

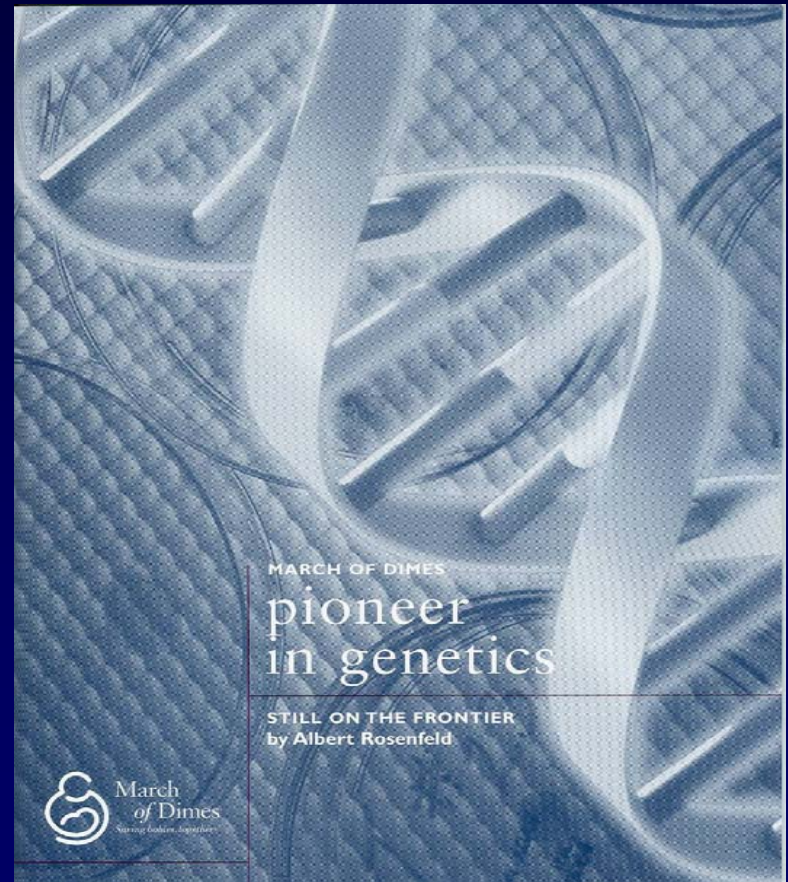
Genetics at March of Dimes

**Margaret Comerford Freda,
EdD, RN, CHES, FAAN**

**Professor, Albert Einstein
College of Medicine,
Montefiore Medical Center**

**Editor, MCN The American
Journal of Maternal Child
Nursing**

**Chair, National Nurse Advisory
Council, MOD**



Genetics at the MOD

- **Mission**
- **History**
- **Prematurity and Birth Defects**
- **Research**
- **Education**

Genetics at the MOD

- **Mission**

- To improve infant health by preventing birth defects, premature birth and infant mortality
- Strategic goal to expand MOD's mission investment in emerging opportunities such as genetic research, gene therapy and genetic services
- Priority of MOD to serve as an information resource on genetics, pregnancy, birth defects and other areas of MCH

Genetics at the MOD

- **History**

- MOD has long been a pioneer in genetics, funding innovative biomedical research, gene mapping, clinical applications, genetic services, professional and public genetics education, community service and policy initiatives

Genetics at the MOD

- **Prematurity and Birth Defects**
 - Almost all birth defects have a genetic component
 - Genetics likely plays a role in preterm birth
 - MOD has a multi-year commitment to prevention of prematurity

Prematurity and Genetics



**Preterm Birth can be viewed as a
Common Complex Disorder**

Complex Disorders are characterized by:

- 1. Genetic contribution**
- 2. Environmental influences**
- 3. Gene-environment interactions**

Genetics and Preterm Birth

- **There IS a genetic contribution to preterm birth**
- **Up to 1/3 of preterm birth is heritable**
- **What do we know about inheritance patterns?**
- **Data needed on multi-generational inheritance patterns in families**
- **Family history research opportunities should include preterm birth**

International Preterm Birth Collaborative (PREBIC)

- **March of Dimes participation in PREBIC**
- **Purpose of the collaborative is to “support and enhance international networking among researchers in preterm birth and the establishment of multinational research projects on preterm birth”**
- **Network of ~ 60 researchers**
- **<http://www.prebic.org/index.asp>**

Contact:

Karla Damus, RN, MSPH, PhD



Genetics at the MOD

- **Research**

MOD funded biomedical research that laid the foundation for the Human Genome Project:

- **1940's: Max Delbruch: founding father of molecular biology**
- **1950's: James Watson: identified structure of DNA**

Genetics at the MOD

- **Research**

- Many grantees have developed new genetic tests and discovered genetic markers or gene mutations for disease

- **1961: Robert Guthrie: first neonatal genetic screening test**
 - **1993 Research Ambassadors: first gene therapy recipients**
 - **Markers or gene mutations for: Sickle Cell, Fragile X, Marfan Syndrome, Cystic Fibrosis, Dwarfism, Retinitis pigmentosa, some forms of heart disease, familial breast and colon cancer**

The 2006 PRI Grantees

- **Maternal and Infant Genetic Contributions to Preterm Birth: The Inflammatory Response**
 - Martin Kharrazi, MPH, PhD
 - California Department of Health Services, Richmond, CA
- **Abruption-induced Preterm Delivery Elicits Functional Endometrial Progesterone Withdrawal by Up-regulating the Transcriptionally Active B Form and Down-regulating the Transcriptionally Inactive C Form of the Progesterone Receptor**
 - Charles J. Lockwood, MD
 - Yale University School of Medicine, New Haven, CT
- **Progesterone Receptor Dysregulation and Preterm Birth**
 - Errol R. Norwitz, MD, PhD
 - Yale University School of Medicine, New Haven, CT

The 2006 PRI Grantees

- **Cytokines from Periodontal Disease Induce Premature Birth**
 - Johnny R. Porter, PhD
 - Louisiana State University Health Sciences Center, New Orleans, LA
- **A Comprehensive Study of Genetic Susceptibility to Preterm Delivery**
 - Timothy Mark Frayling, PhD
 - Peninsula Medical School, University of Exeter, United Kingdom
- **Pharmacological Investigation of Novel Anti-inflammatory Therapeutic Strategies for the Treatment and Prevention of Preterm Birth using Human Ex-vivo Models**
 - Jeffrey Andrew Keelan, PhD, MSc
 - Liggins Institute, University of Auckland, New Zealand

Preterm Birth and Genetics International Alliances (PREGENIA)

- **A subgroup of PREBIC**
- **Vision is to elucidate the cause of preterm birth and to understand the mechanisms behind to facilitate prevention in an early stage**
- **Develop ways to synthesize information, promote methodology and reporting standards, collaborate on meta-analyses**
- **Apply for grant funding and publish review articles**
- **<http://www.prebic.org/pregenia/vision.asp>**

Contact:

Karla Damus, RN, MSPH, PhD

Bruce Lin, MPH



Current Genetics Activities

- **Research**
- **Public Affairs**
 - Impact player in area of Newborn Screening
- **Professional and Consumer Education Programs:**
 - Nursing Modules
 - Chapter Community Grants (MIOP)
 - Genetics & Your Practice (G&YP)
 - Genetics Education Needs Evaluation (GENE) Project
 - Consumer Genetics Education Network (GEN) Project

Genetics Education Projects

- **For Consumers**

- **Genetics Education Needs Evaluation (GENE) Project:** 5-year cooperative agreement that investigated ways to improve consumer access to genetics information (2000-2005).

www.marchofdimes.com/geneproject

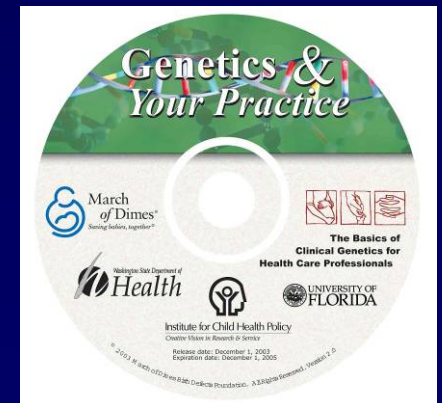
- **Consumer Genetics Education Network (CGEN) Project:** 5-year cooperative to increase genetic literacy in four underserved populations as direct extension of GENE Project (2005-2010)

■ For Professionals

www.marchofdimes.com/gyponline

-Free resources and tools that can be shared with partners at health departments, professional meetings, educational programs

CD-ROM



Contact Information

GENE Project/ Genetics Education Network

Diane M. Ashton, MD, MPH

Associate Medical Director

dashton@marchofdimes.com

EmyLou A.S. Rodriguez

Manager, Community Genetics Education

erodriguez@marchofdimes.com

Genetics & Your Practice

Bruce K. Lin, MPH

Manager, Public Health
Initiatives

blin@marchofdimes.com

Diane Gross, MPH

Manager, Special Initiatives

dgross@marchofdimes.com



Why Has the March of Dimes Endorsed the Genetics Competencies?

- “Advances in genetics are rapidly changing the face of medicine and have major implications for all health care providers. Nurses need to have a basic understanding of genetic science and how it applies to the patients they care for.”**

» Janis Bierman, Vice President, Education and Programs, March of Dimes