

## Neonatal Ichthyosis

by Alyx Rosen, MD and Kate Oberlin, MD

Disease	Gene Defect	Clinical Presentation	Additional Manifestations	Prognosis/Outcome
<b>Harlequin ichthyosis</b>	AR ABCA12	Thick membrane with deep cracks and fissures forming plate-like scales, ectropion, eclabium	Premature delivery; respiratory distress; ear deformities; fluid imbalance	Survival rate ~50%; oral retinoids may help prolong survival
<b>Congenital ichthyosiform erythroderma (CIE)</b>	AR TGM-1 ALOX12B ALOXE3	Generalized erythema and fine scale; collodion membrane at birth; involves flexures; nail dystrophy	Heat intolerance; scarring alopecia; rare ectropion	No improvement with age
<b>Lamellar ichthyosis</b>	AR TGM-1 ABCA12	Thick, brown scale involving flexural areas and trunk; eclabium, ectropion	Heat intolerance; hypernatremia	No improvement with age
<b>Epidermolytic hyperkeratosis (EHK); Bullous CIE</b>	AD K1/K10	Widespread blistering and erythema at birth; involves flexures, palmoplantar keratoderma	Failure to thrive; hypernatremia; recurrent infections	Evolves into verrucous hyperkeratotic plaques
<b>Ichthyosis vulgaris</b>	AD Filaggrin	Generalized fine, adherent scale sparing flexures	Atopic diathesis; keratosis pilaris	Improves with age
<b>X-linked ichthyosis</b>	XLR Steroid sulfatase	Generalized fine scale and desquamation at birth	Corneal opacities; cryptorchidism; prolonged labor	Brown "dirty" scales sparing flexures
<b>Netherton syndrome</b>	AR SPINK5	Trichorrhexis invaginata, ichthyosis linearis circumflexa, atopic dermatitis	Failure to thrive; infections; elevated IgE; food allergies	Pruritus; eczematous plaques
<b>Refsum disease</b>	AR PEX7 PHYH	Skin resembles ichthyosis vulgaris with mild scaling	Retinitis pigmentosa; cerebellar ataxia	Accumulation of phytanic acid; deafness
<b>Sjögren-Larsson syndrome</b>	AR FALDH	Generalized erythema and scale at birth, then dark scales on trunk	Parafoveal glistening white dots on retina	Mental retardation, seizures and spastic paralysis
<b>Conradi-Hünermann-Happle syndrome</b>	XLD EBP gene	Severe ichthyosiform erythroderma and scale following lines of Blaschko	Chondrodysplasia punctata; cataracts; deafness; alopecia	Follicular atrophoderma and scarring
<b>KID syndrome</b>	AD GJB2	Keratitis, ichthyosis, deafness, keratoderma	Photophobia; leukonychia	Increased risk of cutaneous SCC
<b>Trichothiodystrophy (PIBIDS)</b>	AR ERCC2 ERCC3	Photosensitivity, ichthyosis, brittle hair "tiger tail", intellectual impairment, decreased fertility, short	Sulfur deficiency in hair; diffuse alopecia	Photosensitivity decreases with age
<b>CHILD syndrome</b>	XLR NSDHL	Congenital hemidysplasia with ichthyosiform erythroderma, limb defects	Ipsilateral organ aplasia; stippled epiphyses	Erythema fades into hyperkeratosis

DDx "Collodion Baby": Lamellar Ichthyosis, Congenital Ichthyosiform Erythroderma (CIE), Netherton syndrome, Harlequin Ichthyosis, Self-healing collodion baby (SHCB), Trichothiodystrophy (PIBIDS), Sjögren-Larsson syndrome, Infantile Gaucher disease, Neutral lipid storage disease with ichthyosis, Hay-Wells syndrome, Conradi-Hünermann-Happle syndrome

### References:

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## Neonatal Infectious Dermatoses

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Disease	Clinical Presentation	Extracutaneous Manifestations	Work-Up	Treatment
<b>Syphilis</b>	Macules or papules on palms and soles, trunk, extremities; also erosions, bullae and petechiae; mucous patches	IUGR, HSM, osteochondritis, "snuffles" or rhinitis	Maternal and neonate serology; darkfield examination	High dose penicillin G to prevent late complications
<b>Candidiasis</b>	Diffuse red eruption with satellite red papules and pustules on the folds and scrotum	Oral thrush, paronychia, systemic infection	Gram stain or KOH reveals pseudohyphae or budding yeast	Topical nystatin, ciclopirox, imidazoles, PO/IV if septic
<b>Toxoplasmosis</b>	Varies depending on timing of infection; non-specific, diffuse macules or papules, ecchymosis	Chorioretinitis, hydrocephalus, deafness, preterm birth, IUGR	Serologic testing for antigen, PCR, or antibody (IgG, IgM)	Pyrimethamine with Sulfadiazine and leucovorin
<b>Rubella</b>	"Blueberry muffin" macules due to extramedullary hematopoiesis	Cataracts, congenital heart disease, deafness, HSM, microcephaly	Viral cultures of pharynx, CSF, urine, conjunctiva, rubella antibody	Neuroimaging; supportive care
<b>CMV</b>	Generalized petechiae or purpura within 48 hours of birth, "blueberry muffin" macules	IUGR, deafness, HSM, thrombocytopenia, chorioretinititis	Viral culture, PCR, antibody testing	Ganciclovir, valganciclovir, foscarnet
<b>Scabies</b>	Vesicles and papules with rare burrows on the palms, soles, trunk and genitalia	Pruritus, irritability, poor feeding	Mineral oil prep demonstrating mite or eggs	Permethrin cream
<b>HIV</b>	Thrush, widespread seborrheic dermatitis	Bacterial and viral infections	Serology testing, PCR	Zidovudine; anti-retroviral therapy
<b>HSV</b>	Grouped vesicles on red base; diffuse erosions	Ocular and CNS involvement, sepsis	Viral cultures, PCR, Tzanck prep	Intravenous acyclovir
<b>Varicella</b>	Cicatricial dermatomal lesions with hypoplasia (congenital); generalized papules (neonatal)	Ocular and CNS abnormalities	Antibody testing, PCR, Tzanck prep	Varicella-zoster immunoglobulin; acyclovir
<b>Impetigo neonatorum</b>	Pustules, vesicles or bullae on an erythematous base in folds and groin; second week of life	Rarely complicated by sepsis, osteomyelitis and pneumonia	Gram stain with Gram-positive cocci in clusters and neutrophils	Dicloxacillin or cephalaxin for a 10-day course
<b>Staphylococcal Scalded Skin Syndrome (SSSS)</b>	Scarlatiniform eruption with erythroderma in intertriginous sites; periorificial crust, +Nikolsky sign, spares mucosa	Irritability, fever, skin tenderness, rhinorrhea	Histology (frozen) with granular layer cleavage; bacterial cultures	$\beta$ -lactamase-resistant antibiotics; decolonization

**Abbreviations:** HSM=hepatosplenomegaly; CMV=Cytomegalovirus; HSV=Herpes simplex virus; HIV=Human immunodeficiency virus; CNS=Central nervous system; IUGR=intrauterine growth restriction

DDx of "Blueberry muffin baby": CMV, varicella, zoster, rubella, toxoplasmosis, langerhans cell histiocytosis, syphilis, leukemia cutis, neuroblastoma, parvovirus, hemolytic anemia, twin-twin transfusion, myelodysplasia, rhabdomyosarcoma, neonatal lupus

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