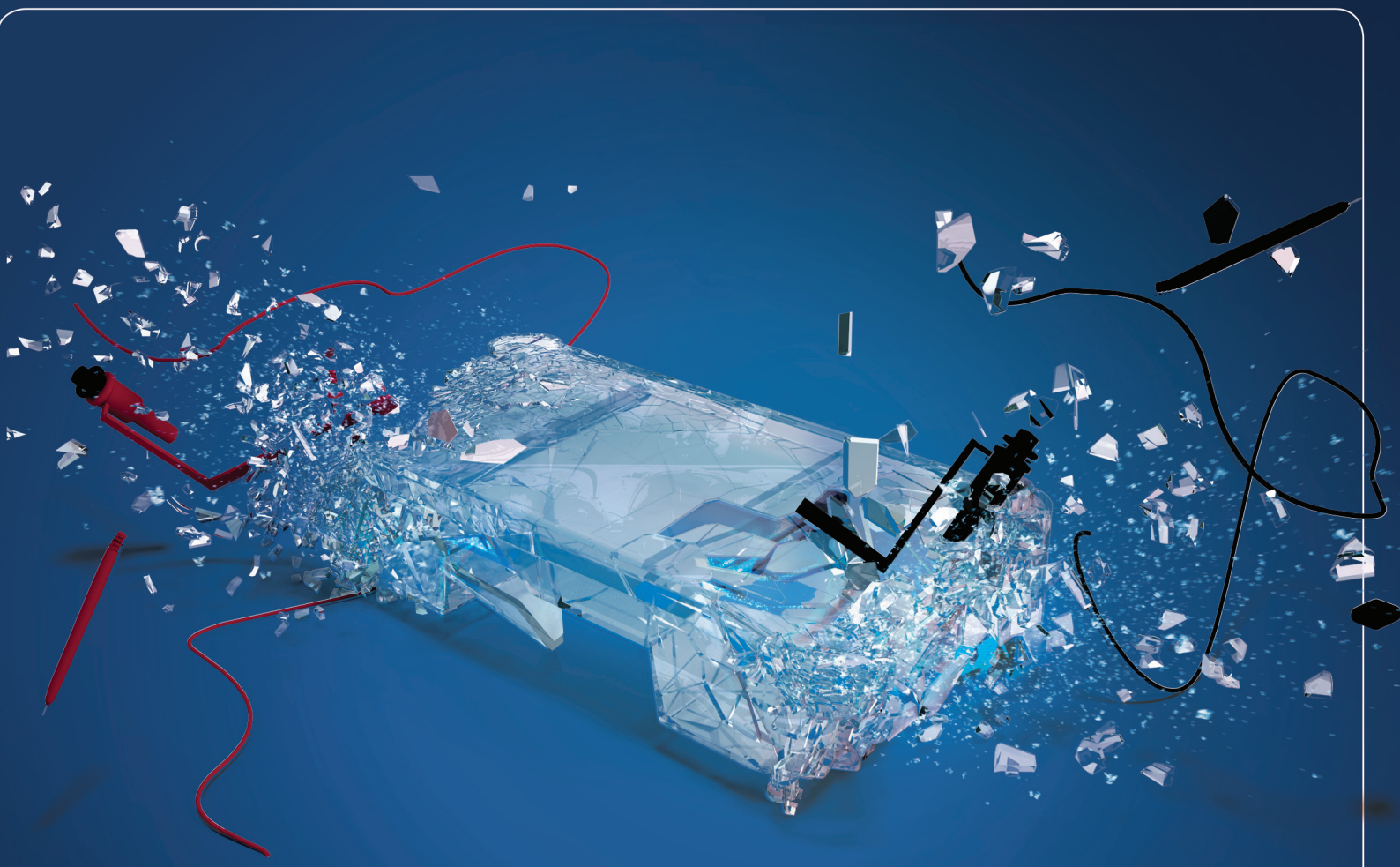

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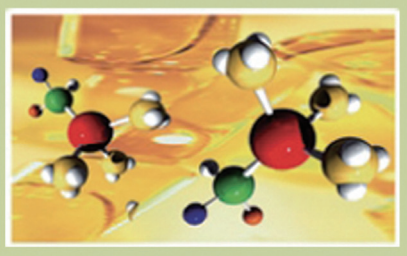

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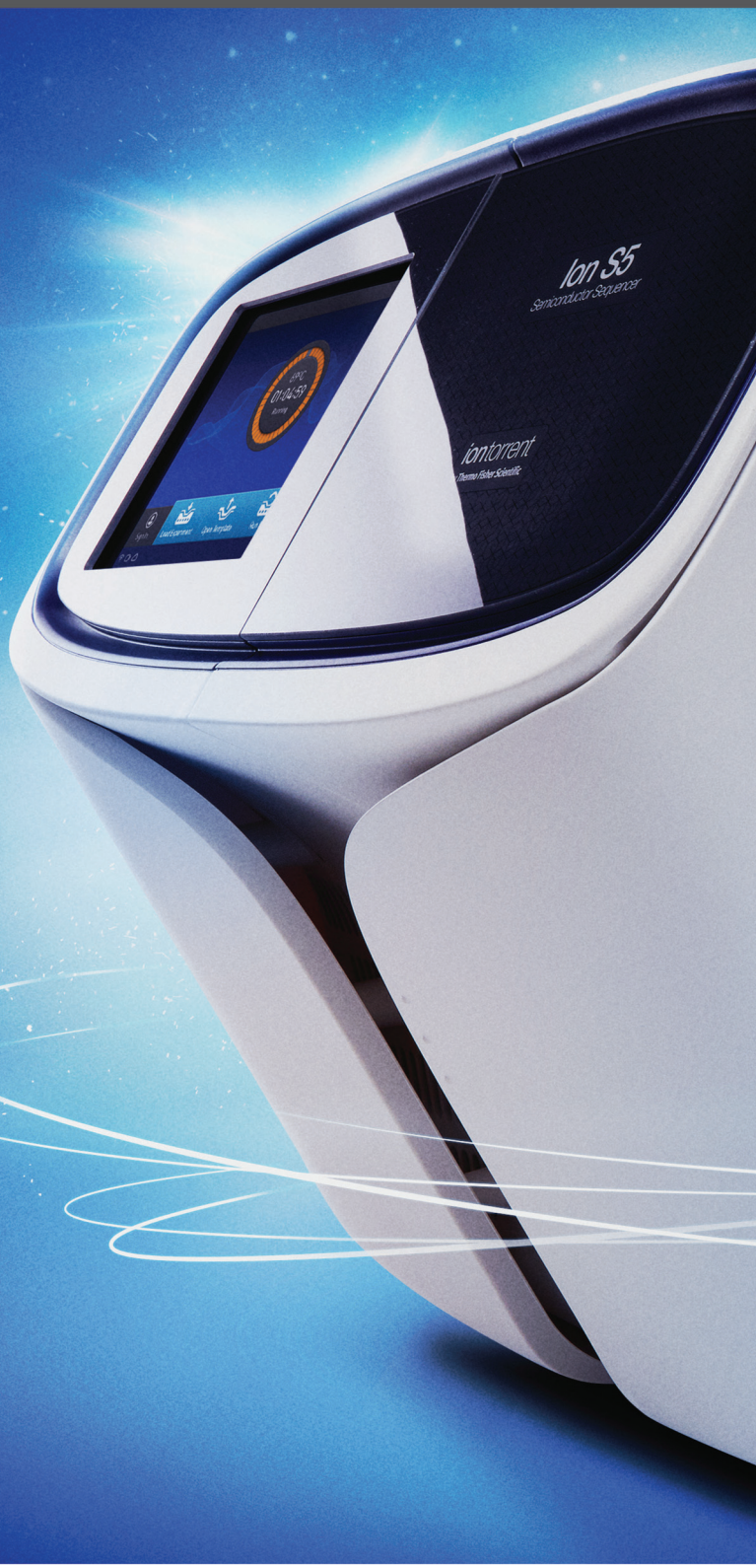

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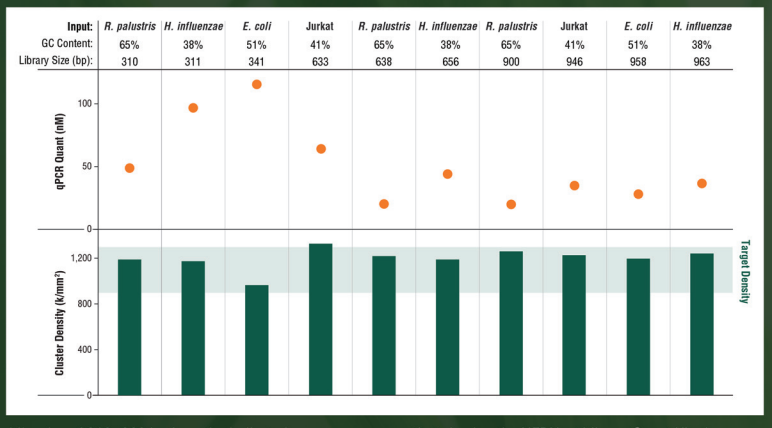

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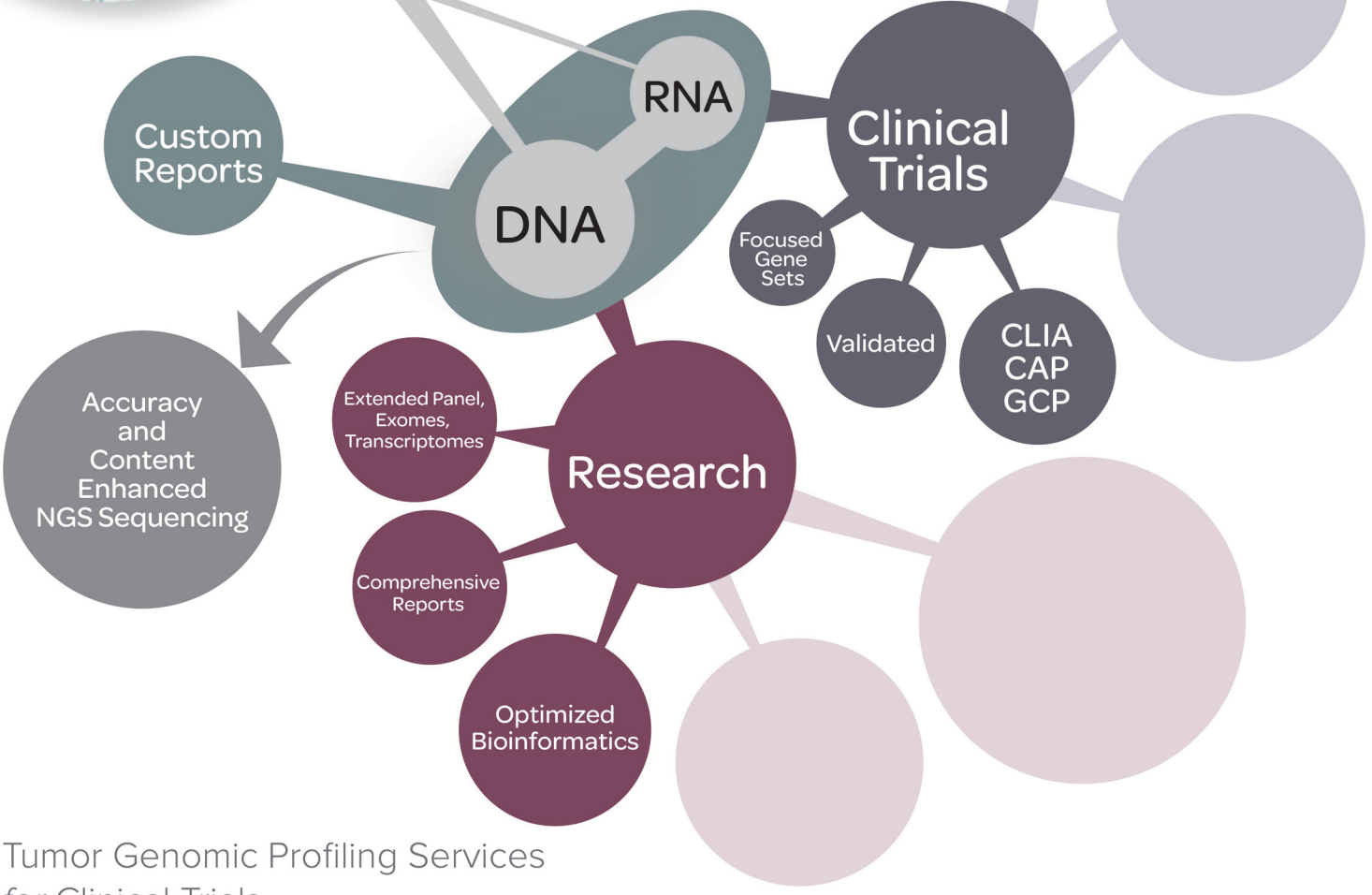

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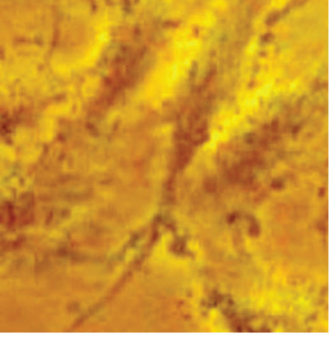

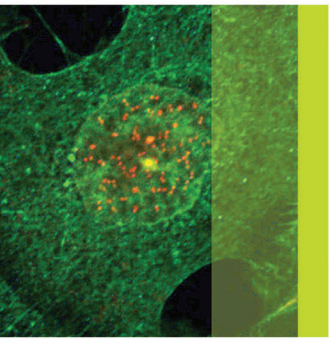

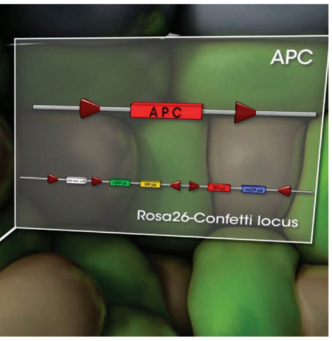

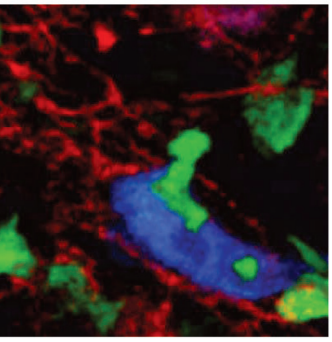

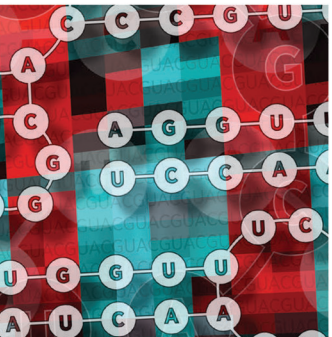

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AACR-NCI-EORTC International Conference on Molecular Targets and Cancer Therapeutics
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Advances in Pediatric Cancer Research: From Mechanisms and Models to Treatment and Survivorship
Co-Chairpersons: 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

11th Annual Personalized Medicine Conference
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November 18-19, 2015 • Boston, MA

Developmental Biology and Cancer
Co-Chairpersons: Suzanne Baker, Hans Clevers, and Stuart Orkin
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
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Noncoding RNAs and Cancer
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December 4-7, 2015 • Boston, MA

San Antonio Breast Cancer Symposium
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December 8-12, 2015 • San Antonio, TX

Fourth AACR-IASLC International Joint Conference: Lung Cancer Translational Science- From the Bench to the Clinic
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January 4-7, 2016 • San Diego, CA

The Function of Tumor Microenvironment in Cancer Progression
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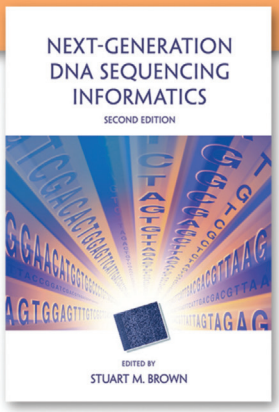

Edited by Stuart M. Brown, *New York University School of Medicine*

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.

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CONTENTS

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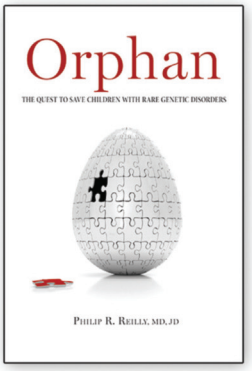
Orphan

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
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Contents

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.

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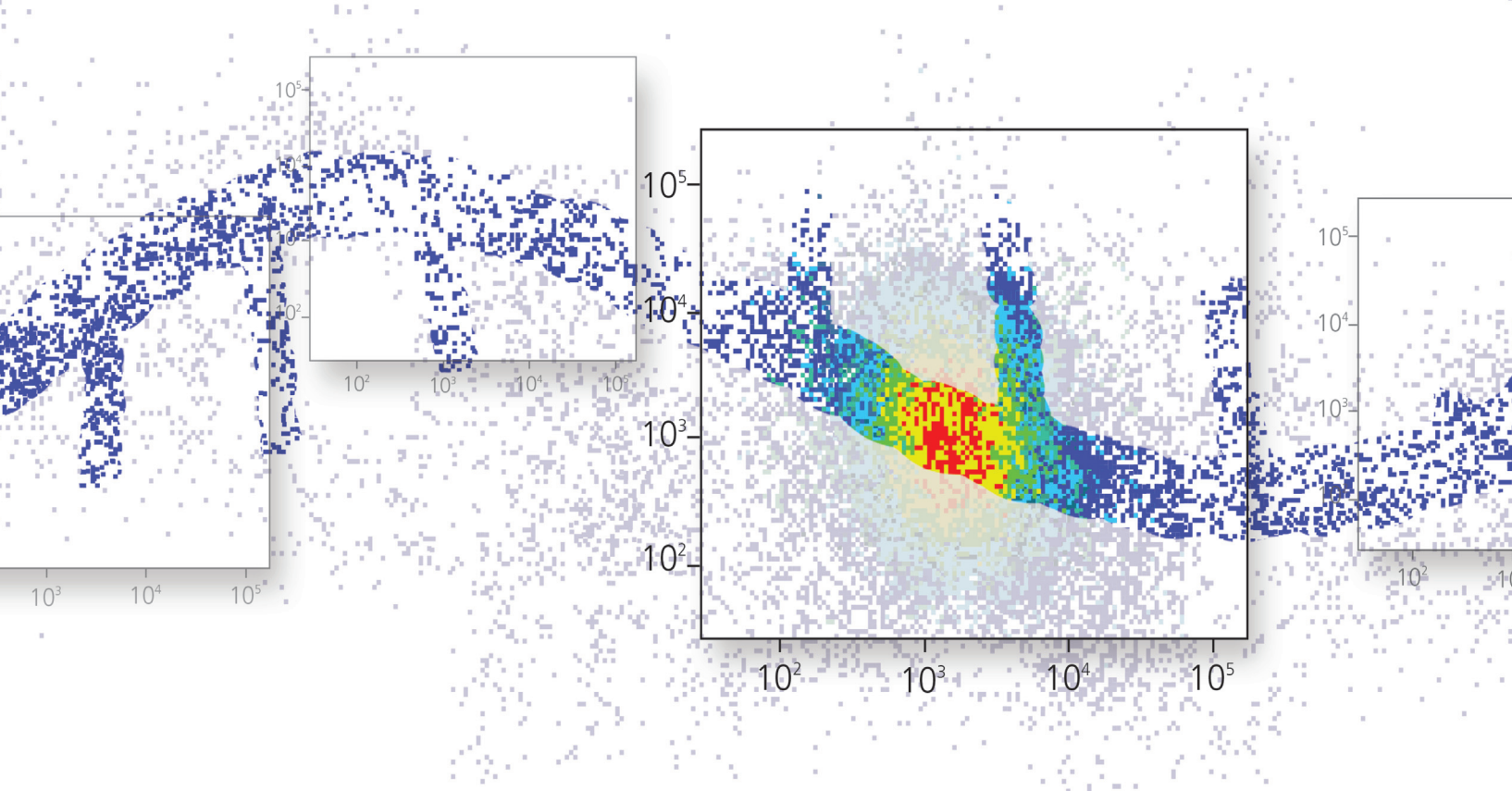

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