Specific Operationnal Criteria - Icthyosis and Palmoplantar Keratoderma

The requested information will be used to define the specific criteria for our project proposal for a European Reference Network (ERN) for Rare and Undiagnosed Skin Disorders. Please note that, each health care provider member of our ERN will have to fulfil these criteria. These criteria have to be realistic/reasonable while ensuring a high level patient management. These criteria have to be based on the evidence and consensus of the scientific, technical and professional community.

Icthyosis and Palmoplantar Keratoderma

Rare Diseases(s)	Short description of the rare disease	Code/ ICD/ Orphacode	Epidemiology	Incidence	Prevalence
1. Hereditary Ichthyosis	Inherited ichthyoses are monogenic disorders of cornification due to mutations in genes involved in skin barrier function. Skin changes involve most of the tegument, and are characterized by scales of various forms and severities, and these are often associated with erythema. The classification should be clinically based, and distinguishes syndromic from nonsyndromic ichthyoses.	79354	Rare disease, no sexual or racial predominance, start at birth or in infancy		Approximately 13.3 per million people in Europ for moderate to severe form
2. Hereditary Palmoplantar keratoderma	Palmoplantar keratodermas comprise a diverse group of hereditary disorders marked by excessive thickening of the epidermis of palms and soles. They are genetically heterozygous.	307141	Rare disease, no sexual or racial predominance, start at birth or in infancy		No studies were performed on prevalence

Rare Diseases(s)	Specific challenges associated with the recognition of the condition	Specific challenges associated with the diagnosis	Specific challenges associated with the treatment	Specific challenges associated with care of these patients over their lifespan - Quality of life issues -

1. Hereditary Ichthyosis	The recognition of the condition is usually straighforward	The diagnosis of the form of ichthyosis is usually very difficult	There is no curative treatment. The treatment is symptomatic (local or systemic) and usually not enough effective and burdensome	A transition from childhood through to adulthood must be organized. Adolescence is a difficult period	Quality of life severely affected as demonstrated by several publications
2. Hereditary Palmoplantar keratoderma	The recognition of the condition is usually straighforward	The diagnosis of the form of palmoplantar keratoderma is usually very difficult	There is no curative treatment. The treatment is symptomatic (local or systemic) and usually not enough effective and burdensome	A transition from childhood through to adulthood must be organized. Adolescence is a difficult period	Quality of life severely affected but no publications

Rare Diseases(s)	**Key Diagnostic Tests**	**Key Treatment, Resources or Procedures**
1. Hereditary	Dermatopathology, electron microsopy,	Emollients, keratolytics, acitretin
2. Hereditary Palmoplantar keratoderma	Dermatopathology, electron microsopy, immunohistochemistry and molecular analysis (usually not available)	Emollients, keratolytics, acitretin

Please state the minimum/optimum thresholds that Healthcare Providers within the network will need to meet to maintain competence and					
Rare Diseases(s)	**Minimum Number of patients treated per year at each HCP**			**Minimum Number of new patients**	
	Adults	Paediatric*	Rationale for the threshold	Adults	Paediatric*
1. Hereditary Ichthyosis	5	5	Ichthyosis are rare diseases, therefore no large numbers of patients can be expected in a single center	2	2
2. Hereditary Palmoplantar keratoderma	4	4	Palmo plantar keratodermas are rare diseases, therefore no large numbers of patients can be expected in a single center	2	2

Please list the necessary human resources and the profesional qualifications essential to the quality of patient care within the Network's				
Rare Diseases(s)	**Health Care Professional (type)**	**Training & Qualifications**	**Minimun of number of procedures per patient per year**	**Rationale**

1. Hereditary Ichthyosis	Expert Dermatologist, psychologist, specialized nurse, ophtalmologist, pediatrician, ENT specialist, clinical Geneticist	Clinical training in rare skin disorders	10	Ichthyosis are very rare diseases, therefore no large numbers of patients can be expected in a single center
2. Hereditary Palmoplantar keratoderma	Expert Dermatologist, psychologist, specialized nurse, clinical Geneticist	Clinical training in rare skin disorders	8	Palmoplantar keratoderma are very rare diseases, therefore no large numbers of patients can be expected in a single center

Please list the specialised equipment, infrastructure, and information technology required to support the rare or complex disease(s), condition(s) or highly specialised intervention(s) and describe the importance of each

Rare Diseases(s)	Specialised equipment, infrastructure, and information technology	Threshold	Rationale
1. Hereditary Ichthyosis	- Service / Unit Dermatology with dedicated locals and specialized nurses - Service / Pediatric Unit. - Service / neonatal unit, with experience in the care of patients with ichthyosis. - Service / Unit nutrition and dietetics. - Service / ophthalmology unit. - Service / otolaryngology unit. - Psychologist - Pathology and skin research lab (for collecting skin or hair samples, perform keratinocyte culture, immunohistochemistry, RNA extraction etc....) - Service / Unit social workers.	- Service / Unit Dermatology with dedicated locals and specialized nurses	
2. Hereditary Palmoplantar keratoderma	- Service / Unit Dermatology with dedicated locals and specialized nurses, psychologist, social workers; - Pathology and skin research lab (for collecting skin or hair perform keratinocyte, immunohistochemistry, RNA extraction etc....)	- Service / Unit Dermatology with dedicated locals and	

Please provide a summary explaining the approach or plans your group will undertake to produce good practice guidelines and implement outcome

1- Subgroup "clinical management": Our group has the following plan Organization of European guidelines for the therapy of ichthyosis.
These guidelines are currently under process under the coordination of the 2 coordinators of the ERN project (J Mazereeuw-Hautier J and A Hernàndez-Martin). European experts scattered in Europe have been identified and will participate to these guidelines. The topics were distributed into 5 subgroups. The guidelines will take place in Toulouse in April 2016. These guidelines will then be published and diffused among European colleagues. The next step will be to implement outcome measures and quality control. We plan to organize a survey in order to see if the guidelines are respected. We also plan to update the guidelines every 5 years
2- Subgroup "research": enclosed document