Dermatologic findings in pediatric genetic diseases

Phenylketonuria: albinism, eczema
Wiscot-Aldrich: eczema, petechia (thrombocytopenia)
Autosomal dominant hyper IgE (Job): eczema
Ataxia-telengiectasia: dilated skin vessels
Chediak-Higashi: albinism
Homocystinuria: fair hair, skin: (homocysteine inhibits tyrosinase)
Alkaptonuria: blue skin
Fabry: angiokeratomas (trunk, genitals, buttocks), hypohidrosis
IPEX syndrome: eczema
Waardenburg: albinism (and hearing loss)

Eczema
PKU
Autosomal dominant hyper IgE (Job)
Wiscot
IPEX

Cafe au lait spots
NF1/2, tuberous sclerosis, Fanconi anemia, McCune-Albright, Noonan, Bloom, ataxia-telangiectasia
(Fanconi, Bloom, ataxia-telengiectasia involve DNA repair defects, increased cancer risk)

Nevus flammeus (Port wine stain)
Sturge-Weber syndrome
Beckwith-Wiedeman syndrome
Klippel-Trenaunay syndrome
congenital cutaneous capillary malformations (port wine stain), varicose veins, soft tissue/bony overgrowth in lower limbs
*Sturge-Weber and Beckwith-Wiedeman also have brain tumors and renal/hepatic tumors respectively

Xanthomas/xanthelasmas: familial dyslipidemias
Eruptive xanthomas on back, buttocks: type 1 dyslipidemia
Tendinous xanthomas: type 2 dyslipidemia
Palmar xanthomas: type 3 dyslipidemia

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Immunodeficiency plus neurological symptoms plus skin findings
Ataxia-telengiectasia: cerebellar dysfunction; dilated skin vessels
Chediak-Higashi: peripheral neuropathy; albinism

Immunodeficiency plus poor wound healing, absent pus at infection sites
Leukocyte adhesion deficiency 1

Immunodeficiency plus recurrent skin abscesses and candida skin infections
Chronic granulomatous disease (abnormal dihydrorhodamine test)

Immunodeficiency plus recurrent candida skin infections
Chronic mucocutaneous candidiasis (abnormal skin antigen test)

Immunodeficiency plus recurrent candida skin infections
Myeloperoxidase deficiency (low hypochlorite)

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Neurocutaneous syndromes

Tuberous sclerosis: angiofibromas, ash leaf spots, shagreen patches, cafe au lait spots
Von Hippel-Lindau: angiomatosis of skin
NF 1/2: neurofibromas, cafe au lait spots

NF 1: axillary freckling
Sturge-Weber: Nevus flammeus (Port wine stain)
Ataxia-telengiectasia: telengiectasias (often ocular)

Hypopigmented macule: ash leaf spot
Hyperpigmented macule: cafe au lait spot

Seizures plus skin lesions
Sturge-Weber, NF1, tuberous sclerosis
(PKU: eczema, albinism)

Skin lesions and intracranial calcifications
Tuberous sclerosis, Sturge-Weber, Gorlin syndrome

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Hereditary hemorrhagic telangiectasia (skin, lips, mouth)

Peutz-Jeghers: hyperpigmented macules on the lips/buccal mucosa, hands, genitals

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Collagen disorders
Ehlers-Danlos (type 3, 5 collagen)
Osteogenesis imperfecta (type 1 collagen)
thin, translucent skin; easy bruising
Menkes (copper deficiency [impaired lysyl oxidase])
Marfans (fibrillin defect but collagen also affected)

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Hemochromatosis
bronzing (hyperpigmentation) of the skin:
(hemochromatosis, an iron overload state, is a risk factor for porphyria cutanea tarda)

Porphyria cutanea tarda: blistering of the skin

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Platelet pathology

petechia, purpura, gingival bleeding
Von Willebrand, Bernard-Soulier, Glanzmann thrombasthenia

Kasabach-Merritt : thrombocytopenia and hemangiomas

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Hemophilia A: easy bruising, bleeding from mucosal surfaces, delayed wound healing

Men 2b: mucosal neuroma (bumps on lips, tongue, buccal)

Nevoid basal-cell carcinoma syndrome (Gorlin syndrome)
autosomal dominant; early development of basal cell carcinomas, medulloblastomas, intracranial calcifications (falx cerebri), and skeletal abnormalities