DISCLOSURE STATEMENT

Before their talks, all speakers and moderators will disclose the existence of any financial interest and/or other relationship(s) they might have with the manufacturer(s) or provider(s) of any commercial product(s) or service(s) to be discussed during their presentation: honorarium/expenses, grants or research support, employee, a consultant role, speakers' bureau member, stock ownership, or any other special relationships. When unlabeled uses are discussed, these will also be indicated.

Friday, March 19

All sessions in Regency Ballroom.
All breaks in Riverfront Hall.

7:15 am–5:00 pm
Registration
Promenade Lower Level

7:30 am–8:30 am
Continental Breakfast
Riverfront Hall

Exhibits and Posters:
7:15 am–8:30 am; 10:30 am–2:00 pm; 3:00 pm–5:30 pm

Theme: Sex Determination and Differentiation in Humans

Sex determination and differentiation underlie many serious, but potentially correctable, birth defects. The human embryo starts as a bisexual organism whose primordial organs can mature into those of one or the other sex. These processes are under the governance of specific genes. Thus genetic sex is determined at the time of fertilization, but post-fertilization influences can affect this process in an adverse manner. Speakers will address the mechanisms of sex determination and differentiation, their aberrations, and possible therapeutic interventions.

▶ After attending the plenary sessions on March 19, participants will:

1. Be familiar with the clinical problems that arise when sex differentiation goes awry.
2. Understand which aspects of this problem are urgent, when to seek appropriate consultation, and what advice to offer the parents.

OPENING ANNOUNCEMENTS

8:15 am–8:30 am

Michael Katz, MD, *March of Dimes Birth Defects Foundation, White Plains, NY;* John M. Opitz, MD, DSci, FACMG, *Univ. of Utah School of Medicine, Salt Lake City*

PRUZANSKY LECTURE

8:30 am–9:30 am

X Inactivation, Dosage Compensation, and the Sex Chromosomes
Huntington F. Willard, PhD, *Case Western Reserve Univ. School of Medicine, Cleveland, OH*

PLENARY SESSION I

9:30 am–10:30 am

Moderator: John M. Opitz, MD, DSci, FACMG
Univ. of Utah School of Medicine, Salt Lake City

Mutations of the Anti-Mullerian Hormone and Receptor Genes in Persistent Mullerian Duct Syndrome: An Update
Nathalie Josso, MD, *Unite de Recherches sur L'Endocrinologie, Montrouge, France*

Genetic Disorders of the Androgen Receptor
Leonard Pinsky, MD, FACMG, *McGill Univ., Montreal, Quebec, Canada*

10:30 am–11:00 am
Break

PLENARY SESSION II

11:00 am–12:00 pm

Moderator: Joe Leigh Simpson, MD, FACMG
Baylor College of Medicine, Houston, TX

Congenital Adrenal Hyperplasia
Maria I. New, MD, *Cornell Univ. Medical Center, New York, NY*

Male Pseudohermaphroditism due to LH Receptor Mutations
Jean D. Wilson, MD, *Univ. of Texas Southwest Medical Center, Dallas*

12:00 pm–2:00 pm
Break

PLENARY SESSION III

2:00 pm–3:00 pm

Moderator: John M. Opitz, MD, DSci, FACMG
Univ. of Utah School of Medicine, Salt Lake City

Role of the Mammalian Y Chromosome in Sex Determination and Fertility
Colin E. Bishop, PhD, *Baylor College of Medicine, Houston, TX*

Autosomal Genes Involved in Male Sex Determination
Felix A. Conte, MD, *Univ. of California San Francisco Medical Center*

3:00 pm–3:30 pm
Break

PLENARY SESSION IV

3:30 pm–5:30 pm

Moderator: Melvin M. Grumbach, MD
Univ. of California San Francisco

Lessons Learned from the RSH (So-called Smith-Lemli-Opitz) Syndrome
John M. Opitz, MD, FACMG, *Univ. of Utah School of Medicine, Salt Lake City*

Disorders of Ovarian Development and Gametogenesis
Joe Leigh Simpson, MD, FACMG, *Baylor College of Medicine, Houston, TX*

Genetics of Human Hypogonadism
Lawrence C. Layman, MD, FACMG, *Univ. of Chicago, IL*

Panel Discussion

COLONEL HARLAND SANDERS AWARD

5:30 pm–6:00 pm

6:30 pm–8:00 pm

Opening Reception for Registrants and Their Guests
Riverwalk (Rain Site: Jasmine Room)

Saturday, March 20

All sessions in rooms as indicated.
All breaks in Riverfront Hall.

7:30 am–5:00 pm
Registration
Promenade Lower Level

7:30 am–8:30 am
Continental Breakfast
Riverfront Hall

Exhibits and Posters:
7:30 am–8:30 am; 9:45 am–2:00 pm;
3:30 pm–6:00 pm (posters only)

ANNOUNCEMENTS

8:10 am–8:15 am
Regency Ballroom

PLENARY SESSION V

8:15 am–12:15 pm
Regency Ballroom

Legislative and Regulatory Impact on Genetic Services
Moderators: R. Rodney Howell, MD, FACMG, *Univ. of Miami School of Medicine, FL*; Maimon M. Cohen, PhD, FACMG, *Greater Baltimore Medical Center, MD*

Recent successes in medical genetics research have provided a wealth of new information about the identification, mapping and function of disease-associated human genes. This genetic information has greatly influenced approaches to identifying individuals at risk or those with a predisposition to a rapidly increasing number of inherited disorders, creating a variety of both medical and non-medical concerns. This session will examine the topic of legislative strategies and efforts concerning the issues of privacy and confidentiality of genetic information and various ways of dealing with the emerging issues.

▶ After attending this session, participants will be able to:

1. Understand the potential problems involved in the handling of "genetic information."
2. Appreciate the efforts underway by legislative bodies and regulatory agencies in addressing these issues.
3. Deal with the issues in the provision of services for their patients.

8:15 am

Genetics, Discrimination and Privacy
Francis S. Collins, MD, PhD, *National Human Genome Research Inst., NIH, Bethesda, MD*

9:00 am

Can/Should Genetic Information Be Protected?
Robert Gellman, JD, *Privacy & Information Policy Consultant, Washington, DC*

9:45 am
Break

10:15 am

Back to the Future: Population Screening as a Paradigm for Prediction and Prevention of Genetic Disease
Louis J. Elsas, MD, FACMG, *Emory Univ. School of Medicine, Atlanta, GA*

11:00 am

PRESIDENTIAL ADDRESS
R. Rodney Howell, MD, FACMG, *Univ. of Miami School of Medicine, FL*

11:30 am–1:00 pm
Break & Focus Groups (see page 11)

POSTER SESSION I

(Authors present, odd-numbered boards)
1:00 pm–2:00 pm
Riverfront Hall

CONCURRENT WORKSHOPS A

2:00 pm–3:30 pm

A1. Speech, Language and Reading Disorders
Hibiscus Room
Moderator: Robert L. Nussbaum, MD, FACMG, *National Human Genome Research Inst., NIH, Bethesda, MD*

Human communication is an extremely complex process. Disorders of communication are highly pleiotropic abnormalities that are commonly encountered by developmental pediatricians and geneticists. This workshop will highlight three facets of human communication: speech, language and reading. Experts have been chosen to provide basic information on phenotypic characterization of these disorders and the role of heredity in their etiology.

▶ After attending this session, participants will be able to:

1. Recognize the phenotypes presented by patients with abnormalities in speech, language and reading.
2. Gain an appreciation for current research designed to uncover the genetic mechanisms that predispose to these fascinating and debilitating forms of communication dysfunction.

Genetics and Functional Speech Disorders: Where Are We Now?
Susan Felsenfeld, PhD, CCC-SLP, *Duquesne Univ., Pittsburgh, PA*

Genetics of Specific Reading Disabilities
John C. DeFries, PhD, *Univ. of Colorado, Boulder*

Assessment of Language Phenotype(s)
Mabel L. Rice, PhD, *Univ. of Kansas, Lawrence*

A2. Genetics of Hemostasis and Thrombophilia
Orchid Room BCD

Moderator: Robin Dawn Clark, MD, FACMG, *USC/Norris Comprehensive Cancer Center, Los Angeles, CA*

This workshop will provide a review of normal hemostasis and describe defects leading to abnormally enhanced thrombosis (the thrombophilias). Thrombophilia type I is comprised of defects that increase the risk of venous thrombosis, such as disorders of antithrombin III, proteins S and C, Factor V Leiden, and prothrombin II. Thrombophilia II includes defects that increase risk of both venous and arterial thrombosis, such as hyperhomocystinemia and the labile 5-10 methylene tetrahydrofolate reductase. Emphasis will be on diagnosis, counseling and therapy.

▶ After attending this session, participants will be able to:

1. Identify the major genetic disorders that increase risk for arterial and venous thrombosis.
2. Interpret and use tests of the hemostatic system in patients at risk for thrombosis due to family history or personal history.
3. Counsel patients appropriately about risks and interventions appropriate to their own hemostatic disorder.

Introduction to Hemostasis and Coagulation Testing
Howard Leibman, MD, *USC/Norris Comprehensive Cancer Center, Los Angeles, CA*

Heritable Errors of Hemostasis: Clinical Aspects
Scott Goodnight, MD, *Oregon Health Sciences Univ., Portland*

DNA Testing for Inherited Hypercoagulability
Annette K. Taylor, MS, PhD, *Kimball Genetics, Inc., Denver, CO*

Biochemical Testing in Hemostasis
Jo Ellen S. Lee, PhD, FACMG, *Quest Diagnostics, Nichols Institute, San Juan Capistrano, CA*

A3. Paradigms for Designing Informed Consent for Genetic Testing and Research
Jasmine Room

Moderators: Judith Benkendorf, MS, CGC, *Georgetown Univ. Medical Center, Washington, DC*; Lisa Baumbach-Reardon, PhD, FACMG, *Univ. of Miami School of Medicine, FL*

Over the past half-century the research community has learned—or should have learned—a great deal about the key components of informed or valid consent. These lessons are of crucial importance to geneticists. This hands-on workshop will begin with short presentations concerning the core elements of the valid consent process: adequate information, absence of coercion, and competence. We will highlight the process and content of valid consent as it applies to genetic testing and in research. We will address the following questions from the consumer's perspective: What does the patient/subject/participant want to know? What does s/he need to know? With faculty guidance, attendees will then work in small groups on actual genetic research protocols to outline prototype consent forms. Workshop participants will receive a packet of sample consent forms and other resource materials.

▶ After attending this session, participants will be able to:

1. Identify the three core elements of valid consent and valid refusal.
2. Translate and apply these three core components to practical situations involving genetic counseling, research, testing and screening.
3. Address special challenges, conflicts and tensions facing subjects and investigators in the era of the new genetics.

Introduction to Informed Consent
Kenneth W. Goodman, PhD, *Forum for Bioethics and Philosophy, Univ. of Miami, FL*

Conducting Research on Informed Consent in Genetics
Gail Geller, ScD, *Johns Hopkins University, Baltimore, MD*

A Difficult Balance: Protecting Consumers Without Impeding Research Progress
Brad Margus, MBA, *A-T Children's Project, Boca Raton, FL*

3:30 pm–4:00 pm
Break

CONCURRENT WORKSHOPS B: PLATFORM PRESENTATIONS

4:00 pm–5:30 pm

Numbers preceding titles of talks are the numbers of the abstracts as they appear later in this volume.

B1. Biochemical Genetics

Hibiscus Room

Moderator: John A. Phillips III, MD, FACMG, *Vanderbilt Univ. School of Medicine, Nashville, TN*

4:00 pm

1. Five Years' Experience of Cholesterol Treatment for the Smith-Lemli-Opitz Syndrome (SLOS): What Have We Learned, Where Are We Going?
Ellen Roy Elias, MD, *Children's Hosp., Boston, MA*

4:15 pm

2. Cholesterol Supplementation Enhances Growth of Phallus in Smith-Lemli-Opitz Syndrome
Mira Irons, MD, FACMG, *Children's Hosp., Boston, MA*

4:30 pm

3. Autism Associated with Elevated Glutamine and Glycine Levels and Clinical Response to Dextromethorphan
Rizwan Hamid, MD, PhD, *Vanderbilt Univ. School of Medicine, Nashville, TN*

4:45 pm

4. Diagnosis of Gaucher Disease by a Flow Cytometric Assay
Hyon J. Kim, MD, FACMG, *Ajou Univ. College of Medicine, Suwon, Korea*

5:00 pm

5. Biochemical Discrimination of Heterozygotes for Cystathionine β-Synthase (CβS) Deficiency
Mark T. Steen, MD, PhD, *Emory Univ. School of Medicine, Atlanta, GA*

5:15 pm

6. An Epidemiologic Assessment of the Relationship Between the G985A Medium Chain Acyl-coA Dehydrogenase Deficiency (MCADD) Allelic Variant and Sudden Infant Death Syndrome (SIDS)
Sophia S. Wang, PhD, *Centers for Disease Control and Prevention, Atlanta, GA*

B2. Clinical Genetics

Orchid Room BCD

Moderator: Debra Freedenberg, MD, PhD, FACMG, *Genetics Inst. of Austin, TX*

4:00 pm

7. A Unique Point Mutation in the PMP22 Gene Is Associated with Deafness, Charcot-Marie-Tooth and Anticipation
Virginia E. Kimonis, BM, MRCP, *Southern Illinois Univ. School of Medicine, Springfield*

4:15 pm

8. Prenatal Diagnosis and Carrier Detection of Albinism
Ada Rosenmann, MD, PhD, *Hadassah Univ. Hospitals, Jerusalem, Israel*

4:30 pm

9. Novel Protein Truncation Test to Detect *COL2A1* Nonsense Mutations in Stickler Syndrome
Ravi Savarirayan, MD, FRACP, *University of Melbourne, Australia*

4:45 pm

10. Gestational Age and Risk for Congenital Defects: A Population-Based Study
Sonja A. Rasmussen, MD, MS, *Centers for Disease Control and Prevention, Atlanta, GA*

5:00 pm

11. Assessment of Quality of Genetic Counseling Services by Study of Adverse Events
Rodney Harris, MD, FRCP, *St. Mary's Hosp., Manchester, England*

5:15 pm

12. Variable Expressivity of Familial Medullary Thyroid Carcinoma (FMTC) due to a *RET* V804M (GTG→ATG) Mutation in Two Families: Reluctance of Gene Carriers to Accept Prophylactic Thyroidectomy
Gerald L. Feldman, MD, PhD, FACMG, *Henry Ford Hosp., Detroit, MI*

B3. Cytogenetics
Jasmine Room
Moderator: Stuart Schwartz, PhD, FACMG, *Case Western Reserve Univ., Cleveland, OH*

4:00 pm

13. A Dual FISH Assay for Detecting Deletions Associated with VCFS/DiGeorge Syndrome I and DiGeorge Syndrome II Loci
Sue Ann Berend, PhD, *Baylor College of Medicine, Houston, TX*

4:15 pm

14. A Patient with Gorlin-Chaudhry-Moss Syndrome and del(9)(q22.1q22.3)
Chun-Hui Tsai, MD, *Henry Ford Hosp., Detroit, MI*

4:30 pm

15. Chromosome Abnormalities due to Maternal Translocations Not Detected with Routine Amniocentesis
Mary Kukolich, MD, FACMG, *Cook Children's Clinical Genetics Service, Ft. Worth, TX*

4:45 pm

16. Karyotyping Miscarriage Tissue in the Managed Care Environment
Evelyn M. Karson, MD, PhD, FACMG, *Columbia Hosp. for Women, Washington, DC*

5:00 pm

17. A Retrospective FISH Study of HER-2/neu Oncogene Amplification in Relapsed and Non-Relapsed Node-Negative Breast Cancer
Hon Fong L. Mark, PhD, FACMG, *Rhode Island Hosp., Providence*

5:15 pm

18. HER-2/neu Oncogene Amplification in Prostate Cancer
Hon Fong L. Mark, PhD, FACMG, *Rhode Island Hosp., Providence*

B4. Molecular Genetics
Regency Ballroom/Brickell
Moderator: Richard A. King, MD, PhD, FACMG, *Univ. of Minnesota, Minneapolis*

4:00 pm

19. Identification of Candidate Genes Involved in Genitourinary Malformations
Joan Overhauser, PhD, *Thomas Jefferson Univ., Philadelphia, PA*

4:15 pm

20. Exclusion of Linkage to Chromosome 3q in Some Familial Cases of the Cornelia deLange Syndrome
Ian D. Krantz, MD, *The Children's Hosp. of Philadelphia, PA*

4:30 pm

21. Recent Progress in Identification of the X-Linked Spinal Muscular Atrophy (XL-SMA) Gene Confirms a Major Disease Locus Between Xp11.3-Xp11.2
Lisa L. Baumbach-Reardon, PhD, FACMG, *Univ. of Miami School of Medicine, FL*

4:45 pm

22. Characterization of a Novel Candidate Gene in Xp.22.3 with Homology to *Drosophila msl3*
Ignatia B. Van den Veyver, MD, *Baylor College of Medicine, Houston, TX*

5:00 pm

23. Transmission of a Fragile X Full Mutation from a Premutation Male to His Two Daughters
Peter J. Bridge, PhD, FCCMG, FACMG, *Alberta Children's Hosp., Calgary, Canada*

5:15 pm

24. Genetic Analysis of Patients with Pancreatitis and a History of Alcohol Abuse
Kristin G. Monaghan, PhD, *Henry Ford Hosp., Detroit, MI*

5:45 pm–6:45 pm
ACMG Membership Meeting
Regency Ballroom

8:00 pm–10:00 pm
DIAGNOSTIC DILEMMAS (UNKNOWNS)
Orchid Room C

Presentations by participants to the panel of consultants: Helga V. Toriello, PhD, FACMG, *Genetic Services, Grand Rapids, MI*; Marilyn C. Jones, MD, FACMG, *Children's Hosp., San Diego, CA*; and David B. Flannery, MD, FACMG, *Medical College of Georgia, Augusta*

Sunday, March 21

All sessions and breaks in locations as indicated.

8:00 am–5:00 pm
Registration
Promenade Lower Level

8:00 am–8:30 am
Continental Breakfast
Promenade Lower Level

Posters only: 9:50 am–2:00 pm

ANNOUNCEMENTS

8:25 am–8:30 am
Jasmine/Hibiscus Rooms

PLENARY SESSION VI

8:30 am–11:40 am
Jasmine/Hibiscus Rooms

Medical Genetics Update

Moderators: R. Rodney Howell, MD, FACMG, *Univ. of Miami School of Medicine, FL*; Robert L. Nussbaum, MD, FACMG, *National Human Genome Research Inst., NIH, Bethesda, MD*

This session will feature four diverse lectures in areas of interest to medical geneticists and counselors. The session will begin with a presentation by Dr. Joseph Nadeau entitled "Functional Genomics." Dr. Nadeau is a specialist in the area of human and mouse genome research and his talk will focus on the interplay between genetic studies in both organisms and how they illuminate human development and disease. The second presentation will be from Dr. Huda Zoghbi, a renowned pediatric neurologist and neurogeneticist, who will present her work on the pathogenesis of the triplet repeat disorder spinocerebellar ataxia I and its important implications for our understanding of neurodegeneration. The third presentation, from Dr. Aubrey Milunsky, a highly respected human geneticist and a Fellow of the ACMG, will be a first-hand overview of how medical geneticists can function optimally as experts in medical malpractice litigation. Finally, Dr. Craig Basson, one of the leaders in the field of the molecular analysis of congenital heart disease, will present his latest findings on atrial septal defects and other forms of congenital heart disease.

▶ After attending this session, participants will be able to:

1. Appreciate the latest advances in research into the pathogenesis of genetic disorders causing congenital heart defects and nervous system degeneration.
2. Discuss legal aspects of medical genetics expert testimony and how the specialty of medical genetics operates in the courtroom.
3. Become *au courant* with the interplay between the human and mouse genome projects and how comparisons between the two organisms provide powerful insights into human development and disease.

8:30 am

Functional Genomics
Joseph H. Nadeau, PhD, *Case Western Reserve Univ., Cleveland, OH*

9:10 am

Pathogenesis of Trinucleotide Repeat Disorders
Huda Y. Zoghbi, MD, *Baylor College of Medicine, Houston, TX*

9:50 am
Break
Riverfront Hall

10:20 am

Litigation and Clinical Genetics
Aubrey Milunsky, MD, DSc, FACMG, *Boston Univ. School of Medicine, MA*

11:00 am

Genetics of Congenital Heart Disease: Cardiac Malformation in Holt-Oram Syndrome
Craig T. Basson, MD, PhD, *Cornell Univ. Medical College-New York Hosp., NY*

11:40 am–2:00 pm
Break

POSTER SESSION II

(Authors present, even-numbered boards)
1:00 pm–2:00 pm
Riverfront Hall

CONCURRENT WORKSHOPS C

2:00 pm–3:30 pm

C1. Genetics of Ocular Disorders

Hibiscus Room

Moderator: Bassem A. Bejjani, MD, *Baylor College of Medicine, Houston, TX*

This workshop will review recent advances in the genetics of eye diseases. The past few years have witnessed a rapid expansion of our understanding of the molecular basis of eye diseases. The study of age-related macular degeneration, and glaucoma in particular, has resulted in a flurry of publications as well as interesting paradigms for the genetics of complex disorders. Additionally, we will review how the group of diseases lumped under the term "retinitis pigmentosa" are proving to be more diverse at the molecular level than previously expected.

▶ After attending this session, participants will be able to:

1. Appreciate the genetic commonality between Stargardt disease and age-related macular degeneration.
2. Become familiar with the genetic basis of various forms of glaucoma.
3. Understand the genetic heterogeneity of retinitis pigmentosa.

Population and Family Screening of CYP1B1 and TIGR Mutations in Different Forms of Primary Glaucoma

Mansoor Sarfarazi, PhD, *Univ. of Connecticut Health Center, Farmington*

Simple Approaches to Complex Retinal Disorders: The Stargardt-AMD Story

Richard A. Lewis, MD, MS, *Baylor College of Medicine, Houston, TX*

Molecular Genetic Diagnosis of Patients with Ocular Disorders

Roberta A. Pagon, MD, FACMG, *Univ. of Washington, Seattle*

C2. Lumping and Splitting: Controversies in Clinical Genetics Caused by Molecular Delineation of Genetic Syndromes

Orchid Room BCD

Moderators: Nathaniel H. Robin, MD, FACMG, *Case Western Reserve Univ. School of Medicine, Cleveland, OH*; Helga V. Toriello, PhD, FACMG, *Genetic Services, Grand Rapids, MI*

This session contains a series of lectures that highlight how recent advances in understanding the molecular basis of many clinically defined genetic syndromes have forced clinicians to revisit long-held beliefs about many of these disorders. Examples include the crainosynostosis syndrome, Pallister-Hall and Greig syndromes, Stickler syndrome and Beckwith-Wiedemann syndrome, as well a talk on "Nosology in the Molecular Age."

▶ After attending this session, participants will be able to:

1. Understand the impact that molecular genetics has had in understanding the relationship of these genetic syndromes.
2. Recognize how these advances have improved our ability to accurately diagnose and counsel families and patients with these syndromes.
3. Recognize that for many syndromes, how we clinically define these entities does not reflect their molecular basis.

The Molecular Delineation of Craniosynostosis: Fusion and Overlap of Clinical Syndromes

Nathaniel Robin, MD, FACMG, *Case Western Reserve Univ. School of Medicine, Cleveland, OH*

Lumping and Splitting: Human Developmental GL13 Disorders

Leslie G. Biesecker, MD, *National Human Genome Research Inst., NIH, Bethesda, MD*

The Growing Confusion Surrounding Overgrowth Syndromes

Rosanna Weksberg, MD, PhD, FACMG, *Hosp. for Sick Children, Toronto, Ontario, Canada*

Stickler Syndrome, Marshall Syndrome and Other Collagenopathies

Matthew L. Warman, MD, *Case Western Reserve Univ. School of Medicine, Cleveland, OH*

Nosology in the Molecular Age

John Opitz, DSci, MD, FACMG, *Univ. of Utah School of Medicine, Salt Lake City*

C3. Dilemmas in Genetic Counseling: The Mentally Handicapped Individual in the Genetic Counseling Setting

Jasmine Room

Moderators: Elizabeth A. Balkite, MS, CGC, *Glaxo Wellcome Research and Development, Research Triangle Park, NC*; Brenda Finucane, MS, CGC, *Elwyn, Inc., Elwyn, PA*

With the expansion of genetic services and testing to an increasing number of genetic disorders, genetic counselors encounter clients who are themselves mentally handicapped, or have a mentally handicapped relative whose informed consent is necessary for genetic studies. This workshop is designed to educate attendees about the medical, ethical, legal and culturally sensitive issues involved when counseling these clients. Case studies will be used to demonstrate how to apply specific recommendations when counseling mentally handicapped patients and/or their relatives in prenatal, pediatric and cancer genetic counseling settings.

▶ After attending this session, participants will be able to:

1. Understand the issues involved when providing genetic counseling for mentally handicapped individuals and/or their relatives.
2. Learn through case studies how to apply specific recommendations when counseling mentally handicapped patients and/or their relatives.
3. Learn how to obtain real informed consent for genetic testing when the proband or his/her relative is mentally handicapped.

Working with Women Who Have Mental Retardation: Guidelines for Genetic Counseling
Brenda Finucane, MS, CGC, *Elwyn, Inc., Elwyn, PA*

Case Presentations: Decisionmaking and the Pregnant Patient
Rachel Baughman, MS, CGC, *Univ. of North Carolina, Chapel Hill*

HD Testing and Mental Impairment
Catherine Walsh-Vockley, MS, CGC, *Mayo Clinic, Rochester, MN*

Cancer Predisposition Testing: Patients and Families
Jessica B. Mandell, MS, *Sarah Lawrence College, Bronxville, NY*

Critical Elements: Genetic Counseling Mentally Impaired Individuals
Elizabeth A. Balkite, MS, CGC, *Glaxo Wellcome Research & Development, Research Triangle Park, NC*

3:30 pm–4:00 pm
Break
Promenade Lower Level

CONCURRENT WORKSHOPS D

4:00 pm–6:00 pm

D1. Down Syndrome: Mysteries and Revelations

Hibiscus Room

Moderator: Ronald P. Bachman, MD, FACMG, *Kaiser Permanente Medical Center, Oakland, CA*

A comprehensive (and rapid) review of controversial issues in Down syndrome: prenatal to clinical to treatment to pathologic changes. A brief review of current molecular and relevant research will be included. Attendees will learn that they did not know everything they thought they knew about Down syndrome, either on a clinical or a research level.

▶ After attending this session, participants will be able to:

1. Describe the controversial clinical issues and the approach to solving them.
2. Describe the treatment of Down syndrome: standard approach and "non-orthodox" approach.

Prenatal Ultrasound Detection: Nuance or Nuisance
Rebecca Smith-Bindman, MD, *UCSF–Mt. Zion Medical Center, San Francisco, CA*

Counseling Controversies and Issues in Down Syndrome
Cam Brasington, MS, *Carolinas Medical Center, Charlotte, NC*

Clinical Conundrums
Tracy L. Trotter, MD, *San Ramon Valley Primary Care, San Ramon, CA*

Alternative Therapies in Down Syndrome
Lawrence G. Leichtman, MD, FACMG, *Genetics & Disabilities Diagnostic Care Center, Virginia Beach, VA*

Mapping and Models: Molecular Update on Down Syndrome
Gregory P. MacDonald, MD, *Kaiser-Permanente Medical Center, Vallejo, CA*

Fetal and Neonatal Pathology of Down Syndrome
Geoffrey A. Machin, MD, PhD, *Permanente Medical Group, Oakland, CA*

D2. Heritable Disorders of Connective Tissue: Bench to Bedside

Orchid Room BCD

Moderator: Reed E. Pyeritz, MD, PhD, FACMG, *Allegheny General Hosp., Pittsburgh, PA*

Knowledge of the molecular bases for heritable disorders of connective tissue is accruing rapidly. However, most clinicians are unsure when to order molecular diagnostic information and whether it is available.

▶ After attending this session, participants will be able to:

1. Discuss the molecular bases of several common heritable disorders of connective tissue.
2. Indicate how molecular information might, and might not, be useful in diagnosis and management.
3. Present the current state of clinical availability of molecular testing.

Osteogenesis Imperfecta: Prospects for Molecular Therapeutics
Joan C. Marini, MD, PhD, FACMG, *HDB/NICHD/NIH, Bethesda, MD*

Molecular Basis of Marfan Syndrome and Related Disorders
Dianna M. Milewicz, MD, PhD, *Univ. of Texas Health Science Center, Houston*

Elastin and the Williams Syndrome Phenotype
Colleen A. Morris, MD, FACMG, FAAP, *Univ. of Nevada School of Medicine, Las Vegas*

D3. Diagnosis, Management and Population Screening Issues for Hemochromatosis

Jasmine Room

Moderator: Alan E. Donnenfeld, MD, FACMG, *Pennsylvania Hosp., Philadelphia*

Hereditary hemochromatosis (HH) is a recessive disorder that affects 30-50 per 10,000 individuals in white populations, and results in significant morbidity and mortality. However, because symptoms can mimic other common conditions, conventional clinical approaches have been relatively ineffective in accurately diagnosing HH in a timely manner. Treatment is simple and can prevent clinical manifestations. Screening strategies (mutational analysis and transferrin saturation testing) have been proposed, and test performance data are available. Therefore, HH would seem to be an ideal candidate for population-based screening. However, uncertainties remain, including the age-related penetrance of the disorder, the relationship between *HFE* genotype, iron status markers and clinical presentation, and the most effective programmatic design (educational materials, systems for appropriate diagnostic testing and follow-up, and psychosocial impact).

▶ After attending this session, participants will be able to:

1. Provide medically relevant information about the natural history, genetics, clinical presentation, diagnosis and management of HH.
2. Provide background information about the discovery of the *HFE* gene and two missense mutations (C282Y and H63D), review current data on population frequency and penetrance, and evaluate the role of mutational analysis in the diagnosis of HH.
3. Compare and contrast mutational analysis and transferrin saturation testing as proposed strategies for population screening, underscore the differences between screening in the asymptomatic general population and case-finding among symptomatic individuals or families of probands, and review the issues and controversies remaining to be resolved prior to implementation of screening.

Clinical Features of Hemochromatosis with Emphasis on Clinical Diagnosis, Management and Prognosis
Corwin Q. Edwards, MD, *Univ. of Utah College of Medicine, Salt Lake City*

Molecular Genetic Approaches to Diagnosis of Hemochromatosis, Including Population Frequency Data
Wylie Burke, MD, PhD, *Univ. of Washington, Seattle*

Issues and Controversies Surrounding Population Screening for Hemochromatosis
Linda A. Bradley, PhD, FACMG, *Foundation for Blood Research, Scarborough, ME*

Monday, March 22

POST-MEETING WORKSHOP

8:30 am–12:30 pm

Regency Ballroom/Brickell

Billing, Collections and Compliance in 1999. (See announcement, page 14.)