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UCI receives second five-year designation as Cornelia de Lange Syndrome Center of Excellence



<http://www.bio.uci.edu/wp-content/uploads/2013/05/Anne-Calof-preferred-image-rev.jpg>Irvine, Calif. The Cornelia de Lange Syndrome (CdLS) Foundation recently designated the University of California, Irvine, as a Cornelia de Lange Syndrome Center of Excellence for a second five-year period. This designation recognizes continued excellence and outstanding achievement in research work related to CdLS by five UC Irvine faculty.

"The research overseen by Anne Calof, Ph.D., Arthur Lander, M.D., Ph.D., Thomas Schilling, Ph.D., Kyoko Yokomori, Ph.D., and Ali Mortazavi, Ph.D., continues to further what we know about CdLS and, in turn, brings new hope to families affected by this syndrome," said Marie Malloy, Executive Director of the CdLS Foundation.

Cornelia de Lange Syndrome is a disabling multisystem genetic disease that affects about one in 10,000 children, although recent research suggests that the incidence may be higher. Individuals with

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Upcoming Events

Experimental Pathology Research Seminar Series: RiP Talks by Graduate Students
Friday, May 31, 2013, 9:00AM [More Info](#)

MB&B Spring 2013 Seminar
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CdLS show a wide range of physical and cognitive problems, some of which can be quite severe and can have a profound impact on quality of life and the families of individuals with the disorder.

Dr. Calof, together with her husband and collaborator Arthur Lander (Donald Bren Professor of Developmental and Cell Biology and Director for the Center of Complex Biological Systems), became involved with the CdLS Foundation and research on the syndrome in 2000, when Calof gave birth to a daughter, Isabel, who was diagnosed with CdLS. They then worked together with researchers at Children's Hospital of Philadelphia to identify the causative gene for Cornelia de Lange Syndrome in 2004 (http://helix.bio.uci.edu/public/press/Arthur_and_Anne.html) . The discovery of the gene, NIPBL, has led to the development of tools for molecular diagnosis of CdLS and has spawned a large body of biomedical research on CdLS and related syndromes.



(http://www.bio.uci.edu/wp-content/uploads/2013/05/lander_p081020_01_daa.jpg)

As part of this effort, Drs. Calof, Lander, and Schilling have developed animal models of CdLS that are being used to find ways to prevent and/or treat this disorder

(<http://www.ncbi.nlm.nih.gov/pubmed/19763162>; <http://www.plosbiology.org/article/info%3Adoi%2F10.1371%2Fjournal.pbio.1001181>).

The effort has expanded in the past few years to include Drs Yokomori and Mortazavi, whose work on genomics is helping to increase the understanding of how mutations in NIPBL affect the expression of other genes in tissues throughout the body. (<http://www.ncbi.nlm.nih.gov/pubmed/21454523>)

To learn more about the CdLS Foundation, please visit <http://www.cdlsusa.org/> (<http://www.cdlsusa.org/>).

Photo at top: Anne Calof in her laboratory at UC Irvine.

Bottom photo: Arthur Lander, courtesy of UC Irvine Strategic Communications

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