# Classification des maladies dermatologiques rares

## Disorders of the epidermis

Ichthyoses
Bullous ichthyosiform erythroderma congenita
Skin peeling syndrome
Ichthyosis congenita, harlequin type
Ichthyosis bullosa of Siemens
Ichthyosis hystrix
    I*chthyosis hystrix, Curth-Macklin type*
    *Ichhtyosis hystrix gravior*
Dominant ichthyosis vulgaris
Lamellar ichthyosis
    *Lamellar ichthyosis, classical form*
    *Lamellar ichthyosis type 1*
    *Lamellar ichthyosis type 2*
Congenital ichthyosiform erythroderma, non bullous
CHILD syndrome
Ichthyosis linearis circumflexa
Dermopathy restrictive lethal
Ichthyosis, X-linked
Sjögren-Larsson syndrome
Gaucher disease type 2
Lipidosis with triglycerid storage disease
Netherton disease
Refsum disease
X-linked dominant chondrodysplasia punctata
PIBIDS syndrome
IBIDS syndrome

Erythrokeratodermas
Erythrokeratodermia variabilis, Mendes da Costa type
Erythrokeratoderma "en cocardes"
Erythrokeratodermia ataxia
Pityriasis rubra pilaris
Erythrokeratodermia, progressive symmetric

Acrokeratoderma
Acrokeratoelastoidosis of Costa
Acrokeratosis verruciformis of Hopf

Hereditary palmoplantar keratodermas
Hyperkeratosis palmoplantar, localized, epidermolytic
Thost-Unna palmoplantar keratoderma
Palmoplantar keratoderma with tonotubular keratin
Diffuse palmoplantar keratoderma, Norrbotten dominant type
Greither's disease
Keratosis palmoplantaris maculosa, papulosa, nummularis
Keratosis palmoplantaris striata
Hereditary painful callosities
Palmoplantar keratoderma punctate, hereditary
    *Keratosis palmoplantaris papulosa*
    *Porokeratosis punctata palmaris et plantaris (PPPP)*
    *Keratoderma, palmoplantar punctate type I*
    *Keratoderma, palmoplantar punctate type II*
Porokeratosis plantaris palmaris and disseminata
Acrokeratoelastoidosis of Costa
Tyrosinemia type 2
Keratoderma palmoplantar - deafness

Keratosis palmaris et plantaris – clinodactyly
Keratoderma palmoplantar - sclerodactyly
Diffuse palmoplantar keratoderma – acrocyanosis
Knuckle pods - leuconychia - sensorineural deafness
Epidermolysis bullosa, epidermolytic
Erythrokeratodermia variabilis, Mendes da Costa type
Keratoderma hereditarium mutilans
*Keratoderma – ichthyosiform dermatosis – elevated beta-glucuronidase*
Vohvinkel syndrome with ichthyosis
Olmsted syndrome
Dyskeratosis congenita
Epidermolysis bullosa, dystrophic
Epidermolysis bullosa simplex with mottled pigmentation (rare form)
Keratoderma palmoplantar spastic paralysis
Dermatopathia pigmentosa reticularis
Clouston syndrome
Woolly hair - palmoplantar keratoderma - dilated cardiomyopathy
Cardiofaciocutaneous syndrome
Keratosis, focal palmoplantar and gingival
Rolled and spiral hairs – palmoplantar keratoderma
Palmoplantar keratoderma – amyotrophy
Keratosis palmoplantaris - oesophageal carcinoma
Howell-Evans syndrome
Naegeli-Franceschetti-Jadassohn syndrome
Poikiloderma, hereditary acrokeratotic, Weary type
KID syndrome
Pachyonychia congenita
Mal de Meleda
Papillon-Lefevre syndrome
Keratosis palmoplantaris - periodontopathia - onychogryposis
Hereditary palmoplantar keratoderma, Gamborg-Nielsen type
Schopf-Schulz-Passarge syndrome
Naxos disease
Tyrosinemia type 2
CEDNIK syndrome
Alopecia congenita keratosis palmoplantaris
Leukoencephalopathy palmoplantar keratoderma
Palmoplantar porokeratosis of Mantoux

<u>Porokeratoses</u>
Porokeratosis of Mibelli
Porokeratosis plantaris palmaris and disseminata
Disseminated superficial actinic porokeratosis
Porokeratosis palmaris et plantaris
Papulosis, malignant atrophic

<u>Other disorders of the epidermis</u>
Fingerprints absence syndactyly milia
Acanthosis nigricans
Darier disease
hereditary painful callosities
Keratosis follicularis spinula decalvans
Knuckle pods leuconychia sensorineural deafness
Dupuytren contracture, familial
Hyperkeratosis lenticularis perstans
Ulerythema ophryogenesis
Keratosis pilaris atrophicans

<u>Syndromic disorders</u>
CHILD syndrome

X-linked dominant chondrodysplasia punctata
Ichthyosis and male hypogonadism
KID syndrome autosomal recessive
KID syndrome autosomal dominant
Neu laxova syndrome
Lipidosis with triglycerid storage disease
Refsum disease
Tyrosinemia type 2
Sjögren-Larsson syndrome

Epidermolysis bullosa
Epidermolysis bullosa, epidermolytic
*Epidermolysis bullosa simplex, Dowling-Meara type*
*Epidermolysis bullosa simplex with mottled pigmentation*
*Epidermolysis bullosa simplex with muscular dystrophy*
*Epidermolysis bullosa simplex, Köbner type*
*Epidermolysis bullosa simplex, Weber-Cockayne type*
*Epidermolysis bullosa simplex, Ogna type und*
*Epidermolysis bullosa simplex, autosomal recessive, without muscular dystrophy*
*Epidermolysis bullosa simplex superficialis*
Epidermolysis bullosa, junctional
*Epidermolysis bullosa, generalized atrophic benign*
*Epidermolysis bullosa, junctional - pyloric atresia*
*Epidermolysis bullosa, junctional, Herlitz type*
*Epidermolysis bullosa, junctional, inversa*
*Epidermolysis bullosa, junctional, late-onset*
*Epidermolysis bullosa, junctional, non-Herlitz*
*Epidermolysis bullosa, junctional, non-Herlitz*
Epidermolysis bullosa, dystrophic
*Epidermolysis bullosa, dystrophic, autosomal dominant*
*Epidermolysis bullosa, dystrophic, autosomal recessive, Hallopeau-Siemens type*
*Epidermolysis bullosa, dystrophic, inversa*
*Epidermolysis bullosa, dystrophic, pre-tibial*
*Transient bullous dermolysis of the newborn*
*Epidermolysis bullosa, dystrophic, autosomal recessive, non-Hallopeau-Siemens type*
*Epidermolysis bullosa, dystrophic, centripetalis*
*Epidermolysis bullosa, dystrophic, pruriginose*
Logic syndrome
Kallin syndrome

Pemphigus, benign chronic familial

## Disorders of epidermal appendages

***Hair group***

Alopecias
Loose anagen syndrome
Atrichia
Hypotrichosis simplex
Ichthyosis follicularis atrichia photophobia syndrome
Marie Unna congenital hypotrichosis
Congenital alopecia, X linked

Hirsutism
Fibromatosis gingival hypertrichosis
Barber-Say syndrome
Hypertrichosis lanuginosa congenita
Cataract hypertrichosis mental retardation
Ambras syndrome
Leprechaunism
Rabson-Mendenhall syndrome

Hypertrichosis cubiti short stature
Cervical hypertrichosis - peripheral neuropathy
Stein-leventhal syndrome

Hair shaft abnormalities, isolated
Monilethrix
Ringed hair disease
Pili torti
Pili torti with enamel defects
Björnstadt syndrome
Uncombable hair syndrome
Woolly hair
*Woolly hair nevus*

Hair shaft abnormalities, syndromic
Menkes syndrome
Tricho-dento-osseous syndrome
Trichodental syndrome
Langer-Giedion syndrome
BIDS syndrome
IBIDS syndrome
Onycho-tricho-dysplasia and neutropenia
PIBIDS syndrome
Sabinas - brittle hair syndrome
Pollitt syndrome
Woolly hair – hypotrichosis- everted lower lip- outstanding ears
Woolly hair - palmoplantar keratoderma -cardiac anomaly

***Nails group***
Nail disorders, isolated
Nails dysplasia

Nail disorders, syndromic
Nail-patella syndrome
Anonychia with flexural pigmentation
Onychodystrophy - deafness
Deafness onychodystrophy dominant form
Deafness onychodystrophy recessive form
Kumar levick syndrome
Anonychia, congenital
Onychodysplasia, congenital
Onycho-tricho-dysplasia and neutropenia
Pachyonychia congenita
    *Pachyonychia congenita, Jackson-Lawler type*
    *Pachyonychia congenita, Jadassohn-Lewandowsky type*

***Sweats glands group***
Hidradenitis suppurativa

***Sebaceous glands groups***
Nevus comedonicus syndrome
Orofaciodigital syndrome, type1
Steatocystoma multiplex
Steatocystoma multiplex natal teeth

***Ectodermal dysplasia syndrome***
Ankyloblepharon - ectodermal defects - cleft lip palate
Clouston syndrome
EEC syndrome
    *EEC syndrome, type 1*

*EEC syndrome, type 2*
*EEC syndrome, type 3*
Facial ectodermal dysplasia
GAPO syndrome
Christ-Siemens-Touraine syndrome
Rapp hodgkin syndrome
Hypodontia dysplasia of nails
Marshall-Smith syndrome
Focal facial dermal dysplasia
Sparse hair - short stature – Hypoplastic thumbs – Hypodontia – skin anomaly
Trichodysplasia - amelogenesis imperfecta
Ellis Van Creveld syndrome
Coffin-Siris syndrome
Incontinentia pigmenti
*Incontinentia pigmenti type 1*
*Incontinentia pigmenti type 2*
Dubowitz syndrome
Langer-Giedion syndrome
BIDS syndrome
IBIDS syndrome
Amelo-cerebro-hypohidrotic syndrome
ADULT syndrome
Alopecia contractures dwarfism mental retardation syndrome
Alopecia congenita keratosis palmoplantaris
Tetraamelia ectodermal dysplasia
Amelo-onycho-hypohidrotic syndrome
Cerebellar ataxia ectodermal dysplasia
Bartsocas-Papas syndrome
Ectodermal dysplasia absent dermatoglyphics
Book syndrome
Tricho-retino-dento-digital syndrome
Cardiofaciocutaneous syndrome
Cataract hypertrichosis mental retardation
CHAND syndrome
Anonychia - onychodystrophy with hypoplasia or absence of distal phalanges
Cortes-Lacassie syndrome
Cote-Katsantoni syndrome
Cranioectodermal dysplasia
Dahlberg - Borer - Newcomer, syndrome
Dermatoosteolysis kirghizian type
Dermo-odonto dysplasia
Digitorenocerebral syndrome
Door syndrome
Hidrotic ectodermal dysplasia type christianson fourie
Hidrotic ectodermal dysplasia Halal type
Ectodermal dysplasia mental retardation cns malformation
Oculodentoosseous dysplasia recessive
Ectodermal dysplasia alopecia preaxial polydactyly
Ectodermal dysplasia, hypohidrotic - hypothyroidism - ciliary dyskinesia
Ectodermal dysplasia neurosensory deafness
Ectodermal dysplasia berlin type
Ectodermal dysplasia tricho odonto onychial type
Facial ectodermal dysplasia
Ectrodactyly ectodermal dysplasia
Clefting - ectropion - conical teeth
Fibromatosis gingival hypertrichosis
Scalp ear nipple syndrome
Focal dermal hypoplasia
Gombo syndrome
Gorlin-Chaudhry-Moss, syndrome

Hallermann-Streiff-Francois syndrome
Hypertrichosis cubiti - short stature
Hypertrichosis lanuginosa congenita
Ichthyosis – alopecia – eclabion – ectropion - mental retardation
Johanson-Blizzard syndrome
Johnson neuroectodermal syndrome
Lacrimoauriculodentodigital syndrome
Martinez-Monasterio-Pinheiro syndrome
Pyramidal molar – glaucoma - upper abnormal lip
Oculodentoosseous dysplasia dominant
Brachymetapody – anodontia – hypotrichosis - albinoidism
Oculo-tricho-dysplasia
Odontoonychodermal dysplasia
Odonto onycho dysplasia with alopecia
Odontotrichomelic hypohidrotic dysplasia
Taurodontia absent teeth sparse hair
Onycho-tricho-dysplasia and neutropenia
Orofaciodigital syndrome, type1
Papillon-Lefevre syndrome
Pili torti onychodysplasia
Pilodental dysplasia with refractive errors
Pilodentoungular dysplasia - microcephaly
Polyposis skin pigmentation alopecia fingernail changes
Ichthyosis - male hypogonadism
Sabinas brittle hair syndrome
Schinzel-Giedion midface retraction syndrome
Stern-Lubinsky-Durrie syndrome
Deafness, enamel hypoplasia, nail defects
Deafness onychodystrophy dominant form
Zlotogora-Ogur syndrome
Trichodental syndrome
Trichodentoosseous syndrome
Trichodermodysplasia dental alterations
Tricho-odonto-onychial dysplasia
Tricho odonto onycho dermal syndrome
Trichorhinophalangeal syndrome, type 1
Trichomegaly - retina pigmentary degeneration - dwarfism
Odonto-onycho-hypohidrotic dysplasia, midline scalp defects
Xeroderma talipes enamel defects
Zunich-Kaye syndrome
Ectodermal dysplasia, hypohidrotic, autosomal recessive
Ectodermal dysplasia, hypohidrotic, autosomal dominant
Ito hypomelanosis
Schopf-Schulz-Passarge syndrome
Odonto-tricho-ungual-digito-palmarn syndrome
Ectodermal dysplasia with natal teeth, Turnpenny type
Ectodermal dysplasia, "pure" hair-nail type
Limb-mammary syndrome
Ectodermal dysplasia - skin fragility syndrome
Naegeli-Franceschetti-Jadassohn syndrome
Anonychia with flexural pigmentation
Pollitt syndrome
Ectodermal dysplasia, anhidrotic, with immunodeficiency, osteopetrosis, and lymphedema
Metaphyseal chondrodysplasia, recessive type

## Disorder of pigmentation

Hyperpigmentation
Dowling-Degos disease
Dyskeratosis congenita
*Dyskeratosis congenita, X-linked*

*Dyskeratosis congenita, autosomal dominant*
*Dyskeratosis congenita, autosomal recessive*
*Hoyeraal-Hreidarsson syndrome*
*Zinsser-Cole-Engman syndrome*
*Dyskeratosis congenita, Scoggins type*
Fanconi anemia
Hemochromatosis familial
LEOPARD syndrome
Moynahan syndrome
linear and whorled nevoid hypermelanosis
McCune-Albright syndrome
Naegeli-Franceschetti-Jadassohn syndrome
Carney complex
NAME syndrome
Neurofibromatosis type 1
*Neurofibromatosis, familial segmental*
*Neurofibromatosis, familial spinal*
Watson syndrome
Neurofibromatosis-Noonan syndrome
Neurofibromatosis type 6
Phacomatosis pigmentovascularis
*Phacomatosis cesioflammea*
*Phacomatosis cesiomarmorata*
*Phacomatosis pigmentovascularis type I*
*Phacomatosis pigmentovascularis type II*
*Phacomatosis pigmentovascularis type III*
*Phacomatosis pigmentovascularis type IV*
*Phacomatosis spilorosea*
Peutz-Jeghers syndrome
Familial progressive hyperpigmentation

Hypopigmentation
Albinism deafness syndrome
Oculocutaneous albinism
*Hermansky-Pudlak syndrome*
*Oculocutaneous albinism type 1A, OCA-1A*
*Oculocutaneous albinism type 2, OCA-2 (Oculocutaneous albinism, tyrosinase-positive)*
*Oculocutaneous albinism type 3, OCA-3 (Rufous/Red/Xanthous oculocutaneous albinism)*
*Oculocutaneous albinism type 1B, OCA-1B*
*Oculocutaneous albinism type 4, OCA-4*
Oculocerebral hypopigmentation syndrome cross type
Ito hypomelanosis
Piebaldism
Vitiligo
Waardenburg syndrome type 1
Waardenburg syndrome type 2
*Waardenburg syndrome type 2A*
*Waardenburg syndrome type 2B*
Waardenburg syndrome type 3
Waardenburg-Shah syndrome
*Hirschsprung disease with pigmentary anomaly*

## Disorder of the dermis

Collagen
Amniotic bands
Buschke-Ollendorff syndrome
Ehlers-Danlos syndrome type 7C
Ehlers-Danlos syndrome, classic type
Ehlers-Danlos syndrome type 3
Ehlers-Danlos syndrome type 4

Ehlers-Danlos syndrome type 6
Ehlers-Danlos, syndrome, type 8
Ehlers-Danlos syndrome, progeroid type
Ehlers-Danlos syndrome, type 5
Ehlers-Danlos syndrome, type 10
Reactive perforating collagenosis
Costello syndrome
Cutis laxa
Pseudoxanthoma elasticum
Elastosis Perforans Serpiginnosa

Vascular
Ataxia telangiectasia
*Ataxia-telangiectasia-like disorder*
Blue rubber bleb nevus
Cutis marmorata telangiectatica congenita
Fabry disease
Rendu-Osler-Weber disease
*Rendu-Osler-Weber disease 2*
*Rendu-Osler-Weber disease 3*
Angio-osteohypertrophic syndrome
Cobb syndrome
Nevi flammei
Enchondromatosis
Sturge-Weber syndrome
Wyburn-Mason syndrome
Cerebral cavernous malformations

Mixed
Circumscribed cutaneous aplasia of the vertex
Adams oliver syndrome
Scalp defects postaxial polydactyly
Scalp ear nipple syndrome
Midas syndrome
Focal dermal hypoplasia
Tuberous sclerosis
*Tuberous sclerosis, type 1*
*Tuberous sclerosis, type 2*

Others disorders of the dermis
Albright hereditary osteodystrophy
*Pseudohypoparathyroidism type 1A*
*Pseudohypoparathyroidism type 1C*
*Pseudopseudohypoparathyroidism*
Ectopic ossification familial type
Cutis verticis gyrata
Cutis verticis gyrata mental deficiency
*Cutis verticis gyrata thyroid aplasia mental retardation*
Pachydermoperiostosis
Cutis gyrata acanthosis nigricans craniosynostosis
Familial dysautonomia
Dermochondralcorneal dystrophy
Synovitis granulomatous uveitis cranial neuropathies
Lipoid proteinosis
Pterygia syndrome, lethal forms
*Multiple pterygium syndrome, Aslan type*
*Multiple pterygium syndrome, X-linked*
*Multiple pterygium syndrome, lethal form*
Multiple pterygium syndrome, autosomal recessive
*Escobar syndrome*

Bartsocas-Papas syndrome
Pterygium syndrome antecubital
Pterygium multiple, syndrome, autosomal dominant
Fibromatosis juvenile hyaline

## Disorders of subcutaneous tissue

Xanthomatosis cerebrotendinous
Encephalo cranio cutaneous lipomatosis
Adiposis dolorosa
Familial symmetric lipomatosis
Fibrodysplasia ossificans progressiva
Farber lipogranulomatosis
Lipodystrophy, familial partial, Dunnigan type
Lipodystrophy, Berardinelli type

## Urticaria

Familial cold urticaria
Pruritic urticarial papules and plaques of pregnancy
Angioneurotic edema
    *Angioneurotic edema, acquired*
    *Angioneurotic edema, hereditary*
Melkersson-Rosenthal syndrome
Muckle-Wells syndrome
Cutaneous mastocytosis
    *Cutaneous mastocytoma*
    *Diffuse cutaneous mastocytosis*
    *Urticaria pigmentosa*

## Other disorders

Erythromelalgia
Erythrothermalgia
Michelin tire baby syndrome
Stiff skin syndrome

## Tumors/Hamartomas

Gorlin syndrome
Bazex-Dupre-Christol syndrome
    *Congenital hypotrichosis milia*
    *Follicular atrophoderma-basal cell carcinoma*
Rombo syndrome
Giant pigmented hairy nevus
Melanosis neurocutaneous
Cowden syndrome
Melanoma, familial
CDK4 linked melanoma
Melanoma type 1
Melanoma type 2
Atypical mole
    *Atypical mole syndrome*
    *Dysplastic nevus syndrome*
    *Familial atypical multiple mole melanoma syndrome (FAMMM)*
Nevus sebaceus syndrome
Linear nevus syndrome
Linear inflammatory verrucous epidermal nevus
Verrucous nevus
Verrucous nevus acanthokeratolytic
Gardner syndrome
Keratoacanthoma familial
    *Multiple keratoacanthoma, Ferguson-Smith type*

*Muir-Torre syndrome*
Infantile myofibromatosis
Multiple endocrine neoplasia, type 2
Leiomyomatosis, familial
*Leiomyomatosis, familial, with renal carcinoma*
*Multiple cutaneous and uterine leiomyomas*
Proteus syndrome
Bannayan-Riley-Ruvalcaba syndrome
*Riley-Smith syndrome*
*Ruvalcaba-Myhre-Smith syndrome*
Nevus sebaceus syndrome
Encephalo cranio cutaneous lipomatosis
Calcinosis, tumoral

## Metabolic disorders

Porphyrias
Porphyria, acute intermittent
Porphyria, congenital erythropoietic -Gunther disease
Protoporphyria, erythropoietic
Hereditary coproporphyria
Porphyria cutanea tarda, familial type
Porphyria cutanea tarda, sporadic type
Variegate prophyria

Mucoplysaccharidoses
Mucopolysaccharidosis type 2

Other metabolic disorders
Acrodermatitis enteropathica, zinc deficiency type
Alkaptonuria
Multiple carboxylase deficiency
*Multiple carboxylase deficiency, due to biotinidase deficiency*
*Multiple carboxylase deficiency, due to holocarboxylase synthetase deficiency*
Amyloidosis
Amyloid lichen

## Premature aging

Cockayne syndrome
Progeroid syndrome de barsy type
Hallermann streiff francois syndrome
Progeria
Werner syndrome
*Atypical Werner syndrome*
Flynn aird syndrome

## Photosensitivity

Bloom syndrome
Hartnup syndrome
Poikiloderma of Kindler
Xeroderma pigmentosum
Rothmund-Thomson syndrome
*Poikiloderma atrophicans-cataract*
De sanctis cacchione syndrome

## Immune deficiency diseases

Chediak-Higashi syndrome
Griscelli disease
*Griscelli syndrome type 1 (hypopigmentation - neurologic impairment)*
*Griscelli syndrome type 2 (hypopigmentation - immunodeficiency, with or without neurologic impairment)*

*Griscelli syndrome type 3*
Granulomatous disease, chronic
Lutz-Lewandowsky epidermodysplasia verruciformis
Candidiasis, chronic mucocutaneous
Job syndrome
Mucoepithelial dysplasia
Wiskott-Aldrich syndrome

## Toxic epidermolysis

Lyell syndrome
Stevens-Johnson syndrome

## Bullous autoimmune diseases

Pemphigus vulgaris
*Pemphigus vegetans*
Bullous pemphigoid
Pemphigus superficial
*Pemphigus erythematosus (pemphigus seborrheic, Senear-Usher syndrome)*
*Pemphigus foliaceus*
Acquired epidermolysis bullosa
Gestationis pemphigoid
Cicatricial pemphigoid
Dermatitis herpetiformis
Linear IgA dermatosis
Pemphigus paraneoplastic