Birthmarks
When to worry?

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No relevant conflict of interest

- Advisory Board member for :
- Penthrox
- Johnson&Johnson
- SanofiGenzyme
- Galderma

Outlines

- To discuss the most common birthmarks
- To discuss the natural course of these birthmarks
- To highlight red flags

Birthmark : Definition

- Birthmarks are defined as:
- Persistent visible marks on the skin that are evident at birth
- However may appear or first noticed long after birth (delayed birthmark)

Birthmarks

- Red
- Yellow
- Hypo or hyper pigmented
- Brown
- Blue

Red Birth marks

Case 1:

- Healthy newborn
- Red spots on glabella and upper eyelids

http://www.pcds.org.uk/ee/images/made/ee/images/uploads/clinical/salmonpatch_300_242_70.jpg

Nevus Simplex

- An extremely common birthmark
- Pale pink to bright red macules and patches with indistinct border
- Classic locations: nape, glabella, upper eyelids
- The nose, upper lip, occipital or parietal scalp are relatively frequent sites of involvement
- The lower lip, upper back, and lumbosacral skin can also be affected

Juern et al. J Am Acad Dermatol 2010;63:805-14

Nevus Simplex : natural course

- Transient capillary malformation so they usually resolve within 1-2 years
- On the glabella may take several years
- On the nape around 50% resolve
- Pulsed dye laser in persistent cases

When to worry?

- Usually not worrisome
- When prominent and persistent can be seen in some syndromes
- Usually the other signs and symptoms of associated syndrome are evident

Beckwith-Wiedemann syndrome	A congenital overgrowth syndrome w/ major features of anterior abdominal wall defects, macroglossia, and gigantism	Autosomal dominant pattern w/ incomplete penetrance in 15% of patients. Most cases are sporadic.
Nova syndrome	Familial communicating hydrocephalus, posterior cerebellar agenesis, and mega cisterna magna	Autosomal dominant pattern w/ incomplete expression
NS w/odontodysplasia	Defects in dentin pulp, enamel, and dental follicle; failure of full eruption of affected teeth	Nonhereditary
Macrocephaly–capillary malformation syndrome	Facial and limb asymmetry, somatic overgrowth, syndactyly or polydactyly, and developmental delay	Unknown
Roberts-SC syndrome	Symmetric limb defects, craniofacial abnormalities, prenatal and postnatal growth	Autosomal recessive

When to worry?

- If on the lumbosacral area and associated with other localized skin changes such as hypertrichosis, a dermal sinus or pit, lipoma, or deviated gluteal cleft :
- Rule out Occult dysraphism

When to worry?

- If on the scalp and associated with a nodule, aplasia cutis, a tuft of hair :
- An underlying neural tube closure defect, should be ruled out

Case 2

- Healthy newborn
- Red patch on left hemiface since birth

Port Wine Stain (PWS)

- Well-demarcated color of port wine patches
- Present at birth
- 0.3 % of all newborns
- Mostly seen on face and neck
- Usually unilateral and segmental (~85 %)

PWS

- Persistent through life
- May lighten over the first 3-6 months of life
- No proliferative phase in infancy
- Gradual darkening and thickening later in life

When to worry?

- Facial PWS involving forehead including upper eyelid
- Hemifacial PWS
- Mildline PWS
- Increased risk of Sturge Weber Syndrome
- Characterized by
  - Facial PWS
  - CNS involvement
  - Eye involvement

  - Refer to :
    Dermatology ,Ophthalmology ,Neurology

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Pediatric Dermatology, Volume: 35, Issue: 1, Pages: 30-42, First published: 16 October 2017, DOI: (10.1111/pde.13304)

PWS Treatment

- Pulsed Dye laser
- Early treatment may prevent thickening
- May need several treatment
- 70-80% lightening is considered a very good response

Case 3

- 3 months old baby
- Red mark on face
- Parents just noticed faint red mark at birth
- Lesion started to grew at 2 weeks of age

Infantile Hemangioma

- The most common vascular tumour of childhood (2.5-5% of newborns)
- Etiology:

- Caused by unsatisfied wishes of craving for strawberries during pregnancy

- Proliferation of immature endothelial cells stimulated by angiogenic factors

Infantile Hemangiomas: Morphological Classification

- Superficial type :Bright red papules, plaques, nodules
- Deep type :Subcutaneous, partially compressible bluish nodules
- Mixed type :Combined features of both

https://d1niluoi1dd30v.cloudfront.net/01406736/S0140673617X00287/S0140673616006450/gr4.jpg?Signature=SlIyFxsl-Pb9VN45%7ETZ9YHHSL03JaFAD4MwEvd39jOSv8cUsH1hoEIiY1arBYT6B-ZMncjKzwPci6TMKwlnsz4srb9RxRgo%7E1VShI0A8%7E2tywBHEF1peYKHdXkRAdEXpu3kZM87JUHEwVIiC-fylWGHWfTGz2d61QceDmeW--JU_&Expires=1543350975&Key-Pair-Id=APKAICLNFGBCWWYGVIZQ

Infantile Hemangioma

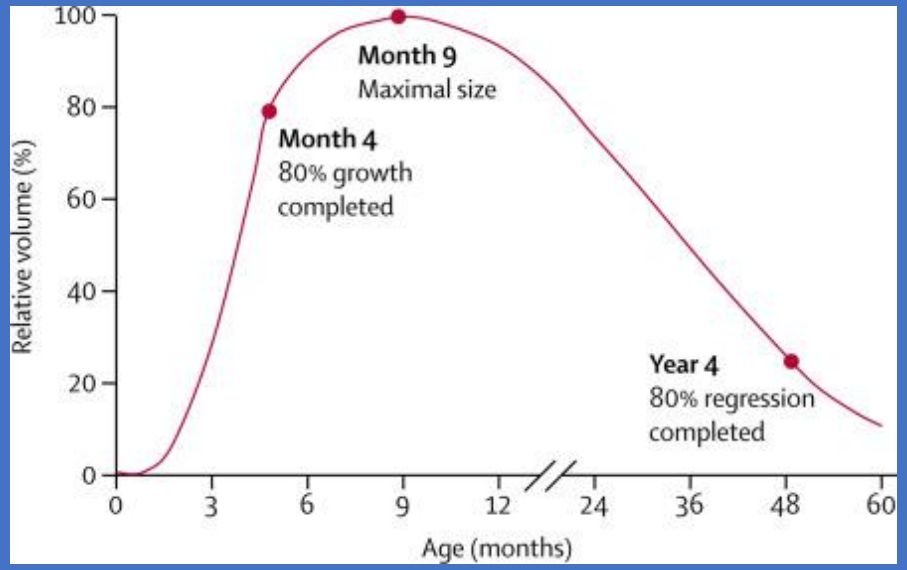
- Usually absent at birth

- Precursor lesion at birth in 30-50% of cases: bruise, ulcer, palor, telangiectasia

https://pocketdentistry.com/wp-content/uploads/285/B9781455753161000282_f028-001ac-9781455753161.jpg

Infantile Hemangiomas: Growth Characteristics

- Proliferative phase: Starts around 2-3 weeks
- Accelerated growth in first 3 months
- Plateau phase: Stable size
- Involution phase: Spontaneous regression over years
- 80% involution by 4 years



http://pediatrics.aappublications.org/content/pediatrics/122/2/360/F7.medium.gif

Infantile Hemangiomas: Involution

- Use caution with the term “will go away”
- Hemangioma disappears BUT skin is not always normal
- 59% of hemangiomas will leave a residual skin deformity
- Epidermal atrophy, dyspigmentation
- Telangiectasia, fibrofatty tissue and skin laxity

https://www.researchgate.net/profile/Swee_Tan2/publication/309665003/figure/fig3/AS:424548249149443@1478231598187/Serial-photographs-showing-a-typical-infantile-hemangioma-progression-at-8-months-of-age.png

When to worry?

- Most infantile haemangiomas regress spontaneously

- At risk of ulceration or ulcerated
- At risk of causing a significant functional impairment
- At risk of disfigurement
- Hemangioma associated with other abnormalities

Obstruction and functional impairment

http://www.reviewofophthalmology.com/CMSImagesContent/2013/6/RP-0613-Peds_Fig2_250W.JPG

http://www.reviewofophthalmology.com/CMSImagesContent/2013/6/RP-0613-Peds-Fig1_250W.JPG

Ulceration

- Large, superficial, and segmental haemangiomas more likely to ulcerate
- Lip, the head and neck area, and the intertriginous regions

Disfigurement

- Centrofacial hemangiomas including nose and lip haemangiomas

- Large haemangiomas affecting the breast area

http://www.e-ijd.org/articles/2013/58/4/images/IndianJDermatol_2013_58_4_330_113997_u1.jpg

Breast hemangioma

Theiler M, Hoffman WY, Frieden IJ. Breast hypoplasia as a complication of an untreated infantile hemangioma. Pediatr Dermatol 2016; 33: e129–e130.

Multiple hemangioma

- Any patient with ≥5 hemangiomas
- Liver hemangioma should be ruled out
- Abdominal ultrasound

http://www.hemangiomaeducation.org/images/thumbnails/50_300.jpg

Lumbosacral/Perineal hemangioma

- Spinal cord abnormalities
- Urogenital abnormalities
- Anorectal abnormalities

http://www.hemangiomaeducation.org/images/examples/M-2.jpg

Large facial hemangioma

http://www.chw.org/~/media/Images/MedicalCare/birthmarksandvascularanomalies/PHACE%201.png

http://sketchymedicine.com/wp-content/uploads/2014/02/PHACE_syndrome-400x322.jpg

Beard distribution

http://www.hemangiomaeducation.org/images/thumbnails/TERN69.jpg

Yellow birth marks

Case 4

- Newborn with yellow orange alopetic lesion on scalp

Sebaceous nevus

- Occurs in 0.3% of newborns
- Mostly seen on the scalp or face
- Solitary, oval to linear smooth yellow/orange hairless patch
- Can get bumpy, warty, scaly at puberty

Sebaceous nevus: natural course

- Very low risk of malignant transformation
- As long as it does not change, treatment is typically not required

Sebaceous nevus : when to worry?

- Suspicious changes including :Focal growth /ulceration
- Extensive Sebaceous nevus

NS syndrome

- Association of extensive naevus sebaceus with
- CNS abnormalities
- Ocular anomalies
- Skeletal defects
- Hypophosphatemic vitamin D–resistant rickets with increased growth factor 23 (FGF23) levels

Brown birthmarks

Case 5

- Newborn with dark black lesion

https://www.consultant360.com/articles/giant-congenital-melanocytic-nevus

Congenital melanocytic nevus

- Present at birth or appear in first 2 years of life (tardive)
- Very common
- 1-3% of newborns

Classification

- Depends on projected adult size
- Small (< 1.5 cm)
- Medium (1.5–20 cm)
- Large (20-40 cm)
- Giant (> 40 cm)

Projected adult size

- Multiply the diameter at infancy by:
- 1.7 for head lesions
- 2.8 for hands, feet, torso, forearms, arms, hips
- 3.4 for thigh lesions
- 3.3 for leg lesions

Natural course

- Proportional growth to the child growth

Pediatr Dermatol. 2018 Sep;35(5):690-692. doi: 10.1111/pde.13540. Epub 2018 Jun 22.

Grow coarse dark hair

http://bp2.blogger.com/_iCcaHsxVvTs/SDRea92hyUI/AAAAAAAAANs/7xroQ5k6TbY/s1600/congenital+melanocytic+nevus2.gif

Get raised, darkened with pebbly, verrucous surface

Can it turn to skin cancer?

- Lifetime risk of developing melanoma in general :1-2%
- Large congenital nevi: lifetime risk of 5%
- Small/intermediate congenital nevi : 1% lifetime risk (mostly after puberty)

Management
Small/medium CMN

- Excision can be considered for aesthetic improvement or in case of suspicious changes

Prophylactic removal of benign appearing/behaving small and medium CMN to* prevent cancer *is no longer warranted.

Large congenital melanocytic nevi

- Excise or not?
- Case by case
- No good evidence that excision decreases the risk of melanoma
- Complete removal of all nevus cells impossible
- Risk of melanoma in CNS
- Aesthetic improvement ??

Neurocutaneous melanosis

- Proliferation of nevomelanocytes within CNS associated with CMN
- Can be seen with large CMN or multiple small to medium sized CMN
- Brain MRI in first 6 months : So refer to dermatology
- If involved : Risk of CNS melanoma

Congenital melanocytic nevi : when to worry and refer?

- Large and giant congenital melanocytic nevi
- 2 or more congenital nevi of any size
- Changing nevus but not all changing nevi are problematic

Case 6

- Neonate with brown spots on trunk

Café au lait Macule

- Well demarcated, uniformly darkened color and variable size macule/patch

- Although CALM are reminder of NF but non pathologic CALM are very common specially in skin of color

CALMs which need dermatology referral?

- 1-3 café au lait usually normal : Refer more than 3 although 6 or more is the criteria for NF
- Any number of café au lait and signs and symptoms suggestive of associated syndromes
- Darker café au lait with irregular borders

- https://assets.cureus.com/uploads/figure/file/52088/article_river_69b1e99001b011e99e4d893eed01a4b3-figure-2.png

Case 7
Hypo and hyperpigmented patterned birthmarks

Indian Pediatrics 2004;401

https://d3i71xaburhd42.cloudfront.net/178ed593eee4ef675a5ee0173569f4b8651f8b47/3-Figure2-1.png
Egyptian dermatology online journal vol 4 No.2;7 Dermatology 2008

Hypo and hyperpigmented patterned birthmarks

- They herald mosaicism
- Their genetic make up is different from surrounding skin
- Mutation in a single cell in a developing fetus
- Clonal expansion of mutated cell leads to patterned pigmentation

Pigmentary mosaicism

- Any systemic association?
- The incidence of systemic involvement is 4-30%
- The more widespread a pigmentary anomaly is, the higher risk of systemic problems
- A careful examination and review of systems should be performed and pertinent clinical findings should direct further evaluation
- Parents to be reassured that serious complications, if present, are typically evident clinically early in infancy

Case 8

Mongolian spot

- Hippocrates believed that a blow to the pregnant mother's abdomen manifests as a mark at the corresponding place in the newborn
- Entrapment of melanocyte in the dermis during migration from neural crest
- Present at birth or appear in first few weeks with fading in first few years
- Very common in African-American and Asian
- Most commonly on buttocks and sacrum

- Document these unusual birthmarks in the medical record
- Can be confused for bruises of child abuse

Mongolian spot: when to worry?

- Extensive Mongolian spots
- Association with inborn errors of metabolism
- Most commonly GM1 gangliosidosis and mucopolysaccharidosis typeI(Hurler’s disease)


THE SLIDE MAN – QUESTION MARK