

CdLS Emergency Care Card



Cornelia de Lange Syndrome

CdLS Emergency Care Card

Patient Details

Name.....

DoB..... Gender.....

Address.....
.....
.....

Phone.....

Family Doctor Details

Name.....

Practice/Clinic Name.....

Address.....
.....
.....

Phone.....

Email.....

Cornelia de Lange Syndrome

The Patient this card refers to has

General information

Cornelia de Lange Syndrome (CdLS) is characterised by intellectual disability, typical facial features, upper limb anomalies, 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.

Last Updated (/ /)

Emergency Contacts

Name.....

Relation to Patient.....

Address (if different).....
.....
.....

Phone.....

Email.....

Name.....

Relation to Patient.....

Address (if different).....
.....
.....

Phone.....

Email.....

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

Main medical problems in 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 in 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 problems in CdLS

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

Further information on CdLS can be obtained from World Federation of CdLS Support Groups

www.cdlsworld.org

Using your CdLS Emergency Care Card

The CdLS Emergency Care Card enables the multi-disciplinary team necessary for best management of CdLS to be aware of the actions of other specialists. As well as filling out personal and medical details, carers should record details every time the patient sees a specialist. By listing the contact details and the date of visit, other specialists can get in touch. By listing medications, your doctors can be confident that there are no conflicting treatments being recommended. Listed specialists may include: geneticist, gastroenterologist, cardiologist, neurologist, dentist, optician, psychologist, occupational therapist, anaesthetist, paediatrician, social worker, surgeon and speech therapist. Carers should make sure they indicate the speciality of the professional. When the card has been filled-up, extra copies can be downloaded from www.cdlsworld.org

