



METABOLIC OUTREACH SERVICE *December 2007*

The Metabolic Outreach Service is based at Tufts-New England Medical Center's Floating Hospital for Children, Boston, MA

Highlights...

- MOS Launch a Success
- What is MOS?
- MOS Goals
- Current Highlights
- First Year Plan
- Plan for the Future
- Patient-as-Teacher Project
- Thank you to Sponsors

We've begun!

After 3 years of planning, the Metabolic Outreach Service has finally begun! The first programs began in October 2007 with visits to **Baystate Medical Center** in Springfield, MA and **Vermont Children's Hospital** in Burlington, VT. **Boston Medical Center** began their MOS programming the following month in November. **Dartmouth-Hitchcock Medical Center** (Lebanon, NH) and **Eastern Maine Medical Center** (Bangor, ME) are scheduled to begin their programming during the next month or two.

The MOS is seeking a sixth participating center. **Maimonides Medical Center** in Brooklyn, NY is the newest candidate center. Negotiations are underway to determine its eligibility as an MOS site.

What is the MOS?

The Metabolic Outreach Service is an educational and consultative assistance program for academic medical centers that don't have an on-site metabolic service. It represents the largest and most comprehensive education program about metabolic disease ever undertaken in North America.

The MOS was developed as a response to the existing shortage of clinicians certified in the field of clinical biochemical genetics (aka metabolic disease) and the inadequate number of fellows training in this subspecialty. It is clear that non-genetic physicians need to participate more in the diagnosis and management of patients with metabolic disease. The MOS provides physicians and medical students and trainees at all levels with educational opportunities to improve their knowledge base about metabolic disease and learn how to effectively approach a patient's symptoms from a metabolic perspective.

The aim of the MOS is not just to increase awareness about metabolic disease but to change clinical practice, enabling clinicians to "think metabolic."



Mark S. Korson, MD
Division of Genetics and Metabolism
Tufts-New England Medical Center
750 Washington Street, Box 434
Boston, MA 02111

Phone: 617-636-5454
Email: mkorson@tufts-nemc.org

Current MOS Highlights (number of attendees in parentheses)

BAYSTATE MEDICAL CENTER, Springfield, MA:

Oct/2007 –

Resident rounds: “Metabolic approach to metabolic acidosis, hyperammonemia, and hypoglycemia” (27 attendees).
 Ward rounds: Case discussion – Metabolic causes of seizures in infancy (12).
 Office Hours: Review of cases including patient with high ammonia levels; patient with mild methylmalonic acid; management of short chain acyl CoA dehydrogenase (SCAD) deficiency; case of Apo CII deficiency with lipid deposits in the brain.

Nov/2007 –

NICU rounds – “Metabolic approach to metabolic disturbances in the neonate” (12);
 “Newborn screening” (10).
 Office Hours: Review of cases including patient with seizures; a patient with abnormal odor to urine; a patient with hypernatremia and metabolic acidosis.

Dec/2007 (planned)–

Patient Forum: “Pyruvate dehydrogenase complex deficiency (mitochondrial disease).”

BOSTON MEDICAL CENTER, Boston, MA:

Nov/2007 –

Case of the Week – “Tyrosinemia type I” (74).
 Pediatric Morning Report: “Metabolic causes of failure to thrive” (8).
 Neurology rounds – “Metabolic causes of seizures in early childhood,” with focus on glucose transporter defect (23).
 Office Hours: Review of cases including a family with seizures and sudden death in multiple siblings; a patient with intractable seizures; a patient with mental retardation; a patient with coarse-appearing facies.

Dec/2007 (planned)–

Neurology rounds – “A case of high ammonia levels and recurring coma” (N-acetylglutamate synthetase deficiency)

VERMONT CHILDREN’S HOSPITAL, Burlington, VT:

Oct/2007 –

Office Hours: Review of cases including management of mild hyperphenylalaninemia; management of galactosemia.

In addition, at Tufts University School of Medicine, Boston, MA:

Oct/2007 –

Patient Forum, 1st year students – “Pyruvate dehydrogenase complex deficiency” (95).

Nov/2007 –

Patient Forum, 1st year students – “Ornithine transcarbamylase deficiency” (109).



MOS GOALS

- 1) Improve the level of metabolic disease awareness at medical centers that currently do not have metabolically-trained professionals on staff. This will be done through a combination of encounters, including:
 - Workshops
 - Patient forums
 - Participating in ward and divisional rounds
 - Office hours (reviewing cases with clinicians)
- 2) Develop mechanisms for long-distance consultation with regard to the diagnosis and management of metabolic patients, supervised by an established metabolic center;
- 3) Provide educational opportunities in metabolism for medical students and residents and encourage formal training in biochemical genetics.



First Year Plan

The following represents only some of the upcoming workshop topics to be developed in MOS.

Metabolic approach to:

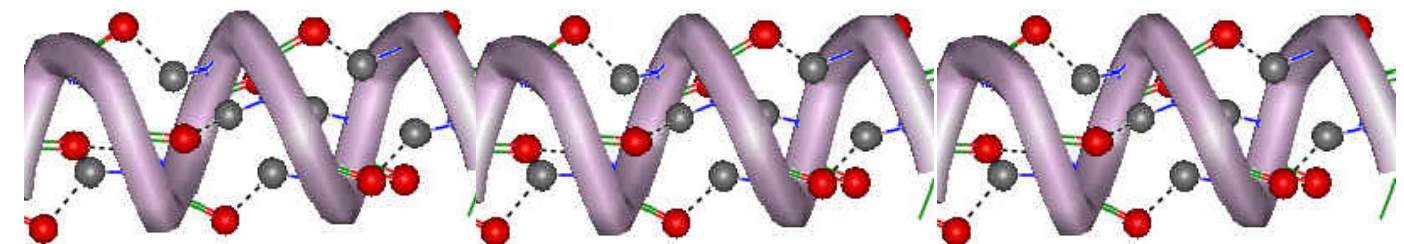
- Liver disease in neonates
- Hyperammonemia
- Liver disease in childhood
- Hypoglycemia
- Neonatal seizures
- Patient with coarse-appearing facies
- Autism and autistic-like features
- Recurrent vomiting
- Abdominal pain
- Patients with pain (non-abdominal)
- Lactic acidemia
- Metabolic disease associated with ENT manifestations
- Metabolic considerations for patients with an elevated CPK level
- “Metabolic clue” – The dietary history
- “Metabolic clue” – The routine urinalysis

PATIENT FORUMS PLANNED to date:

- Mitochondrial disease
- Fatty acid oxidation disorder
- Urea cycle disorder
- Fabry disease
- Hunter syndrome

ADDITIONAL LECTURES:

The management of Gaucher disease: A vision of future therapy options. This lecture will discuss the innovative treatment options for Gaucher disease (already in use or under development) including enzyme replacement, substrate inhibition, and pharmacologic chaperoning, as powerful options for the future treatment of other genetic diseases. Speaker TBA.



Plan for the Future

In an effort to bring the educational tools developed within the MOS program to a wider audience, negotiations are underway to create separate symposia targeting pediatric neurologists and pediatric gastroenterologists. These symposia will offer a selection of case-based workshops of greatest relevance to these particular subspecialty groups. It is anticipated that these symposia will occur during year 4 of the MOS. As these symposia are developed and the logistical details figured out, additional conferences can be designed to target other specialist groups.

Patient-As-Teacher Project

The **Patient-As-Teacher Project** is a separate but parallel effort to the MOS to bring patients and family members into the teaching process. One particularly effective and highly-regarded session within the MOS is the “**Patient Forum**” in which patients or family members (usually parents) discuss the disease presentation, their diagnostic journey, and the challenges they have faced in their encounters with the health care system. Patients and family members can be very effective teachers; their stories are more personal and more compelling than anything a lecturer can convey. These forums are a regular component of the MOS program at each participating center.

In addition, part of the efficacy of the MOS derives from the use of patient cases to highlight important aspects of disease or treatment practice. They are adapted and used for practice exercises in the metabolic workshops. The MOS needs many cases to be able to offer a variety of teaching lessons.

Patients and families are being recruited through a number of avenues including advertising through disease foundation bulletins and web sites.

Thank You to our Sponsors!

The MOS is grateful for the generous support by the following companies and foundations during Year 1 of the program. These sponsors have shown considerable vision to recognize the value and importance of metabolic education:

Genzyme Therapeutics
Shire Human Genetic Therapies
Hyperion Therapeutics, Inc
(formerly Ucyclid Pharma)
Sigma-Tau, Inc
Actelion Pharmaceuticals
The Baby Lorenzo Gregory Scavio Fund

We continue to rely on the generosity of sponsors for the necessary funding to sustain and grow over the upcoming years. For more information about how you or your organization/corporation can support the Metabolic Outreach Service, please contact:

Mark Korson, MD
Division of Genetics and Metabolism
Tufts-New England Medical Center
750 Washington Street, Box 434
Boston, MA 02111

Phone: 617-636-5454
Email: mkorson@tufts-nemc.org

Erin Henry
Office of Development
Tufts-New England Medical Center
750 Washington Street, Box 231
Boston, MA 02111

Phone: 617-636-4685
Email: ehenry@tufts-nemc.org



Tufts-New England Medical Center
Floating Hospital for Children