

# The Smith Family BReaK Thru Fund

We have established the Smith Family **BReaK** Thru Fund in honor of three of our children, **Braden**, **Riley** and **Keaton**, who have been diagnosed with Niemann-Pick Type C disease (NPC). Braden lost his fight with NPC in October 2006 at the age of 10. Riley and Keaton continue battle the effects of NPC daily.

The Smith Family BReaK Thru Fund is a "donor directed" research fund that has been established within the framework of the Ara Parseghian Medical Research Foundation (APMRF). The Smith Family BReaK Thru Fund is 100% directed by us along with Chandler, our oldest daughter, with input from the NPC Scientific Community to fund research to fight NPC. APMRF is a 501(c)(3) non profit organization named in honor of Ara Parseghian, the well-known Notre Dame football coach, who had three grandchildren pass away from NPC.

NPC disease is a genetic, neurodegenerative disorder which causes progressive deterioration of the nervous system. It usually affects children by interfering with their ability to metabolize cholesterol. Adult onset may also occur. Large amounts of cholesterol accumulate within the liver, spleen, and brain. This metabolic disorder leads to a series of neurological problems that are ultimately fatal...until a treatment is found.

The classic form of the disease accounts for 50 to 60 percent of all cases. In this form, prenatal development and early childhood are usually normal, although jaundice may be present at or shortly after birth. Concern about behavioral problems may develop upon entry into school. The child will progressively develop loss of intellectual function, clumsiness, and difficulty with upward and downward eye movements.

In childhood, spleen or liver enlargement may be noted. Difficulties with speech (such as slurring), problems with swallowing, and in some cases, seizures may occur. Psychiatric disturbances may develop. Other symptoms may include sudden loss of muscle strength, which may vary from head nodding to complete collapse, abnormal posturing of the limbs, and lung complications. Death from complications of the disease usually occurs in the teenage years or early adulthood

Our initial golf outing in September 2008 raised over \$45,000. Those funds allowed us to support our first research project this year. We provided support to Drs. Eric Barker and Val Watts in the Department of Medicinal Chemistry and Molecular Pharmacology at the Purdue University School of Pharmacy and Pharmaceutical Sciences. This pilot project is designed to develop a new process to identify drugs that could potentially treat NPC. A fundamental problem in NPC is an inability of cells to use cholesterol properly. Drs. Barker and Watts are developing a novel method of identifying compounds that restore normal cholesterol metabolism. It is their hope to develop this assay and use it for drug screening.

For additional information, you can email us at [info@breakthrufund.org](mailto:info@breakthrufund.org).

We hope to see all of you on August 15 as we continue the fight against NPC

Trent and Julie Smith

