New therapy for Fabry’s disease patients tested at Mayo Clinic

A study (see item in Protocols) underway at Mayo Clinic in Rochester will measure the effectiveness of new therapy for patients with Fabry’s disease. Fabry’s disease is an inherited condition known as a lysosomal storage disorder. It is caused by an enzyme deficiency that results in the body’s inability to break down certain naturally occurring glycolipids, primarily GL-3. These glycolipids accumulate in the body, primarily in the lining of blood vessels within the kidney, heart and other internal organs. Passed to children by their parents, Fabry’s disease is an X-linked recessive genetic disorder. While women can pass the trait for Fabry’s disease on to their children, the disease primarily affects males. Symptoms include pain in the hands and feet, angiokeratoma (spotted, dark red skin rash), renal dysfunction, decrease in ability to perspire, stroke and cardiac arrest. Due to severe organ complications, Fabry’s disease is often fatal by age 40.

Some of the female carriers exhibit signs of the condition, especially cloudiness of the cornea. In addition to the eye manifestations, males characteristically have burning sensations in their hands and feet that is worse with exercise and hot weather. Most of the males have small, raised, reddish-purple blemishes on their skin.

Mayo Clinic testing new pediatric refractory cancer agent

Mayo Clinic is among 20 centers in the United States that are conducting Phase I trials of investigational new agents for treating children with refractory cancer. The new anti-cancer agents, which are being evaluated, have a different mechanism of action than many of the traditional chemotherapeutic agents. These include: a radioisotope monoclonal antibody for recurrent lymphoma, for which the initial adult trials were conducted at Mayo Clinic; a proteasome inhibitor which inhibits degradation of most short and long lived proteins, inhibitor of epidermal growth factor; histone deacetylase inhibitor; a modulator of tumor cell apoptosis through an antisense mechanism directed against bcl-2; and a fusion protein of an anti-GD2a antibody (directed against multiple types of cancer, in particular neuroblastoma). Also work is ongoing on the radioisotope samarium in refractory osteosarcoma with bone metastases. Mayo Clinic’s participation in pediatric Phase I clinical trials is through the Pediatric Phase 1 Consortium, which is comprised of a select number of institutions participating in the Children’s Oncology Group, a National Cancer Institute-supported clinical trials cooperative group devoted exclusively to childhood and adolescent cancer research. Although there are nearly 240 member institutions involved with the Children’s Oncology Group, Mayo Clinic is among a small number involved in the Phase I trials.
Detection of cystic fibrosis at birth is key to long-term care

Cystic fibrosis, if detected in childhood, can be treated effectively into adulthood to allow patients a full, more active life.

Each year approximately 3,200 Caucasian babies are born in the United States with cystic fibrosis. The disease is much less common among African-American and Asian-American children. Two-thirds of the infants born with cystic fibrosis will be diagnosed within the first three years of life.

Mayo Clinic’s cystic fibrosis clinic has been in operation for 10 years. All patients enrolled in the clinic (two-thirds are in the pediatric age-range; and one-third adults) receive care from a multi-specialty group of physicians, nurses and support staff. The multi-specialty team includes pulmonologists, infectious disease specialists, gastroenterologists, endocrinologists, medical geneticists, ear, nose and throat specialists, nutritionists, respiratory therapists, social workers and a nurse coordinator.

Mayo Clinic uses the traditional screening test, the sweat chloride test, but also uses genetic testing to confirm the presence of the disease. The genetic testing takes place at Mayo Clinic, so the turnaround time is more rapid than sites where the testing is done elsewhere.

The Mayo Clinic program is smaller than those at other large medical institutions, allowing for more individualized attention to each patient’s needs, including the determination of inpatient or outpatient treatment. Services range from diagnosis to treatment and may include lung transplant.

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Mayo Clinic study advises consideration of untethering procedure of tethered cord patients

Mayo Clinic researchers say strong consideration should be given to an untethering procedure in patients with repaired meningomyelocele at the time of the onset of symptoms of tethered cord.

In a study that appeared in the May 2002 issue of Neurosurgery, Mayo Clinic researchers performed a retrospective review of 45 patients with a history of neonatal meningomyelecele repair, who developed symptoms of tethered cord. Symptoms of tethered cord in this cohort consisted of the development of bladder spasticity or orthopedic foot deformity. None of the patients were treated with cord untethering; instead, they were treated symptomatically.

On follow-up, 40 (88.9 percent) of these patients subsequently required additional orthopedic or urological procedures because of further symptoms of tethered cord. The incidence of progression of tethered cord syndrome is 27.5, 40, and 60 percent at one, two and five years, respectively.

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New Protocol

Here is a Mayo Clinic study recruiting patients.

Mayo Clinic in Rochester is one of 20 medical centers taking part in a major international study of a new treatment for Fabry’s disease, a rare genetic disorder that affects an estimated 1 in 40,000 males worldwide.

The goal of this trial is to determine the safety and efficacy of Recombinant Human alpha-galactosidase A (Fabrazyme™) on the progression of renal disease and significant clinical events in patients with Fabry’s disease. This will be a Phase IV multi-center, multinational, randomized, double-blind, placebo-controlled trial.

In this study, patients will be treated for at least 12 months. To be eligible for this study, patients must have a clinical presentation consistent with Fabry’s disease, be over 16 years of age, and meet certain other eligibility criteria. Patients who have undergone or are currently scheduled for kidney transplantation, are currently on dialysis, or have unconfirmed Fabry’s disease are not eligible. Female patients who are pregnant or lactating are also not eligible.

For more information about the study of a new treatment for Fabry’s disease, patients can call Mayo Nephrology Collaborative Group at 507-266-1047 or visit the Web site at http://www.mayo.edu/research/trials/trials/trial_259.html

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If you have a question about Dialogue, or know of a physician who would like to be added to the mailing list, please call 507-284-9258.