

Annan intressant forskning

Dr. Maria Fuller

" and improved clinical management for the mucopolysaccharidosis"

North Adelaide, SA, Australia

Although the underlying genetic defects have been determined for many MPS, the disease process remains poorly understood. The diverse array of clinical symptoms in MPS suggests that many cellular processes are altered. A major one is likely to be the fat composition and distribution in cells. Fats have been shown to be altered in the MPS and this project proposes to examine the types of fats that are altered and their location in the cell. Once we understand the changes in fats, we will attempt to correct these changes using conventional drugs and fatty acid manipulation. Successful studies performed in cells in this project will pave the way for further studies in animal models to see if the pathology in MPS can be treated with diet and drugs.

Dr. Synthia Mellon

"Neurosteroid treatment of MPS IIIA"

University of California, San Francisco

We have identified a potential treatment for a lysosomal storage disorder that involves a class of biological compounds called neurosteroids. These compounds are synthesized in the brain in a developmentally programmed fashion. Among their many effects, they have effects on development of new neurons, survival of neurons, protection against toxicity to neurons. We showed that treatment of a mouse model of the lysosomal storage disorder Niemann Pick Type C (NP-C) with the neurosteroid allopregnanolone doubles lifespan, delays loss of motor function, and rescues neurons that die in NP-C. We now have preliminary data in MPS IIIA mice that a similar treatment with allopregnanolone will enhance lifespan, delay loss of motor function, increase muscle strength, and reduce aggressive behavior. We now propose to expand these studies to include more mice to assess the effect of allopregnanolone treatment on MPS IIIA 1. longevity 2. locomotor function 3. neuronal survival 4. neuronal storage and 5. begin to assess peripheral markers of disease progression and effective allopregnanolone treatment. Successful completion of these aims should provide preliminary data for submission of a larger grant to the NIH.