Specific Operationnal Criteria

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.

Group Cutaneous Mosaicism (Nevi and Nevoid Overgrowth Disorders)	The number of cutaneous disorders of mosaicism is obviously much higher than the few examples that have been considered in this table. However, these have been selected for being more representative of the disorders in the group. It should be noted that a complete list of all mosaic skin disorders is impossible to include in this table.	**The true incidences of these diseases are not known and some of the diagnoses overlap- these are approxiate estimates and therefore also equated with approximate prevalences**

Rare Diseases(s)	Short description of the rare disease	Code/ ICD/ Orphacode	Epidemiology	Incidence	Prevalence
Congenital melanocytic naevus syndrome and other melanocytic naevus syndromes	Neurocutaneous Multisystem Mosaic Disorder	OMIM #137550	Sporadic, genetic, all races	1 in 10-20,000	1 in 10-20,000
Schimmelpenning Syndrome	Neurocutaneous Multisystem Mosaic Disorder	OMIM #163200	Sporadic, genetic, all races	1 in 50,000	1 in 50,000
Phakomatosis Pigmentokeratotica	Neurocutaneous Multisystem Mosaic Disorder	ORPHA 2874	Sporadic, genetic, all races	1 in 50,000	1 in 50,000
Inflammatory Linear Verrucous Epidermal Naevus	Cutaneous Mosaic Disorder	ORPHA 79466	Sporadic, genetic, all races	1 in 50,000	1 in 50,000
Other epidermal naevi	Cutaneous Mosaic Disorder	OMIM #162900	Sporadic, genetic, all races	1 in 20,000	1 in 20,000
Hypomelanosis of Ito and other hypopigmentary mosaic disorders	Neurocutaneous Multisystem Mosaic Disorder	OMIM #300337	Sporadic, genetic, all races	1 in 50,000	1 in 50,000
Naevoid and whorled hypermelanosis and other hyperpigmentary mosaic disorders	Neurocutaneous Multisystem Mosaic Disorder	OMIM %61423	Sporadic, genetic, all races	1 in 50,000	1 in 50,000

Extensive Dermal Melanocytosis	Neurocutaneous Multisystem Mosaic Disorder		Sporadic, genetic, all races	1 in 20,000	1 in 20,000
Proteus Syndrome	Multisystem Mosaic Disorder	OMIM #176920	Sporadic, genetic, all races	1 in 100,000	1 in 100,000
PIK3CA related overgrowth syndromes (incl. CLOVES syndrome)	Multisystem Mosaic Disorder	OMIM #602501 and 612918	Sporadic, genetic, all races	1 in 10,000	1 in 10,000
Klippel-Trenaunay-Weber syndrome	Vascular Mosaic Disorder	OMIM %149000	Sporadic, genetic, all races	1 in 20,000	1 in 20,000
Megalencephaly-capillary malformation polymicrogyria syndrome	Multisystem Mosaic Disorder	OMIM #602501	Sporadic, genetic, all races	1 in 50,000	1 in 50,000
Capillary malformation congenital	Vascular Mosaic Disorder	OMIM #163000, #600998	Sporadic, genetic, all races	1 in 2000	1 in 2000
Venous malformations	Vascular Mosaic Disorder	OMIM #600195, 112200, ORPHA 211252	Sporadic, genetic, all races	1 in 2000	1 in 2000
PHACE association	Vascular Mosaic Disorder	OMIM #606519	Sporadic, genetic, all races	1 in 100,000	1 in 100,000
Sturge Weber syndrome	Vascular Mosaic Disorder	OMIM #185300	Sporadic, genetic, all races	1 in 50,000	1 in 50,000
Phakomatosis Pigmentovascularis	Neurocutaneous Multisystem Mosaic Disorder	ORPHA 79483	Sporadic, genetic, all races	1 in 50,000	1 in 50,000
Lymphatic malformations and lymphangiomatosis	Multisystem Mosaic Disorder	OMIM #185300	Sporadic, genetic, all races	1 in 10,000	1 in 10,000
Cutaneous arteriovenous malformations	Vascular Mosaic Disorder	ORPHA 211266, ORPHA 156230	Sporadic, genetic, all races	1 in 50,000	1 in 50,000
Capillary malformation-arterio venous malformation syndrome	Vascular Mosaic Disorder	OMIM #608354	Sporadic, genetic, all races	1 in 50,000	1 in 50,000
Maffucci syndrome	Vascular Mosaic Disorder	OMIM % 614569	Sporadic, genetic, all races	1 in 50,000	1 in 50,000
Undiagnosed mosaic paediatric dermatology conditions			Sporadic, genetic, all races		
Glomuvenous malformation, segmental	Vascular Mosaic Disorder	OMIM # 138000	Sporadic, genetic, all races	1 in 50,000	1 in 50,000
Cutis marmorata telangiectatica congenita	Vascular Mosaic Disorder	OMIM 219250	Sporadic, genetic, all races	1 in 20,000	1 in 20,000

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
Congenital melanocytic naevus syndrome and other melanocytic naevus syndromes	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Schimmelpenning Syndrome	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Phakomatosis Pigmentokeratotica	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Inflammatory Linear Verrucous Epidermal Naevus	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Other epidermal naevi	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available

Hypomelanosis of Ito and other hypopigmentary mosaic disorders	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Naevoid and whorled hypermelanosis and other hyperpigmentary mosaic disorders	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Extensive Dermal Melanocytosis	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin biopsy and sequencing	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Proteus Syndrome	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin biopsy and sequencing	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
PIK3CA related overgrowth syndromes	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin biopsy and sequencing	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available

Klippel-Trenaunay-Weber syndrome	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin biopsy and sequencing	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Megalencephaly-capillary malformation polymicrogyria syndrome	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin biopsy and sequencing	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Capillary malformation congenital	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin biopsy and sequencing	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Venous malformations	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin biopsy and sequencing	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available

PHACE association	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Sturge Weber syndrome	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Phakomatosis Pigmentovascularis	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Lymphatic malformations and lymphangiomatosis	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Cutaneous arteriovenous malformations	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Capillary malformation-arteriovenous malformation syndrome	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available

Maffucci syndrome	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Undiagnosed mosaic paediatric dermatology conditions	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Glomuvenous malformation, segmental	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available
Cutis marmorata telangiectatica congenita	Diagnosable only by experts as rare	Need full assessment of the child, skin and other systems, genetic diagnosis only from skin	Untreatable currently, need multidisciplinary management of complications	Need an expert to coordinate the different strands of care, and to watch for serious complications, often poor transition to adult care as not available

Rare Diseases(s)	**Key Diagnostic Tests**	**Key Treatment, Resources or Procedures**
Congenital melanocytic naevus syndrome and other melanocytic naevus syndromes	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, ophthalmology, neurology, radiology, plastic surgery, genetics
Schimmelpenning Syndrome	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, ophthalmology, neurology, radiology, plastic surgery, genetics
Phakomatosis Pigmentokeratotica	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, ophthalmology, neurology, radiology, plastic surgery, genetics

Inflammatory Linear Verrucous Epidermal Naevus	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, ophthalmology, neurology, radiology, plastic surgery, laser serivces
Other epidermal naevi	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, Laser services, Genetics if appropriate, ophthalmology, neurology, photography, radiology
Hypomelanosis of Ito and other hypopigmentary mosaic disorders	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, ophthalmology, neurology, radiology, plastic surgery, genetics
Naevoid and whorled hypermelanosis and other hyperpigmentary mosaic disorders	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, ophthalmology, neurology, radiology, plastic surgery, genetics
Extensive Dermal Melanocytosis	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, ophthalmology, neurology, radiology, plastic surgery
Proteus Syndrome	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, Laser services, Genetics if appropriate, ophthalmology, neurology, photography, radiology, plastic surgery, oncology
PIK3CA related overgrowth syndromes	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, Laser services, Genetics if appropriate, ophthalmology, neurology, photography, radiology, plastic surgery
Klippel-Trenaunay-Weber syndrome	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, Sclerotherapy, Genetics if appropriate, ophthalmology, neurology, photography, radiology, plastic surgery

Megalencephaly-capillary malformation polymicrogyria syndrome	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, Laser services, Genetics if appropriate, ophthalmology, neurology, photography, radiology
Capillary malformation congenital	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, Laser services, Genetics if appropriate, ophthalmology, neurology, photography, radiology
Venous malformations	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, Sclerotherapy, Genetics if appropriate, ophthalmology, neurology, photography, radiology, plastic surgery
PHACE association	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, Cardiology, Neurology, Radiology, Genetics, ophthalmology, neurology
Sturge Weber syndrome	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, ophthalmology, neurology, radiology, plastic surgery
Phakomatosis Pigmentovascularis	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, ophthalmology, neurology, radiology, plastic surgery
Lymphatic malformations and lymphangiomatosis	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, Laser services, Genetics if appropriate, ophthalmology, neurology, photography, radiology

Cutaneous arteriovenous malformations	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, Interventional Radiology, Imaging, Haematology, Photography, Plastic surgery, genetics if appropriate
Capillary malformation-arterio venous malformation syndrome	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, Genetics, ophthalmology, neurology, photography, radiology
Maffucci syndrome	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology, Sclerotherapy, Genetics if appropriate, ophthalmology, neurology, photography, radiology, plastic surgery
Undiagnosed mosaic paediatric dermatology conditions	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology in first instance, plus Genetics
Glomuvenous malformation, segmental	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology in first instance, plus Genetics
Cutis marmorata telangiectatica congenita	Skin biopsy, Histopathology, DNA extraction from whole skin, next generation deep sequencing, imaging	Need access to expert Dermatology in first instance, plus Genetics

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

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*
Congenital melanocytic naevus syndrome and other melanocytic naevus syndromes	These conditions are all usually diagnosed in childhood	10	Based on prevalence and difficulty in management		2
Schimmelpenning Syndrome		1	Based on prevalence and difficulty in management		0-1

Phakomatosis Pigmentokeratotica		1	Based on prevalence and difficulty in management		0-1
Inflammatory Linear Verrucous Epidermal Naevus		2	Based on prevalence and difficulty in management		0-1
Other epidermal naevi		5	Based on prevalence and difficulty in management		1
Hypomelanosis of Ito and other hypopigmentary mosaic disorders		5	Based on prevalence and difficulty in management		2
Naevoid and whorled hypermelanosis and other hyperpigmentary mosaic disorders		5	Based on prevalence and difficulty in management		2
Extensive Dermal Melanocytosis		5	Based on prevalence and difficulty in management		2
Proteus Syndrome		1	Based on prevalence and difficulty in management		0-1
PIK3CA related overgrowth syndromes		10	Based on prevalence and difficulty in management		5
Klippel-Trenaunay-Weber syndrome		10	Based on prevalence and difficulty in management		5
Megalencephaly-capillary malformation polymicrogyria syndrome		5	Based on prevalence and difficulty in management		1
Capillary malformation congenital		20	Based on prevalence and difficulty in management		5
Venous malformations		20	Based on prevalence and difficulty in management		5
PHACE association		5	Based on prevalence and difficulty in management		0-1
Sturge Weber syndrome		5	Based on prevalence and difficulty in management		1
Phakomatosis Pigmentovascularis		5	Based on prevalence and difficulty in management		1
Lymphatic malformations and lymphangiomatosis		20	Based on prevalence and difficulty in management		10

Cutaneous arteriovenous malformations		2	Based on prevalence and difficulty in management		0-1
Capillary malformation-arteriovenous malformation syndrome		5	Based on prevalence and difficulty in management		1
Maffucci syndrome		1	Based on prevalence and difficulty in management		0-1
Undiagnosed mosaic paediatric dermatology conditions		5			2
Glomuvenous malformation, segmental		5	Based on prevalence and difficulty in management		1
Cutis marmorata telangiectatica congenita		5	Based on prevalence and difficulty in management		1

Please list the necessary human resources and the profesional qualifications essential to the quality of patient care within the Network's area of expertise.

Rare Diseases(s)	Health Care Professional (type)	Training & Qualifications	Minimun of number of procedures per patient per year	Rationale	
Congenital melanocytic naevus syndrome and other melanocytic naevus syndromes	Physicians, nurses	Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease	5	Patients attended by specialists at least once per year	

Schimmelpenning Syndrome	Physicians, nurses	Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease	5	Patients attended by specialists at least once per year
Phakomatosis Pigmentokeratotica	Physicians, nurses	Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease	5	Patients attended by specialists at least once per year
Inflammatory Linear Verrucous Epidermal Naevus	Physicians, nurses	Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease	5	Patients attended by specialists at least once per year
Other epidermal naevi	Physicians, nurses	Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease	5	Patients attended by specialists at least once per year

Hypomelanosis of Ito and other hypopigmentary mosaic disorders	Physicians, nurses	Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease	5	Patients attended by specialists at least once per year
Naevoid and whorled hypermelanosis and other hyperpigmentary mosaic disorders	Physicians, nurses	Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease	5	Patients attended by specialists at least once per year
Extensive Dermal Melanocytosis	Physicians, nurses	Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease	5	Patients attended by specialists at least once per year
Proteus Syndrome	Physicians, nurses	Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease	5	Patients attended by specialists at least once per year

PIK3CA related overgrowth syndromes		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year
Klippel-Trenaunay-Weber syndrome		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year
Megalencephaly-capillary malformation polymicrogyria syndrome		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year
Capillary malformation congenital		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year

Venous malformations		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year
PHACE association		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year
Sturge Weber syndrome		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year
Phakomatosis Pigmentovascularis		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year

Lymphatic malformations and lymphangiomatosis		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year
Cutaneous arteriovenous malformations		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year
Capillary malformation-arterio venous malformation syndrome		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year
Maffucci syndrome		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year

Undiagnosed mosaic paediatric dermatology conditions		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year
Glomuvenous malformation, segmental		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year
Cutis marmorata telangiectatica congenita		Formally recognised national qualifications in their relavent subspecialty, with experience in the field of this disease		Patients attended by specialists at least once per year

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

Congenital melanocytic naevus syndrome and other melanocytic naevus syndromes	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Schimmelpenning Syndrome	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Phakomatosis Pigmentokeratotica	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Inflammatory Linear Verrucous Epidermal Naevus	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Other epidermal naevi	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Hypomelanosis of Ito and other hypopigmentary mosaic disorders	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging

Naevoid and whorled hypermelanosis and other hyperpigmentary mosaic disorders	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Extensive Dermal Melanocytosis	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Proteus Syndrome	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
PIK3CA related overgrowth syndromes	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Klippel-Trenaunay-Weber syndrome	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Megalencephaly-capillary malformation polymicrogyria syndrome	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging

Capillary malformation congenital	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Venous malformations	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
PHACE association	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Sturge Weber syndrome	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Phakomatosis Pigmentovascularis	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Lymphatic malformations and lymphangiomatosis	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging

Cutaneous arteriovenous malformations	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Capillary malformation-arterio venous malformation syndrome	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Maffucci syndrome	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Undiagnosed mosaic paediatric dermatology conditions	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	
Glomuvenous malformation, segmental	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging
Cutis marmorata telangiectatica congenita	Appropriate office; computers; photo camerea; physical examination tools; imaging (ultraosund, CT scan, MRI, angio MRI), access to genetic testing	All required	Need for protocolized follow-up, recording and imaging

Please provide a summary explaining the approach or plans your group will undertake to produce good practice guidelines and implement outcome
Regular meetings will be organized for the lead members in the group, with invitation to attend for collaborators. A committee for guidelines will be nominated composed of 3-4 experts, that will produce guidelines for the proper management and testing for the best characterized disorders in the group of Cutaneous Mosaicism. These guidelines must be accepted by all lead members, and will be circulated to all the collaborators. The committee for guidelines for every disease will be responsible fr checking yearly the convenience and quality of those guidelines. Every 4 years, the guidelines will be subject for review.