

Indications™

Newsletter of The Lysosomal Disease Network™

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*Keys to Understanding
the Lysosomal Disease Network*

What are the Lysosomal Disease Network's Current Research Studies?

By Evelyn S. Redtree, M.S.

The Lysosomal Disease Network (LDN) is in the fifth year of its first five-year NIH research contract (contract number U54NS065768). Research protocols which have been pursued during this funding cycle include several pilot studies and numerous longitudinal studies. Research goals include expansion of knowledge of the genetic causes, structural and functional sequelae, and natural history of rare inherited metabolic diseases. Some also are exploring routes to enhancement of the efficacy and benefit of standard-of-care treatments.

Pilot research studies, all now completed, during this funding cycle have included:

LDN Protocol # 6707: Characterizing the neurobehavioral phenotype(s) in MPS III
Principal Investigator: Michael Potegal, Ph.D.

LDN Protocol # 6708: Pulmonary disease and exercise tolerance in boys with Fabry disease
Principal Investigator: William R. Wilcox, M.D., Ph.D.

LDN Protocol # 6710: Lysosomal storage disease: Health, development, and functional outcome surveillance in preschool children
Principal Investigator: Michael E. Msall, M.D.

LDN Protocol # 6711: Lysosomal Disease Network: Expanded screening for the Fabry trait
Principal Investigators: Raphael Schiffmann, M.D., and Michael Mauer, M.D.

LDN Protocol # 6718: Gene therapy for Tay-Sachs disease (Phase 1: natural history data gather)
Principal Investigator: Chester B. Whitley, Ph.D., M.D.

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Meet Our Patient Advocacy Groups

The LDN considers itself lucky to have dedicated patient advocacy groups as members of the Network. This issue, we present with gratitude . . .



Fabry Support & Information Group

<http://www.fabry.org/FSIG.nsf/Pages2/HomePage>

FSIG was started in 1996 as a "kitchen table" support group formed by two Fabry patients and a supportive family member. Their aim was to share their particular understanding of this disease, combined with experience at gathering information and working with doctors, in order to benefit others. Since then, FSIG has been a continually growing and expanding organization constantly looking for new opportunities to assist in improving patient's lives. FSIG is led by its Executive Director Jack Johnson, one of the original co-founders.

FSIG pursues many avenues to assist those in the Fabry community. Primary among these are spreading infor-

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