Specific Operationnal Criteria - Epidermolysis Bullosa High Level Patient Management Group
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. NB: A sample of healthcare providers will be selected for on-site audits to validate the information.
Epidermolysis Bullosa High Level Patient Management Group

Rare Diseases(s)	Short description of the rare disease	Code/ ICD/ Orphacode	Epidemiology	Incidence	Prevalence
1.Epidermolysis bullosa	Epidermolysis Bullosa (EB) is a rare group of inherited disorders that manifest as blistering or erosion of the skin and, in some cases, the epithelial lining of other organs, in response to little or no apparent trauma.		The exact prevalence of EB is unknown. Mild variants have been estimated to occur as frequently as 1 per 20,000 births. The more severe varieties are believed to occur in 1 per 500,000 births annually.	The estimated incidence of all forms of EB is 1 per 20,000 life births.	The estimated prevalence of all forms of EB in the EU is 50,000 to 80,000 cases.

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 - Gaps accross the care continuum	
1.Epidermolysis bullosa	Mild cases may have nail only or lichen-planus-like presentation. Late-onset blisters are often attributed to other diseases. Differentiation of different types of EB can be difficult on clinical grounds alone in the neonatal period	Diagnosis is increased in complexity like the mantles of an onion: clinical - EM blister level - protein deficiency - DNA (RNA)	Multiple organs are affected, and therefore multiple disciplines are involved. The challenge is to integrate the management of all disciplines. In severe EB, patients may be faced with numerous treatments or interventions on a daily basis which can affect compliance.	The care needs continuity from childhood to adulthood; services need to provide appropriate transitional care between paediatric and adult settings	Pain - itch - misery - immobility - nutritional compromise - cancer.

Rare Diseases(s)	Key Diagnostic Tests	Key Treatment, Resources or Procedures
1.Epidermolysis bullosa	IF antigen mapping. EM of skin biopsy. DNA Sanger or Next Generation Sequencing. RT-PCR of RNA. Cell culture.	Multidisciplinary team. Wound care. Pain management. Hand surgery. Gastrostomy. Oesophageal dilatation. Oncology.

Please state the minimum/optimum thresholds that Healthcare Providers within the network will need to meet to maintain competence and expertise. List

Rare Diseases(s)	Minimum Number of patients treated per year at each HCP			Minimum Number of new patients diagnosed per year at each HCP	
	Adults	Paediatric*	Rationale for the threshold	Adults	Paediatric*
1.Epidermolysis bullosa	10	10	Minimal exposure to clinical presentation.	3	4

Please list the necessary human resources and the profesional qualifications essential to the quality of patient care within the

Rare Diseases(s)	Health Care Professional (type)	Training & Qualifications	Minimun of number of procedures per patient per year	Rationale
1.Epidermolysis bullosa	Dermatologist	2-3 years experience	10 patients	minimal experience
	Paediatrician	2-3 years experience	10 patients	
	1. or 2. above OR physician experienced in EB MDT care	5 years experience		
	Plastic surgeon	2-3 years experience	1 hand surgery	
	General or paediatric surgeon	2-3 years experience	1 gastostomy	
	Occupational therapist	2-3 years experience	5 splints	
	EB nurse	2-3 years experience	10 patients	
	Ophthalmologist	2-3 years experience	5 patients	
	Physiotherapist	2-3 years experience	5 patients	
	Psychologist	2-3 years experience	3 patients	
	Dentist	2-3 years experience	2 patients	
	Pain physician	2-3 years experience	3 patients	
	Anaesthetist	2-3 years experience	3 patients	
	Dietitian	2-3 years experience	10 patients	
	Disgestive Surgeon or Interventional radiologist	2-3 years experience	3 oesophageal dilatations	
	Pathologist	2-3 years experience	3 patients	
	Neonatalogist (not for centers where only adults are seen)	2-3 years experience	3 patients	

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.Epidermolysis bullosa	Out-patient clinic and in-patient beds	4 clinics per year and as required admissions	minimal experience

Please provide a summary explaining the approach or plans your group will undertake to produce good practice guidelines and implement outcome
Best clinical practice guidelines for different elements of EB have already been initiated via DEBRA International. Published guidelines exist for wound management, dental care in children, pain management and cancer management. Others e.g. nutrition, are in development.