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FOR IMMEDIATE RELEASE

Local Nonprofit to Observe Rare Disease Day

CT Rare Disease Community to gather during Capitol event

(Avon, CT, February 22, 2013)-----The Cornelia de Lange Syndrome (CdLS) Foundation 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."

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."

This year, Rare Disease Day activities in Connecticut will include a collaborative event at the Legislative Office Building in Hartford, including patients, parents, researchers and legislators.

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

CdLS occurs in about 1 in 10,000 births. An estimated 20,000 people in the U.S. have CdLS but remain undiagnosed and/or without support services. Individuals often have similar physical characteristics including small size, hands and feet; thin eyebrows that meet in the middle. Some

individuals have limb differences, including missing fingers or arms. Common medical problems include GERD, bowel obstruction, hearing loss and congenital heart defects.

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.

Activities around 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.

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.

"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 more information about the CdLS Foundation or to make a donation, call the CdLS Foundation at 800-753-2357 or visit www.cdlsusa.org.

About the CdLS Foundation

Founded in 1981, the Cornelia de Lange Syndrome Foundation is a national family support organization that exists to ensure early and accurate diagnosis of CdLS, promote research into the causes and manifestations of the syndrome, and help people with a diagnosis of CdLS and their families make informed decisions throughout their lifetime.

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