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Foundation funds two research projects

AVON, CT (June 15, 2011) – As part of its 2011 small grants program, the Cornelia de Lange Syndrome (CdLS) Foundation awarded a total of \$15,000 to researchers studying various aspects of CdLS. The following projects were funded:

- *Immunodeficiency in Cornelia de Lange Syndrome*, Soma Jyonouchi, M.D., Children’s Hospital of Philadelphia. This study explores the idea that immune dysfunction could be a clinical feature of CdLS, which can lead to recurrent upper respiratory and lower respiratory infections. The hope for this study is improved care of patients with CdLS who are at risk for complications from underlying immunodeficiency.
- *Molecular Origins and Mechanisms of Cornelia de Lange Syndrome*, Julie Woodman, pre-doctoral student, University of Colorado, Denver. This research project aims to study the underlying mechanisms involved in targeting cohesin to particular regions of the chromosome. The study anticipates providing insight into the molecular etiology of CdLS, hopefully resulting in potential treatments for the disorder.

The researchers will present their findings at the fifth biennial CdLS Scientific Symposia, slated for June 21, 2012, in Lincolnshire, IL.

About CdLS:

CdLS is a genetic syndrome that 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. Although individuals with CdLS range from mildly to severely affected, most have similar physical characteristics: small size, hands and feet; thin eyebrows that meet in the middle; long eyelashes; upturned nose; and thin, downturned lips. Some individuals have limb differences, including missing fingers or arms, and partial joining of the toes.

Common medical problems include gastroesophageal reflux, bowel obstruction, hearing loss, and congenital heart defects.

About the Foundation:

Founded in 1981, the Cornelia de Lange Syndrome Foundation is a 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 diagnosis of CdLS make informed decisions throughout their lifetime. For more information, call 800 – 753 – 2357 or go to www.cdlsusa.org.

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