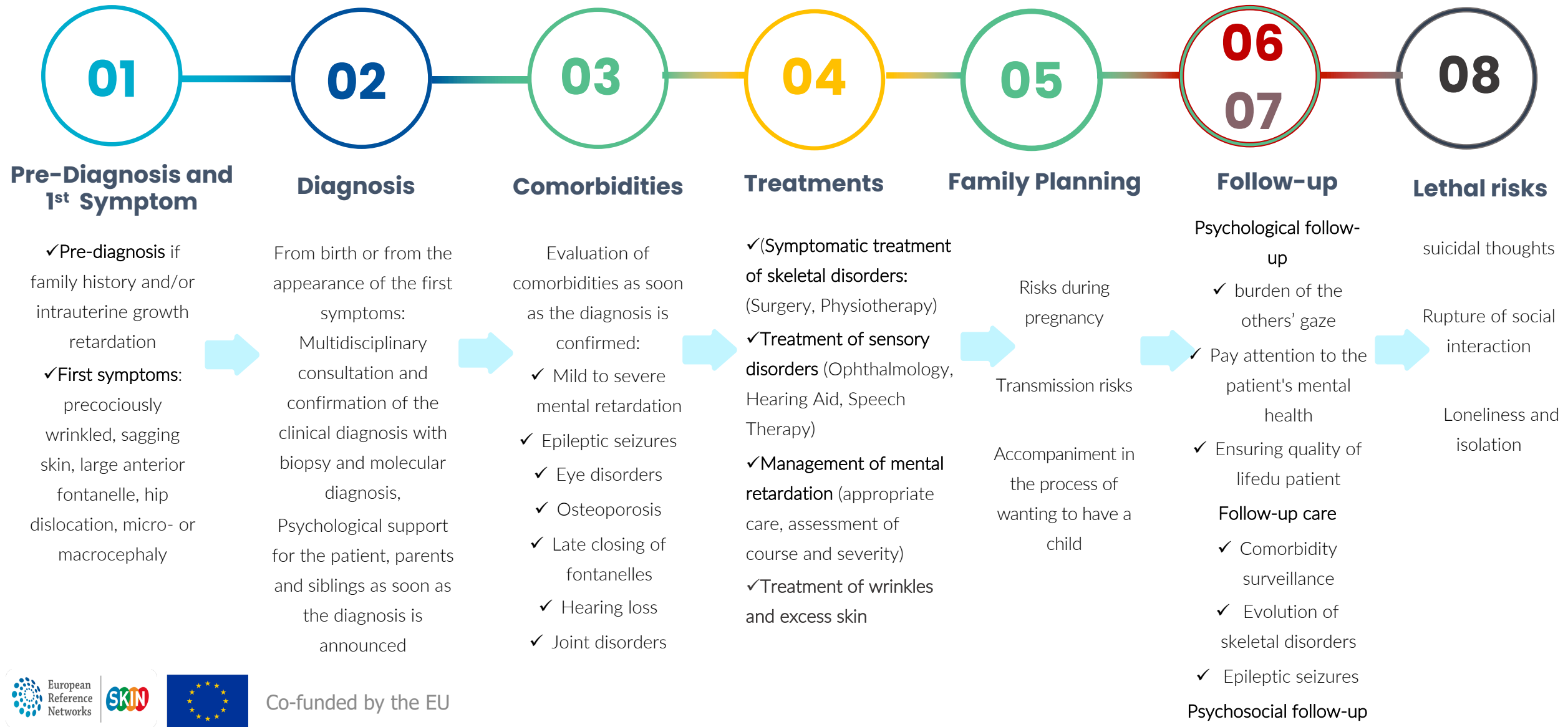




CONGENITAL CUTIS LAXA with Neuro and Skeletal symptoms

2025

Overview of Patient Journey



Pre-Diagnosis and First Symptoms

Clinical Presentation

- From birth, precociously wrinkled, sagging skin, large anterior fontanelle, hip dislocation, micro- or macrocephaly

Actions needed

- multidisciplinary consultation (genetics, cardiology, pulmonology, dermatology, etc.)
- Connecting with Cutis Laxa experts

Patient Needs

- Access to experts in and/or outside the country of birth
- Patient-friendly and plain language information
- Taking into account the whole family (parents and siblings)
- Psychological support

Ideal situation & Support

- Prenatal test in case of family history
- Facilitating clinical diagnosis: training of general practitioners, specialists and other health professionals in the identification of symptoms
- Peer Support (Patient Organisation)
- Quick determination of the exact type by molecular diagnosis

Clinical Presentation

- From birth, precociously wrinkled, sagging skin, large anterior fontanelle, hip dislocation, micro- or macrocephaly

Patient Needs

- Announcement of the diagnosis followed by psychological care
- Be listened to, understood and your opinion taken into account
- Have reliable answers to your questions
- Psychological support for parents and siblings
- Quick medical and social care
- Symptomatic Treatment Options

Actions needed

- Establishment of a multidisciplinary consultation
- Clinical diagnosis confirmed by biopsy and molecular diagnosis to define the precise type of CL

Ideal Situation & Support

- Quick determination of the exact type of Cutis Laxa for rapid assessment of possible comorbidities
- Information and announcement of the diagnosis made in clear, understandable and attentive language,
- All patient/parent questions are heard and answered
- Immediate implementation of care (medical, psychological and medico-social)
- Peer Support (Patient Organisation)

Comorbidities

Clinical Presentation

- Mild to severe mental retardation
- Epileptic seizures
- Stunting
- Eye disorders
- Osteoporosis
- Late closing of fontanelles
- Hearing loss
- Joint disorders

Patient Needs

- Be accompanied and supported during the delivery of the results of internal assessments
- Find the experts
- Be supported in the psychological impact of comorbidities (impact of mental retardation)
- Be properly informed, in one's mother tongue, about the treatment options for comorbidities
- Be informed about the consequences of comorbidities on quality of life (lifestyle, sports practice, etc.)
- Psychological care for the patient, parents and siblings

Actions needed

- Complete internal check-up
- Hearing tests
- Ophthalmological tests
- Neurological Assessment
- Orthopedic Evaluation
- Assessment of the level of joint pain

Ideal situation & 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 and financial support for this support
- Access to adapted sports activities

Treatment(s)

Clinical Presentation

According to the comorbidities identified

Patient Needs

- Be properly referred to the CL experts for each of the associated comorbidities
- Find expert specialists
- Psychological support
- Medico-social support
- Continued follow-up in the transition from child to adult
- Information on options and reservations for the treatment of wrinkles and excess skin

Actions Needed According to the Comorbidities Identified:

- Symptomatic treatment of skeletal disorders (Orthopaedic surgery, Physiotherapy)
- Treatment of sensory disorders (Ophthalmology, Hearing Aid, Speech Therapy)
- Management of mental retardation (appropriate care, assessment of evolution and severity)
- Treatment of wrinkles and excess skin

Ideal situation & support

- International map of experts according to identified comorbidities
- Multidisciplinary care throughout life
- Reliable information on reconstructive surgery options
- Accompaniment and tutoring, adapted to mental retardation
- Medical and social support in adulthood
- Psychological support for the patient and his or her family
- Sharing experience with peers (disease association)
- Surveillance/follow-up of comorbidities

Planning Familial

Clinical Presentation

- Puberty
- Desire to have a child
- Questioning 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,
- Procedure to follow in case of desire to have a child (prenatal test, invitro fertilization, preimplantation diagnosis, etc.)
- Psychological support for decision-making

Actions needed

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

Ideal situation & support

- Consideration and support for future parents' choices
- Expert obstetrician-gynecologists
- Patient informed of options and consequences
- Information on the risks associated with the comorbidities identified

Psychological Follow-up and Follow-up Care

Clinical Presentation

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

Besoins du Patient

- Information on the various therapies available and support in self-esteem
- Coordinated child/adult transition without a break in follow-up
- Improvement of quality of life on all levels (relational, professional, family, intimate, etc.)
- Knowledge of the tools/resources available at each age of life (sports, social activities)
- Specific emergency card

Actions needed

- 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)
- Comorbidity surveillance
- Epileptic seizures
- Psychosocial support

Ideal Situation & 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

Lethal Risks

Clinical Presentation

- Suicidal thoughts
- Rupture of social interaction
- Solitude
- Isolation

Patient Needs

- Appropriate support and care
- Support for the patient and those around him
- Specially trained counselors

Actions needed

- Pay attention to the psychological impact of the disease
- Ensure that the patient is properly cared for

Ideal Situation & Support

- Connection with specialized services/experts
- Regular support from expert peers
- Support from loved ones

Further Information

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