Lumps, Bumps and Spots of the Skin:

*When there is more than meets the eye*

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Aplasia cutis

- Congenital defect
  - Localized absence epidermis, dermis ± subcutaneous tissue
- Scalp, face, trunk, extremities
- Clinical diagnosis
  - Erosion, glistening membrane ("membranous"), well-formed scar
  - Hair collar sign
- Sporadic
- Potential associations
  - Teratogens, limb abnormalities, epidermal nevi, underlying embryologic malformations, epidermolysis bullosa, malformation syndromes, infections
- Etiology unknown

Aplasia cutis

- **Scalp** – most common location
  - 70% scalp lesions are isolated, 20% double, 8% 3+ defects
  - 80% occur in close proximity to hair whorl

- **Complications (rare)**
  - Hemorrhage, venous (sagittal sinus) thrombosis, meningitis

- **Associated defects (rare)**
  - Cleft lip/palate, ophthalmologic defects, limb reduction defects, cardiac anomalies, GI tract malformations, spinal dysraphism, hydrocephalus, defects underlying skull, congenital midline porencephaly, spastic paralysis, seizures, mental retardation, vascular anomalies

- **Prognosis**
  - Excellent with proper wound care for larger lesions
  - Large and/or deep lesions may require surgery with grafting to prevent potential complications

Dermoid cyst

- Nontender, firm subcutaneous nodule, fixed to bone
- Most common location – orbital ridge
  - No association with deep extension
- Nasal midline (3%)
  - Potential for deep extension/CNS communication
- Differential diagnosis
  - Midline - Encephaloceles, gliomas, hemangiomas
  - Eyebrow ridge – epidermal cyst, pilomatricoma
- Imaging - MRI or CT for midline location
- Treatment - Surgical excision

Dermal sinus

- Developmental epithelium-lined tract or cyst
  - Extends inward from surface of skin
- Majority seen in occipital and lumbosacral regions
  - Midline fusion of ectodermal and neuroectodermal tissue occurs at cephalic and caudal ends of neural tube
- Difficult to visualize
  - Localized thickening of scalp, hypertrichosis, dimpling in midline of neck/back
- Complications - abscesses, osteomyelitis, meningitis
- MRI or CT - underlying lesion and/or CNS connection
- Treatment – surgery
Branchial cleft cyst/sinus

- Formed along course of first and second branchial clefts
  - Improper closure during embryonic development
- Lower third of lateral aspect of neck near anterior border of sternocleidomastoid muscle
- Unilateral/bilateral
- Open to cutaneous surface or drain into pharynx
- Present as child or adult – recurrent inflammation

Treatment
  - Complete surgical removal or marsupialization when problematic
Thyroglossal cysts

- **Mucus production** into incompletely obliterated thyroglossal duct
- **Painless soft mass** moves upwards on swallowing and on protrusion of tongue
  - Lie close to hyoid bone in or near midline of neck
  - Located at any site along pathway of thyroid anlage
- **Complication**
  - Discharging sinus
    - Occurring spontaneously or after insufficient removal
  - Infection
- **Treatment** – surgery when problematic
Thyroglossal duct cyst

Branchial cleft cyst

Bronchogenic cyst
Preauricular pits, sinuses and tags

- Imperfect fusion of auditory tubercles of first two branchial arches
- Prevalence 0.5-1.0%
- Associations (rare)
  - Hearing or GU defects
  - branchio-oto, branchio-oto-renal syndromes, oculoauriculovertebral (Goldenhar), Treacher Collins
- Complications
  - Pits – infected, chronic ulcerations, retention cysts
- Treatment – surgical excision if problematic
**Midline skin stigmata**

**Risks**

- **Underlying mass**
  - Lipoma, meningomyelocele, hemangioma

- **Tethered cord syndrome**

- **Spinal dysraphism**
  - Incomplete fusion of midline mesenchymal, bony or neural elements of spine

- **Associated symptoms of spinal cord dysfunction**
  - Genitourinary and/or rectal problems
    - Recurrent infections or incontinence
  - Abnormal gait, abnormal arching or ulcers of feet

Guggisberg D *et al.* Arch Dermatol 2004;140:1109-1115
Midline Skin Stigmata

*Low risk*

- **Simple dimple**
  - Soft tissue depression <5mm diameter
  - up to 2.5cm above anus or within coccygeal proximity

- **Deviated gluteal fold**
  - Bifid, split symmetrical, any other degree of deviation without an underlying mass

- **Simple capillary malformation (port wine stain)**

- **Dermal melanocytosis (Mongolian spot)**

- **Light Hair**

Guggisberg D et al. Arch Dermatol 2004;140:1109-1115
Midline Skin Stigmata

High risk

- Nonsimple dimple
- Midline protrusion (lipoma)
- Pedunculated skin tag (human tail)
- Hypertrichosis – long, silky hair (faun tail)
- Giant congenital melanocytic nevus
  - Multiple satellite lesions
- Infantile hemangioma (segmental)
- Two or more congenital midline skin lesions

Guggisberg D et al. Arch Dermatol 2004;140:1109-1115
Midline skin stigmata

Management

Imaging
- Ultrasound
  - Limited by vertebral ossification relative to age
  - Up to 6 months
  - Operator dependent
- MRI
  - Gold standard for diagnostic screening
  - Costly, General anesthesia

Treatment - surgical

Guggisberg D et al. Arch Dermatol 2004;140:1109-1115
Nevus depigmentosus

- Hypopigmented macules or patches
  - Wood’s light – off-white accentuation
- Congenital, stable over time
- Sporadic
- Present before 3 years of age
  - Untanned skin, color contrast not readily visible
- Developmental defect of fetal melanocytes
  - Transfer of melanosomes from melanocytes to keratinocytes

Segmental Pigmentation Disorder

- Patch of hypopigmentation or hyperpigmentation
  - Segmental – localized, unilateral, well-circumscribed
  - Block-like patch with midline demarcation
  - Dermatomal, checkerboard
- Onset 3-4 months
- Risk of associated abnormalities is low
- Somatic mutation reflecting disparate pigmentation properties
- Torso most common location – extremities, face, neck

Frieden IJ, Hogeling M. Br J Dermatol 2010;162(6):1337-1341
Tuberous sclerosis

- Autosomal dominant
  - Up to 75% new mutation
  - TSC1 encoding hamartin
  - TSC2 encoding tuberin

- Hamartomas
  - Skin, brain, eye, heart, kidneys, lungs and bone

- Skin lesions
  - Hypopigmented macules, adenoma sebaceum, fibrous tumors, periungual and gingival fibromas

Narayanan V. Pediatr Neurol 2003;29(5):404-409
Tuberous sclerosis

**Systemic symptoms**

- **Seizures**
  - Focal/generalized seizures – 2-3 yrs

- **Developmental delay**
  - Correlates with age of seizures onset

- **Tubers – 50-75%**
  - Periventricular/subependymal nodules on CT/MRI
  - Not malignant, may cause obstructive hydrocephalus

- **Renal hamartomas (angiomyolipomas) 15%**
  - Loss of heterozygosity of TSC1 (or TSC2) gene

- **Multiple bilateral renal cysts**
Tuberous sclerosis

**Systemic symptoms**

- Cardiac rhabdomyomas
  - Infants, regress spontaneously
  - Usually asymptomatic – assoc CHF, murmurs, cyanosis, arrhythmias, sudden death
- Eyes – gliomas (phakomas) (50%)
- Cystic lesions in lungs
- Osseous manifestations (85%)
  - Cysts, periosteal thickenings
- Tooth pits
  - (punctate, round, oval, 1-2mm enamel defects)
Tuberous sclerosis

Management

- Monitor for cutaneous manifestations
- Family history
- Neurological evaluation
- Genetics evaluation/counseling
- MRI/CT brain
- Renal ultrasound
- Cardiac echocardiography
- Ophthalmologic examination
- Bone survey
- Chest radiography for honeycombing of lungs

www.tsalliance.org
Café au lait spots (CALS)

- Tan macules with distinct edges, 5mm-15cm
  - Increased melanin content in melanocytes & basal keratinocytes

Solitary CALS

- 3% infants
  - 25% healthy children

>3 CALS

- 0.2-0.3% children with no known evidence of underlying disorder

6+ CALS

- ~75% pts present to NF1 clinic meet criteria for NF1
  - Majority pts meet diagnostic criteria by 6 yrs

Neurofibromatosis I

Criteria

- 6 or more CALS
  - >5mm prepubertal, >15mm postpubertal
- 2 or more neurofibromas of any type
  or 1 plexiform neurofibroma
- Freckling in axillary or inguinal region (Crowe sign)
- Optic glioma
- 2 or more Lisch nodules (iris nevi)
- Distinctive osseous lesion
- First-degree relative with NF1

www.ctf.org

NIH diagnostic criteria for neurofibromatosis type 1 NF-1
(Consensus Development Conference of 1988)
Neurofibromatosis I

Evaluation and management

- Multidisciplinary NF1 clinic
  - Neurology, ophthalmology
  - Genetics - Molecular DNA mutation analysis – ch 17
  - Dermatology, Oncology

- Neuroimaging - controversial
  - Optic pathway or other gliomas (15%)
  - Distinctive hyperintense T2-weighted signals

- Whole body MRI
  - Clinically unapparent plexiform neurofibromas (paraspinal regions)

Riccardi VM. Arch Dermatol 2009; 145(8):929-930
Café au lait spots

Associated syndromes

- Legius syndrome
  - Mutation in *SPRED1* locus (ch. 15)
  - Phenotypic features similar to NF1
    - CALMs, +axillary/inguinal freckling
    - +family history, macrocephaly

- McCune-Albright

- Ring chromosome syndromes

- Watson syndrome

- Bloom syndrome

Messiaen L et al. JAMA 2009;302:2111-8
Lumps, bumps and spots

Summary

The majority of cutaneous lesions are only “skin deep”

Certain skin stigmata are markers for underlying involvement or signs of systemic disease

Midline lesions – consider CNS connection

White and tan spots alone do not make a diagnosis of a genodermatosis