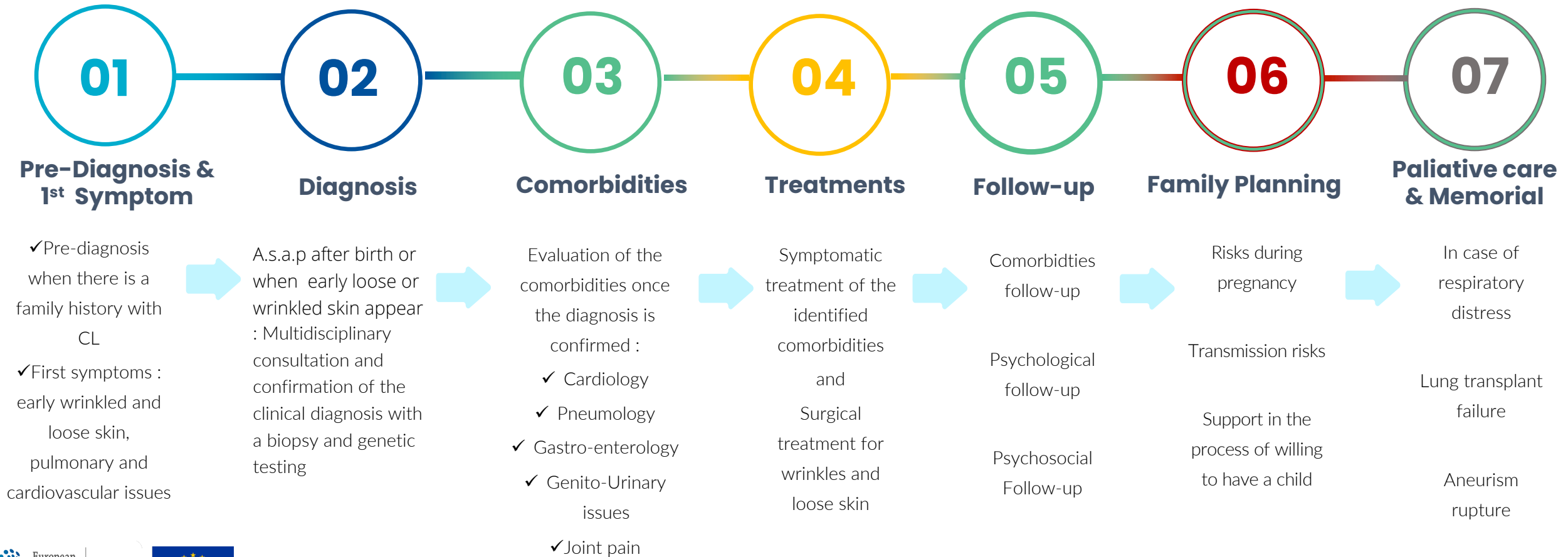




## CONGENITAL CUTIS LAXA with Cardiovascular and pulmonary symptoms

*2025*

# Overview of Patient Journey



# Pre-Diagnosis & First Symptoms

## Clinical Presentation

- Soon at birth, precociously wrinkled, loose, skin associated with respiratory, cardiovascular and gastro-Intestinal symptoms,

## Necessary Actions

- Multidisciplinary consultation (genetics, cardiology, pneumology, dermatology, etc)
- Turn to Cutis Laxa experts

## Patient Needs

- Access to experts in and/or out of the native country
- Information in clear language adapted to the patient
- Taking in consideration the whole family (parents and siblings)
- Psychological support

## Ideal Outcome & Support

- Prenatal testing in case of family history with CL
- Facilitate the clinical diagnosis : Training general practitioners, specialists and other health professionals to identify symptoms
- Peer support (patient organisation).

# Diagnosis

## Clinical Presentation

- Soon at birth, precociously wrinkled, loose, skin associated with respiratory, cardiovascular and gastro-Intestinal symptoms,

## Patient Needs

- Psychological care is organised immediately after announcing the diagnosis
- Being listened to, understood and advice/opinion heard/trusted.
- Getting reliable answers to their questions
- Psychological care for parents and siblings
- Quickly organised medico-social care

## Necessary Actions

- Organising a multidisciplinary consultation
- Clinical diagnosis is confirmed with a biopsy and genetic testing to define the right type of CL,

## Ideal Outcome & Support

- Early identification of the exact type of Cutis Laxa to allow an early evaluation of the possible comorbidities,
- Information and diagnosis given in a clear, understandable and full of consideration language
- All patient's /parents' questions are heard and answered
- Immediate medical, psychological and medico-social care
- Peer support (patient organisation).

# Comorbidities

## Clinical Presentation

- Cardiology (aneurisms, stenosis, bicuspid aortic valva )
- Pulmonary (emphysema)
- Gastrointestinal (Inguinal/diaphragmatic hernias diverticulae, gastric reflux).
- Genito-Urinary (diverticulae)
- Joint pain

## Patient Needs

- Being accompanied and supported during the delivery of the results of internal assessments
- Finding experts
- Being supported in the psychological impact of comorbidities (death anxieties)
- Being properly informed, in one's mother language, about the treatment options for comorbidities
- Being informed about the consequences of comorbidities on quality of life (lifestyle, sports practice, etc.)

## Necessary Actions

- Total internal check-up
- Tpulmonary tests, radiology and/or CT scan
- Cardiovascular MRI
- Scan of the digestive system
- Scanning of the genitourinary system
- Assessment of the level of joint pain

## Ideal Outcome & Support

- Access to national or international expertise
- Receive all the necessary information on comorbidities (consequences, possible treatments, impact on quality of life) in their mother tongue
- Psychological support for the patient and his or her family
- Medico-social support for the impact of comorbidities on quality of life
- Access to adapted sports activities
- Peer support (patient organization)

# Treatment(s)

## Clinical Presentation

- Depending on the identified comorbidities

## Patient Needs

- Being properly referred to the CL experts for each of the associated comorbidities
- Finding Expert Surgeons
- Having full access to transplant waiting lists
- Information on wrinkle treatment options and reservations,
- Consideration of pain relief

## Necessary Actions

- Surgery and/or treatment of identified comorbidities according to the usual procedures for the treatment of these cardiac, respiratory, gastrointestinal symptoms
- Wrinkle treatments (facelift, reconstructive surgery)

## Ideal Outcome & Support

- International map of experts according to identified comorbidities
- Multidisciplinary care throughout life
- Reliable information on options for reconstructive surgery/wrinkle reduction, as well as the consequences and long-term results
- Financial coverage of reconstructive surgery
- Psychological support of the patient and his entourage
- Sharing experience with peers (disease association)
- Supervision/follow-up of comorbidities

# Psychological and long term follow-up

## Clinical Presentation

- Monitoring of the patient's psychological state and evolution over the years,
- Impact of information received (comorbidities)
- Burden of the others' gaze
- Social and professional impact
- Patient's Mental Health

## Patient Needs

- Information on the various therapies available (hypnosis, behavioural therapy, psychotherapy, family therapy, etc.)
- Support in self-esteem
- Coordinated child/adult transition without a break in follow-up
- Improvement of quality of life at all levels (relational, professional, family, intimate, etc.)
- Specific emergency card

## Necessary Actions

- Taking into account psychosocial challenges (school, work, social life)
- Consideration of quality of life
- Skin Health Education
- Environmental impact and "risky" behaviour (tobacco, diet, sports)

## Ideal Outcome & Support

- Long-term support in acceptance and self-image
- Access and coverage of various possible therapies
- Informed social environment
- Support for the whole family
- Access to information in one's own language
- Psychotherapists who are experts in the impact of dermatological diseases on quality of life and their psychosocial burden

# Planning Familial

## Clinical Presentation

- Puberty
- Desire to have a child
- Genetic transmission
- Pregnancy

## Patient Needs

- Be informed from puberty of the impact of the disease on pregnancy (transmission, risks to the mother, possible options)
- Sharing of experience and advice on family planning,
- Information on the procedure to follow in case of desire to have a child (prenatal test, invitro fertilization, preimplantation diagnosis, etc.)
- Psychological support for decision-making

## Necessary Actions

- Answering questions, even anticipating them
- Provide the necessary information
- Patient education/training in genetic transmission
- Information on the risks associated with pregnancy

## Ideal Outcome & Support

- Taking into account and supporting the choices of future parents
- Expert obstetrician-gynecologists
- Patient informed of options and consequences
- Information on the risks associated with the identified comorbidities



## Clinical Presentation

- Respiratory distress
- Lung Transplant Failure
- Aneurysm rupture

## Necessary Actions

- End of life
- Palliative care

## Patient Needs

- Accompaniment
- Support for the patient and those around him
- Specially trained counsellors

## Ideal Outcome & Support

- Connection with specialized services/experts
- Daily support from expert peers
- Collectively commemorating the deceased

# Further Information

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