

Genoderm Buzzwords

By Sara Brooks, M.D.

BUZZWORD	GENODERM
Eye buzzwords	
Comma shaped corneal opacities	X-Linked Ichthyosis
Perifoveal glistening white dots	Sjogren-Larsson Syndrome
Retinitis pigmentosa with salt and pepper retinal pigmentation	Refsum Disease Cockayne Syndrome
Dendritic keratitis with corneal ulceration	Richner-Hanhart Syndrome
Tyrosine crystals on slit-lamp examination	Richner-Hanhart Syndrome
Lisch nodules (iris hamartoma)	Neurofibromatosis Type I
Optic gliomas	Neurofibromatosis Type I
Juvenile posterior subcapsular lenticular opacities	Neurofibromatosis Type II
Retinal hamartoma	Tuberous Sclerosis
Retinal hemangioblastoma	Von Hippel Lindau
Upward lens displacement	Marfan Syndrome
Downward lens displacement	Homocystinuria
Angioid Streaks	Pseudoxanthoma Elasticum
Congenital hypertrophy of the retinal pigment epithelium (CHRPE)	Gardner Syndrome
Lester Iris (hyperpigmentation of pupillary margin of iris)	Nail Patella Syndrome
Heterochromia irides	Waardenburg Syndrome
Blue grey sclera	Alkaptonuria
Blue sclera	Osteogenesis Imperfecta (Type I, III, IV); Ehlers-Danlos Syndrome
Corneal opacities in whorl-like configuration	Fabry Disease
Cherry Red Spot	Niemann-Pick Disease
Optic Atrophy	Multiple Carboxylase Deficiency
Dystopia Canthorum	Waardenburg Syndrome (Type I, III, IV)
Kayser-Fleischer Ring (calcium deposition in Decemet's Membrane of the cornea)	Wilson Disease
Musculoskeletal buzzwords	
Stippled epiphysis	Conradi-Hunermann Syndrome
Polyostotic fibrous dysplasia	McCune-Albright Syndrome
Osteopathia striata	Goltz Syndrome (Focal Dermal Hypoplasia)
Osteopoikilosis	Buschke-Ollendorff Syndrome
Bifid Ribs	Gorlin Syndrome
Dysostosis multiplex	Mucopolysaccharidoses (Hurler's, Scheie's, Hunter's, Maroteaux-Lamy's)
Ehrlenmeyer Flask deformity	Gaucher Disease
Posterior iliac horns	Nail Patella Syndrome
Absent/hypoplastic patella	Nail Patella Syndrome
Lobster claw deformity	Ectrodactyly-Ectodermal Dysplasia-Cleft lip/palate
Metaphyseal widening	Menkes Disease
Wormian bones in sagittal and lambdoid sutures	Menkes Disease
Radial ray defects(absent/hypoplastic radius and thumbs)	Rothmund-Thompson Syndrome Fanconi Anemia
Acro-osteolysis	Haim-Munk Disease
Multiple enchondromas	Maffucci Syndrome
Hemihypertrophy	Beckwith-Wiedemann Syndrome
Broad thumbs	Rubinstein-Taybi Syndrome
Tufted terminal phalanges	Hydrotic Ectodermal Dysplasia
Scissor gait	Sjogren-Larson Syndrome
Occipital Horns (exostosis at insertion of trapezius and sternocleidomastoid muscles)	Occipital Horn Syndrome
Oral buzzwords	
Hypodontia/Anodontia	Ectrodactyly-Ectodermal dysplasia-Cleft lip/palate Ankyloblepharon-Ectodermal Dysplasia-Cleft lip/palate Hypomelanosis of Ito Incontinentia Pigmenti
Peg teeth/Conical Teeth	Hypohidrotic Ectodermal Dysplasia Incontinentia Pigmenti
Jaw cysts	Gorlin Syndrome
Enamel pits	Junctional Epidermolysis Bullosa Tuberous Sclerosis
Retained primary teeth (double row of teeth)	Hyper IgE Syndrome
Thickened lips	Multiple Endocrine Neoplasia Syndrome Type IIb
Natal teeth	Pachyonychia Congenita Type II
Cobblestoned oral mucosa	Cowden Syndrome
Wooden tongue	Lipoid Proteinosis
Grimacing smile	Rubinstein-Taybi Syndrome
Oral Leukoplakia	Pachyonychia Congenita Type I (not premalignant) Dyskeratosis Congenita (pre-malignant)
Gingival Fibroma	Tuberous Sclerosis



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CNS buzzwords	
Temporal and hippocampal calcification	Lipoid proteinosis
Falx cerebri calcification	Gorlin Syndrome
Dural calcification	Papillon-Lefevre Syndrome
Basal ganglia calcification	Cockayne Syndrome
Sphenoid wing dysplasia	Neurofibromatosis I
Tram-track calcifications beneath leptomeningeal lesions	Sturge-Weber Syndrome
Frontal Bossing	Gorlin Syndrome Conradi-Hunermann Syndrome Progeria Syndrome Rothman-Thompson Syndrome Hypohidrotic Ectodermal Dysplasia
Urine buzzwords	
Maltese Cross under polarized light	Fabry Disease
Mousy Odor	Phenylketonuria
Dark Urine	Alkaptonuria
Heme buzzwords	
Crumpled tissue paper macrophages	Gaucher Disease
Foam cells on bone marrow biopsy	Niemann-Pick Disease
NBT (nitroblue tetrazolium) reduction assay	Chronic Granulomatous Disease
Cold abscesses	Hyper IgE Syndrome
Absent thymic shadow	Severe Combined Immunodeficiency
Sexual buzzwords	
Precocious puberty	McCune-Albright Syndrome
Calcifying Sertoli-cell (testicular) tumor	Carney Complex
Other buzzwords	
Low-pitched cry at birth	Cornelia de Lange Syndrome
Hoarse cry at Birth	Lipoid Proteinosis
Eyelid string of pearls	Lipoid Proteinosis
Linear earlobe crease	Beckwith-Wiedemann Syndrome
Pain insensitivity	Riley-Day Syndrome
Left-sided cardiomyopathy	Carvajal Syndrome
Right-sided cardiomyopathy	Naxos Disease
Exuberant granulation tissue	Junctional epidermolysis bullosa, Herlitz type