

Cutis Laxa with Neuro and/or Skeleton Symptoms Patient Journey (1)

...Diagnosis... ... Treatments...





Pre-diagnosis & main symptoms...

... Diagnosis follow-up...



1. Pre-diagnosis

During pregnancy, intrauterine growth delay, specially patients with family history with CL.

1. Main symptoms

Start at birth: wrinkles and/or lax skin; wide anterior fontanelle; hip dislocation; micro or macrocephaly.

2. Diagnosis

. A.s.a.p after birth or when the first symptoms appear.

3. Diagnosis follow-up

Possible complications at birth or after diagnosis:

Mild to severe mental delay Seizures

Developmental delay
Joint contractures
Ocular issues
Osteoporosis

Late closing of the fontanelles Hearing loss

4. Treaments

Treatment of skeleton issues during lifetime:

Physiotherapy Orthopedic surgery

Care for mental delay during lifetime:

Appropriated care and evaluation of the severity

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

Need: Amniocentesis search for known mutations during pregnancy. Need to find a dermatologist expert CL. **Ideally:** Prenatal screening and appropriate prenatal follow-up during pregnancy.

Training for the General

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.

Need: Multidisciplinary consultation is mandatory (geneticist and dermatologist experts in CL). Information on treatment options and on psychological impact. Psychosocial aspects underestimated.

Ideally: Correct information on treatment options in own language. More/new evidence of medical and psychological impact. Psychological care and global and complete medical care. Psychological support for parents and siblings. Patients and/or families' advice/opinion heard/trusted.

Need: Find the expertise. Symptomatic treatments.

Ideally: Map the expertise. Access to international consultation (CPMS?) and access to information in own language.

Need: Find the expertise, in particular too few experienced plastic surgeons. Information limited, specially for the facelift surgery (costs? duration of the results? burden of surgery on patients and families, risks of the surgery). Too high hope for the results (wrinkles will not totally disappear).

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



Cutis Laxa with Neuro and/or Skeleton Symptoms Patient Journey (2)



5. Family Planning

During early adulthood, gene mutation transmission in dominant or recessive form.

6. Psychological follow-up

During lifetime due to others' gaze burden and to medical follow-up burden.

7. Follow-up care

During lifetime, comorbidities evolution, skeleton evolution and seizures

8. Palliative care and Memorial

Need: 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: No overview on types of psychological therapies (hypnose, behavioural therapy, therapy, analytical therapy, family therapy, etc.). Expert psychotherapists for Cutis Laxa (there are none as of today).

Ideally: Psychological guidance on selfacceptance, 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. 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). Patients' social care. Support for family helpers.

Ideally: 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. Support for family helpers. Respite Centers with global and total care of the patient.

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

Ideally: Connection with medical experts and with close peer expertise for daily support.

Collectively commemorate people who passed away.

