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## PREFACE

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Over the past few decades, advances in genetics and genomics have exceeded our greatest expectations and have revolutionized the way we think about health. While genetics has been traditionally associated with pregnancy, birth defects, and newborn screening, almost every disease is influenced in part by an individual's genetic make-up. Therefore, it is important to consider the impact of genetics for any condition throughout a patient's lifetime.

The purpose of this manual is to provide a genetics educational resource for patients and health professionals in the New England states and to increase awareness of specialty care in genetics. The manual opens with a basic introduction to genetic concepts followed by a description of the different types and applications of genetic tests. We also provide information about diagnosis of a genetic disease, family-history taking, newborn screening, and genetic counseling. We included helpful resources to assist in patient care, patient and professional education, and specialty genetics services in New England. At the end of each section, we provide a list of references to use if you desire additional information. In addition, we provide a series of consumer fact sheets to be copied and offered to patients. These take-home resources are critical to helping patients understand some of the basic concepts and applications of genetics.

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Genetic Alliance is an international coalition comprised of more than 600 advocacy, research, and healthcare organizations that represent more than 14 million people. With a 20-year history as a 501(c)(3) not-for-profit organization, Genetic Alliance is dedicated to improving the quality of life for everyone who is living with a genetic condition. Strategically situated at the crossroads of the genetics community, Genetic Alliance provides technical assistance to advocacy organizations, builds and sustains robust information systems to empower an active and dynamic network of stakeholders, and actively works for public policies that promote the translation of basic research into therapies and treatments. In particular, Genetic Alliance identifies solutions to emerging problems and works to reduce obstacles to rapid and effective translation of research into accessible technologies and services that improve human health.

With the increasing need for current knowledge about genetics, the New England Public Health Genetics Education Collaborative, made up of representatives from New England's six state public health departments, has committed to joint efforts and sharing resources in order that all in the region will have knowledge of genetics and its effects on health—toward the goal of improved health outcomes. A subcommittee of NERGG, Inc., the Collaborative supports the NERGG, Inc. mission to promote health of both children and adults by increasing the awareness of genetic concerns, the understanding of the role of genetics in healthcare, and the availability of appropriate services. Funding for this manual was provided by the U.S. Department of Health and Human Services, Health Resources and Services Administration, Grant U22MC03959-03-00 for Heritable Disorders to New England Regional Genetics Group (NERGG, Inc.).

The manual is available online at the NERGG, Inc. website, the Genetic Alliance website, and the individual state departments of public health websites in New England.

