



WaihonaPedia
See and Learn. Know and Share.

COMMUNITY SUPPORT & PLATFORM



GOALS

- ✓ Learn about a rare disease by **Self Studies** -> Discover needs (Questionnaires & Scorecards)
- ✓ Organize online **theme-rooms** to discuss topics
- ✓ **Share** individual Patient (& family) information easily
- ✓ Publish Experiences & Patient Journey's -> Discover needs
- ✓ Ask **Questions** and Receive **Answers** -> Discover needs
- ✓ Publish information
- ✓ **International**

PATIENT AND THEIR COMMUNITIES IN CONTROL!



AMBITIONS

- ✓ Support more communities
- ✓ Support more families
- ✓ Activate experts from Educational, Care and Medical services, Therapists, Researchers, Supporters
- ✓ Make information accessible -> empower families and supporters
- ✓ Become Sustainable

PATIENT AND THEIR COMMUNITIES IN CONTROL!



PATIENT CENTRAL

- ✓ Patient **needs** based
- ✓ **Bi Directional**
- ✓ Disease info -> **Standard where possible, Specific where needed**
- ✓ Immediate benefits -> **See our Alert-Card**



CURRENT STATE, SOME STATS

- ✓ **21** ITHACA diseases
- ✓ **7** active communities (ITHACA)
- ✓ **117** patients Pitt-Hopkins syndrome (very rare), **15 countries**,
two languages
- ✓ **9** patients Marshall Syndrome (ultra rare), **7 countries**
- ✓ **200** patients Cornelia de Lange syndrome (rare), ~ **500 people**, **30 countries**
- ✓ Growth: Rubinstein-Taybi > **25 registrations in 1 week** to participate
in a study on Sleeping Problems
- ✓





WaihonaPedia

See and Learn. Know and Share

What is WaihonaPedia?

WaihonaPedia is a online platform of and for families and friends of people with rare syndromes. The goal is let them be who they are with a smile on their and our faces.

Why WaihonaPedia?

We dream of a beter life and want to be there for them! As family or friend of people with a rare syndrome we face and have faced the challenges we find on our path. With this platform we try to ease the path by listening and learning from all.



FAMILIES AND FRIENDS

from novice to inspirator
from working hard to working with our heart

from alone to together

- contact with other families to exchange experiences
- to be seen and listened to
- Build our personal plans

from fear and uncertainty to secure and powerful
from seeker to sharer

SCHOOLS AND CARE PROVIDERS

develop to the full potential

- Doelen stellen en mijlpalen halen
- Beter inzicht in mogelijkheden
- Passend onderwijs
- Goede indicatie onderbouwing

from unknowing to knowing
be together in our strength

exceed expectations

HOSPITALS AND EXPERTS

from alone to with all expertises
from question to diagnose

- Open insight in the heath condition
- Better measurements during therapy
- Knowledge development & centre's of expertise

from unhealthy to feeling free

Researchers

Sharing is multiplying
from the unknown to the known

- Collect a lot and high quality data during the lifetime of the travel
- Use WaihonaPedia as a review channel collecting direct feedback of the families, schools and other experts
- Grow to become a super guide

from opinion to science

INSURANCES AND PUBLIC SERVICES

- Fundraising
- Horizon 2020
- National plan for rare deceases



TOGETHER ON OUR PATH TO A LIFE MORE BEAUTIFUL



Stories of experience



Question & Answer

Health

Problem, diagnose, treatment, smile

Quality of Life

mourn, celebrate, friendship, living



Development

communication, behavior, learn, achieve



Knowledge Development



Questionnaire & ScoreCards



OUR ALERT CARD -> DIRECT BENEFIT



Rai Koekkoek

Cornelia de Lange syndrome

The Patient this card refers to has

Vereniging Cornelia de Lange
Syndroom

General Information
Cornelia de Lange syndrome (CdLS) is a rare genetic disorder. Many people have never heard of it. It poses various physical, cognitive and medical challenges.

Health Care Information about Cornelia de Lange syndrome (cdls) for Professionals

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 gastroesophageal 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 (gastroesophageal 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
- Intestinal malrotation, duodenal atresia, annular pancreas
- Perthes disease, hip dislocations
- Scoliosis
- Barrett esophagus
- Renal malformations
- Immunological problems
- Dental crowding, caries
- Nystagmus, strabismus
- Cleft palate

More information

Further information on Cornelia de Lange syndrome can be obtained from cdls www.waihonapedia.org (<https://www.waihonapedia.org>)

Using your cdls Emergency Care Card

Using your cdls Emergency Care Card. The cdls Emergency Care Card enables the multi-disciplinary team necessary for best management of cdls. 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 your WaihonaPedia website (<https://www.waihonapedia.org>).

Introduction Rai Koekkoek

Address: Nowhere
Oxygen saturation (%): 95
Heart rate (bpm): 120
E-mail: no@gmail.com
Blood pressure (mmHg): 60

Temperature regulation: 35
Length: 150 (24-07-2020)
Weight: 36 (23-07-2020)

NG tube G-tube type and size:
 inoperative Tracheostomy
 inoperative Mechanical ventilation Vascular access device:

Allergies

Foods Allergy (Sea Food Allergy)

Major Malformations cdls

- Genital anomalies: No, neither of the testicles are descended (they are for example high in the scrotum, in the groin or in the belly); this has been this way from birth. The penis seems smaller than average. It is green.
- Cardiac anomaly: VSD
- Upper limb malformation

Psychomotor/Cognitive Development cdls

Degree of delay: Normal Borderline Disabled
 Mild Moderate Severe Profound

Verbal communication cdls

Near normal Limited Strongly limited Absent

Behavioural problems cdls

Likes
 Dislikes

Medical Complications cdls

Gastroesophageal reflux
 Feeding problems
 Constipation
 Seizures

"big" seizures (tonic-clonic seizures that involve arms and legs)
Fenytoine (Diphantoine) Fenytoine (Diphantoine)
on both sides
My child had/will have a hearing aid
My child had/will have grommets in his/her ears

Hearing loss
 Visual impairment

My child has had limited sight in both eyes from birth
Glasses for to see well both on a distance and close-up

Small airways
 Pneumonia
 Ear infections (frequent)
 Hip problems

The joint dislocation occurs a few times a year
My child was/is treated under general anaesthesia
 Did /does your child allow a dentist to give a treatment?

Other medical problems

Medications for Rai

Type	Medication	Dosage	Frequency	Started
Pain medications	Paracetamol	1 x 50 mg	2x p dag	17-07-2019 - 31-07-2020
Pain medications	aspirine 3	2 tablet	1x daily	23-07-2020

Treatments for Rai

Name	Specialty	Location	Contact
Raoul Hennekam	General practitioner	Amsterdam UMC	gerritjan.koekkoek@gmail.com
02/Apr/2002	Treatment of the airways or earinfection		
Dr. Jan Widdershoven	General practitioner	JBZ, 's-Hertogenbosch	jw@gmail.com
02/Nov/2005	Gastro Endoscopy	(073) 12345678	

CdLS Emergency Care Card

Emergency Care Card

Name: Rai Koekkoek
DoB: 02-11-1999
Gender: Boy/Male

Family Doctor Details

Name: Bas van Son
Practice/Clinic Name: Huisartsenpraktijk Van Son
Address: Groen van Prinstererlaan 142b, 5237 CH 's-Hertogenbosch, NETHERLANDS
Phone: (073) 6421122, NETHERLANDS
Name: Dr NO

Last Updated (12-Nov-2020)

Emergency Contact 1

name: Gerritjan Koekkoek
Relation to Patient: father
Address: Burchtenlaan 97 5235 GH 's-Hertogenbosch, NETHERLANDS
Phone: (073) 6400541
E-mail: gerritjan@cdlsworld.org

