Care Card **F**Wergency



Syndrome Cornelia de Lange

Deing MIPBL. deues' the most common one barpodenic variants in one of six symptoms. It can be caused by large variety of other signs and drowth disturbances, and a teatures, upper limb anomalies, intellectual disability, typical facial (CdLS) is characterised by Cornella de Lange Syndrome

Cornelia de Lange Syndrome

The Patient this card refers to has

CdLS Emergency Care Card

Patient Details

Name	
Birth Date	Gender
Address.	
Phone.	

Family Doctor Details						
Name						
Practice/Clinic Name						
Address						
Phone						
Fire all						

Last Updated (Year-Month-Day)

Emergency Contacts

	Name.
	Relation to Patient
	Address (if different)
Folc	Phone.
ω	Email.
	Name
	Relation to Patient
	Address (if different)
	·
	Dhono
	Phone
	Email

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 gastrooesophageal 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 (gastrooesophageal 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

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

Typical Vital Parameters of Patient	Medical Complications	Medications			
Outroe Seturation (0/)	Gastroesophageal Reflux:	Туре	Dosage	Frequency Reas	son
Oxygen Saturation (%)	Surgery: Date (Year-Month-Day)				
Heart Rate (bpm)	☐ Feeding Issues				
Blood Pressure (mmHg)	Constipation: Occasional Often/Frequent				
Temperature Regulation	☐ Food Intolerance: ☐ Lactose				
Height (cm) Date (Year-Month-Day)	Gluten Dther				
Weight (kg)Date (Year-Month-Day)	Special Diet				
☐ NG Tube ☐ G-tube: Type and Size					
$\ \ \square$ Tracheostomy $\ \ \square$ Mechanical Ventilation	☐ Seizures: ☐ Frequent ☐ Rare	Considiate Toss	Line Call C Davison	<u> </u>	
☐ Vascular Access Device	Type	Specialists Trea	ting your CdLS Person		
Allergies	Medication:	Name	Clinic/Hospital	Phone	Email
Allergies	☐ Hearing Loss: ☐ Sensorineural	Specialty	Date of Visit	Treatment	
	☐ Conductive ☐ Mild ☐ Moderate	Name	Clinic/Hospital	Phone	Email
Major Malformations	☐ Severe ☐ Hearing aids	Specialty	Date of Visit	Treatment	
Cleft Palate	☐ Visual Impairment: Type	Name	Clinic/Hospital	Phone	Email
	Glasses	Specialty	Date of Visit	Treatment	- "
Genital Anomalies: Type Intestinal Malrotation:	Ptosis: Surgery; Date (Year-Month-Day)	Name	Clinic/Hospital	Phone	Email
Surgery; Date (Year-Month-Day)	_	Specialty	Date of Visit	Treatment	Free:I
Cardiac Anomaly: Type	Small Airways	Name	Clinic/Hospital	Phone	Email
Last Evaluation Date (Year-Month-Day)	☐ Pneumonia (recurrent); Dates	Specialty Name	Date of Visit Clinic/Hospital	Treatment Phone	Email
Surgery; Date (Year-Month-Day)		Specialty	Date of Visit	Treatment	LIIIdii
Upper Limb Malformation	Ear Infections (frequent)	Name	Clinic/Hospital	Phone	Email
	☐ Sinus Infections	Specialty	Date of Visit	Treatment	Eman
Psychomotor/Cognitive Development	Renal Disorder: Type	Name	Clinic/Hospital	Phone	Email
□ Normal □ Borderline □ Disabled		Specialty	Date of Visit	Treatment	
Degree of Delay: Mild Moderate	☐ Hip Problems: Type	Name	Clinic/Hospital	Phone	Email
Severe Profound	☐ Dental Anomalies: ☐ Cavities	Specialty	Date of Visit	Treatment	
Verbal Communication	☐ Crowding ☐ Allows Inspection	Name	Clinic/Hospital	Phone	Email
Absent Strongly limited	Other Medical Conditions:	Specialty	Date of Visit	Treatment	
Limited Near normal	Type	Name	Clinic/Hospital	Phone	Email
Behavioural Issues		Specialty	Date of Visit	Treatment	
☐ Anxiety ☐ Aggression ☐ Self-injurious	Gene Change	Name	Clinic/Hospital	Phone	Email
Hyperactivity Autism Spectrum Disorder	\square Undetermined \square Clinically Diagnosed	Specialty	Date of Visit	Treatment	
Likes:	☐ ANKRD11 ☐ BRD4 ☐ HDAC8 ☐ NIPBL	Name	Clinic/Hospital	Phone	Email
Dislikes:	☐ RAD21 ☐ SMC1A ☐ SMC3	Specialty	Date of Visit	Treatment	