

Mount Sinai adds ML4 to Carrier Screening Panel

Following is the text of a letter from the Mount Sinai School of Medicine, Department of Human Genetics, dated March 2002:

Dear Referring Physician:

The Mount Sinai Center for Jewish Genetic Diseases is dedicated to providing exceptional care for patients with genetic disorders that are prevalent in the Jewish community and to conducting research focused on the development of improved diagnostic treatment. As part of its preventative medicine approach, the Center conducts a Jewish Genetic Disease screening program, which provides genetic counseling and reproductive screening to individuals at risk for being carriers.

We are pleased to inform you of the recent addition of Mucopolysaccharidosis Type IV (MLIV) to the Ashkenazi Jewish carrier screening panel. MLIV is a severe neurodegenerative condition characterized by a variable degree of growth and psychomotor retardation. Most patients never develop the ability to speak or walk and remain

at the developmental level of a child age 1-2 years. MLIV resembles cerebral palsy with moderate to severe mental retardation. The carrier frequency of MLIV in the Ashkenazi Jewish population is approximately 1/125, and the detection rate is 96%. With the addition of this test, we are currently offering carrier screening for nine conditions more frequently found in this population of patients: Tay-Sachs disease, Canavan disease, Niemann-Pick disease, Gaucher disease, cystic fibrosis, Fanconi anemia, Bloom's syndrome, familial dysautonomia and MLIV.

We are in the process of making new test requisitions to accommodate the addition and as soon as they become available, we will send them to you. In the interim, please request MLIV testing by handwriting "MLIV" on the current test requisition. The cost for the entire panel (9 tests) is \$512.50 per person. We appreciate your patience.

As you know, screening is held on Monday mornings at 9am or Thursday evenings at 5pm, where couples are educat-

ed about the disease, what it means to be a carrier, and their reproductive options. The education session is led by a genetic counselor and conducted in a group setting, where individuals can feel free to ask questions. After counseling, each person can decide to be screened for all the diseases or for selected diseases. Blood is drawn from each individual at this time.

Results from the screening take 2-3 weeks and are sent to the couples' referring physician. All carriers will receive a phone call from a genetic counselor at Mount Sinai. Carrier couples are offered an individual counseling session in order to fully understand the disease and review their pregnancy risks and options.

For more information or to schedule an appointment for Jewish Genetic Screening, call us at (212) 241-6947. We are located on Madison Ave. and 100th Street in the Annenberg Building, room 17-90. Thank you for your time.

See The Presidents Message on page 2 for additional testing sites. ML4



NIH Awards Harvard 5 Year Grant

Dr. Susan Slaughaupt of Harvard Medical Center has been awarded an additional five year grant by the National Institutes of Health, to study ML4. Congratulations to Dr. Slaughaupt and her devoted team of researchers.