



## Cornelia de Lange Syndrome Foundation, Inc.

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### **Researchers identify third gene responsible for Cornelia de Lange Syndrome** *Finding indicates many children going undiagnosed*

AVON, CT (February 5, 2007)—For the third time in three years, a gene responsible for Cornelia de Lange Syndrome (CdLS) has been identified. The most recent discovery was announced in January by researchers at the Children’s Hospital of Philadelphia (CHOP). Their finding will be published in the March edition of the *American Journal of Human Genetics*.

The new gene—known as *SMC3*—seems to cause a somewhat milder form of CdLS. “Many of the children who have this newly identified gene change are so mildly affected that researchers believe other children with this change are being misdiagnosed or not diagnosed at all,” says Julie Mairano, executive director of the national CdLS Foundation, based near Hartford, CT.

Researchers are finding that children with CdLS who are on the mild side have the *SMC3* gene change or the *SMCIA* gene change, which was identified by Italian researchers in 2006. (*NIPBL*, the first gene to be identified (also by CHOP), is found in about half of tested individuals and seems to correlate with a more moderate to severe form of the syndrome.)

“Many of the children we are now finding these new gene changes in would likely be called “isolated developmental delay” or “mental retardation” by many physicians,” says Dr. Ian Krantz, a geneticist who heads up the CdLS research team at the Children’s Hospital of Philadelphia. “These children were only diagnosed with CdLS by very astute clinicians, and the diagnosis was not certain until a test confirmed it.”

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Although children with CdLS range from mildly to severely affected, most have similar physical characteristics: small hands and head, thin eyebrows that meet, long eyelashes, upturned nose, and thin, downturned lips. Children develop physically and mentally much slower than their peers. Some have limb differences, missing limbs or partial joining of the toes. Common medical problems include gastroesophageal reflux, bowel abnormalities, heart defects, seizures, and cleft palate.

“This latest gene finding reinforces our belief that tens of thousands of individuals have CdLS but don’t know it,” says Mairano.

The CdLS 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 a diagnosis of CdLS, and others with similar characteristics, make informed decisions throughout their lifetime. For more information on CdLS, call 800-753-2357 or go to [www.CdLSusa.org](http://www.CdLSusa.org).

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To read the CHOP news release, go to the CHOP media room, <http://www.prnewswire.com/micro/CHOP>.