Sound-alikes in dermatology

by Jeffrey Kushner, DO, and Kristen Whitney, DO

Disease Entity	Description
Actinic granuloma/ Annular elastolytic giant cell granuloma	Variant of granuloma annulare on sun-damaged skin; annular erythematous plaques with slightly atrophic center in sun-exposed areas, which may be precipitated by actinic damage.
Actinic prurigo	PMLE-like disease with photodistributed erythematous papules or nodules and hemorrhagic crust and excoriation. Conjunctivitis and cheilitis are commonly found. Seen more frequently in Native Americans (especially Mestizos).
Actinomycetoma	"Madura Foot"; suppurative infection due to *Nocaria, Actinomadura,* or *Streptomyces* resulting in tissue tumefaction, draining sinuses and extrusion of grains.
Actinomycosis	"Lumpy Jaw"; *Actinomyces israelii*; erythematous nodules at the angle of jaw leads to fistulous abscess that drain purulent material with yellow sulfur granules.
Acrokeratosis verruciformis	Multiple skin-colored, warty papules on the dorsal hands and feet. Often seen in conjunction with Darier disease.
Acrodermatitis enteropathica	AR; SLC39A4 mutation; eczematous patches on acral, perineal and periorificial skin; diarrhea and alopecia; secondary to zinc malabsorption.
Atrophoderma	1) Atrophoderma vermiculatum: Pitted atrophic scars in a honeycomb pattern around follicles on the face; associated with Rombo, Nicolau-Balus, Tuzun and Braun-Falco-Marghescu syndromes. 2) Follicular atrophoderma: Icepick depressions at follicular orifices on dorsal hands/feet or cheeks; associated with Bazex-Dupré-Christol and Conradi-Hünermann-Happle syndromes. 3) Atrophoderma of Pasini and Pierini: Depressed patches on the back with a "cliff-drop" transition from normal skin. 4) Atrophoderma of Moulin: Similar to Pasini/Pierini, except lesions follow the lines of Blaschko.
Anetoderma	Localized area of flaccid skin due to decreased or absent elastic fibers; exhibits "buttonhole" sign.
Bart's syndrome	AD; collagen VII mutation; aplasia cutis congenita of the lower extremities, plus dominant dystrophic epidermolysis bullosa.
Bart-Pumphrey syndrome	AD; GJB2 mutation; diffuse PPK with knuckle pads, leukonychia, and deafness.
Bazex syndrome/ Acrokeratosis paraneoplastica	Paraneoplastic disorder with a psoriasiform dermatitis involving the hands, feet, ears, and nose; associated with upper aerodigestive tract malignancies.
Bazex syndrome/Bazex–Dupré–Christol syndrome	XLD; follicular atrophoderma, milia, multiple BCCs, hypotrichosis, and hypohidrosis.
Cheilitis glandularis	Inflammatory hyperplasia of the lower labial salivary glands due to chronic sun exposure or irritation; characterized by swelling and eversion of the lower lip.
Cheilitis granulomatosa	Non-caseating granulomatous inflammation resulting in swelling of the lip; associated with facial nerve palsy and fissured tongue in Melkersson-Rosenthal syndrome.
Chilblain lupus	AD; TREX1 mutation; cutaneous form of chronic cutaneous lupus with red to dusky purple papules and plaques on the fingers and toes associated with acrocyanosis.
Chilblains pernio	Abnormal inflammatory and vascular response to cold temperatures resulting in erythematous to violaceous macules, papules, and nodules on acral skin.
Cockayne-Touraine	AD; COL7A1 mutation; DDEB with bullae localized to extremities resolving with milia and scarring.
Cockayne syndrome	AR; ERCC8 and ERCC6 mutations; cachectic dwarf with photosensitivity, salt-and-pepper retinal pigmentation, facial lipoatrophy, CNS demyelination, deafness.
Weber-Cockayne	AD; K5, K14 mutations; localized form of EBS with palmoplantar bullae and callouses.
Darier's disease	AD; ATP2A2 mutation; hyperkeratotic papules and plaques in a seborrheic distribution, acrokeratosis verruciformis of hopf, red-white longitudinal nail bands with V-shaped nicks, oral cobblestoning.
Darier's sign	Rubbing of lesions in patients with mastocytosis that leads to erythema, pruritus and swelling.
Dowling-Meara/ EBS herpetiformis	AD; K5, K14 mutations; most severe form of EBS with widespread "herpetiform" bullae, PPK, blistering/erosions of oral cavity and esophagus, nail dystrophy and early death; clumped tonofilaments on EM.

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Disease Entity	Description
Dowling-Degos disease	AD; K5 mutation; reticulated hyperpigmentation in flexural sites with comedone-like lesions on the neck and back.
Degos disease/ malignant atrophic papulosis	Vaso-occlusive disorder with characteristic lesions having a umbilicated, porcelain-white center with surrounding telangiectasias; death due to GI perforation and peritonitis.
Ecthyma	*S. pyogenes* or *S. aureus*; deep form of impetigo with punched out ulcers and thick, overlying yellow crusts.
Ecthyma contagiosum	*Orf* virus; associated with exposure to sheep/goats; skin lesion progresses through six stages: maculopapular, targetoid, acute, regenerative, papillomatous, and regressive.
Ecthyma gangrenosum	*Pseudomonas aeruginosa*; hemorrhagic pustules evolving into necrotic black ulcers in septic immunosuppressed patients.
Erythrokeratoderma variabilis	AD; GJB3, GJB4 mutations; fixed hyperkeratotic plaques on the face and extremities with transient, migratory erythematous patches.
Epidermodysplasia verruciformis	AR; EVER1, EVER2 mutation; sporadic form associated with HIV, immunosuppression; abnormal susceptibility to HPV 5 & 8 resulting in multiple verrucous lesions with significant risk of malignant transformation.
Goltz syndrome/ Focal dermal hypoplasia	XLD; PORCN mutation; linear atrophy following Blaschko's lines with fat herniation, osteopathia striata, lobster claw deformity, syndactyly, coloboma.
Gorlin syndrome/ Basal cell nevus syndrome	AD; PTCH gene; numerous BCC's, palmoplantar pits, odontogenic keratocysts, calcification of falx cerebri, medulloblastomas, bifid ribs.
Greither syndrome	AD; K1 mutation; transgrediens PPK, hyperhidrosis, hyperkeratotic plaques on shins, knees and elbows.
Gunther disease	A.K.A. Congenital Erythropoietic Porphyria (CEP); AR; uroporphyrinogen III synthase mutation; photosensitivity with scarring, erythrodontia, hypertrichosis, hemolysis, red urine.
Pemphigus vegetans – Hallopeau subtype	P. vegetans classically occurs in two subtypes: Hallopeau subtype is less severe and begins with pustules while Neumann subtype is more severe and begins with flaccid bullae and erosions; both forms develop into vegetative plaques.
Hallopeau-Siemens	AR; COL7A1 mutation; RDEB; severe, generalized bullae with atrophic scarring; mitten deformity of hands/feet, SCCs.
Jackson-Lawler	AD; K6b and K17 mutations; Type II Pachyonychia Congenita; subungual hyperkeratosis, focal PPK, steatocystoma multiplex, epidermoid cysts, natal teeth.
Jadassohn-Lewandowsky	AD; K6a and K16 mutations; Type I Pachyonychia Congenita; subungual hyperkeratosis, focal PPK, oral leukokeratosis.
Jadassohn-Pellizzari anetoderma	Subtype of primary anetoderma with preceding inflammatory lesions.
Livedo reticularis	Mottled, reticular reddish-blue vascular pattern typically on the extremities with a variety of causes.
Livedo racemosa	Irregular, branching vascular pattern with broken circular segments that are fixed and do not vary with temperature; can be associated with Sneddon's syndrome or antiphospholipid syndrome.
Livedoid vasculopathy/livedoid vasculitis	A.K.A. Atrophie Blanche; painful, punched out ulcers on lower extremities that heal with atrophic hypopigmented scars.
Lupus pernio	Form of cutaneous sarcoidosis that presents with indurated, violaceous nodules and plaques on the nose, ears and cheeks.
Lupus vulgaris	Form of cutaneous tuberculosis in previously sensitized individuals; appears as a red-brown plaque on the head/neck; "apply-jelly" color on diascopy.
Lupus miliaris disseminatus faciei	Granulomatous rosacea variant with red to brown papules frequently on malar cheeks.
Majocchi's disease/ Purpura annularis telangiectodes	Type of pigmented purpuric dermatosis with annular plaques and punctate telangiectasias.
Majocchi's granuloma	Granulomatous folliculitis due to dermatophyte infection of the hair follicle often due to *T. rubrum.*

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Necrolytic acral erythema	Acral, pruritic, hyperkeratotic plaques; associated with HCV and altered zinc metabolism.
Necrolytic migratory erythema	Paraneoplastic disorder associated with underlying glucagon-secreting tumor of the pancreas; erythematous, crusted patches often found on the face, groin and abdomen.
Olmsted syndrome	AD; TRPV3 mutation; mutilating PPK with periorificial plaques.
Omens syndrome	AR; RAG1 and RAG2 mutations; form of SCID with erythroderma.
Rothmund-Thomson syndrome	AR; RECQL4 mutation; poikiloderma, premalignant acral keratosis, photosensitivity, nail dystrophy, hypoplastic/absent thumbs, risk of osteosarcoma.
Rubinstein-Taybi syndrome	Sporadic; CREB-binding protein mutation; capillary malformation, broad thumbs, craniofacial abnormalities, MR, cryptorchidism.
Trichodysplasia spinulosa	Skin-colored, spiny papules typically on the face; seen in immunosuppressed organ transplant patients; associated with polyomavirus.
Trichostasis spinulosa	Asymptomatic comedo-like lesions containing keratin and multiple vellus hairs on the face.
McCune-Albright syndrome	Sporadic; GNAS1 mutation; "Coast of Maine" CALMs, polyostotic fibrous dysplasia, precocious puberty, hyperthyroidism.
Albright's hereditary osteodystrophy	AD; GNAS Gs subunit mutation; pseudohypoparathyroidism, short fourth and fifth metacarpals, subcutaneous calcifications, short stature, round face, mental retardation.
Endemic typhus	Organism – *Rickettsia typhi*; Vector – rat flea (*Xenopsylla cheopis*); headache, fever, myalgias, transient maculopapular truncal eruption.
Epidemic typhus	Organism – *Rickettsia prowazekii*; Vector – body louse (*Pediculus humanus corporis*); HA, fever, myalgias; macular lesions evolve into truncal petechiae sparing the face.
Typhoid fever	Organism – *Salmonella typhi*; fecal-oral transmission; "rose-spots" on trunk, fever, abdominal pain, weakness, diarrhea.
Vaccinia	Injection site reaction to administration of live vaccine for smallpox; eczema vaccinatum is a more exuberant reaction in atopic patients.
Variola	Virus associated with smallpox; results in vesicles/pustules all at the same stage of development.

References

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