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DELIVRABLE 4: CLINICAL DECISIONS SUPPORT DOCUMENTS

ERN-Skin

Specific Agreement number: 838437 — ERN-Skin — HP-ERN-SGA-2018

EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW PREVALENCE AND COMPLEX DISEASES

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DOCUMENT INFORMATION

DELIVERABLE 4: CLINICAL DECISIONS SUPPORT DOCUMENTS

ERN: ERN-Skin

Call: Specific Agreement number: 838437 – ERN-Skin – HP-ERN-SGA-2018

Type of action: HP-SGA-PJ

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Short Description

Guidelines / recommendations / healthcare pathways for each thematic group of diseases

DISSEMINATION LEVEL

PU	Public	✓
IN	Internal use only	
CO	Confidential, only for members of the consortium (including Commission Services)	

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TABLE OF CONTENTS

<i>Short Description</i>	2
1. GUIDELINES	4
1.1. GUIDELINES ENDORSED BY THE ERN-SKIN	5
1.2. GUIDELINES ELABORATED WITH THE PARTICIPATION OF ERN-SKIN MEMBERS	7
1.3. GUIDELINES ELABORATED BY AN ERN-SKIN MEMBER.....	7
1.4. GUIDELINES AND CLINICAL DECISION TOOLS ELABORATED BY THE ERN-SKIN	7
1.5. GUIDELINES ACTUALIZED WIT THE ERN-SKIN SUPPORT	8
1.6. SURVEYS AND BIBLIOGRAPHIC STUDIES.....	8
2. EMERGENCY CARDS, TUTORIALS	8
2.1. EMERGENCY CARDS	8
2.2. TUTORIALS	9
3. HEALTHCARE PATHWAYS AND PATIENT JOURNEYS	10
3.1. UNDERSTAND THE HEALTHCARE PATHWAYS AND PATIENT JOURNEYS	10
3.2. THE PATIENT JOURNEYS’ DEFINITION AND METHODOLOGY.....	10
3.3. THE ERN-SKIN PATIENT JOURNEYS	10
4. OTHER ACTIVITIES LINKED TO GUIDELINES/CLINICAL DECISION TOOLS	11
4.1. PARTICIPATION TO THE ERN-COORDINATOR GROUP ON KNOWLEDGE GENERATION	11
4.2. RECEIVE SUPPORT FROM THE CPGs AND CDSTs PROGRAMME.....	11
5. DISSEMINATION	12
5.1. OBJECTIVES	12
5.2. TARGET GROUPS	12
5.3. INFORMATION	12
5.4. ACTIVITIES.....	12
6. NEXT STEPS	12
6.1. ADPAT AND MAKE AVAILAIBLE GUIDELINES AND CLINICAL DECISION TOOLS IN EVERY LANGUAGE	13
6.2. PRODUCE ADDITIONAL MATERIALS TO IMPROVE MANAGAMENT AND PATIENT EMPOWERMENT	13
6.3. RECEIVE SUPPORT FROM THE CPGs AND CDSTs PROGRAMME.....	13

INTRODUCTION

OFFER THE PATIENT HIGH LEVEL MANAGEMENT

The aim of the ERN-Skin is to offer patients highly specialised healthcare and support in their daily care while enabling them to remain in their home environment.

The ERN-Skin healthcare providers via the 9 ERN-Skin thematic groups and the Skin ePAG worked together

- to assess, endorse, actualize, adapt or develop guidelines to improve diagnosis and management.
- to develop clinical decision tools to help healthcare providers and patients to face the day to day management and care

The ERN-Skin conducted additional activities not initially planned in the project but part of the ERN coordinator group (ERN coordinator group on Clinical Guidelines and Knowledge Generation) and of the European Commission programme to support the ERN (CPGs and CDSTs Programme).

OBJECTIVES

- To provide high level patient management for every condition covered by the ERN-Sin
- To identify available guidelines and recommendations at the ERN-Skin level, at the ERN level, at the Scientific Societies level, at patient support group level
- To elaborate an ERN-Skin methodology for developing / accrediting these guidelines and recommendations
- To share and spread harmonized best practices thanks to the adaptation and development of guidelines and recommendations for health care professionals

1. GUIDELINES

The ERN-Skin thematic/disease groups, as well as specialists, paramedical staff and patient groups work together according to complementary approaches:

- Guidelines endorsed by the ERN-Skin: each disease group assess the available guidelines at the local and national level. When complying with standard evidence-based procedure, the ERN-Skin board via each disease group endorses these guidelines.
- Guidelines elaborated with the participation an ERN-Skin or several members : An ERN-Skin or several members contribute to the development of a new guideline with other healthcare providers, European task forces or networks such as the European Academy of Dermatology and Venereology Task Force or the European Dermatology Forum
- Guideline developed by an ERN-Skin or several members and then endorsed by the ERN-Skin thematic group: the guideline is first developed at the national level. Then, the ERN-Skin disease group assess the guideline and endorses it.
- Guidelines developed by the ERN-Skin. These guidelines follow the standard evidence-based procedure by involving multidisciplinary group of experts, analyzing literature and taking into account experience of experts, including SKIN ePAG advocates. They are updated every 5 years or before in case of therapeutic innovation.
- Guidelines actualized with the ERN-Skin
- Surveys and bibliography studies to initiate the development or the revision of a guideline

1.1. GUIDELINES ENDORSED BY THE ERN-SKIN

Each disease group assess the available guidelines at the local and national level. When complying with standard evidence-based procedure, the ERN-Skin board via each disease group endorses these guidelines.

- Psychosocial Guidance for social and healthcare professionals/ epidermolysis bullosa (EB) teams
 - Recommended by the ERN-Skin
 - Available in English
- Occupational therapy for epidermolysis bullosa: clinical practice guidelines
 - Recommended by the ERN-Skin
 - Available in English
- Preventative nutritional care guideline
 - Recommended by the ERN-Skin
 - Available in English
- Psychosocial recommendations for the care of children and adults with epidermolysis bullosa and their family: evidence based guidelines
 - Recommended by the ERN-Skin
 - Available in English
- Foot care in epidermolysis bullosa: evidence-based guideline
 - Recommended by the ERN-Skin
 - Available in English
- Skin and wound care in epidermolysis bullosa
 - Recommended by the ERN-Skin
 - Available in English
- Management of cutaneous squamous cell carcinoma in patients with epidermolysis bullosa: best clinical practice guidelines
 - Recommended by the ERN-Skin
 - Available in English
- Management of cutaneous squamous cell carcinoma in patients with epidermolysis bullosa: best clinical practice guidelines
 - Recommended by the ERN-Skin
 - Available in English
- Multicentre consensus recommendations for skin care in inherited epidermolysis bullosa
 - Recommended by the ERN-Skin
 - Available in English
- Oral Health Care for Patients with Epidermolysis Bullosa – Best Clinical Practice Guidelines
 - Recommended by the ERN-Skin
 - Available in English
- Updated S2K guidelines on the management of pemphigus vulgaris and foliaceus initiated by the European academy of dermatology and venereology (EADV)
 - Recommended by the ERN-Skin
 - Available in English
- S2k guidelines (consensus statement) for diagnosis and therapy of dermatitis herpetiformis initiated by the European Academy of Dermatology and Venereology (EADV).
 - Recommended by the ERN-Skin
 - Available in English

- European guidelines (S3) on diagnosis and management of mucous membrane pemphigoid, initiated by the European Academy of Dermatology and Venereology – Part I.
 - Recommended by the ERN-Skin
 - Available in English
- European Guidelines (S3) on diagnosis and management of mucous membrane pemphigoid, initiated by the European Academy of Dermatology and Venereology – Part II.
 - Recommended by the ERN-Skin
 - Available in English
- Diagnosis and management of pemphigus: Recommendations of an international panel of experts
 - Recommended by the ERN-Skin
 - Available in English
- Pemphigus. S2 Guideline for diagnosis and treatment–guided by the European Dermatology Forum (EDF) in cooperation with the European Academy of Dermatology and Venereology (EADV)
 - Recommended by the ERN-Skin
 - Available in English
- Management of bullous pemphigoid: the European Dermatology Forum consensus in collaboration with the European Academy of Dermatology and Venereology
 - Recommended by the ERN-Skin
 - Available in English
- French national diagnosis and treatment protocol for Xeroderma Pigmentosum
 - Recommended by the ERN-Skin
 - Available in French
- European S1 guideline for the treatment of hidradenitis suppurativa/acne inversa
 - Recommended by the ERN-Skin
 - Available in English
- Ehlers-Danlos syndromes: state of the art on clinical practice guidelines
 - Recommended by the ERN-Skin
 - Available in English
- Supportive Care Guidelines for the Management of SJS/TEN in Adults
 - Recommended by the ERN-Skin
 - Available in English
- Management of ocular involvement in the acute phase of Stevens-Johnson syndrome and toxic epidermal necrolysis: French national audit of practices, literature review, and consensus agreement
 - Recommended by the ERN-Skin
 - Available in English
- Epidermal necrolysis French national diagnosis and care protocol (PNDS; protocole national de diagnostic et de soins)
 - Recommended by the ERN-Skin
 - Available in 2 languages: English and French
- British Association of Dermatologists guidelines for the management of Stevens-Johnson syndrome/toxic epidermal necrolysis in children and young people
 - Recommended by the ERN-Skin
 - Available in English
- Severe cutaneous adverse reactions to drugs
 - Recommended by the ERN-Skin

- Available in English
- U.K. guidelines for the management of Stevens-Johnson syndrome/toxic epidermal necrolysis in adults
 - Recommended by the ERN-Skin
 - Available in English

1.2. GUIDELINES ELABORATED WITH THE PARTICIPATION OF ERN-SKIN MEMBERS

An ERN-Skin or several members contribute to the development of a new guideline with other healthcare providers, European task forces or networks such as the European Academy of Dermatology and Venereology Task Force or the European Dermatology Forum

- Clinical practice guidelines for laboratory diagnosis of epidermolysis bullosa
 - Elaborated by the ERN-Skin with other healthcare providers
 - Recommended by the ERN-Skin
 - Available in English
- French Guidelines on Albinism
 - Elaborated by French healthcare providers
 - Recommended by the ERN-Skin
 - Available 2 languages: English and French
- Hidradenitis suppurativa/acne inversa: a practical framework for treatment optimization – systematic review and recommendations from the HS ALLIANCE working group
 - Elaborated with the participation of ERN-Skin members
 - Recommended by the ERN-Skin
 - Available in English
- Management of congenital ichthyoses: European guidelines of care, part one and part two
 - Elaborated with the participation of ERN-Skin members
 - Recommended by the ERN-Skin
 - Available in English
- First German guidelines on the treatment of Adamantiades-Behçet’s disease
 - Elaborated by the Deutsches Register Morbus Adamantiades-Behçet’s e.V.
 - Available in German

1.3. GUIDELINES ELABORATED BY AN ERN-SKIN MEMBER

The ERN-Skin members develop a guideline and the ERN-Skin thematic group assess the guideline and endorses it.

- Management of children with Epidermolysis Bullosa: Therapeutic education for parents and carers
 - Elaborated by the Ospedale Pediatrico Bambino Gesù and endorsed by the ERN-Skin
 - Available in 2 languages: English, Italian

1.4. GUIDELINES AND CLINICAL DECISION TOOLS ELABORATED BY THE ERN-SKIN

These guidelines follow the standard evidence-based procedure by involving multidisciplinary group of experts, analyzing literature and taking into account experience of experts, including SKIN ePAG advocates. They are updated every 5 years or before in case of therapeutic innovation.

- Emergency management in epidermolysis bullosa: consensus clinical recommendations from the European reference network for rare skin diseases

- Elaborated by the ERN-Skin
- Available in English
- Multidisciplinary consensus recommendations from a European network for the diagnosis and practical management of patients with incontinentia pigmenti
 - Elaborated by the ERN-Skin
 - Available in English
- Mosaic abnormalities of the skin – review and guidelines from the European Reference Network for rare skin diseases (ERN-Skin).
 - Elaborated by the ERN-Skin
 - Available in English
- Supportive care in the acute phase of Stevens–Johnson syndrome and toxic epidermal necrolysis: an international, multidisciplinary Delphi-based consensus
 - Elaborated by the ERN-Skin
 - Available in English
- Epidermal necrolysis – Stevens-Johnson syndrome and Toxic Epidermal Necrolysis (Lyell syndrome) – Form for Emergency Department Use
 - Elaborated by the ERN-Skin
 - Available in English

1.5. GUIDELINES ACTUALIZED WITH THE ERN-SKIN SUPPORT

- Partial support of the actualization of the European guideline for Hidradenitis Suppurativa Treatment
 - Actualized with the ERN-Skin and the European Hidradenitis Suppurativa Foundation
 - Available in English

1.6. SURVEYS AND BIBLIOGRAPHIC STUDIES

- To initiate the development or the revision of a guideline, thematic groups have engaged in
 - Delphi Survey for SJS/TEN guidelines
 - Trichothiodystrophy bibliography
 - Ectodermal Dysplasia / P63 bibliography
 - Validation of severity score for bullous pemphigoid study

2. EMERGENCY CARDS, TUTORIALS

2.1. EMERGENCY CARDS

DEFINITION

Emergency cards have all the essential information on the management of the patient with a rare skin disease. They are produced by the ERN-Skin experts, along with SKIN ePAG advocates. Rare skin diseases are unknown to most medical staff. In an emergency situation, it is vital to provide adequate care to avoid any complications. These emergency cards are the size of a business card and are written in the national language so that patients can carry them at all times.

ERN-SKIN EMERGENCY CARDS



The ERN-Skin has developed emergency cards for

- ectodermal dysplasias in 3 languages: English, French, German
- epidermolysis bullosa in 6 languages, with the participation of 11 healthcare providers
- severe cutaneous drug reactions in 2 languages: English and French
- ichthyosis and palmoplantar keratoderma in English and French
- mendelian connective tissue disorders in English

They are available on the ERN-Skin website.

2.2. TUTORIALS

DEFINITION

ERN-Skin tutorials are educational clips on rare conditions. These tutorials are developed by ERN-Skin partners for a specific condition and a specific care. Once produced by the ERN-Skin partner they are validated by the ERN-Skin thematic groups responsible for the specific condition

ERN-SKIN TUTORIALS

The ERN-Skin has developed educational tutorials on

- Learning about photoprotection for patients with albinism and xeroderma
 - Produced by FIMARAD, the French national network for rare dermatological diseases
 - Validated by the ERN-Skin Cutaneous Diseases Related to DNA Repair Disorders Thematic Group
 - Available in 2 languages: English, French.
- Hand mobility exercises for children suffering from epidermolysis bullosa in 2 languages: English, French
 - Produced by FIMARAD, the French national network for rare dermatological diseases
 - Validated by the ERN-Skin Inherited Epidermolysis Bullosa and skin fragility syndromes - Darier, Hailey-Hailey Thematic Group
 - Available in 2 languages: English, French
- Training on specific hand care for patients with epidermolysis bullosa in 2 languages: English, French
 - Produced by FIMARAD, the French national network for rare dermatological diseases
 - Validated by the ERN-Skin Inherited Epidermolysis Bullosa and skin fragility syndromes - Darier, Hailey-Hailey Thematic Group
 - Available in 2 languages: English, French
- Epidermolysis Bullosa and breastfeeding
 - Produced by the Ospedale pediatrico Bambino Gesù, Rome, Italy
 - Validated by the ERN-Skin Inherited Epidermolysis Bullosa and skin fragility syndromes - Darier, Hailey-Hailey Thematic Group
 - Available in 2 languages: English, Italian
- Epidermolysis Bullosa and oral hygiene
 - Produced by the Ospedale pediatrico Bambino Gesù, Rome, Italy

- Validated by the ERN-Skin Inherited Epidermolysis Bullosa and skin fragility syndromes - Darier, Hailey-Hailey Thematic Group
- Available in 2 languages: English, Italian
- Red Baby Movie

They are available on the ERN-Skin website.

3. HEALTHCARE PATHWAYS AND PATIENT JOURNEYS

3.1. UNDERSTAND THE HEALTHCARE PATHWAYS AND PATIENT JOURNEYS

The healthcare pathway is the patient's journey through the healthcare system. It refers to all the stages a patient experiences in the management of his or her disease, from pre-diagnosis to palliative care.

3.2. THE PATIENT JOURNEYS' DEFINITION AND METHODOLOGY

DEFINITION

Patient Journeys are visual testimonies that reflect the natural history/needs of patients and their families for a specific rare disease. They represent the collective perspective on the burden of the disease and the needs of people with first-hand experience of living with a rare skin disease.

It aims to :

- Engage the wider patient community to consolidate common needs for a specific rare disease
- Engage in a discussion with the clinicians to highlight the different (unmet) needs of a specific rare disease, both medical and psychological, so the clinicians can respond to these needs
- Keep in mind both the professional and patient are experts

METHODOLOGY

The patient journey methodology is

- Mapping the journey and identifying the needs and recommendations on ideal care for each specific skin disorder across the different stages from the first symptoms at birth to the end of life care
- Reviewing by both ePAG advocates and clinical experts (clinical presentation validated by some clinicians)
- Organising the information in easy reading graphics (table and illustration)
- Identifying the needs that are common for all 'skin disorders' (to be develop in the next steps)

3.3. THE ERN-SKIN PATIENT JOURNEYS

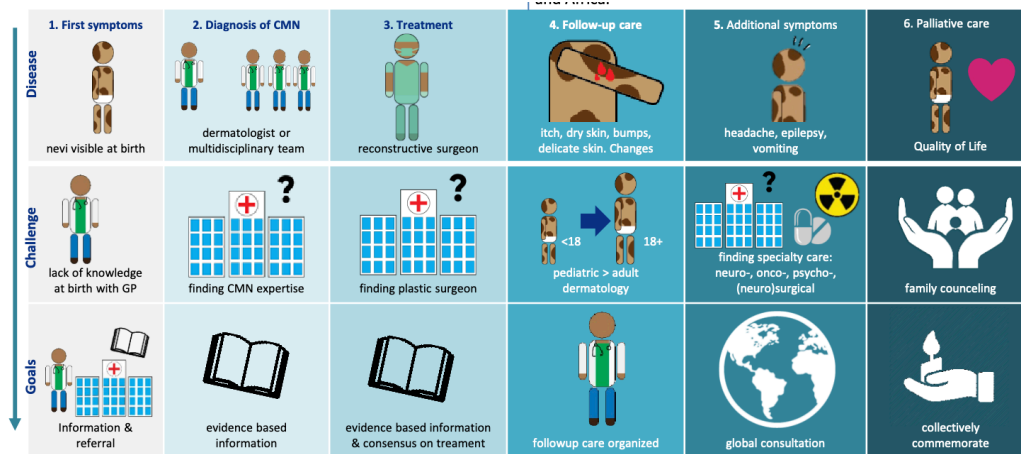
THE KEY ROLE OF THE SKIN ePAG

European Patient Advocacy Groups (ePAGs) bring together rare disease patient advocates who are actively involved in the work of the European Reference Networks. Each ePAG has a number of 'ePAG advocates', who represent their disease area in the ERN and the interests of the wider patient community.

The Skin ePAG is the ePAG advocate group of the ERN-Skin. Each member is a representative of a patient organisation. Within the ERN-Skin, ePag advocates are present in each thematic and transversal group and play a key role in governance.

The Skin ePAG have developed the patient journeys with the support of some thematic groups.

THE ERN-SKIN PATIENT JOURNEYS



The Skin ePAG have developed the patient journeys with the support of some thematic groups:

- Congenital Melanocytic Naevus Syndrome
- Cutis Laxa with Arteries and Lung Symptoms
- Cutis Laxa with Neuro and Skeleton Symptoms
- Cutis Marmorata Teleangiectatica Congenita
- Ectodermal Dysplasia
- Netherton Syndrome
- Pemphigus Vulgaris

They are available on the ERN-Skin website.

4. OTHER ACTIVITIES LINKED TO GUIDELINES/CLINICAL DECISION TOOLS

4.1. PARTICIPATION TO THE ERN-COORDINATOR GROUP ON KNOWLEDGE GENERATION

The ERN-Skin is member of the ERN coordinator group on Clinical Guidelines and Knowledge Generation.

4.2. RECEIVE SUPPORT FROM THE CPGs AND CDSTs PROGRAMME

In the framework of the ERN Clinical Practice Guidelines (CPGs) and Clinical Decision Support Tools (CDSTs) Program, the ERN-Skin has conducted a survey among the 9 ERN-Skin thematic groups to prioritize the diseases for which healthcare providers and patient need the most CPG and CDST development or revision.

The ERN-Skin thematic group identified

- 7 CPG to develop:
 - Palmo-plantar keratoderma
 - Syndromic Hidradenitis
 - P63 related ectodermal dysplasias
 - Cutis laxa
 - Pseudoxanthoma elasticum
 - Ehlers-Danlos syndrome
 - Trichothiodystrophy
 - Poikiloderma
 - Bullous pemphigoid
- 3 CDST to revise:
 - Inherited ichthyosis
 - Syndromic Hidradenitis
 - Bullous pemphigoid

5. DISSEMINATION

Make available the guidelines and clinical decision tools to every healthcare provider, patient representatives and patients

5.1. OBJECTIVES

The dissemination objectives were

- to ensure high level patient management of every rare skin disease patient (guidelines, emergency cards, tutorials)
- to ensure patient are properly referred by Healthcare professionals (patient journey)
- to increase awareness on ERNs and rare skin diseases

5.2. TARGET GROUPS

The target groups were:

- Healthcare professionals
- Academic Communities and Scientific Societies
- Public Health Authorities
- Other ERNs
- Other European and international networks
- European Commission
- Patient representatives
- Patients

5.3. INFORMATION

The following information was shared:

- List of available guidelines and link to the publications
- Guidelines
- Emergency cards
- Tutorials
- Patient journeys

5.4. ACTIVITIES

The ERN-Skin coordination shared information via:

- Scientific publication in peer review journals
- Website
- Newsletter
- Posters
- Presentation in congress and conferences
- E-Mailings
- Printing and distribution of emergency cards to each ERN-Skin member dealing with the diseases

6. NEXT STEPS

6.1. ADAPT AND MAKE AVAILABLE GUIDELINES AND CLINICAL DECISION TOOLS IN EVERY LANGUAGE

The next step is to adapt to the national environment and translate in the national language the available guidelines and clinical decision tools. ERN-Skin healthcare providers and Skin ePAG have started to translate these documents.

6.2. PRODUCE ADDITIONAL MATERIALS TO IMPROVE MANAGEMENT AND PATIENT EMPOWERMENT

The next step is to develop guidelines for each condition covered by the ERN-Skin. To have a real impact, the ERN-Skin has to

- Hire additional staff: a methodologist and medical writer
- Engage national, European and international scientific societies and other ERNs
- Organize expert meetings

6.3. RECEIVE SUPPORT FROM THE CPGs AND CDSTs PROGRAMME

The ERN-Skin has been selected to receive technical support and guidance from the ERN Clinical Practice Guidelines (CPGs) and Clinical Decision Support Tools (CDSTs) Programme for the development of an ERN-Skin guideline on p63.

ABBREVIATIONS

- CDST: Clinical Decision Support Tool
- CPG: Clinical Practice Guideline
- ERN-Skin: European Reference Network for Rare, Low Prevalence and Undiagnosed Skin Disorders



European
Reference
Networks

<https://ec.europa.eu/health>



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