

# Emergency Care Card



# Cornelia de Lange Syndrome

## CdLS Emergency Care Card

### Patient Details

Name.....

Birth Date..... Gender.....

Address.....

Phone.....

### Family Doctor Details

Name.....

Practice/Clinic Name.....

Address.....

Phone.....

Email.....

Fold 2

Last Updated (Year-Month-Day)

### Emergency Contacts

Name.....

Relation to Patient.....

Address (if different).....

Phone.....

Email.....

Name.....

Relation to Patient.....

Address (if different).....

Phone.....

Email.....

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## Cornelia de Lange Syndrome

The Patient this card refers to has

Cornelia de Lange Syndrome (CdLS) is characterised by intellectual disability, typical facial features, upper limb anomalies, growth disturbances, and a large variety of other signs and symptoms. It can be caused by pathogenic variants in one of six genes, the most common one being NIPBL.

## Information for Health Care Professionals about Cornelia de Lange Syndrome (CdLS)

### Main Medical Conditions Associated with CdLS

- Short stature (specific growth charts available)
- Microcephaly
- Long term feeding difficulties
- Developmental delay, intellectual disability
- Speech problems
- Behavioural problems, especially self-injurious behaviour
- Severe recurrent gastro-oesophageal reflux
- Constipation
- Small hands, missing fingers to absent fingers
- Hearing loss
- Ptosis, recurrent blepharitis, myopia
- Cryptorchidism
- Cutis marmorata, hirsutism

### Acute Life-threatening Complications with CdLS

- Bowel obstruction, volvulus
- Aspiration pneumonia (gastro-oesophageal reflux/swallowing difficulties)

- Seizures
- Cardiac problems
- Bladder infections
- Retinal detachment
- Small airways (anaesthesia risk)

### Less Frequent Medical Conditions Associated with CdLS

- Heart malformations (ventricular septal defect, pulmonary stenosis)
- Diaphragmatic hernia
- Intestinal malrotation, duodenal atresia, annular pancreas
- Perthes disease, hip dislocations
- Scoliosis
- Barrett esophagus
- Renal malformations
- Seizures
- Immunological problems
- Dental crowding, caries
- Nystagmus, strabismus
- Cleft palate

More information can be obtained from the Canadian CdLS Foundation:

[www.canadiancdlsfoundation.com](http://www.canadiancdlsfoundation.com)



### Using your CdLS Emergency Care Card

This Emergency Care Card enables the multi-disciplinary team to be aware of the actions of other specialists, to best manage CdLS. In addition to personal and medical details, caregivers should record details about every visit with a specialist (e.g., type of specialist, date of visit, contact information) so other specialists can get in touch. Types of specialists may include: geneticist, surgeon, anaesthetist, gastroenterologist, cardiologist, neurologist, pediatrician, dentist, optician, psychologist, occupational therapist, social worker, speech therapist. List all medications, so doctors can be confident there are no conflicting treatments being recommended. When the card is full, download extra copies at: [www.canadiancdlsfoundation.com/resources/EDcard](http://www.canadiancdlsfoundation.com/resources/EDcard)

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