RARE DISEASE DAY

FOR IMMEDIATE RELEASE

CADASIL Together We Have Hope to Observe Rare Disease Day

CADASIL Together We Have Hope Non-Profit (Round Rock, Texas) will be joining the National Organization for Rare Disorders (NORD) and others around the world in observing World Rare Disease Day on February 28, 2013. On this day, millions of patients and their families will share their stories to focus a spotlight on rare diseases as an important global public health concern.

"There are nearly 30 million Americans—and millions more around the world—affected by rare diseases," said Peter L. Saltonstall, president and CEO of NORD. "Everyone knows someone with a rare disease. But, while many of these diseases are serious and lifelong, most have no treatment and many are not even being studied by researchers. This leaves patients and families without hope for a better future."

A rare disease is one that affects fewer than 200,000 Americans. There are nearly 7,000 such diseases affecting nearly 30 million Americans.

On Rare Disease Day, people with rare diseases around the world promote awareness of the challenges of living with a rare disease. The global theme for 2013 is "Rare Disorders Without Borders."

World Rare Disease Day was launched in Europe four years ago and last year was observed in more than 60 nations. It is always observed on the last day of February. On that day, patients and patient organizations will post stories, videos and blogs online and host events to raise awareness of these diseases, which are often called "orphans".

This year, the observance has special significance in the U.S. because 1983 is also the 30th anniversary of the Orphan Drug Act, which provides incentives to encourage companies to develop treatments for rare diseases, and of NORD, which was established by patient advocates in 1983.

CADASIL Together We Have Hope represents patients and families affected by CADASIL**.** CADASIL (Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy) is a hereditary autosomal dominant disease affecting all the small cerebral arteries. It causes subcortical infarcts and damages the white matter (leukoencephalopathy) and it is due to various mutations of the Notch3 gene situated on chromosome 19. Some of the symptoms can include Migraine like headaches with aura, TIAs, Small Strokes, Strokes, Short Term Memory Loss and Dementia. The overall course of CADASIL is variable. Unfortunately at this time there are no interventions that can effectively prevent the course of CADASIL or its clinical manifestations. Certain signs and symptoms can be treated, as they appear.

CADASIL Together We Have Hope (CTWHH) was established on May 10, 2005 as a non-profit organization. When CTWHH became a nonprofit in 2005, the Board of Directors decided to have no membership dues, no paid staff, and run solely on donations.

CTWHH is devoted to promoting awareness, education, support, and research for CADASIL patients, families, friends, and health care providers. We are dedicated to enhancing the established communication network among families as well as identifying sources of medical care and social services. We foster advocacy and open communication among all stakeholders as we work collegially to find a treatment or cure for CADASIL. We have an international Scientific Advisory Committee which is comprised of experts from Canada, England, France, Italy, Scotland, and the United States. CTWHH is honored to have such distinguished experts serving on the committee. Over the past 7 years we have 1,301 confirmed cases worldwide on the CADSAIL registry.

Presently CTWHH is encouraging global wide participation in raising awareness for Rare Disease Day. Students at a high school health science academy in Austin, Texas have already planned a fundraiser to benefit the National Organization of Rare Disease (NORD) and are celebrating this day with the entire student body and faculty.

In the U.S., the coalition supporting Rare Disease Day includes patient organizations and advocacy groups, media professionals and associations, government agencies, researchers, and companies developing treatments for rare diseases.

Rare Disease Day 2013 activities in the U.S. will include awareness events at several State Houses, a Rare Disease Research Hall of Fame, a Handprints Across America photo gallery, and an event at the National Institutes of Health (NIH) in Bethesda, MD.

"More than half of the people who have rare diseases are children," Saltonstall said. "Challenges faced by patients and their families include delayed diagnosis, few treatment options, and difficulty finding medical experts. Many rare diseases have no approved treatment. Insurance may not cover treatments that aren't approved. Also, treatments for rare diseases tend to be more expensive than those for common diseases."

In 1983, the *Orphan Drug Act* was passed by Congress to create financial incentives for companies to develop treatments for rare diseases. Since then, more than 400 orphan drugs and biologics have been approved by the Food and Drug Administration (FDA). It is estimated that approximately 15 million Americans benefit from these products, but that still leaves millions more with diseases for which there is no approved treatment.

For more information about Rare Disease Day activities in the U.S., go to www.rarediseaseday.us. For information about global activities, go to www.rarediseaseday.org. For information about CADASIL, go to www.cadasilfoundation.org.

* * *

Media contacts:

Billie Yvonne Duncan-Smith, CADASIL TWHH, info@cadasilfoundation.org,
Office: (512)255-0209 or 1-877-519-HOPE, Mobile: (512) 585-2052

Deborah Ewald, CADASIL TWHH, ewaldcoaster@yahoo.com,
Mobile: (512)809-0206

Mary Dunkle, NORD, mdunkle@rarediseases.org, Office: (203) 744-0100,
Mobile: (203) 482-9934

February 2, 2013