

# Epidemiology of Cornelia de Lange syndrome in Europe - population-based data

## Research networks

### Private database information: (for Eurordis only)

- code: 45
- date: 20070615163430
- IP: 161.53.106.3

Cornelia de Lange syndrome (CdLS) is a rare genetic syndrome characterized by the specific facial dysmorphism, hypertrichosis, upper limb deficiency, intrauterine growth retardation, developmental delay and a variety of associated malformations. The particular clinical features make severe forms of the syndrome easily recognizable.

We have analysed the population-based data extracted from the database of EUROCAT (European Surveillance of Congenital Anomalies), a large European network of birth defect registries that use the same epidemiological methodologies.

The study is based on the 23-year (1980-2002) monitoring covering 8,558,346 births from 16 European countries. We found the prevalence of the classical form of CdLS to be 1.24/100,000 births and estimated the overall CdLS prevalence at 1.6-2.2/100,000 or 1:80,645 births. There were 91.5% (97/106) live born infants with high first week survival (91.4%). Termination of pregnancy following prenatal diagnosis was performed in 5.7% (6/106) cases, and 2.8% (3/106) were foetal deaths. Prenatal detection rate of abnormalities in CdLS cases was 32.1% in the last eleven years.

The most frequent associated congenital malformations were limb defects (73.1%), congenital heart defects (45.6%), central nervous system malformations (40.2%) and cleft palate (21.7%). Almost 70% of infants, born after the 37th week of gestation, weighed  $\leq$  2500 g. Low birth weight correlates with a more severe phenotype. All patients were sporadic. Maternal and paternal ages do not seem to be risk factors for CdLS and no evidence of exposure to consistent teratogenic agents was noted.

### Authors:

Ingeborg Barisic, *Professor of Paediatrics, Chief of the Referral Centre of the Ministry of Health for Surveillance of Congenital Anomalies, Department of Paediatrics, Children's University Hospital Zagreb, Croatia*, Visnja Tokic, *Junior Researcher*, Maria Loane, Fabrizio Bianchi, Eliza Calzolari, Ester Garne, Diana Wellesley, Helen Dolk