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## PREFACE

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Over the past few decades, advances in genetics and genomics have revolutionized the way we think about health. Although genetics has traditionally been associated with pregnancy, birth defects, and newborn screening, almost every disease is influenced in part by an individual's genetic makeup. Therefore, it is important to consider the impact of genetics in health and disease throughout an individual's lifetime.

The purpose of this manual is to provide an educational genetics resource for individuals, families, and health professionals in the New York – Mid-Atlantic region and increase awareness of specialty care in genetics. The manual begins with a basic introduction to genetics concepts, followed by a description of the different types and applications of genetic tests. It also provides information about diagnosis of genetic disease, family history, newborn screening, and genetic counseling. Resources are included to assist in patient care, patient and professional education, and identification of specialty genetics services within the New York – Mid-Atlantic region. At the end of each section, a list of references is provided for additional information. Appendices can be copied for reference and offered to patients. These take-home resources are critical to helping both providers and patients understand some of the basic concepts and applications of genetics and genomics.

The original manual was created by Genetic Alliance with funding from the District of Columbia Department of Health, through U.S. Department of Health and Human Services (HHS) Health Resource and Services Administration (HRSA) Grant #5 H91 MC 00228-03.

Genetic Alliance transforms health through genetics. We promote an environment of openness centered on the health of individuals, families, and communities.

We bring together diverse stakeholders to create novel partnerships in advocacy. Genetic Alliance's network includes hundreds of disease-specific advocacy organizations, as well as universities, companies, government agencies, and policy organizations. The network is an open space for thousands of shared resources, creative tools, and dozens of focused programs.

We revolutionize access to information to enable translation of research into services and individualized decision-making. Genetic Alliance offers technical assistance to organizations, builds and sustains robust information systems, 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. In all we do, we integrate individual, family, and community perspectives to improve health systems.

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NYMAC, the New York – Mid-Atlantic Consortium for Genetic and Newborn Screening Services, is one of seven federally-funded regions in the U.S., created to ensure that individuals with heritable disorders and their families have access to quality care and appropriate genetic expertise and information. It is funded by HRSA Collaborative Agreement #U22 MC 03956.

This manual is available on the Genetic Alliance website, [www.geneticalliance.org/publications](http://www.geneticalliance.org/publications), and on the NYMAC website, [www.wadsworth.org/newborn/nymac/resources.html](http://www.wadsworth.org/newborn/nymac/resources.html).

# GENETIC ALLIANCE MANDATE FOR QUALITY GENETIC SERVICES

*Access to quality genetics services is critical to healthcare.*

1. Individuals and families partner with their healthcare providers to identify needs, develop and monitor treatment plans, and manage their genetic condition.
2. Healthcare providers refer individuals to appropriate specialists, as needed, including those outside of their health insurance plans.
3. Providers and payers consider the psychosocial, as well as the medical, effects of a genetic condition—on both the individual and the individual’s family—at each stage of life.
4. Healthcare insurance plans reimburse genetic testing, diagnosis, and treatment for genetic conditions.
5. Quality resources are available to assist individuals and their families in understanding family health history, signs/symptoms, screening/testing options and their implications, diagnosis, treatment, and long-term follow-up, as needed.
6. A healthcare provider with experience in genetic services is available to all individuals.
7. Providers, payers, and employers create and use policies, guidelines, and procedures to ensure the appropriate use of genetic information.
8. Information about genetic conditions is provided to individuals and families in a culturally-appropriate manner, which may include primary language, appropriate educational level, and various media.
9. Information about genetic research and clinical trials is available to the affected individuals and integrated into clinical practice when appropriate.
10. Referrals to support groups and resources are offered at regular office visits.
11. Outpatient, home, and hospital care for individuals with genetic conditions is available and integrated.

