CLINICAL GENETICS SERVICES
CENTER FOR MOLECULAR MEDICINE AND GENETICS

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Updates in Genetic Medicine

Fifty Years of DNA: From Double Helix to Genomic Medicine

Anne Greb, MS, CGC

April 2003 witnessed the historic culmination of one of the most important scientific projects in history: the sequencing of the human genome. This date also marked the 50th anniversary of James Watson and Francis Crick's Nobel Prize winning description of the molecular structure of DNA. An incredible amount of progress has been made in understanding the genetic basis of human disease in the 50 years spanning these two events, in large part, due to the Human Genome Project.

The **Human Genome Project (HGP)** began in 1990 with the goal of mapping and sequencing all the human genes as well as those of model organisms. The inspiration for this enormous effort was to identify the genetic components of human diseases, to provide diagnostic tests, and then ultimately, to develop better treatments and cures. Because of advances in laboratory techniques , the project progressed well ahead of schedule. In February 2001, the international genome consortium reported the completion of a draft sequence and its implications in both *Science* and *Nature*. As of April 2003, the human genome is 99% complete with 99.99% accuracy. The research generated by the HGP has already had a profound impact on clinical medicine. Genes associated with numerous genetic conditions such as Huntington disease, cystic fibrosis, fragile X, inherited colon cancer, Alzheimer disease and inherited breast cancer are among the 8,000 human genes identified so far.

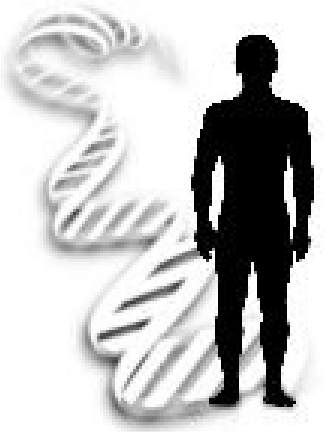

Image provided by the U.S. Dept. of Energy Human Genome Program. www.ornl.gov/hgmis

The identification of the gene(s) responsible for a specific disease allows researchers to better understand the disease process, creates the potential for ge-

(Continued on page 2)

Inside this issue:

Points of Interest

- Know your responsibilities with regard to informed consent for genetic testing. Sample consent enclosed.
- Find out what a health insurance provider can and cannot ask you about your genetic information.
- Learn about Michigan's expanded newborn screening.

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netic testing to identify at-risk individuals, and in some cases, for improving treatment or preventing disease.

For example, the gene for cystic fibrosis, CFTR, was mapped to chromosome 7 and then cloned in 1989. Shortly after, it was determined that the protein made by the gene is a chloride channel. This discovery has increased our understanding of the disease process in cystic fibrosis (CF). Furthermore, identifying the specific mutations a person has can provide some indication of symptomatology. The options of carrier testing, prenatal diagnosis, and preimplantation diagnosis are now available and gene therapy clinical trials are underway.

> ***"(The human genome)...is a giant resource that will change mankind, like the printing press."***
>
> **Dr. James Watson, Co-discoverer of DNA structure.**

Programs to implement population screening for CF have also evolved. Recently the American College of Obstetrics and Gynecology and the American College of Medical Genetics issued statements recommending that CF carrier screening be offered to all Caucasian couples of Ashkenazi Jewish and non-Jewish descent planning a pregnancy or seeking prenatal care. They also recommended that screening be made available to other ethnic and racial groups with informed consent about the limitations of detecting carriers in these populations.

In addition to genes for relatively rare disorders, the HGP has also helped to identify genes for more common conditions. In 1994, the first gene associated with an inherited form of breast cancer, BRCA1, was identified on chromosome 17. BRCA1 gene analysis is an example of how a genetic test can identify people who have a high likelihood of developing a disease long before they have any symptoms. Identification of such persons provides a potential for aggressive surveillance and prophylactic treatment to reduce disease-related morbidity and mortality.

However, such technology has created new concerns. With genetic information comes a potential threat to a person's privacy in the form of genetic discrimination. Also, individual risk identification can reveal information about the rest of the family, some members of which may not want this knowledge. From its inception, those who developed the HGP recognized the need to anticipate and address the societal implications of genetic information. Consequently, a portion of the HGP budget has been spent on efforts to develop programs and policies to address the Ethical, Legal and Societal Issues (ELSI).

In 1994, the Institute of Medicine of the National Academy of Sciences issued a report assessing the potential implications of genetic testing with particular attention to genetic discrimination. This report recommended that people be fully informed of the implications of genetic testing and that genetic counseling precede testing. The issues of informed consent and the societal implications of genetic technology continue to be addressed through the work of Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS).

In summary, the role of genetics in medicine is evolving rapidly. The goal of the clinical genetics service providers at the Detroit Medical Center/Wayne State is to keep abreast of such advances in order to provide patients with comprehensive, up-to-date, informed genetic healthcare.

Informed Consent for Predictive or Presymptomatic Genetic Testing: It's the Law!

Angela Trepanier, MS, CGC

On March 15, 2000, legislation went into effect in Michigan that requires a physician (or an individual to whom the physician has delegated authority) to obtain written informed consent from a patient before ordering a presymptomatic or predictive genetic test. Written informed consent must consist of a signed document executed by the patient or his/her legally authorized representative. The consent form should include language that documents that the patient has been informed and understands, at minimum, all of the following:

- The nature and purpose of the presymptomatic or predictive genetic test
- The effectiveness and limitations of the test (accuracy, specificity, and sensitivity)
- The implications of the test, including the medical risks and benefits
- The future uses of the sample taken from the patient in order to conduct the test and the information obtained from the presymptomatic or predictive test
- The meaning of the genetic test results and the procedure for results notification
- Who will have access to the sample taken from the patient and the information obtained from the test, and the patient's right to confidential treatment of the sample and information

The physician is required to give the patient a copy of the signed consent form. The original form should be kept in the patient's medical record. If the patient signs an informed consent document, he/she is barred from subsequently bringing a civil action for damages against the physician based on failure to obtain informed consent.

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As part of this legislative act, the Michigan Department of Community Health, in consultation with several medical specialists and professional organizations, was charged with developing a model informed consent document. Dr. Gerald Feldman from Wayne State University chaired this committee. A copy of the consent form language is included (page 4). You can make as many copies as you need.

An informed consent brochure, which is written for patients to describe in general terms the benefits, risks, and limitations of genetic testing, has also been developed. If you would like copies of the brochure, please contact Valerie Ewald at ewaldv@michigan.gov or (517) 335-8110.

CONSENT TO OBTAIN A SPECIMEN FOR GENETIC TESTING

PATIENT LAST NAME: FIRST NAME: MI:
(Please Print)

DATE OF BIRTH: / /

HOSPITAL/ID NUMBER:

ORDERED BY:

I REQUEST GENETIC TESTING FOR:

_____
(name of condition)

LABORATORY NAME AND ADDRESS:

_____

_____

_____

SAMPLE TYPE

- ☐ Amniotic fluid
- ☐ Blood
- ☐ Cheek swab
- ☐ Chorionic villus sampling (CVS)
- ☐ Skin
- ☐ Tissue block
- ☐ Other_____

THERE IS A FAMILY HISTORY OF THIS CONDITION: Yes No

The intended purpose is (check all that apply):

- ☐ Carrier status
- ☐ Diagnostic
- ☐ Predictive
- ☐ Prenatal
- ☐ Pre-symptomatic
- ☐ Screening
- ☐ Other _____

1. I have been informed about the purpose of this genetic test.
2. I have received an explanation of the limitations of this genetic test.
3. I have discussed the benefits and risks of this genetic test with my physician and other health care professional. I understand some genetic tests can involve possible medical, psychological, and insurance issues for my family and me.
4. I understand the meaning of possible test results and have been informed how I will receive the result.
5. I understand any leftover sample may be retained by the laboratory, but only used for my future clinical care, with my permission. I understand I may cancel this consent up until the time the test is performed.
6. I have been informed who may have access to the biological sample and genetic test result. I have been informed that the test result will be part of my medical record and remain confidential in accordance with standard medical practice.
7. I have read this consent form and booklet. I received a copy of the form and booklet for my records. My questions have been answered to my satisfaction.

By my signature below, I give consent to have a sample taken for genetic testing on the above-named patient for the condition(s) listed above.

_____
Signature of Patient or Authorized Designee

Circle one: **Self** **Parent(s)** **Legal Guardian** **Durable Power of Attorney for Health Care**

Print Name of Authorized Person Reviewing Consent Form with Patient:

Signature of Authorized Person: Date:

Clinical Genetics Patient Services and Education

Angela Trepanier, MS, CGC

Your 30-year old patient has a sister and mother who developed breast cancer in their 40's. What is her chance of developing breast cancer? Is she at increased risk for any other types of cancer? Is there a genetic test that can determine her cancer risk?

Your pregnant patient has a brother with cystic fibrosis. What is the chance that her baby will have the condition? Can genetic testing determine if the baby will be affected?

Your patient is a 5-year old boy with developmental delay and unusual physical features. His parents want to know what caused his condition, what to expect regarding his future development, and whether any other children they may have will be at risk. How do you attempt to answer their questions?

Your 35-year old patient recently found out that his father had Huntington disease (HD). What is the chance your patient will develop the condition? What are the potential implications if he seeks presymptomatic genetic testing for HD?

Clinical genetics professionals are individuals trained to work with patients and families to answer these types of questions. Providing genetic services requires a team approach. The team includes the following:

Clinical geneticists: MD's with specialized training and board certification in genetics and another discipline (e.g., pediatrics, neurology, internal medicine, or obstetrics/gynecology). Clinical geneticists have expertise in the diagnosis and management of genetic conditions.

Genetic counselors: Master's degree level individuals who have specialized training and usually certification in genetic counseling. Genetic counselors have expertise in identifying people at risk of genetic disease, educating them about genetic risk and testing options, facilitating informed decision making regarding genetic testing and health management options, and helping people cope with the genetic condition in their family. Genetic counselors and geneticists are also involved in providing genetics education to other health care professionals.

Genetics laboratory specialists: These specialists include cytogeneticists, who perform chromosomal analyses; molecular geneticists, who perform gene (DNA and RNA) analyses; and, biochemical geneticists, who analyze and quantify enzyme levels as well as the biochemical products of metabolic pathways.

Other specialists: Specialists, such as neurologists, oncologists, and maternal fetal medicine physicians are an integral part of the team in genetics specialty clinics.

Currently, genetic services at the Detroit Medical Center (DMC) are provided through the following programs: **Cancer Genetic Counseling**, **Neurogenetics**, **Pediatric Genetics**, and **Reproductive Genetics**. The DMC also houses a molecular diagnostics laboratory and a cytogenetics laboratory in the Department of Pathology. The clinics and laboratories support training programs and education in medical genetics. Integration and coordination of the training programs and clinical genetics services is achieved through the Center for Molecular Medicine and Genetics.

A description of the clinical services and educational programs as well as biographical information about the professionals involved with them is included on the following pages.

Clinical Genetics Services
Medical Genetics Education and Training Programs
Center for Molecular Medicine and Genetics (CMMG)

This component of the CMMG is responsible for the following: integration and coordination of all clinical genetics services; administration of a medical genetics residency program, a genetic counseling graduate program, and post-doctoral fellowship programs in clinical molecular genetics, clinical cytogenetics, and PhD medical genetics; and genetics education for medical and graduate students, healthcare professionals, and lay audiences. For more information or to request educational outreach, contact Willia Lake at (313)577-6298.

Gerald Feldman, MD, PhD. Dr. Feldman is the Director of Clinical Genetics Services, Director of the Medical Genetics Residency Program, Medical Director of the Genetic Counseling Graduate Program, and Director of the Molecular Genetics Diagnostic Laboratory. He is an associate professor in the CMMG, and the departments of Pathology, and Pediatrics. Dr. Feldman obtained his PhD and MD from the Medical College of Virginia. He did post-doctoral fellowships in molecular genetics and biochemical genetics as well as a pediatrics residency and clinical genetics fellowship at Baylor College of Medicine. He is board certified in pediatrics, clinical genetics, molecular genetics and biochemical genetics. Dr. Feldman coordinates patient care in all the genetics clinics of the Detroit Medical Center. His research interests include identifying the current practices of genetics professionals in evaluating hearing loss etiology and genotype phenotype correlations in MEN2 and cystic fibrosis. Dr. Feldman, as former chairman of the Michigan Genetics Advisory Committee, has also been involved in ensuring proper use of genetic technology in the state.

Anne Greb, MS, CGC. Anne is the Director of the Genetic Counseling Graduate Program and an assistant professor in the CMMG. Anne received her Masters of Science in Genetic Counseling from the University of Wisconsin and is certified by the American Board of Genetic Counseling. In addition to her role with the genetic counseling program, she is the co-director of the Year 1 Medical Genetics Course for medical students. Anne's interests include improving and expanding medical genetics education and identifying factors that motivate African-American women to seek genetic counseling for cancer risk.

Angela Trepanier, MS, CGC. Angela is the Co-Director of the Genetic Counseling Program and an assistant professor in the CMMG. Angela received her Masters of Science in Genetic Counseling from the University of Minnesota and is certified by the American Board of Genetic Counseling. Her clinic interests include genetic risk assessment and counseling for cancer and other adult onset disorders. Other interests include promoting medical genetics education, developing cancer genetic counseling practice guidelines, and identifying barriers to genetic evaluation for individuals with hearing loss.

Cancer Genetic Counseling Service
Karmanos Cancer Institute

About 5-10% of common cancers, such as breast and colon cancer, are inherited. People with an inherited predisposition tend to develop specific types of cancer at an earlier age than usual, are at increased risk of developing two or more tumors, and may be at risk for developing more than one type of cancer. In addition, their relatives may also have an increased risk of cancer.

The Cancer Genetic Counseling Service identifies individuals and families with an inherited predisposition to cancer by taking and assessing a comprehensive family history, provides them with information about the nature and magnitude of their cancer risk, offers them genetic testing when available and indicated and provides informed consent for genetic testing, and gives them information about how to manage their cancer risk. Identification of those with an inherited predisposition to cancer leads to increased cancer surveillance and a discussion of risk reduction strategies, when available. The ultimate goal is that this will reduce the chance of developing or dying from cancer for the individual as well as his or her at-risk relatives. To make an appointment, contact Wanda Sheard at (313)966-7780.

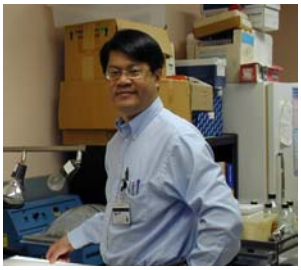
Michael Simon, MD, MPH. Dr. Simon is the Director of the Cancer Genetic Counseling Service of the Karmanos Cancer Institute and is an associate professor in the Department of Internal Medicine. He also has an associate faculty position in the Center for Molecular Medicine and Genetics. Dr. Simon obtained his MD from the University of Illinois and Master's of Public Health (Epidemiology) from the University of Michigan . He completed a residency in internal medicine and a fellowship in hematology/oncology at the University of Michigan and is board-certified in both. Dr. Simon's research interests include identifying factors that affect cancer screening and that motivate African-American women to seek cancer genetic risk counseling. The Centers for Disease Control recently awarded Dr. Simon the prestigious Charles C. Shepard Award for scientific excellence demonstrated by the publication of an article he co-authored, entitled "Oral Contraceptives and the Risk of Breast Cancer".

Nancie Petrucelli, MS, CGC. Nancie is a genetic counselor and the Coordinator of the Cancer Genetic Counseling Service at the Karmanos Cancer Institute. She has an adjunct faculty position in the Center for Molecular Medicine and Genetics. Nancie obtained her Masters of Science in Genetic Counseling at the University of Cincinnati and is certified by the American Board of Genetic Counseling. She has been very active in cancer genetics education and cancer genetics-related activities in the state and is currently the co-chair of the Michigan Cancer Genetics Alliance.

Neurogenetics Program
University Health Center

Half of all genetic diseases affect the nervous or neuromuscular systems. There is also growing acceptance of the role genetics play in predisposition to common diseases, such as stroke, headache, epilepsy and multiple sclerosis. The Neurogenetics Program, which includes the Neurogenetics Clinic and the Charcot-Marie-Tooth Disease (CMT) Clinic, offers comprehensive diagnostic and care services as well as genetic counseling for those who have or who are suspected of having a genetic disorder affecting the nervous system or muscles. Examples of such disorders include Huntington disease, spinocerebellar ataxias, muscular dystrophies, mitochondrial disorders, leukodystrophies, hereditary spastic paraplegia, Charcot-Marie-Tooth disease, neurofibromatosis, tuberous sclerosis, Friedreich ataxia, some types of Alzheimer disease, and amyotrophic lateral sclerosis.

The staff of the Neurogenetics Clinic also provides comprehensive pre- and post-test evaluation and genetic counseling for those who are at risk for, but do not yet manifest symptoms of a disease, such as Huntington disease (presymptomatic testing). This service is offered in recognition that the decision to undergo presymptomatic testing should be made only after careful consideration for one's own need to know this information, along with concerns for one's family, occupation and financial planning. To make an appointment in the Neurogenetics Clinic call Karen Krajewski at (313)577-8317. For appointments in the CMT clinic, call Lisa Rowe at (313)577-1689 .



James Garbern, MD, PhD. Dr. Garbern is the Director of the Neurogenetics Clinic and an associate professor in the Department of Neurology and the Center for Molecular Medicine and Genetics. He obtained his PhD in Neurosciences and Cell Biology and his MD from Baylor University. Following his graduate training, he completed a residency in neurology at the University of Washington and is board-certified in the specialty. Dr. Garbern also did postdoctoral work in molecular neurobiology at the National Institutes of Health. His research interests include Pelizaeus-Merzbacher disease (PMD) and the molecular genetics of myelin disorders. Dr. Garbern is currently a member of the Board of Directors of the PMD Association.

John Kamholz, MD, PhD. Dr. Kamholz is the Co-Director of the Neurogenetics Clinic and a professor in Neurology and the Center for Molecular Medicine and Genetics. He obtained his MD, PhD, and completed a medical residency in neurology at the University of Pennsylvania and is a board-certified neurologist. Dr. Kamholz also did a post-doctoral fellowship in molecular neurobiology at the National Institutes of Health. His research interests include multiple sclerosis, Pelizaeus-Merzbacher disease and the molecular basis of disorders affecting myelin.

Michael Shy, MD. Dr. Shy is the Director of the Charcot-Marie-Tooth Clinic and Co-Director of the Neuromuscular Disease program. He is a professor in the Department of Neurology and the Center for Molecular Medicine and Genetics. Dr. Shy obtained his MD from Albany Medical College. He completed a residency program in neurology followed by a fellowship in neuromuscular diseases at Columbia Presbyterian in New York and is a board-certified neurologist. Dr. Shy has published extensively on the topic of acquired and inherited diseases of the peripheral nervous system, including Charcot-Marie-Tooth disease (CMT). For his work with CMT families, the CMT Disease Association has presented him with an Award of Excellence.

Karen Krajewski, MS, CGC. Karen is a genetic counselor and assistant professor in the Department of Neurology with an associate faculty position in the Center for Molecular Medicine and Genetics. She coordinates the Adult Neurogenetics Clinic and the Charcot-Marie-Tooth (CMT) Clinic. Karen is a graduate of the Indiana University Genetic Counseling Graduate program and is certified by the American Board of Genetic Counseling. Her clinical interests include Huntington disease and other neurogenetic disorders. She is also interested in presymptomatic genetic testing issues for adult onset neurological disorders. Karen is actively involved in research on both CMT and Pelizaeus-Merzbacher disease. She has numerous publications and has presented both nationally and internationally.

Pediatric Genetics Clinic
Children's Hospital of Michigan

Over 4000 single gene disorders have been described, many of which present with symptoms at birth, in infancy or in early childhood. The Pediatric Genetics Clinic offers comprehensive diagnostic and care services for children who have, are suspected of having, or are at risk of having a genetic condition. Examples of conditions commonly seen in the clinic include Down syndrome, Marfan syndrome, neurofibromatosis, hereditary deafness, metabolic disorders, fragile X mental retardation, and chromosome deletion syndromes (e.g., Williams syndrome, Prader-Willi syndrome, Angelman syndrome). Common indications for genetic evaluation include mental retardation or autism (rule out genetic etiology), multiple congenital anomalies, suspected genetic disorder, or family history of inherited disease.

A patient is evaluated for the presence of a genetic disorder by collecting a comprehensive family history and medical history, reviewing relevant medical records and tests, performing a physical examination including a dysmorphology exam, and ordering genetic testing (if available and indicated). Sometimes additional medical tests are needed to establish or rule out a diagnosis. If a diagnosis is established, the clinic staff provides families with information about the condition, the inheritance pattern, and the risk to other relatives, resources, medical management guidelines, and support. To make an appointment, call Sherry Young at (313)745-4513. *(continued on page 10)*

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Erawati Bawle, MD. Dr. Bawle is the Director of the Division of Genetics & Metabolic Disorders at Children's Hospital of Michigan and a consulting physician at St. John's Hospital. She is professor in the Department of Pediatrics and the Center for Molecular Medicine and Genetics. Dr. Bawle obtained her MD from Grant Medical College at the University of Bombay and completed a pediatrics residency at both the University of Bombay and Children's Hospital of Michigan. She is board certified in pediatrics and clinical genetics. Dr. Bawle sees a wide variety of patients in the Pediatric Genetics Clinic and has expertise in dysmorphology and managing patients with metabolic disorders. She is also very active in providing education and training to medical students, genetic counseling students, and residents.

Patients are also seen by Dr. Gerald Feldman, MD, PhD, who is also a board-certified clinical geneticist (see page 5).

Joan Conard, MS, CGC. Joan is a genetic counselor in the Division of Genetic and Metabolic Disorders at Children's Hospital of Michigan. She has an adjunct faculty position in the Center for Molecular Medicine Genetics. Joan obtained her degree in genetic counseling from the University of South Carolina and is certified by the American Board of Genetic Counseling. She provides genetic counseling for a wide variety of indications in the Pediatric Genetics Clinic. In addition, she counsels families about hemophilia and other bleeding disorders in the Hemophilia Treatment Center at Children's Hospital and the Harper Hospital Hemophilia Clinic.

Nicole Teed, MS, CGC. Nicole is a genetic counselor in the Division of Genetic and Metabolic Disorders at Children's Hospital of Michigan. She obtained her degree in genetic counseling from the University of Michigan and is certified by the American Board of Genetic Counseling. Nicole provides genetic counseling for a wide variety of indications in the Pediatric Genetics Clinic. Her research interests include development of a cost-effective approach for evaluating children with developmental delay.

Sujatha Sastry, MS, CGC. Sujatha is a genetic counselor in the Division of Genetic and Metabolic Disorders at Children's Hospital of Michigan. She obtained her degree in genetic counseling from the University of California Berkeley and is Board Certified by the American Board of Genetic Counseling. Sujatha provides genetic counseling for a wide variety of indications in the Pediatric Genetics Clinic. Other clinical interests include issues surrounding presymptomatic testing for Huntington Disease.

Reproductive Genetics Services
Hutzel Hospital

Technological advances have allowed for the prenatal diagnosis of an increasing number of genetic conditions and birth defects and for the identification of couples at risk of having offspring with such conditions. Reproductive Genetics Services uses the latest technologies to accurately diagnose, and in some cases, treat birth defects and genetic conditions detected during pregnancy. Reproductive Genetics offers the following: genetic counseling for both pregnant and non-pregnant patients; management of fetal anomalies; chorionic villus sampling (CVS) and midtrimester genetic amniocentesis for a variety of indications; fetal ultrasound; and other tests and procedures. Common indications for genetic counseling and services include advanced maternal age, abnormal serum screen results, family history of genetic condition, fetal anomalies (referred for diagnostic evaluation and management); teratogen exposure; and pregnancy at risk for genetic condition based on population carrier screening results or family history. To make an appointment, call (313)745-7066.

Marjorie C. Treadwell, MD. Dr. Treadwell is the Acting Director of Reproductive Genetics Services and the Director of Obstetric Ultrasound at Hutzel Hospital. She is an associate professor in the Department of Obstetrics & Gynecology, Division of Maternal Fetal Medicine and Department of Radiology. Dr. Treadwell obtained her MD at the University of Michigan. She completed a residency in Obstetrics & Gynecology and a Fellowship in Maternal Fetal Medicine at Hutzel Hospital/Wayne State University and is boarded in both. Dr. Treadwell has numerous publications on the topic of obstetrical ultrasound evaluation. She has been named one of the Top Doctors in the United States.

Dr. Gerald Feldman and Dr. Erawati Bawle, both board-certified clinical geneticists, provide genetics consultations for Reproductive Genetics patients as needed.

Adel Gilbert, MS, CGC. Adel is the Chief Genetic Counselor and Coordinator of Genetic Services in the Division of Reproductive Genetics, Department of Obstetrics and Gynecology at Hutzel Hospital. She is a graduate of the McGill University Genetic Counseling program and is certified by the American Board of Genetic Counseling. Adel provides prenatal and preconceptional genetic counseling for a wide range of indications. She is interested in fetal therapy and involved in research examining cultural diversity and the uptake of genetic services, benefits of psychological intervention for couples giving birth to babies with congenital anomalies, neonatal outcome post-in utero therapy for complicated monochorionic twin gestations, and the value of a combined didactic/clinical genetics rotation on obstetrics residents' learning.

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Elizabeth Dugan, MS, CGC. Beth is a genetic counselor in the Division of Reproductive Genetics, Department of Obstetrics and Gynecology at Hutzel Hospital. She obtained her degree in genetic counseling from Case Western Reserve University and is board certified by the American Board of Genetic Counseling. Beth provides prenatal and preconceptional genetic counseling for a wide variety of indications. Her interests include preimplantation genetic diagnosis and ethical concerns related to the use of assisted reproductive technology.

Quick Guide to Genetic Services

Service	Telephone Number	Contact
Clinical Genetics Services **Medical Genetics Education & Training Programs** **Center for Molecular Medicine & Genetics**	(313)577-6298	Willia Lake
Cancer Genetic Counseling Service **Karmanos Cancer Institute**	(313)966-7780	Wanda Sheard
Neurogenetics Program **Department of Neurology** • **Charcot Marie Tooth (CMT) Clinic** • **Neurogenetics (NG) Clinic**	 (313)577-1689 (CMT) (313)577-8317 (NG)	 Lisa Rowe (CMT) Karen Krajewski (NG)
Pediatric Genetics Clinic **Children's Hospital of Michigan**	(313)745-4513	Sherry Young
Reproductive Genetics Service **Hutzel Hospital**	(313)745-7066	(multiple)

Genetic Nondiscrimination in Health Insurance: The Status of Protections for Michigan Residents

Angela Trepanier, MS, CGC

On March 15, 2000, the State of Michigan enacted legislation that prohibits a health insurance provider from requiring an insured member or an asymptomatic applicant from undergoing genetic testing prior to having their health insurance policy renewed, continued, or issued. The law also prevents the insurance provider from asking the insured member to disclose whether genetic testing has been conducted or the results of genetic testing or genetic information.

The law *does not* prevent the insurance provider from asking an applicant to answer questions about their family history. Also, the law does not prevent the provider from using genetic test results/genetic information should it be obtained by other means, such as through a medical record review.

The purpose of this legislation was to extend protections against genetic discrimination already afforded by the federal Health Insurance Portability and Accountability Act (HIPAA) of 1996. HIPAA prohibits excluding a person from group coverage because of past or present medical problems, including genetic information. HIPAA also prevents an insurer from raising an individual's premiums within a group based on genetic information, although the whole group's premiums can be raised.

There are two important distinctions between the federal and state laws. One, HIPAA provides its protections only to those insured as part of a group policy whereas the Michigan law protects those with individual policies as well. Two, HIPAA prevents an insurer from **using** genetic information but does not prevent them from collecting it or from asking an person to have a genetic test. The state legislation prevents health insurers from **collecting** genetic information for use in deciding coverage by two means: directly asking an individual to have a genetic test or to disclose genetic information. However, it does not prevent the use of genetic information if it is obtained by other means. Nonetheless, those covered by group policies would still be protected from use of genetic information by HIPAA, regardless of how it is obtained.

It is important to note that both the federal and state legislation only apply to health insurance coverage and not to life or disability insurance.

Definitions of Terms Used in Michigan Legislation

Genetic Test: Analysis of DNA, RNA, chromosomes, and those proteins and metabolites used to detect heritable or somatic disease-related genotypes or karyotypes for clinical purposes. The test must be accepted in the scientific and medical communities as being specifically determinative for the presence, absence, or mutation of a gene or chromosome to qualify under this definition.

Genetic Information: Information about a gene, gene product, or inherited characteristic derived from a genetic test.

Predictive Genetic Test: A test performed for the purpose of predicting the future probability that a person will develop a genetically related disease or disability.

Presymptomatic Genetic Test: A test performed before the onset of clinical symptoms or indications of disease.

Genetic Nondiscrimination in Employment: An Amendment to the Persons with Disabilities Civil Rights Act of 1976

Angela Trepanier, MS, CGC

On March 15, 2000, the state of Michigan enacted legislation that prevents an employer from doing any of the following because of genetic information or a disability that is unrelated to an individual's ability to perform his/her job:

- Failing to refuse to hire, recruit, or promote an individual
- Discharging an individual
- Discriminating against a person with regard to compensation, terms, or conditions of employment
- Limiting, segregating or classifying an employee or applicant for employment in a way that deprives him/her of employment opportunities or adversely affects his/her status as an employee
- Requiring an individual to submit to a genetic test or to provide genetic information as a condition of employment or promotion

Except in situations where an employee voluntarily provides genetic information related to their health and safety in the workplace, employers are prohibited from directly or indirectly acquiring or having access to any genetic information concerning an employee, an applicant for employment or a member of the employee/applicant's family.

MCAD Added to the Michigan Newborn Screening Panel

Angela Trepanier, MS, CGC

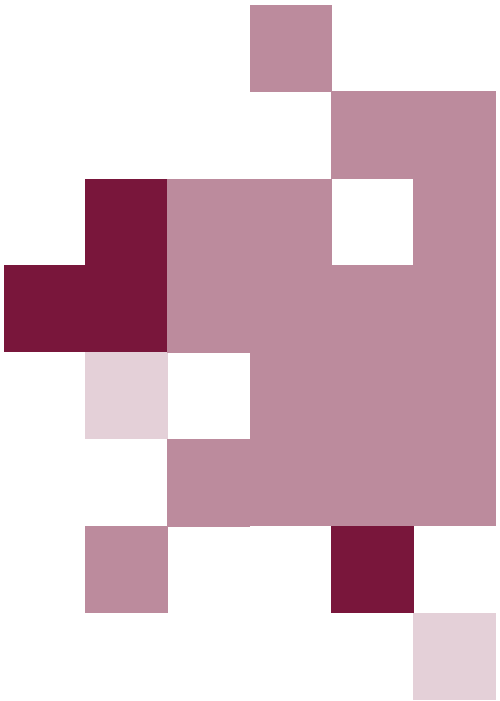
As of April 1, 2003, all infants born in Michigan will be screened for **M**edium-**C**hain **A**cyl-Coenzyme A **D**ehydrogenase deficiency (MCAD) as part of the existing newborn screening program. Below is the list of diseases screened for in the state followed by the year that screening was initiated.

- Phenylketonuria (1965)
- Galactosemia (1984)
- Congenital Hypothyroidism (1977)
- Biotinidase Deficiency (1987)
- Maple Syrup Urine Disease (1987)
- Hemoglobinopathies (1987)
- Congenital Adrenal Hyperplasia (1993)
- MCAD (2003)

MCAD is an autosomal recessive disorder caused by an enzyme deficiency that inhibits the body's ability to use stored fat. Symptoms generally appear between 3-24 months of age and can include vomiting and lethargy. Hypoglycemia may occur during a period of fasting or illness and can lead to coma, encephalopathy, liver failure or death. Children with MCAD are also at risk of speech and developmental delay if they have episodes of significant hypoglycemia and metabolic decompensation.

With early identification and treatment, such episodes can be avoided. Treatment includes a low-fat/high carbohydrate diet and supplemental carnitine. Frequent feedings are needed to avoid fasting. During illnesses, aggressive medical management may be necessary. With proper treatment, outcome is excellent.

For more information visit the Michigan Department of Community Health website (www.michigan.gov/mdch) under the Healthy Children link.



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WAYNE STATE UNIVERSITY
SCHOOL OF MEDICINE

Molecular Medicine and Genetics

For more information about clinical genetics services, medical genetics education, the Genetic Counseling Program, Medical Genetics Residency program or to request educational outreach, please contact Willia Lake at (313)577-6298 or wlake@genetics.wayne.edu.

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