## Emergency Care Card



## Cornelia de Lange Syndrome

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

#### General information

### Cornelia de Lange Syndrome

The Patient this card refers to has

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### **CdLS Emergency Care Card**

### **Patient Details**

Email.

Name	
DoB	Gender
Address	
Phone	
Family Doctor D	<b>Details</b>
Name	
Practice/Clinic Name	
Address	
Phone	

	Last Updated (//)
	Emergency Contacts
	Name
	Relation to Patient
	Address (if different)
Fold 3	Phone
	Name
	Relation to Patient
	Address (if different)
	Phone

# 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 gastrooesophageal 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 (gastrooesophageal 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 <a href="https://www.cdlsworld.org">www.cdlsworld.org</a>

Typical Vital Parameters of Patient	Medical Complications		
Oxygen saturation (%)	☐ Gastroesophageal reflux:		
Heart rate (bpm)	Surgery; Date (/)		
Blood pressure (mmHg)	Feeding problems		
Temperature regulation	Constipation: Occasional		
Height (cm)	Often/Frequent		
Weight (kg) Date (/)	☐ Food intolerance: ☐ Lactose		
☐ NG tube ☐ G-tube type and size	Gluten Other		
☐ Tracheostomy ☐ Mechanical ventilation	Special diet		
☐ Vascular access device.			
Allergies	Seizures: Frequent Rare		
	Type		
	Medication:		
Major Malformations	☐ Hearing loss: ☐ Sensorineural		
☐ Cleft palate	☐ Conductive ☐ Mild ☐ Moderate		
Genital anomalies: Type	Severe Hearing aids		
☐ Intestinal malrotation:			
Surgery; Date (//)	☐ Visual impairment: Type		
Cardiac anomaly: Type	Glasses		
Last evaluation (//)	☐ Ptosis: ☐ Surgery; Date (/		
☐ Surgery; Date (/)			
Upper limb malformation	☐ Small airways		
Psychomotor/Cognitive Development	☐ Pneumonia (recurrent); Dates		
Normal Borderline Disabled			
Degree of delay: Mild Moderate	☐ Ear Infections (frequent)		
Severe Profound	Sinus infections		
Verbal communication	Renal problems: Type		
Absent Strongly limited			
Limited Near normal	Hip problems: Type		
Behavioural problems	☐ Dental anomalies: ☐ Cavities		
☐ Anxiety ☐ Aggression ☐ Self-injurious	☐ Crowding ☐ Allows inspection		
☐ Hyperactivity ☐ Autism spectrum disorder	☐ Crowding ☐ Allows Inspection		
Likes:	Other medical problems:		
Dislikes:	Type.		
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### Medications

Medication	Dosage	Frequency	Reason

## Specialists Treating your CdLS Person

Name	Clinic/Hospital	Phone	Email
Speciality	Date of Visit	Treatment	
Name	Clinic/Hospital	Phone	Email
Speciality	Date of Visit	Treatment	
Name	Clinic/Hospital	Phone	Email
Speciality	Date of Visit	Treatment	
Name	Clinic/Hospital	Phone	Email
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