PERSPECTIVES
In Genetic Counseling

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newsletter of the National Society of Genetic Counselors, Inc.

Vol. 9 No. 3 Fall 1987

Election Results Announced

Deborah Eunpu, Past President I and Nominating Committee Chair, has announced the results of the 1987/1988 elections to the Board of Directors. Congratulations to the following members who will assume their respective duties at the Board meeting on October 6 in San Diego.

- *Debra Collins*, President Elect
- *William Herbert*, Treasurer
- *Andrea Gainey*, Region I Rep.
- *Ron Cadle*, Region III Rep.
- *Jane Congleton*, Region V Rep.

Other NSGC members serving on the nominating committee were: Jodi Rucquoi, Helen Travers, Bonnie Baty and Naomi Nakata.

The 1987/1988 Nominating Committee will be chaired by Beth A. Fine.

on the inside

Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency and its Link to Reye Syndrome/SIDS

by Laura E. Hill, M.S., and James E. Haddow, M.D.
Foundation for Blood Research, Scarborough, ME

Introduction

Sudden Infant Death Syndrome (SIDS) and Reye Syndrome are serious disorders of infancy and childhood for which the etiology is unknown. It is likely that these two diagnostic categories actually encompass a heterogeneous group of conditions. Recent studies implicate an inherited disorder of fatty metabolism involving the mitochondrial enzyme, *medium chain acyl—CoA dehydrogenase* (MCAD), as a cause of some SIDS and Reye Syndrome cases. MCAD deficiency is an autosomal recessive condition and predisposes affected individuals to life-threatening episodes of encephalopathy and hypoketotic hypoglycemia. This finding is of significance to those providing genetic services because it makes possible diagnostic testing to high risk families as well as treatment for those individuals identified as having this metabolic disorder.

The Biochemical Problem

Medium chain acyl-CoA dehydrogenase is involved in the mitochondrial beta-oxidation of straight chain fatty acids to acetyl-CoA and ketone bodies, the major energy producing metabolites during periods of prolonged fasting. In MCAD deficiency, this source of energy is compromised and potentially toxic fatty acids and fatty acid metabolites accumulate within the mitochondria during periods of fasting.

The Molecular Basis

The MCAD gene has been mapped to human chromosome 1p31[1] and its mRNA has been sequenced.[2] Preliminary studies indicate that in some patients with MCAD deficiency, the underlying defect is either a small deletion or a point mutation.[2]

Clinical Presentation

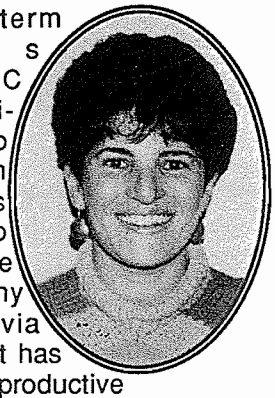
Typically, an infant presents with lethargy, possibly accompanied by persistent vomiting, followed by a period of prolonged fasting. This may be associated with an upper respiratory or other relatively minor viral prodrome. Sudden death may occur, or if the infant survives and the symptoms persist, a diagnosis of Reye Syndrome may be made. Table 1 demonstrates features of MCAD deficiency and Reye Syndrome which are common to both, as well as the characteristics that should help to differentiate the two disorders.

Frequency

At present, the incidence of MCAD deficiency is not known. Bennett et. al.[3] have studied its occurrence retrospectively in SIDS cases and estimate that 5%

continued on p. 6

Corner Thoughts



As my term as NSGC president draws to an end, I am grateful for this opportunity to communicate with all of my colleagues via this column. It has been a busy, productive year—one of growth, excitement and a sense of accomplishment.

In 1986, the Board of Directors voted to form an *ad hoc* committee to investigate the feasibility of establishing a national office staffed with an Executive Director. With our membership exceeding 600, we felt it was time to stop relying on the generous voluntary efforts of our Board members for many administrative activities. I am pleased and proud to welcome Bea Leopold as our Executive Director. Some of you may remember her as the coordinator of local arrangements for our Philadelphia conference.

Bea comes to us with past experience as the Executive Director of the Huntington's Disease Society of America's Delaware Valley chapter, as well as being a consultant to numerous health care organizations. As of this summer, all NSGC correspondence regarding membership, professional information, the job hotline, training programs and NSGC projects have begun to be centralized through Bea's office. Her creativity and efficiency will enhance our Society and our active membership can now direct our collective energies towards new projects. Please work with Bea by contacting your Regional Representatives.

Another exciting development that occurred in the last year is that Ed Kloza has agreed to assume the editorship of *Perspectives*. Joe McInerney has co-edited and edited the newsletter since its inception and has nurtured it over the years. I know you all join me in expressing our gratitude for a job well done. Ed, who has served on the editorial board for about five years, will oversee the overall editing and Bea will manage production and distribution. I look forward to *Perspectives'* continued growth and urge each of you to contribute and work with Ed as a reviewer or solicitor.

I am also pleased to announce that Joe McInerney will continue his active involvement by serving on the finance committee, where his expertise in budgeting and financial planning will benefit all of us.

On a personal note, I would like to thank each of you for your confidence in me and for the opportunity to serve you. I have enjoyed my years of active involvement with the NSGC for several reasons. My professional sense of self has grown along with our Society and our profession. I have learned to work with others despite geographic and philosophic differences and have seen the fruits of our combined labors, including: excellent regional and national conferences, published proceedings of our meetings, *Perspectives* and representation on national committees of various related professional organizations. Professional acquaintances have become colleagues who have been transformed into close personal friends.

In expressing my appreciation to all genetic counselors who contribute to our success by being NSGC members, I also urge you to volunteer for one of our committees, or, if there is a project you would like to establish, contact your Regional Representative. We will all benefit from your efforts, but, on a more selfish note, you will gain far more than the group will.

As I welcome Diane Baker to the Presidency, I wish to be available to all members with questions or concerns. I look forward to seeing old friends and meeting new ones in San Diego.

Beth A. Fine

Beth A. Fine, M.S.
President

EdNotes

The many years that I've spent working with Joe McInerney as he edited *Perspectives* have given me a healthy respect for Joe's patience and energy as well as for the degree of teamwork necessary to produce this newsletter.

As I begin my tenure as editor, I'm pleased that this team will include incumbent editorial board members Joan FitzGerald, Carla Golden and Melonie Krebs. The editorial board welcomes Ann Swinford, who will be covering legislation and funding issues among others under the larger heading of Professional and Personal Advancement.

Joan will be reporting on Technology—new techniques involving diagnosis, analysis or clinical applications. Carla will cover Counseling Approaches and continue to be responsible for case reports. Melonie will continue to bring to the attention of *Perspectives* readers the Resources which our patients or clients might find most helpful.

Another new member of the team is Bea Leopold, who will be responsible for layout, production and distribution of *Perspectives*.

But, as always, the most important team members are you—the *Perspectives* readers. *Perspectives* is the vehicle by which your ideas, projects, insights, questions or criticisms are shared among us.

We hope to introduce several new features in the issues ahead and by doing so, open up new horizons, challenge traditional ways of thinking and perhaps give us (dare I say it?...yes!) new *Perspectives in Genetic Counseling.*

Let us hear from you.

Ed Kloza

Correction

Due to an editing error, Carla Golden was listed as the contributor of Genetic Counseling Case Report No. 6 in *Perspectives* Vol. 9, No. 1. In fact, the case was contributed by Robert Wallerstein and *reviewed* by Carla.

Perspectives regrets this error and any confusion that it may have caused.

Legislative Briefs

Impending State Law has Profound and Far-Reaching Effect

Both houses of the State Legislature of Illinois recently passed House Bill 1415, altering the "Right of Conscience Act." This will allow a physician to refuse to "perform, assist, counsel, suggest, recommend, refer or participate in diagnostic testing to detect fetal abnormalities which may lead to an abortion, regardless of whether such tests are considered routine or normal practice."

This law would relieve physicians of all civil and criminal liability for their refusal to inform a patient about the availability of prenatal testing procedures or their refusal to release the results of testing if such information might lead to a decision to terminate a pregnancy.

At the time of this writing, the Governor of Illinois has not yet taken action on this legislation. Numerous organizations and individuals, including the National Abortion Rights Action League, the American Society of Human Genetics and the Genetics Task Force of Illinois have publicly stated their opposition to this bill.

Many state legislatures at one time or another have considered legislation which attempts to restrict a woman's access to prenatal diagnosis. Considering the far reaching implications of this legislation, it is difficult to understand how HB1415 managed to pass both houses and make its way to the Governor's desk.

Regardless of HB1415's fate, it is clear that the lobbying power of groups resistant to prenatal diagnosis can not be underestimated.

The proposed nomination of Judge Robert Bork further underscores the political climate in which providers of genetic services must operate. For those who are committed to keeping these services available, active participation in the political process may soon become a requirement rather than an option.

Seth Marcus, M.S.
Region IV Representative

Case Report

Case No. 8

Does Group vs. Individual Counseling Influence Amniocentesis Decision-Making?

by Adria Bowin, M.S., Permanente Medical Group, San Jose, CA

Mr. and Mrs. R attended a group amniocentesis lecture for an advanced maternal age of 40. They have two children, ages 15 and 9; the current pregnancy was unplanned.

The design of our group counseling lectures is such that a clerk takes basic family history information over the phone prior to the lecture. If there is no significant family history, couples are generally scheduled for a group counseling lecture to help them decide if they would like to have the procedure. I did not speak with the R's individually since they had a negative family history. During the class, I mentioned that most disorders which can be diagnosed by amnio are not treatable and the only options available after receiving abnormal results are to continue or terminate the pregnancy. I also recommend that, before deciding whether or not to have amnio, couples consider their values and beliefs and how they would utilize the information obtained from the procedure.

A few days after the amnio lecture, the R's scheduled an amnio for 16 weeks gestation. Ultrasound performed prior to amnio revealed that the fetus had anencephaly and I discussed this finding with the Rs. During our discussion, Mrs. R explained that she needed time before she could think about terminating the pregnancy. The R's went home after agreeing to maintain close contact.

Later that same day, Mr. R returned to my office to discuss some additional concerns. At that time, he revealed that he and his wife were against the idea of abortion and had been very involved in the pro-life movement. He added that the diagnosis of anencephaly may present "special circumstances."

During the next few weeks, I talked with the R's several times, in person and by phone, regarding their decision-making. The R's asked to repeat the ultrasound so the anencephaly could be pointed out, but declined amniocentesis. They discussed the diagnosis with clergy, friends and family (including their children) and felt they would be supported regardless of their decision.

After about 5 weeks of what Mr. R described as "agony" they decided to terminate the pregnancy by D&E. Our HMO generally refers patients to an outside clinic for D&E terminations but special arrangements were made for an inhouse termination since the R's did not feel they could handle going to an "abortion clinic." In follow-up discussions after the termination, the R's seemed to be coping well and felt they had made the right decision.

Prior to the amniocentesis, the R's were clearly opposed to pregnancy termination, yet they put themselves in a position of getting information about possible problems in the pregnancy. There certainly are many reasons why a couple opposed to termination may choose to have amniocentesis. However, this case brought up several questions for me: Would the R's have made a different decision regarding prenatal diagnosis if they had been counseled individually? Would they have been "better off" if they had not had prenatal diagnosis? Should more emphasis be placed on the limitations of prenatal diagnosis during counseling sessions? Is amnio for maternal age becoming so common that patients consider it a routine part of prenatal care without considering the implications of the test?

We have a very large patient population and it would be impossible on a practical basis to counsel all amnio patients individually. Should we make an effort to meet individually with all couples attending amnio lectures to discuss their feelings regarding pregnancy termination?

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Related Reading: "An evaluation of three commonly-used counseling formats." R. Young, R. Jorgenson, S. Shapiro, *Perspectives in Genetic Counseling*, Vol. 7, #4, December1985.

Coalition Activities

Counselors Needed for HMHB Coalitions

The National Coalition

The National Healthy Mothers Healthy Babies Coalition is an association of 85 organizations whose purpose is to foster educational efforts for pregnant women through collaborative activities and sharing information and resource. Its goals are to:

- Promote public awareness and education in preventive health habits for all pregnant women and their families.
- Develop networks for sharing information among groups concerned about improving the health of mothers and babies.
- Distribute public education materials on topics related to improving maternal and child health.
- Assist the development of state HMHB coalitions.

Coalition projects are developed by subcommittees addressing several priority areas, among them: policy, adolescent pregnancy, networking and genetics. These subcommittees have prepared a directory of educational materials on infant and maternal care, a national newsletter, market research reports on low income women and breastfeeding, a networking handbook on coalition building and a mass media campaign.

The Genetics Subcommittee

The focus of this subcommittee is: educating ancillary health care personnel (nurses, pediatric educators, etc.) to enable better access for high risk patients to screening, early risk detection and other genetics services; targeting genetics courses in medical, nursing and high schools; and assessing materials used for genetics education and counseling.

State HMHB Coalitions

Each state's HMHB coalition strives to meet the needs of its local population. Unlike national HMHB, some state coalitions coordinate legislative advocacy on maternal and child health issues. The real work of the HMHB Coalition is carried out in these state organizations.

It is important for genetics professionals to join their state coalitions to maximize the attention to genetic issues in prenatal and postnatal care.

For more information, please see the HMHB exhibit at the Annual Education Conference in San Diego.

Jill S. Fonda Allen, M.S.
& Roberta Spiro, M.S.

Letters to the Editor

To the Editor:

One of my initial reactions to the passage of HR1415 in Illinois amending the "Right of Conscience Act" (see Legislative Briefs, p. 3) was how such a bill could get as far as it already has. Perhaps it could have been defeated in legislative committee, or perhaps its introduction onto the floor could have been prevented. We should at least have been aware of its existence long ago. Are we, as individuals and as an organization, aware of what is currently going on in our own state legislatures?...in the federal legislature? Groups such as "pro life" organizations work not only to get bills introduced, but also to lobby tirelessly for their passage.

One of the greatest challenges to our political conscience is now beginning to unfold. As you are aware, Judge Robert Bork's nomination to the Supreme Court can have far-reaching consequences.

Here is the opportunity for us as individuals, and the NSGC as an organization, to assume our political responsibility by becoming aware of legislative activities and current proposals. By taking action on those issues that we feel are relevant, we can make a difference.

Legislation such as HB1415 and the nomination of Judge Bork directly affect us, our jobs and the patients we see. Let's not let our political system happen to us. Let's be an active part of it.

Seth Marcus
Luteran Hospital

Regional Notes

Region II

The Mid Atlantic Regional Human Genetics Network (MARHGN) will sponsor a one-day conference, "Genetics and Deafness," at the Greater Baltimore Medical Center, Baltimore, MD, on January 10, 1988.

Among other topics, Dr. Kathleen Shaver of Gallaudet University Genetic Services Center will address the genetics of deafness, deafness syndromology and a model for the provision of genetic services.

For more information, contact Karen Shirley, MARHGN Coordinator, St. Christopher's Hospital, Genetic Services, 5th & Lehigh Streets, Philadelphia, PA 19133; or call 215-427-5987.

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Region IV

An NSGC Region IV Educational Conference has been scheduled on April 15-16, 1988 at Hotel Sofitel in St. Paul, MN in conjunction with meetings of the Great Plains Clinical Genetics Society, the Midwest Regional Genetic Services Network and the Great Plains Genetics Service Network.

Members interested in helping plan the meeting should contact: Christine K. Barth, M.A., Genetic Diseases Section, Maternal and Child Health, Indiana State Board of Health, 1330 W. Michigan Street, Indianapolis, IN 46206-1964 or Barbara Kunz, M.S., United Hospital, Room 2225, 333 N. Smith Avenue, St. Paul, MN 55454.

Members from outside Region IV are welcome.

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Region VI

An NSGC Region VI Educational Conference has been set for March 23-26, 1988 at the Asilomar conference grounds in Pacific Grove, CA, near Monterey. Presentations on important topics are planned with time to network with other professionals in a casual yet stimulating environment.

Registration materials will be sent to all NSGC Region VI members in late October 1987.

Professionals outside of Region VI are welcome. Please contact Linda Burney, M.A., at 213-533-3759.

Books

How to Have the Healthiest Baby You Can

author: Aubrey Milunsky
publisher: Simon & Schuster, New York. 1987, 352 pp.
price: $17.95
reviewed by: Melonie Krebs, M.S.

In his latest volume, Dr. Milunsky attempts to provide the reader with a comprehensive guide to a healthy pregnancy. It covers everything from preconception planning to delivery complications and breast feeding.

Clearly targeting men and women in the reproductive age group most interested in optimizing their chances of having a healthy child, this book addresses several consumer issues. For example, Chapter 2, "Planning for Pregnancy," includes a practical list of questions such as, "Who will deliver the baby if your doctor is not available?" Prenatal care providers may wish to familiarize themselves with the suggestions contained in this consumer-oriented chapter.

The book describes the many complications which can adversely affect pregnancy. The effects of drugs, infectious agents and other potential environmental teratogens are reviewed. The discussions of the causes of miscarriage as well as genetic and chromosomal abnormalities reflect the current literature.

Each chapter contains a summary of its major points, a successful reinforcement tool. In addition, an appendix lists many organizations available to families with specific reproductive or genetic concerns. It is an excellent resource for any genetic counseling center.

This book is written in a straightforward manner. However, as the title implies, the emphasis is on the avoidance of birth defects. While the author recognizes that couples are generally not responsible for miscarriages and birth defects, relatively little emphasis is made of this point, possibly adding to the burden of their guilt.

Overall, however, this is an excellent resource which should be very useful to any couple wishing to optimize the probability of having a healthy child.

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Planning for a Healthy Baby: A Guide to Genetic and Environmental Risks

author: Richard M. Goodman, M.D.
publisher: Oxford University Press, New York 10016, 269 pp.
price: $16.95
reviewed by: Robert Resta, M.S.

I suspect that more reading gets done during pregnancy than at any other time of life. The increasing sophistication both of obstetrics and of patients has created a voracious appetite among the pregnant public for information. Arcane knowledge possessed only by the medical elite is no longer tolerated.

Dr. Goodman's book is intended for this knowledge-hungry public. The book provides succinct and accurate information on the factors which can produce birth defects. No new information is presented here, but it's a nice compendium. All the major topics are discussed: genes, chromosomes, AFP screening, prenatal diagnosis, maternal illness and drugs. Refreshingly, all medications are not condemned to thalidomide hell. Parts of the book are intended for all pregnant couples, while others are intended for special interest readers, i.e. thyroid disease in pregnancy, thalassemias, G6PD and epilepsy. Genetic counseling plays a big role in planning for a healthy baby and Goodman recommends it throughout the book.

The format of this book is Q&A. I found this format to be tiresome by the 400th question. (Maybe it reminded me of the *Baltimore Catechism.*) The book is certainly not intended to be read from cover to cover, but rather should be used as a reference book to answer many common questions. To this end, the structure works well.

A few quibbles: The definition of genetic counseling is awkward. Recurrence risks are quoted too liberally and may be interpreted incorrectly by the lay reader. Additional references are very technical...How many couples can wade through "Population Structure and Genetic Disorders?". He suggests that the risk of miscarriage after amniocentesis is 1-3 per cent, greater than the risk generally quoted for this occurrence.

These points aside, Goodman's book is certainly a cut above the half-truths found in skimpy chapters on birth defects in many other pregnancy books. You can recommend Dr. Goodman's book to your patients with a fairly clear conscience.

Organizations

The Dystonia Medical Research Foundation (DMRF) offers many services for individuals with neurological conditions known as dystonia, generalized dystonia, spasmodic torticollis, writer's cramp and blepharospasm, as well as for physicians and other care providers.

The DMRF has been in existence since 1976 and is privately funded. A quarterly newsletter is published; literature and a videotape on dystonia are available. Referrals to specialists who evaluate and treat dystonia can be made though the scientific director. The DMRF also funds research on various aspects of dystonia and supports three Dystonia Clinical Research centers.

Chapters are located in Vancouver, Chicago, Denver, Philadelphia, Manchester (MO), Providence (RI), New York City, Montreal, Hermosa Beach (CA), Houston, New Orleans and London.

For information, contact Nancy Harris, Director of Services, Dystonia Medical Research Foundation, 8383 Wilshire Blvd, Beverly Hills, CA 90211; 213-852-1630.

continued from p. 1

of such cases can be explained by MCAD deficiency. The carrier frequency for this condition has been estimated at between 1/30 - 1/50.

Current Approaches to Diagnosis and Treatment

Several methods are available to test for MCAD deficiency in asymptomatic individuals: direct analysis of MCAD activity[4], urinary organic acid analysis following overnight fast[5] or phenylpropionic acid loading[6] and urinary acylcarnitine analysis following L-carnitine load[7]. Carrier detection[4,5] and prenatal diagnosis[8,9] have also been reported for MCAD deficiency. During symptomatic periods, diagnosis is possible via analysis of organic acids[10] or acylcarnitines[7] in urine collected prior to treatment with IV glucose, since this rapidly corrects the metabolic derangement.

Dietary modification to reduce the accumulation of medium chain fatty acids and their metabolites has been recommended as a way to prevent the acute phase of MCAD deficiency[8]. Avoidance of prolonged fasting, a diet low in fats and dietary supplementation with carnitine[7] might be an effective means of avoiding mortality associated with MCAD deficiency.

Discussion

Individuals involved in providing genetic services should be aware of the recently reported association between MCAD deficiency and sudden unexplained sudden infant death and Reye-like syndrome. Although it is possible that neonatal screening for this condition will be available in the future, at present appropriate testing and treatment can only be offered to those individuals recognized as being at high risk as indicated by their family or medical history. Evaluation of SIDS or Reye syndrome should include a thorough review of the family history for the presence of unexplained childhood deaths. Additionally, post mortem findings of fatty infiltration of the liver are strongly suggestive of MCAD deficiency and appropriate genetic counseling and MCAD testing should be considered.

1 Matsubara Y, Kraus JP, Yang-Feng TL, Franke U, Rosenberg LE and Tanaka K. Molecular cloning of cDNAs encoding rat and human medium-chain acyl-CoA dehydrogenase and assignment of the gene to human chromosome 1. 1986. Proc. Natl., Acad. Sci. 83:6543.

2 Kelly DP, Kim J, Billadello JJ, Hainline BE, Chu TW and Strauss AW. Nucleotide sequence of medium-chain acyl-CoA dehydrogenase mRNA and its expression in enzyme-deficient human tissue. 1987. Proc. Natl.Acad.Sci. 84:4068.

3 Bennett MJ, Variend S and Pollitt RJ. Screening siblings for inborn errors of fatty acid metabolism in families with a history of sudden infant death. 1986. Lancet ii: 1470.

4 Coates PM, Hale DE, Stanley CA, Corkey BE and Cortner JA. Genetic deficiency of medium-chain acyl-CoA dehydrogenase: Studies in cultured skin fibroblasts and peripheral mononuclear leukocytes. 1985. Ped. Research. 19:671.

5 Duran M, Hofkamp M, Rhead WJ, Saudubray J and Wadman SK. Sudden child death and "healthy" affected family members with medium-chain acyl-coenzyme A dehydrogenase deficiency. 1986. Pediatrics. 78:1052.

6 Rumsby G, Seakins JWT and Leonard JV. A simple screening test for medium-chain acyl CoA dehydrogenase deficiency. 1986. Lancet ii: 467.

7 Roe CR, Millington DS, Maltby DA, Bohan TP, Kahler SG and Chalmers RA. Diagnostic and therapeutic implication of medium-chain acylcarnitines in the medium-chain acyl-CoA dehydrogenase deficiency. 1985. Ped. Research. 19:459.

8 Bennett MJ, Allison F, Pollitt RJ, Manning NJ, Gray RGF, Green A, Hale DE and Coates PM. Prenatal diagnosis of medium-chain acyl-CoA dehydrogenase in a family with sudden infant death. 1987. Lancet i:440.

9 Bennett MJ, Allison F, Lowther GW, Gray RGF, Johnston DI, Fitzsimmons JS, Manning NJ and Pollitt RJ. Prenatal diagnosis of medium-chain acyl-coenzyme A dehydrogenase deficiency. 1987. Prenat. Diag. 7:135.

10 Duran M, Mitchell G, deKlerk JBC, deJager JP, Hofkamp M, Bruinvis L, Ketting D, Saudubray J and Wadman SK. Octanoic academia and octanylcarnitine excretion with dicarboxylic aciduria due to defective oxidation of medium-chain fatty acid. 1985. J. Pediatrics. 107:397.

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Editor's Note: Further information about testing for MCAD deficiency may be obtained by contacting: Paul M. Coates, Ph.D., Division of Genetics, Children's Hospital of Philadelphia, 215-596-9451; Stephen Goodman, M.D., Dept. of Pediatrics, U. Colorado Health Sciences Center, 303-394-7301; or Charles R. Roe, M.D., Duke University Medical Center, 919-648-2036.

1: Reye Synd. vs. MCAD Deficiency: Shared & Contrasting Characteristics

	RS	MCAD
Shared		
Viral Prodrome	Yes	Yes
Persistent Vomiting	Yes	Yes
Decreased Consciousness	Yes	Yes
Hypoglycemia	Yes	Yes
Elevated Blood Ammonia	Yes	Yes
Dicarboxylic Aciduria	Yes	Yes
Contrasting		
Pernicious Vomiting (First 24 hours)	No	Yes
Rapid Response to IV Glucose	No	Yes
Usual Age of Onset < 2 Years	No	Yes
Previous Clinical Episode(s)	No	Yes
History of Unexplained Sibling Death	No	Yes
Agitation and Disorientation	Yes	No
Abnormal Posturing	Yes	No
Signs of Increased CNS Pressure	Yes	No
Ketosis with Hypoglycemia	Yes	No

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Deadline: Nov. 10

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The University of Connecticut (UConn) School of Medicine, in conjunction with the Connecticut Dept. of Health Services, is seeking a Coordinator for the statewide genetics programs.

This individual will act as liaison among existing genetic programs in the state (UConn and Yale) and the Dept. of Health Services, MCH. Major responsibilities will include: incorporation of genetics knowledge into the State Health Dept. programs, public education, data collection and analysis and overseeing the Tay Sachs, Sickle Cell and Thalassemia screening programs. Input at the universities will be primarily directed at PKU management.

Preferred qualifications include: a Masters Degree in Human Genetics and experience, training or interest in public health. At least 2 years genetics or public health experience is required. Individuals with Masters Degrees in other fields will be considered. Salary range: $25,000 - 33,000, depending on experience. Faculty appointment as Instructor in Pediatrics at UConn School of Medicine is available.

Please send letter of interest and resume to: Suzanne B. Cassidy, M.D., Director, Division of Human Genetics, Dept. of Pediatrics, UConn School of Medicine, Farmington, CT 06032; 203-679-2676.

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The Division of Human Genetics of the University of Medicine and Dentistry of NJ–NJ Medical School is seeking a full-time genetic counselor (preferably Spanish speaking). The position involves a broad range of genetic counseling services in in-hospital and out-reach settings.

This University-based program sees approximately 2000 patient and families per year. Current clinical faculty includes 2 medical geneticists, 4 genetic counselors and 1 project coordinator for the Immigrant Outreach Program. Opportunities exist for clinical research, counseling and case management. Individual should be able to work independently and be comfortable with inner city populations.

The position is available as of July 1. Salary is competitive and based on previous experience. Board Certification/ Eligibility is desired.

Candidates should contact and send resume to: Franklin Desposito, M.D., Division of Human Genetics, F548, New Jersey Medical School, 185 S. Orange Avenue, Newark, NJ 07107-2757; 201-456-4477.

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The Division of Public Health in Central Delaware is seeking a genetic counselor immediately. The position involves prenatal and preconceptual genetic counseling to public and private patients. The counselor will assist clinical geneticist in satellite clinics and will collect medical and family histories prior to appointments and follow-up. The genetic counselor will provide genetic education to parents, community groups and health professionals.

Requirements include a Masters Degree in genetic counseling and Board Certification/Eligibility. No prior experience necessary.

Candidates should contact Barbara Jarrell-Krausz at 302-736-4786.

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Washington, DC Private Practice: Social Worker with genetic training desires genetic associate to join me in private practice. Office space, practice in place.

Contact Gloria W. McNally, Ph.D., 3000 Connecticut Avenue N.W., Suite 439, Washington, DC 20008; 703-536-7434.

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Duke University Medical Center (DUMC) Department of Obstetrics and Gynecology, Division of Perinatology seeks a full-time Board Certified/Eligible genetic counselor to coordinate prenatal genetics clinic. The position involves counseling for amniocentesis and chorionic villus sampling as well as counseling for fetal anomalies discovered on ultrasound. The counselor will coordinate and counsel all patients referred for abnormal alpha-fetoprotein results. This professional will work closely with the cytogeneticist and the pediatrics genetics/metabolism personnel, yet will independently run the prenatal genetics clinic. The position entails considerable involvement in resident and medical student teaching.

The position is open immediately. Salary is negotiable and is commensurate with experience. DUMC provides excellent health and educational benefits.

For more information, please call: Joseph Lanman, Ph.D., *or* Marcy Speer, M.S. DUMC, Box 3390, Durham, NC 27710 *or* Allen P. Killam, M.D., Chief, Division of Perinatal Medicine, DUMC, Box 3122, Durham, NC 27710.

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The University of South Florida Regional Genetics Program in Tampa Bay has a full-time faculty position for a Board Certified/Eligible genetic counselor.

Primary responsibilities include directing patient services—genetics outreach, specialty clinics, prenatal diagnosis counseling and consumer education. A CVS program, molecular genetics laboratory and Teratogen Information Service will be added this fall. Current staff includes 2 clinicians, 4 genetic associates and 1 fellow.

Send CV and 3 letters of recommendation to: Boris G. Kousseff, M.D., Pediatrics/Box 15-G, University of South Florida, 12901 N. 30th Street, Tampa, FL 33612.

The University of South Florida is an Equal Opportunity/Affirmative Action Employer.

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The Indiana Statewide Genetic Services Network has several openings for full-time genetic counselors. Responsibilities include genetic counseling and follow-up support in regional genetic services centers and satellite clinics. Involvement in community education is expected as well as programmatic consultation and evaluation.

The Network is coordinated by the

continued on p. 8

Genetic Diseases Section in the Division of MCH at the Indiana State Board of Health. Genetic evaluation and counseling services are provided at 5 regional centers and 3 satellite clinics. Master's Degree and Board Certification/Eligibility required.

Send resume to: F. John Meaney, Ph.D., Chief, Genetic Diseases Section, Division of Maternal and Child Health, 1330 W. Michigan Street, Indianapolis, IN 46206-1964; 317-633-0805.

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The Lutheran General Perinatal Center (LGPC), located in suburban Chicago, has a position for a full-time genetic counselor. Lutheran General Hospital is affiliated with U. Illinois. The genetics staff of this rapidly-growing perinatal center, includes 3 medical geneticists, 1 fellow and 3 genetic counselors. The position requires Board Certification/Eligibility.

Responsibilities include a wide range of activities: general genetics, prenatal diagnosis, maternal serum AFP screening, specialty clinics, satellite clinics and outreach education.

For more information, please contact: Debra Rita, M.D., c/o LGPC, 1875 Dempster Street, Suite 330, Park Ridge, IL 60068; 312-696-7705.

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Shodair Children's Specialty Hospital (SCSH) in Helena, MT has a full-time position available for Board Certified/Eligible genetic counselor. The job entails general clinical genetics coordination and counseling in an established broadly diverse genetics program with the opportunity for expansion into educational planning and provision.

Send vitae to: John M. Opitz, M.D. or Joan FitzGerald, M.S., Medical Genetics, SCSH, P.O. Box 5539, Helena, MT 59604; 406-442-1980.

SCSH is an Affirmative Action/Equal Opportunity employer.

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Emanuel Hospital & Health Center in Portland, OR, has full-time opening in its genetic counseling program. The Medical Genetics Dept. provides all phases of genetic counseling, including prenatal diagnosis (both chorionic villi sampling and amniocentesis). Applicant should have Masters Degree in genetic counseling and be Board Certified/Eligible. Work experience is preferred. Salary is competitive.

Please send resume to: Wendy Busch, M.S., Medical Genetics, Emanuel Hospital & Health Center, 2801 N. Gantenbein, Portland, OR 97227; 503-280-4726.

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Half-time position in San Luis Obispo and northern Santa Barbara available for individual with Masters Degree in genetic counseling who is Board Certified/Eligible. This position involves both amniocentesis and general genetic counseling. Qualified individuals must have excellent judgment and be able to assume significant responsibility in patient counseling. Travel required.

Please send resume to Lee Neidengard, M.D., Tri-Counties Regional Center, 1428 Phillipos Lane, Suite 203, San Luis Obispo, CA 93401; 805-543-2833.

PERSPECTIVES in Genetic Counseling

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