Teeth

Charya By, MD and Matt Steadmon, MD

Anodontia / hypodontia

Associated diseases	Gene / defect	Other findings
Hypomelanosis of Ito	-	Hypopigmentation following lines of Blaschko, seizures, scoliosis, alopecia, mental retardation, strabismus
Incontinentia pigmenti	NEMO	Cutaneous lesions in lines of Blaschko, scarring alopecia, seizures, delayed psychomotor development, blindness, retinal vascular abnormalities
Hypohidrotic ectodermal dysplasia	EDA , EDA receptor, NEMO	Hypotrichosis, heat intolerance, periorbital hyperpigmentation, saddle nose, everted thick lips, bronchopulmonary infections
Focal dermal hypoplasia	PORCN	Atrophic, telangiectactic streaks in Blaschko's lines; papillomas in lips, axillae, perineum; dystrophic nails; syndactyly; alopecia; colobomas; osteopathia striata; scoliosis; mental retardation; short stature
Ectrodactyly-Ectodermal dysplasia-Cleft lip/palate syndrome	p63	Ectrodactyly (split hand/foot), deafness, nail dystrophy, palmoplantar keratoderma, cleft lip/palate, sparse hair
Ankyloblepharon filiforme adenatum-Ectodermal dysplasia-Cleft palate syndrome	SAM domain of p63	Erosive scalp dermatitis, ankyloblepharon, bacterial infections, hypotrichosis, cleft lip/palate, abnormal granulation tissue
Down syndrome	Trisomy 21	Small ears, epicanthic folds, upslanting palpebral fissures, scrotal tongue, simian crease, mental retardation, heart disease
Hurler syndrome	α-L-iduronidase	Coarse facies, short stature, thick skin, macroglossia, macrocephaly, mental retardation, deafness, heart disease, hepatosplenomegaly
Gorlin syndrome	PTCH1	multiple BCCs, palmoplantar pits, odontogenic keratocysts, frontal bossing, bifid ribs, calcification of falx cerebri
Lipoid proteinosis	ECM1	Yellow papules on face and neck, eyelid "string of pearls", patchy alopecia, hoarse cry, temporal and hippocampal calcification
Progeria	lamin A	Atrophic skin, prominent scalp and thigh veins, loss of subcutaneous fat, mottled hyperpigmentation, sclerodermoid changes on lower trunk / thigh, absent scalp hair, severe atherosclerosis, dystrophic nails, large cranium, frontal / parietal bossing, small ears without lobules, increased urinary hyaluronic acid
Rothmund-Thomson syndrome	RecQL4	Poikiloderma, alopecia, cataracts, skeletal abnormalities, hypogonadism, sarcomas

Pegged teeth

Associated diseases	Gene / defect	Other findings
Hypohidrotic ectodermal dysplasia	EDA , EDA receptor, NEMO	see above
Incontinentia pigmenti	NEMO	see above

Natal teeth

Associated diseases	Gene / defect	Other findings
Pachyonychia congenita type 2	Keratins 6b / 17	Trachyonychia, palmoplantar keratoderma, steatocytomas, epidermal cysts

Retention of primary teeth

Associated diseases	Gene / defect	Other findings
Hyper-IgE syndrome	STAT3	Increased serum IgE, cold abscesses, eosinophilia, coarse facies, eczematous dermatitis, osteopenia, respiratory infections

Polydontia

Associated diseases	Gene / defect	Other findings
Gardner syndrome	APC	Epidermoid cysts, osteomas, congenital hypertrophy of the retinal pigment epithelium, GI adenocarcinomas, hepatoblastoma, papillary thyroid carcinoma

Erythrodontia

Associated diseases	Gene / defect	Other findings
Congenital erythropoietic porphyria	Uroporphyrinogen III cosynthase	Severe photosensitivity, hypertrichosis, red urine, bullae with scarring

Pigmentation

Associated diseases	Gene / defect	Other findings
Tetracycline therapy	-	Tetracycline favors the gingival third of the tooth, minocycline favors the middle third
Alkaptonuria	Homogentisic acid oxidase	Dark urine; hyperpigmenation of skin, sclera, and cartilage of ears, nose, and tendons; black cerumen; arthropathy

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Teeth (continued)

Charya By, MD, and Matt Steadmon, MD

Enamel hypoplasia

Associated diseases	Gene / defect	Other findings
Tuberous sclerosis	TSC1 / TSC2	Ash-leaf macules, angiofibromas, café-au-lait macules, retinal hamartomas, renal angiomyolipomas, cardiac rhabdomyomas, pulmonary lymphangioleiomyomatosis
Sjögren-Larson syndrome	FALDH	Generalized ichthyosis involving flexures, pruritus, palmoplantar keratoderma, spastic ditetraplegia, mental retardation, perifoveal "glistening dots"
Phenylketonuria	phenylalanine hydroxylase	Generalized hypopigmentation, blonde hair, blue eyes, eczematous dermatitis, sclerodermoid changes, mental retardation, urine with mousy odor
Morquio syndrome	hexosamine 6-sulfatase, β-galactosidase	Thick skin, joint laxity, lumbar lordosis, short stature, heart disease
Tricho-dento-osseous syndrome	DLX3	Kinky "steel wool" hair, osteosclerosis of base of skull and mastoid, brittle nails
Incontinentia pigmenti	NEMO	see above
Junctional epidermolysis bullosa - Herlitz	Laminin 5	Widespread bullae, nonhealing granulation tissue, nail dystrophy, mucosal involvment, early death

Notched incisors or mulberry molars

Associated diseases	Gene / defect	Other findings
Congenital syphils (late)	-	Saber shins, rhagades of lips, saddle nose, Higoumenaki's sign, corneal scars, Clutton joints

Periodontitis with early tooth loss

Associated diseases	Gene / defect	Other findings
Papillon-Lefèvre syndrome	Cathepsin C	Palmoplantar keratoderma with transgradiens, psoriasiform skin lesions, calcification of falx cerebri / tentorium cerebelli
Ehlers-Danlos syndrome, hypermobility type	unknown	Hyperextensible skin, ecchymoses, pretibial scarring
Naegeli-Franceschetti-Jadassohn syndrome	Keratin 14	Reticulated pigmentation, palmoplantar hyperhidrosis, palmoplantar keratoderma, absence of dermatoglyphs
Haim-Monk syndrome	Cathepsin C	Palmoplantar keratoderma, onychogryphosis, acro-osteolysis

Floating teeth

Associated diseases	Gene / defect	Other findings
Letterer-Siwe disease	-	Diffuse dermatitis in seborrheic-like distribution
Papillon-Lefèvre syndrome	Cathepsin C	

Delayed eruption of primary teeth

Associated diseases	Gene / defect	Other findings
Progeria	Lamin A	see above

Dental caries

Associated diseases	Gene / defect	Other findings
Dyskeratosis congenita	DKC1, TERC	Reticulated pigmentation, premalignant leukoplakia, fanconi type anemia, palmoplantar hyperhidrosis, palmoplantar keratoderma, absence of dermatoglyphs
Kindler syndrome	KIND1 / FERM	acral blistering, photosensitivity, poikiloderma, palmoplantar hyperkeratosis, wrinkling of hands / feet, phimosis, pseudoainhum
Recessive dystrophic epidermolysis bullosa	Collagen VII	Mitten deformity, generalized bullae, mucosal erosions, dystrophic nails, short stature
Cockayne syndrome	ERCC6 / ERCC8	Subcutaneous fat loss, aged appearance, thin nose, large ears, progressive neurologic deterioration, deafness, retinitis pigmentosa, basal ganglia calcification

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