

Cutis Laxa with Arteries and/or Lung Symptoms (CL) Patient Journey (1)



1. Pre-diagnosis

During pregnancy, specially patients with family history with CL.

1. Main symptoms

Start at birth or when the first symptoms appear: early loose/wrinkled skin, lax skin, respiratory and gastrointestinal issues.

Need: Find the right expertise. Dermatologists familiarized with CL in and/or outside a country.

Ideally: Prenatal screening during pregnancy. Training for the General practitioners, Dermatologists, Pediatricians and other healthcare professionals to identify faster the symptoms and to get a quick and a correct diagnosis.

2. Diagnosis

A.s.a.p after birth or when early loose/wrinkled skin appear, through a multidisciplinary consultation (geneticist and dermatologist with CL expertise)

Need: Be informed on the treatment options and on the psychological impact. Psychosocial aspects are underestimated. Psychological support for siblings and patients due to wrinkles/old aged appearance.

Ideally: Correct information on treatment options (plastic surgery). Psychological care for patients, psychological support for parents and siblings and peer support (patient organisations). Patients and/or families' advice/opinion heard/trusted.

3. Diagnosis follow-up

Possible complications at birth or after diagnosis:

Aortic aneurysm.
Aortic stenosis.
Bicuspid aortic valves.
Vascular involvement distal to the aortic root.
Emphysema.
Cardiorespiratory issues.
Gastro-Intestinal issues (inguinal/diaphragm hernia, diverticulae).
Bladder/urinary issues (diverticulae).

Need: Find the expertise. Treatments: symptomatic for emphysema, gastrointestinal issues and/or aortic aneurysm if severe. Fear for aortic aneurysm rupture (death)

Ideally: Map the expertise. Access to international consultation (CPMS?) and access to information in own language. Psychological support for patients, parents and siblings. Peer support (patient organisations).

4. Treatments

Treatment of Cardiorespiratory issues

during lifetime:
cardiac surgery and lung transplant in adulthood

Treatment of gastrointestinal issues during lifetime:
gastrointestinal surgery

Treatment of wrinkles if the burden of others' gaze is too heavy for the child or in adulthood: facelift

Need: Find the expertise, in particular too few experienced plastic surgeons and waiting lists for transplantation. Fear for death due to aortic aneurysm. Information limited, specially for the facelift surgery (costs? duration of the results? burden of surgery on patients and families)

Ideally: : Map the expertise. Access to international consultation (CPMS?) and access to information in own language. Psychological support for parents and siblings. All financial costs of the facelift covered.

Cutis Laxa with Arteries and/or Lung Symptoms (CL) Patient Journey (2)



Need: Share experience, data and advise on family planning. Impact of information given underestimated. Information on the risks during pregnancy.

Ideally: Patients received information if they want to have a child, specially on Pre Natal testing and in vitro fertilization and risks in case of pregnancy (mother and child). Consultation with an expert counselor.

Need: Information on psychological burden is limited. No overview on types of psychological therapies (hypnose, behavioural therapy, therapy, analytical therapy, family therapy, etc.). Impact of information given underestimated. Expert psychotherapists for Cutis Laxa (there are none as of today).

Ideally: Psychological guidance on self-acceptance, self-esteem. Easy access to the different types of psychological therapies. Psychotherapists trained and expert with the disorder.

Need: Psychosocial challenges (school, work, social life). Psychosocial burden/Quality of Life. No overview on types of psychological therapies (hypnose, behavioural therapy, therapy, analytical therapy, family therapy, etc.). Poor knowledge on material/resources for kids, youngsters, school, teachers, siblings, peers, sports/social activities. Moving to adult care treatment 13-16 and 18> adulthood. Skin care (sun). Daily life habits (diet, smoking, sports).

Ideally: Psychological guidance on self-acceptance, self-esteem. Awareness for social surrounding. Psychological support for patients, siblings and parents. Multi-disciplinary consultation. Well organized transfer to adulthood in medical field (dermatology/cardiology/pulmonology). Access to information in own language on skin care.

Need: Guidance/support and special trained councilors in palliative care.

Ideally: Connection with medical experts and with close peer expertise for daily support. Collectively commemorate people who passed away.