Birthmarks

Fatemeh Jafarian MD.
Assistant professor
McGill University Health Center
70th Annual McGill Refresher Course for Family Physicians

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 discuss the referral criteria

Birthmark : Definition

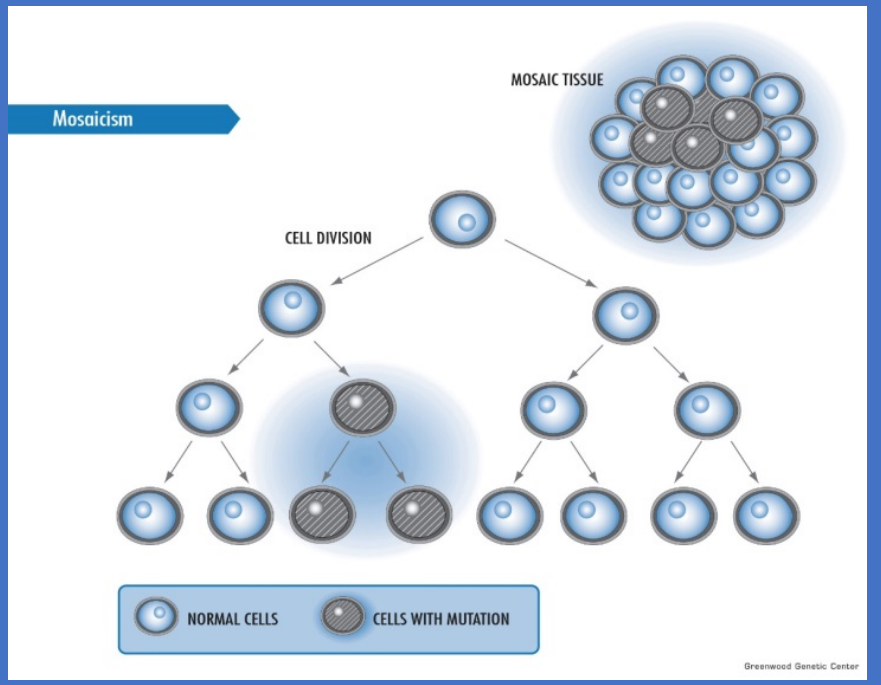
- A persistent visible mark on the skin that is evident at birth
- However may appear or first noticed long after birth (delayed birthmark)

Birthmarks :Myths

- When an expectant mother’s craving was not satisfied
- When an expectant mother experienced a lot of fear
- When an expectant mother touched her skin during a solar eclipse
- When a traumatic injury happened in the past life

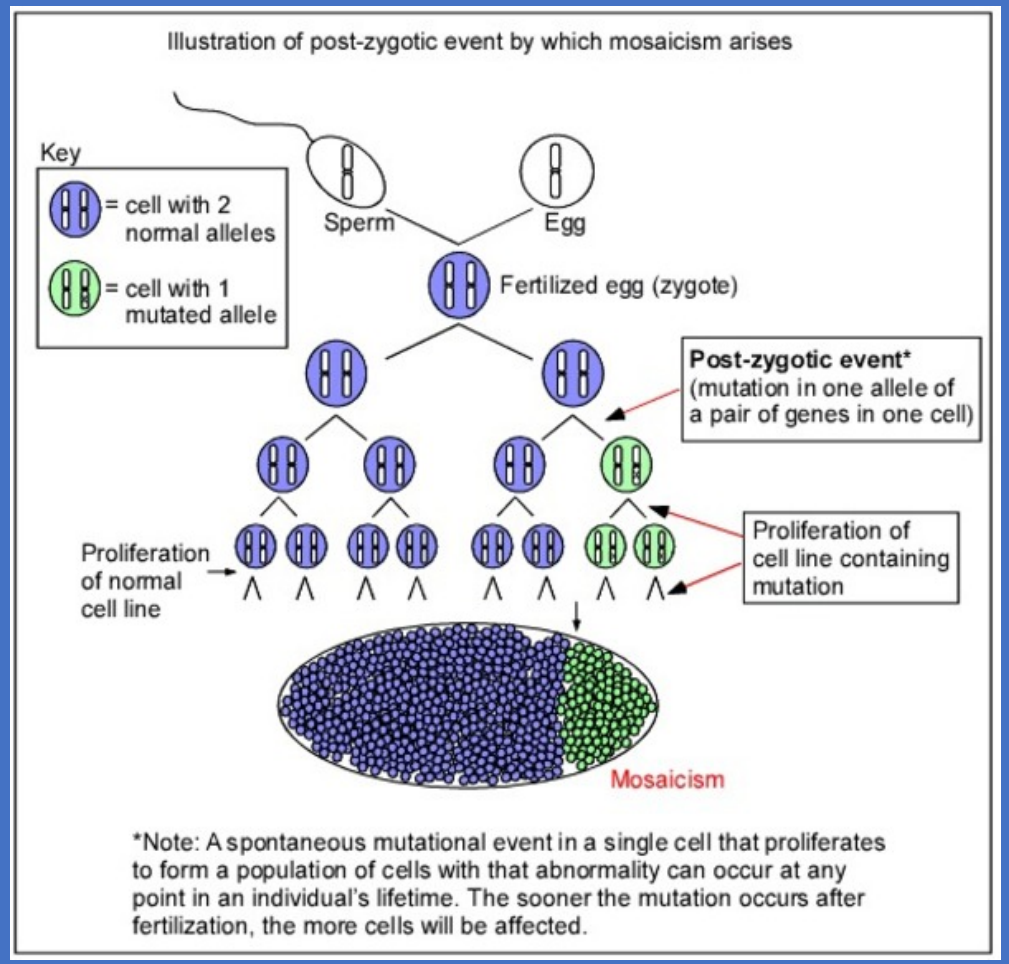
Birthmark :Facts

- Birthmarks herald mosaicism
- Genetic make up of birthmarks are different from surrounding skin



Birthmark :Mosaicism

- Mutation in a single cell in a developing fetus
- Clonal expansion of mutated cell
- Already confirmed for some birthmarks by genetic analysis



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

- Present at birth; often multiple
- Occurs in >15% neonates
- Pale pink to bright red macules and patches with indistinct border
- Classic locations: nape, forehead, glabella, upper eyelids

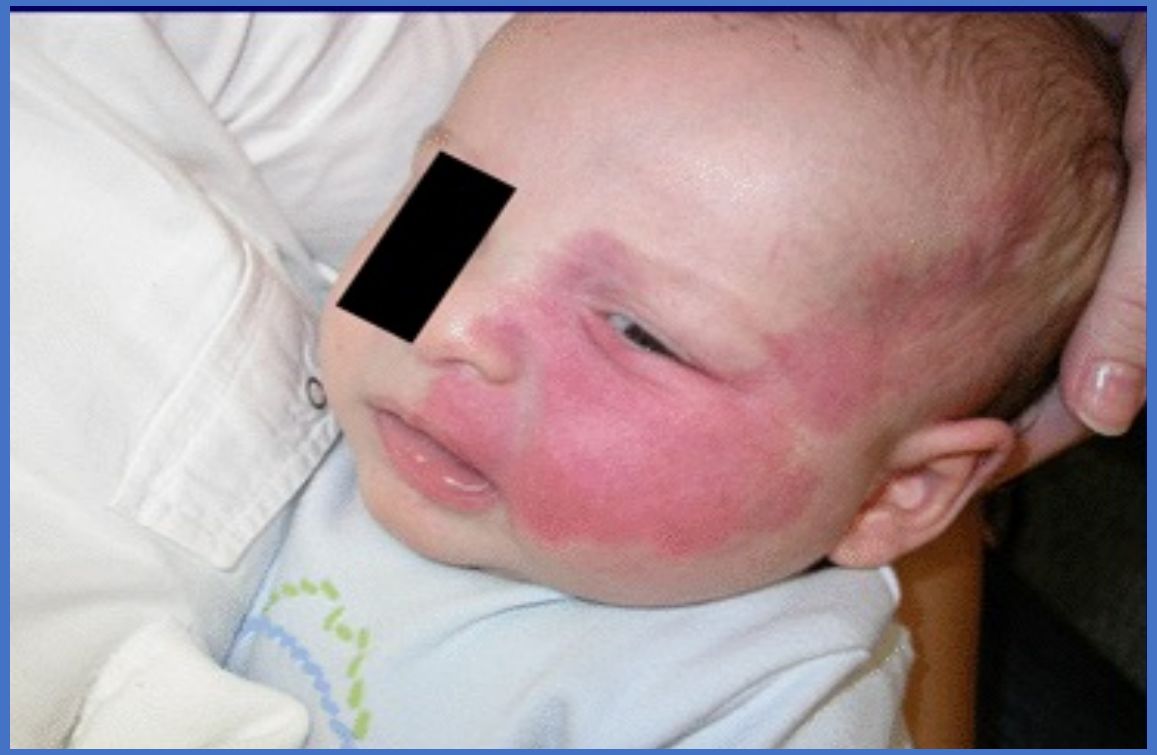
Juern et al. J Am Acad Dermatol 2010;63:805-14

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 recalcitrant cases

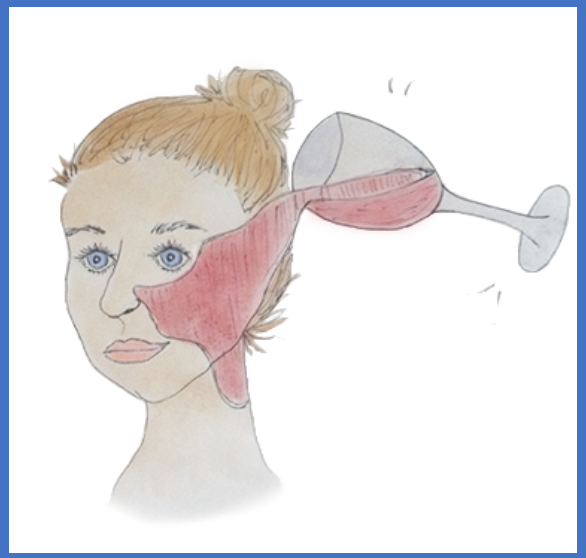
Case 2

- Healthy newborn
- Red patch on left hemi face



Port Wine Stain (PWS)

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

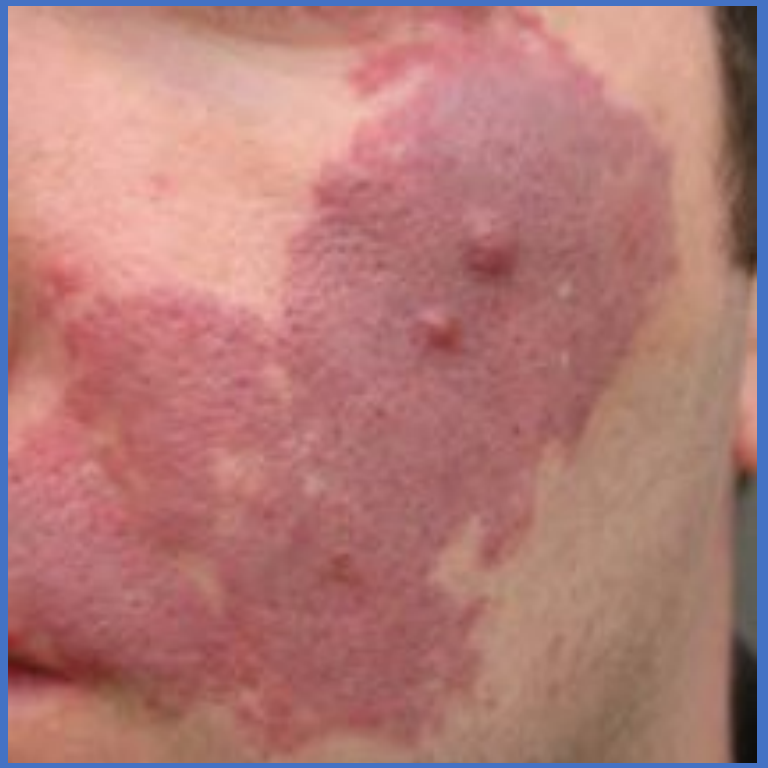


PWS

- 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

Any associations?

- Sturge-Weber syndrome

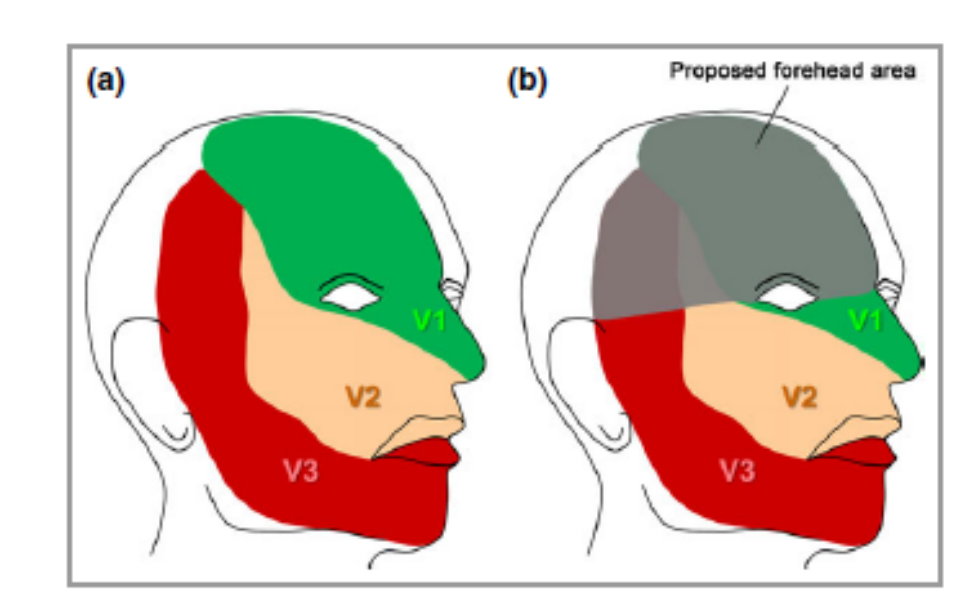


Sturge-Weber Syndrome

- Characterized by
  - Facial PWS
  - CNS involvement
  - Eye involvement

  - Refer to :

    Dermatology ,ophthalmology ,neurology

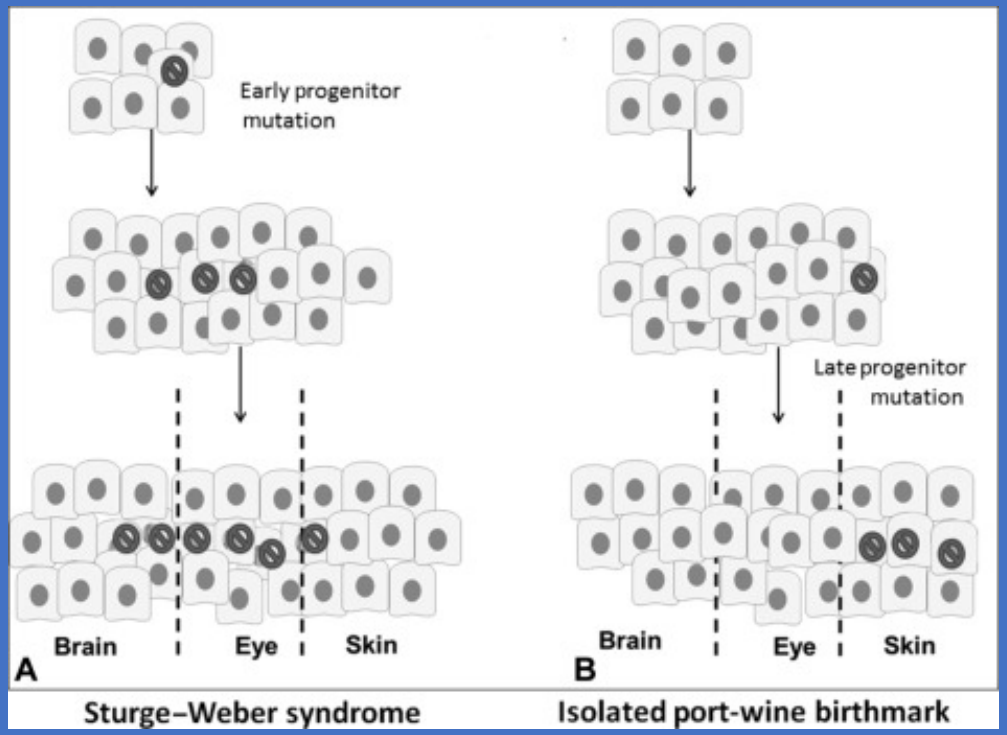


R. Waelchli et al. New vascular classification of port wine stains: improving prediction of Sturge-Weber risk. BJD 2014, Oct;171(4):861-7

ORIGINAL ARTICLE

Sturge–Weber Syndrome and Port-Wine Stains Caused by Somatic Mutation in *GNAQ*

- Genetic analysis of paired DNA samples from affected tissues (skin, brain) and matched unaffected
- A specific mutation in GNAQ in affected tissues
- The extent determined by timing of mutation



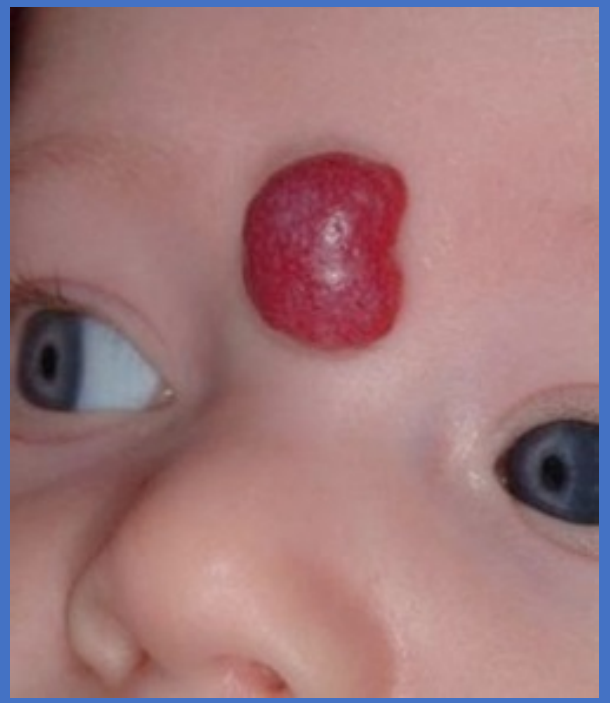
https://ars.els-cdn.com/content/image/1-s2.0-B9780444627025000111-f11-02-9780444627025.jpg

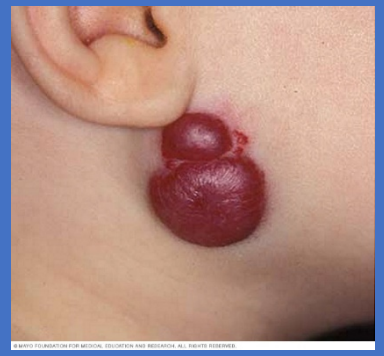
PWS Treatment

- 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

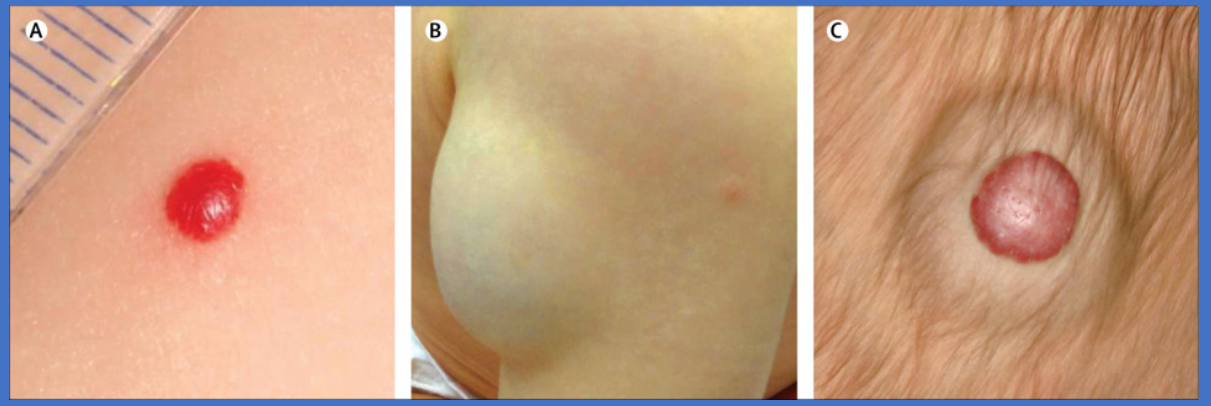
- The most common vascular tumour of childhood (2.5-5% of newborns)
- Etiology:

- Caused by unsatisfied wishes of craving for strawberries during pregnancy

- Proliferation of immature endothelial cells stimulated by angiogenic factors

Infantile Hemangiomas: Morphological Classification

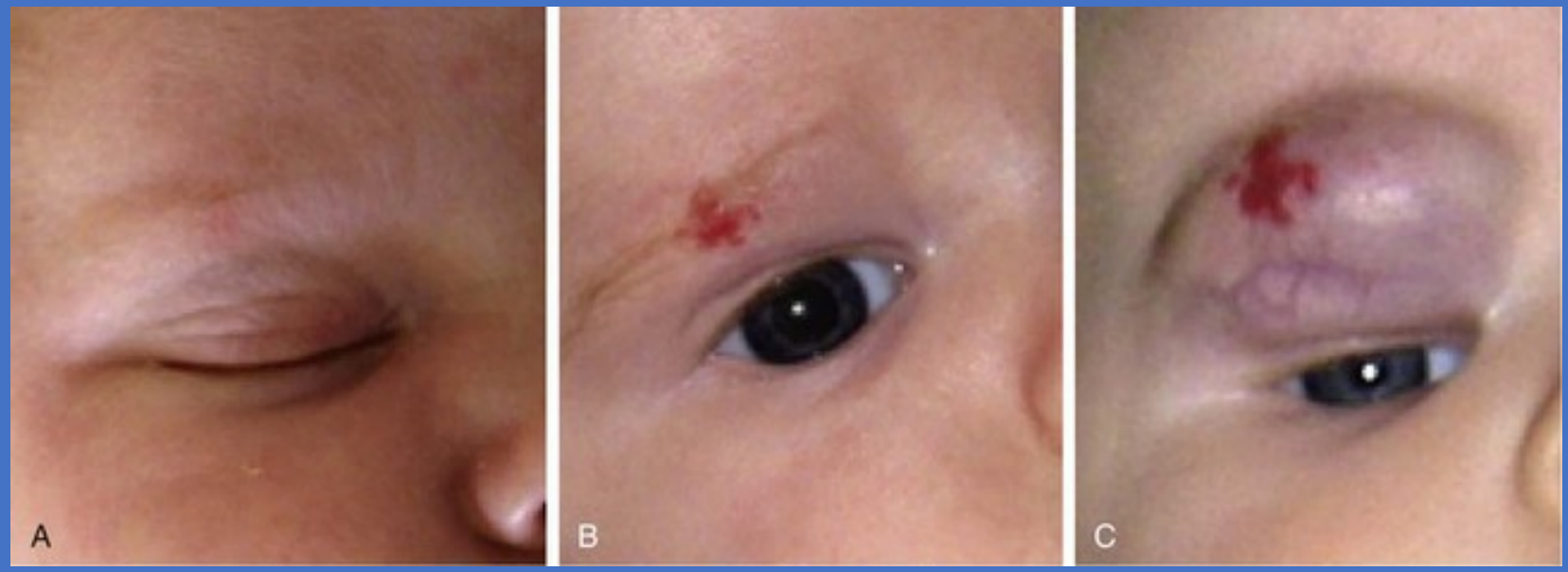
- Superficial type :Bright red papules, plaques, nodules
- Deep type :Subcutaneous, partially compressible bluish nodules
- Mixed type :Combined features of both



https://d1niluoi1dd30v.cloudfront.net/01406736/S0140673617X00287/S0140673616006450/gr4.jpg?Signature=SIIyFxsl-Pb9VN45%7ETZ9YHHSL03JaFAD4MwEvd39jOSv8cUsH1hoEliY1arBYT6B-ZMncjKzwPci6TMKwlnsz4srb9RxRgo%7E1VShI0A8%7E2tywBHEF1peYKHdXkRAdEXpu3kZM87JUHEwVIiC-fylWGHWfTGz2d61QceDmeW--JU_&Expires=1543350975&Key-Pair-Id=APKAICLNFGBCWWYGVIZQ

Infantile Hemangioma

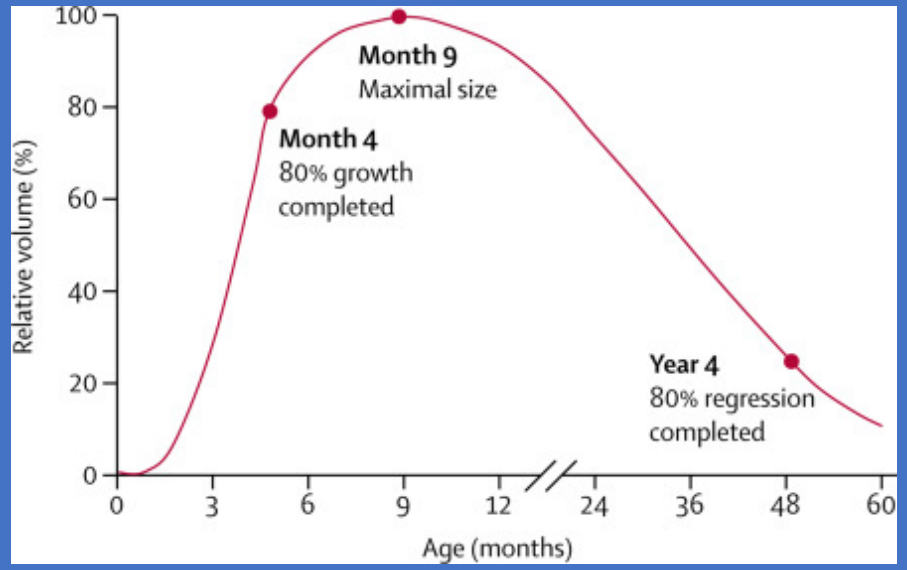
- Usually absent at birth
- Precursor lesion at birth in 30-50% of cases: bruise, ulcer, palor, telangiectasia



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

Infantile Hemangiomas: Growth Characteristics

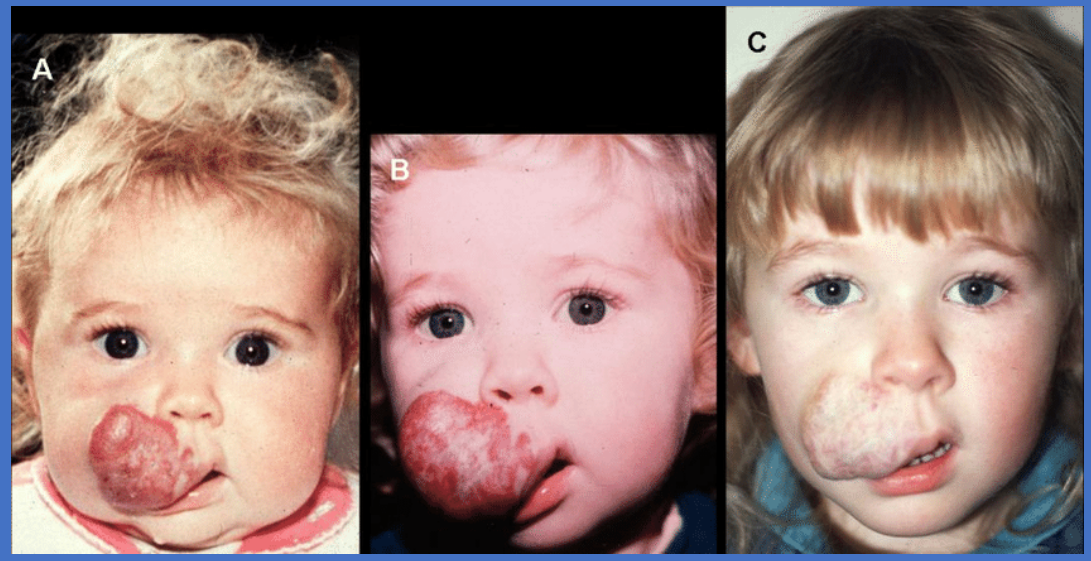
- Proliferative phase: Starts around 2-3 weeks
- Accelerated growth in first 3 months
- Plateau phase: Stable size
- Involution phase: Spontaneous regression over years
- 80% involution by 4 years

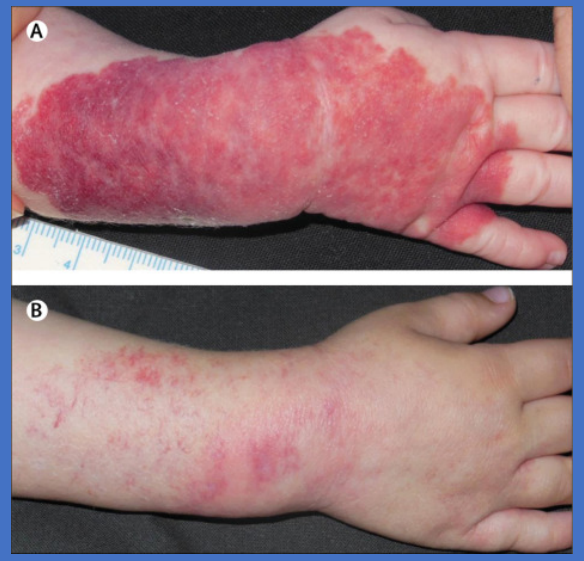


http://pediatrics.aappublications.org/content/pediatrics/122/2/360/F7.medium.gif

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 skin laxity





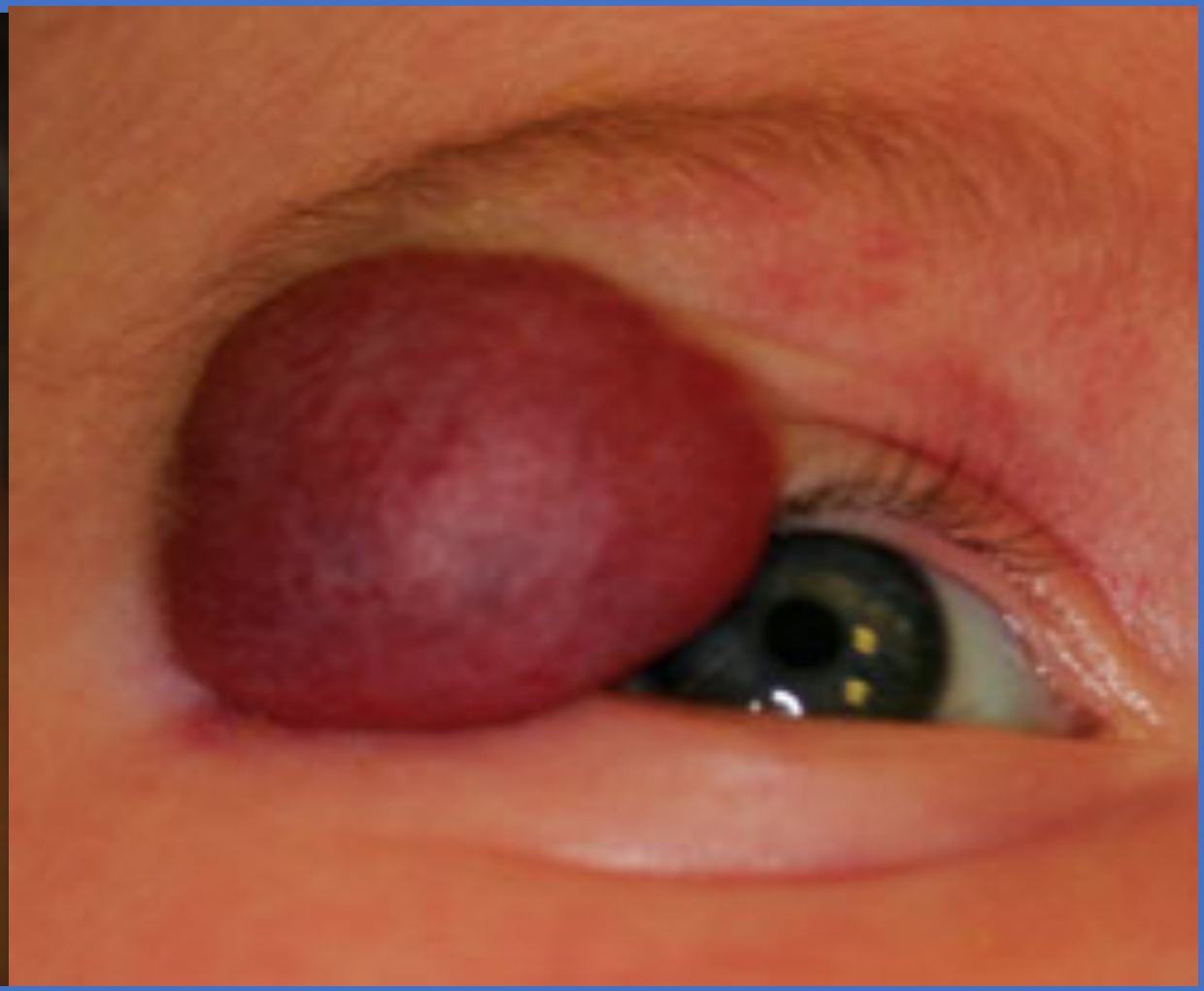
https://www.researchgate.net/profile/Swee_Tan2/publication/309665003/figure/fig3/AS:424548249149443@1478231598187/Serial-photographs-showing-a-typical-infantile-hemangioma-progression-at-8-months-of-age.png

Hemangioma: management ?

- Most infantile haemangiomas regress spontaneously

- **Referral and treatment is required when:**
- 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 face
- Large haemangiomas affecting the chest area in girls
- Nose and lip haemangiomas frequently have incomplete spontaneous regression

Breast hemangioma

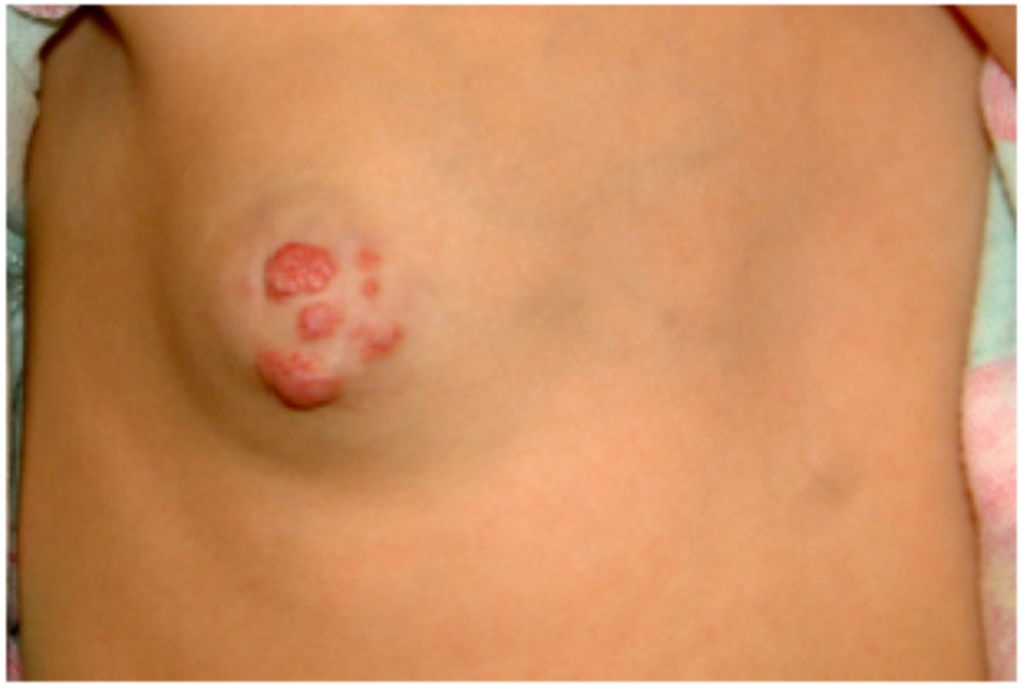


Figure 1. Mixed infantile hemangioma at 3 years of age involving the areola and underlying soft tissue.

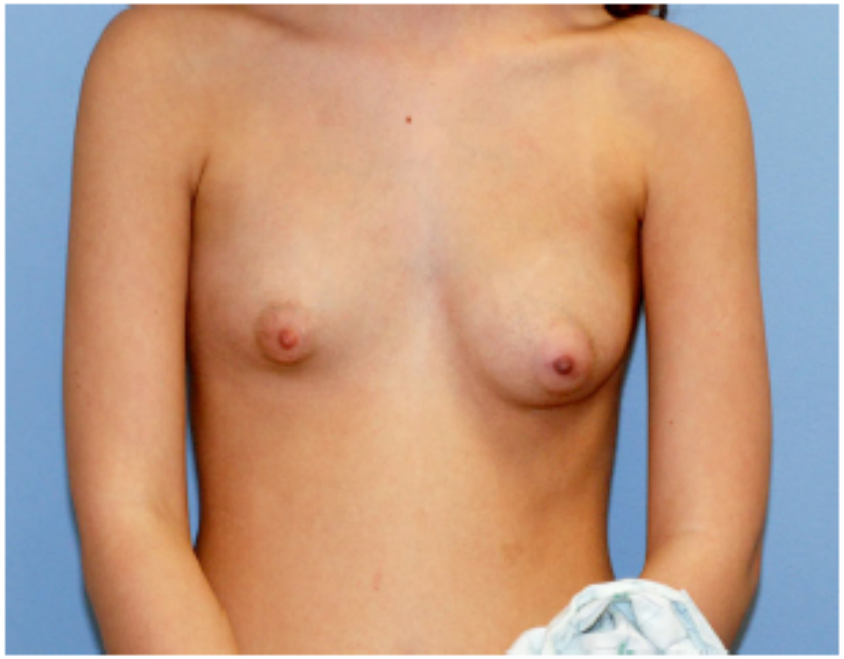
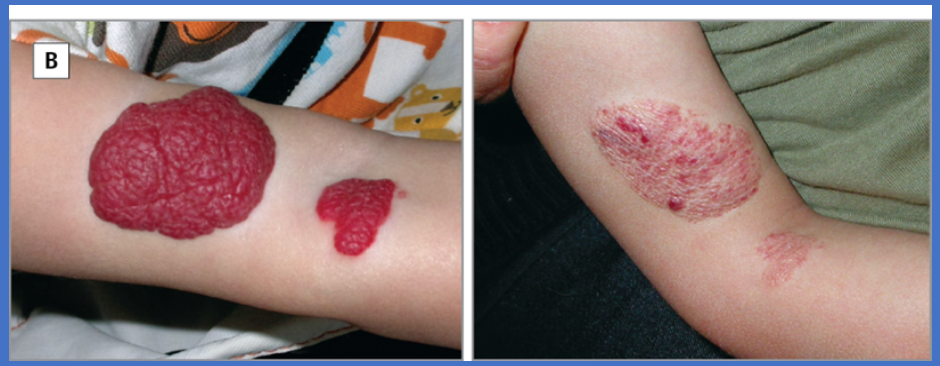


Figure 2. Marked hypoplasia of the right breast at age 14 years.

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

Large Exophytic Hemangioma

- Abrupt steep border
- Cobblestoned surface

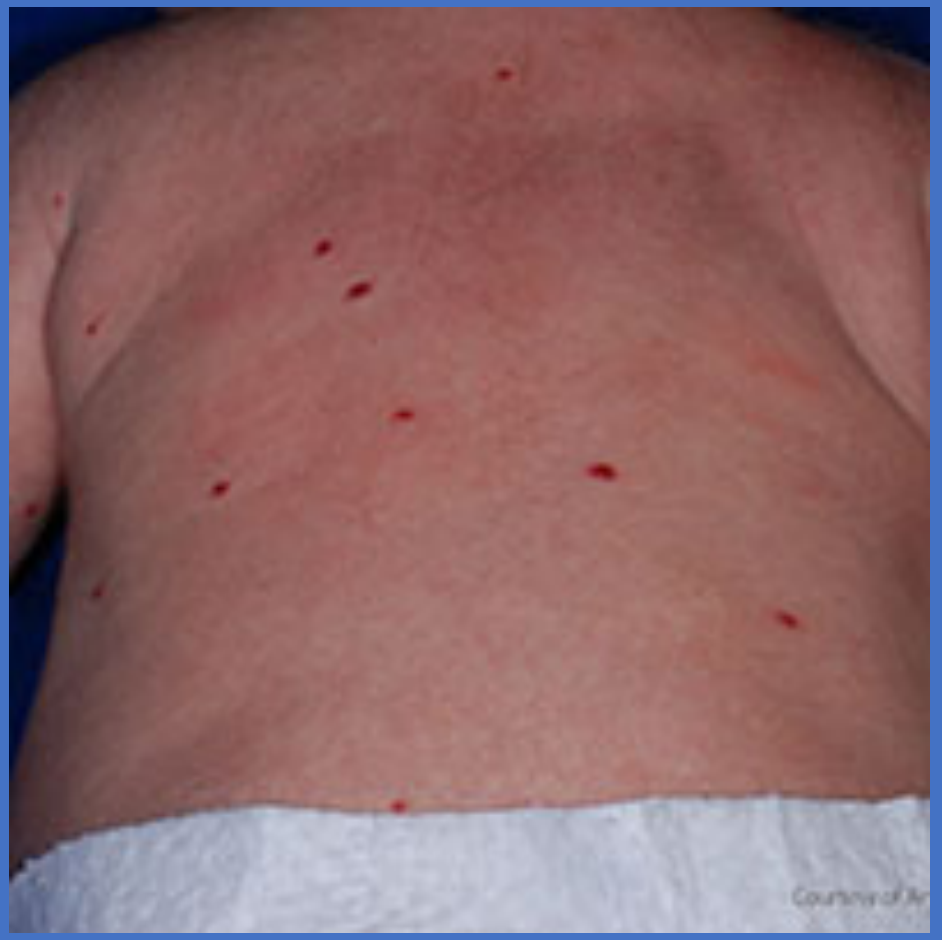


https://jamanetwork-com.proxy3.library.mcgill.ca/data/Journals/DERM/935847/m_doi160042f1.png

Association with systemic problems

Multiple hemangioma

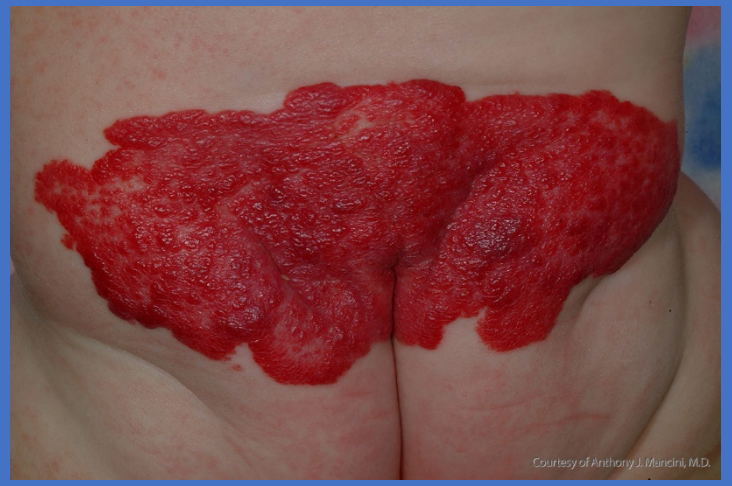
- Any patient with ≥5 hemangiomas
- Thorough physical examination
- Abdominal ultrasound
- Other diagnostic evaluations, as clinically indicated.

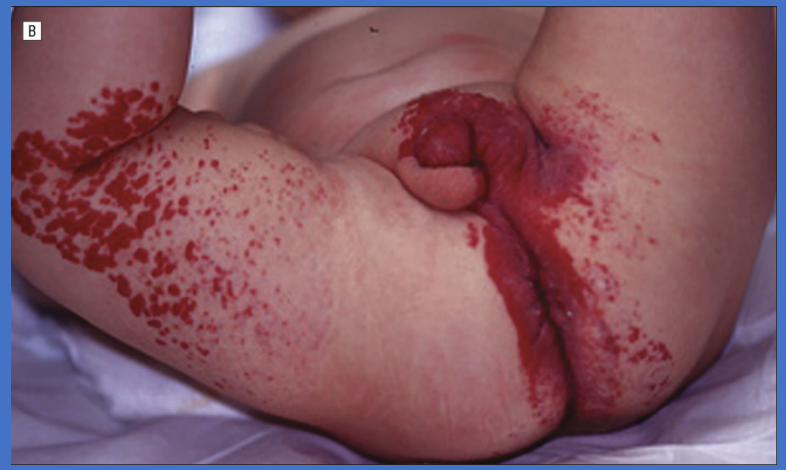


http://www.hemangiomaeducation.org/images/thumbnails/50_300.jpg

Lumbosacral/Perineal hemangioma

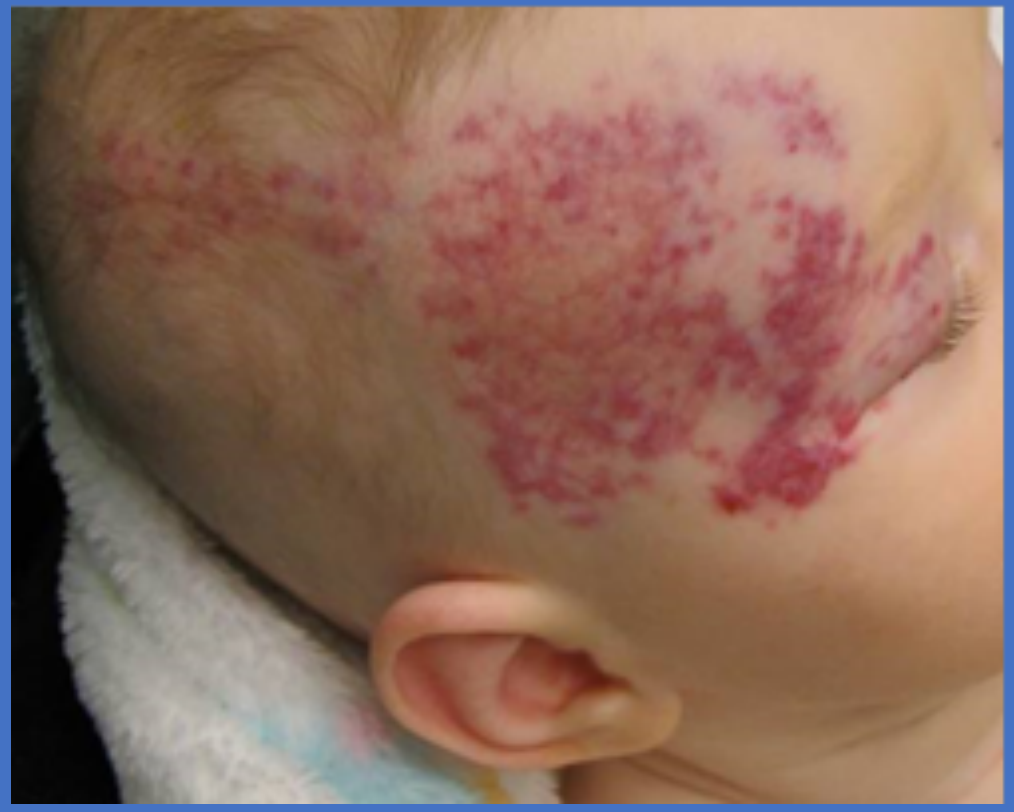
- Spinal cord abnormalities
- Urogenital abnormalities
- Anorectal abnormalities



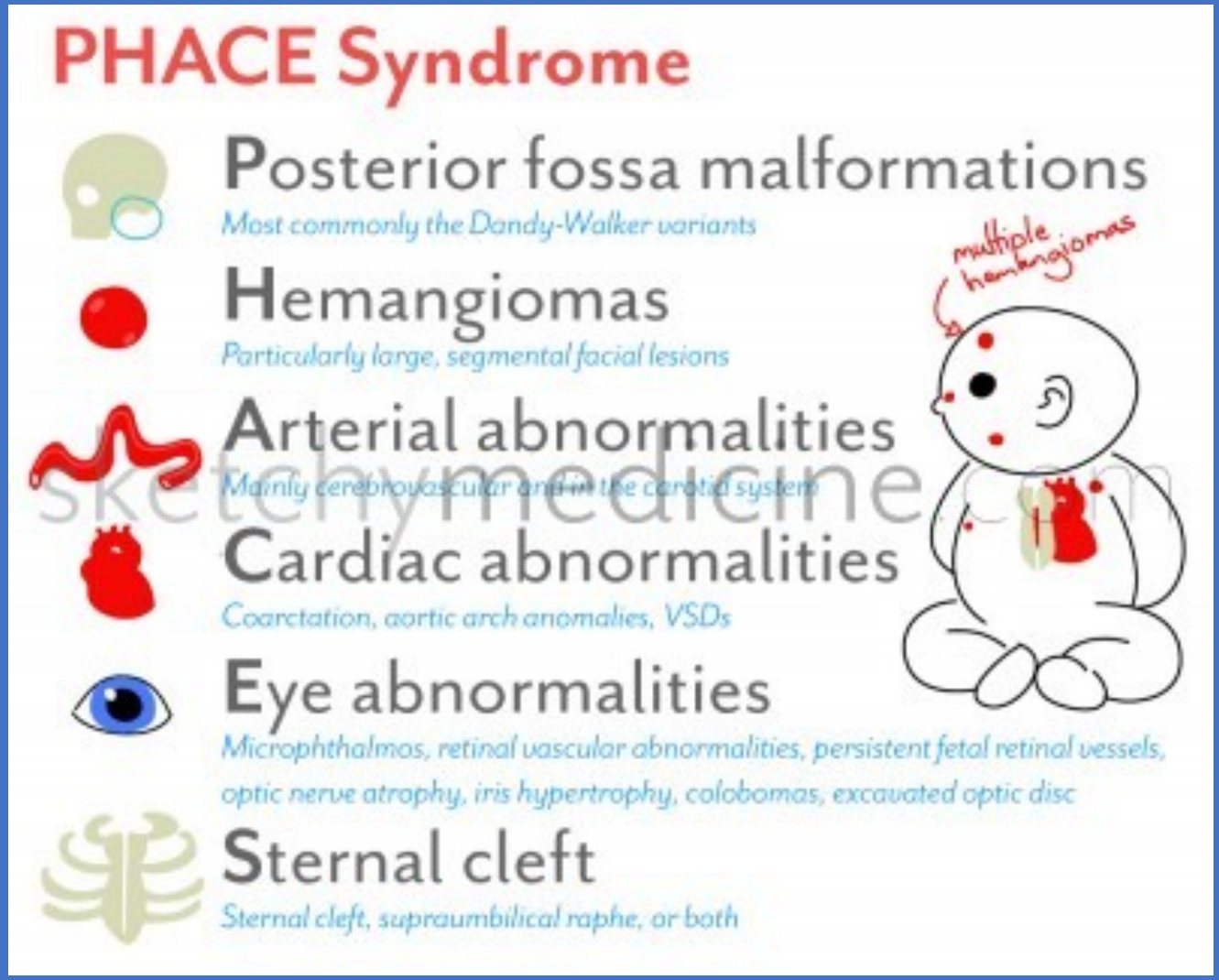


http://www.hemangiomaeducation.org/images/examples/M-2.jpg

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

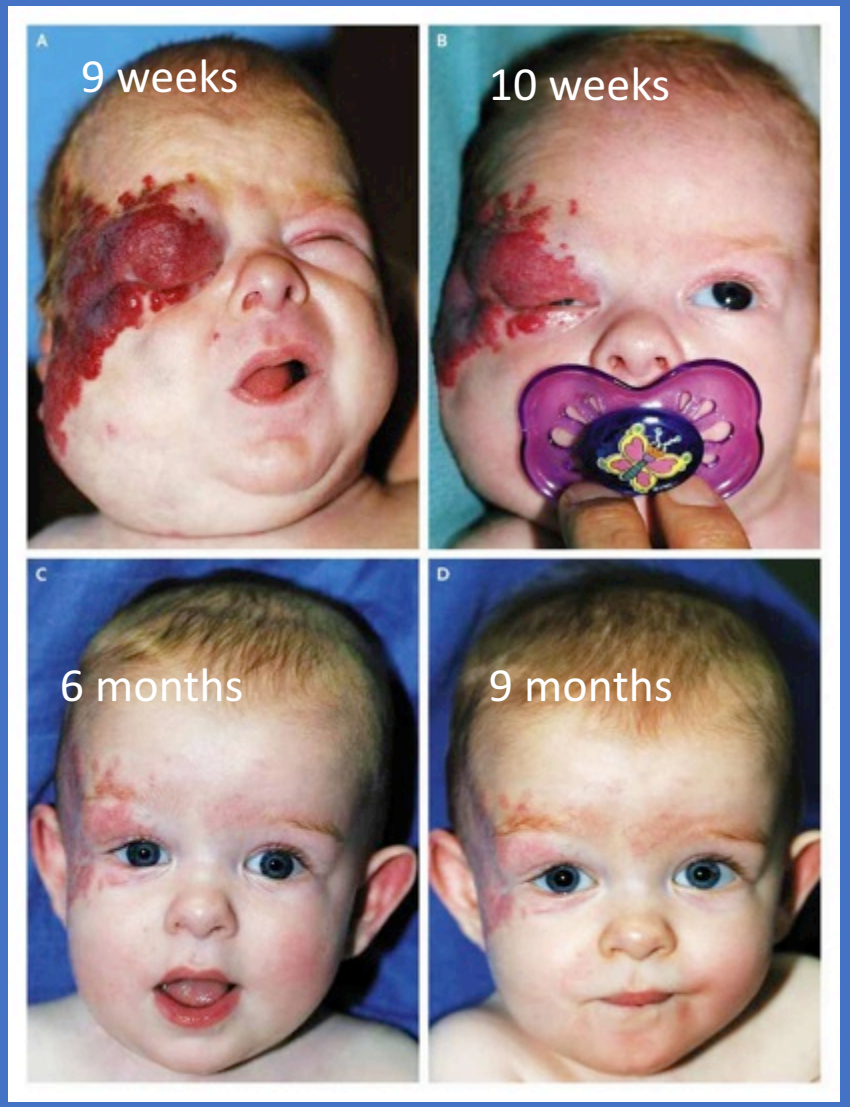
http://www.hemangiomaeducation.org/images/thumbnails/TERN69.jpg

Treatment of Hemangiomas

- Active Nonintervention/close follow up
- Corticosteroids
- Beta-blockers
  - Propranolol
  - Topical Timolol
- Excision
- Pulsed Dye Laser

Discovery of Propranolol

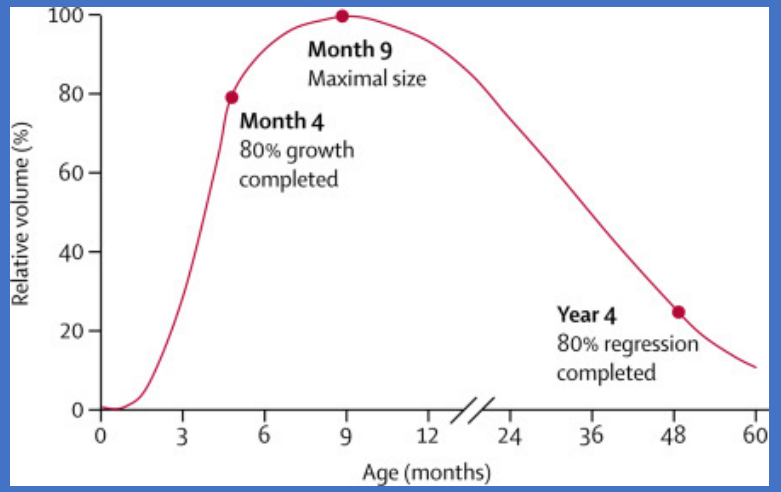
- 9 week old baby with hemangioma on oral steroid
- Baby developes high output cardiac failure
- Started Propanolol
- Hemangioma improves



Propranolol for severe hemangiomas of infancy.
Léauté-Labrèze et al. NEJM 2008;358:2649-2651

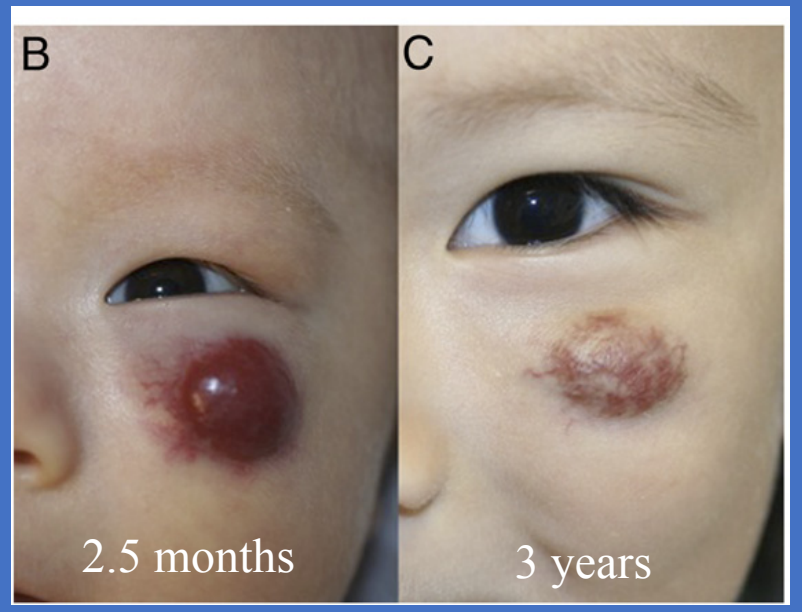
Referral timing

- Potentially problematic haemangiomas should be identified within the **first 2–3 weeks of life**, so they can be treated while in the early stage of proliferation
- Treatment should ideally be started by 3 months of age
- **Ideally refer by 1 month of age**



Hemangioma evolution

- Evolution of an exophytic hemangioma from birth to 3 months.
- Treatment with steroid and propranolol starting at 2.5 months of age
- Residual telangiectasia and fibrofatty skin changes persisted,
- Don’t let potential problematic hrmangioma grow
- You can end up with irreversible skin change



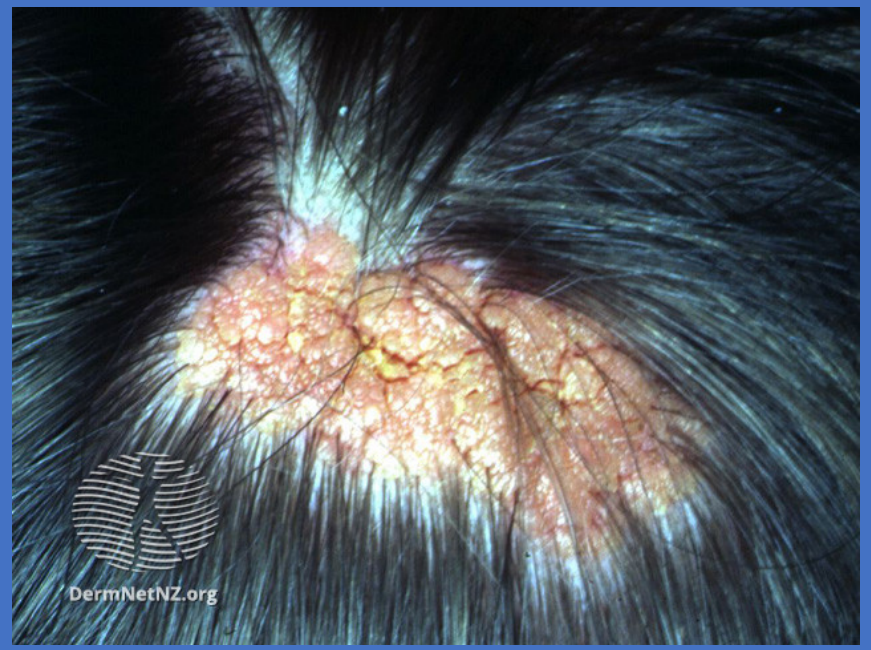
Pediatrics,2012,130

Yellow birth marks

Case 4

- Newborn with yellow orange alopecic 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
- Refer if changing but most growths are benign

Brown birthmarks

Case 5

- Newborn with dark brown lesion

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

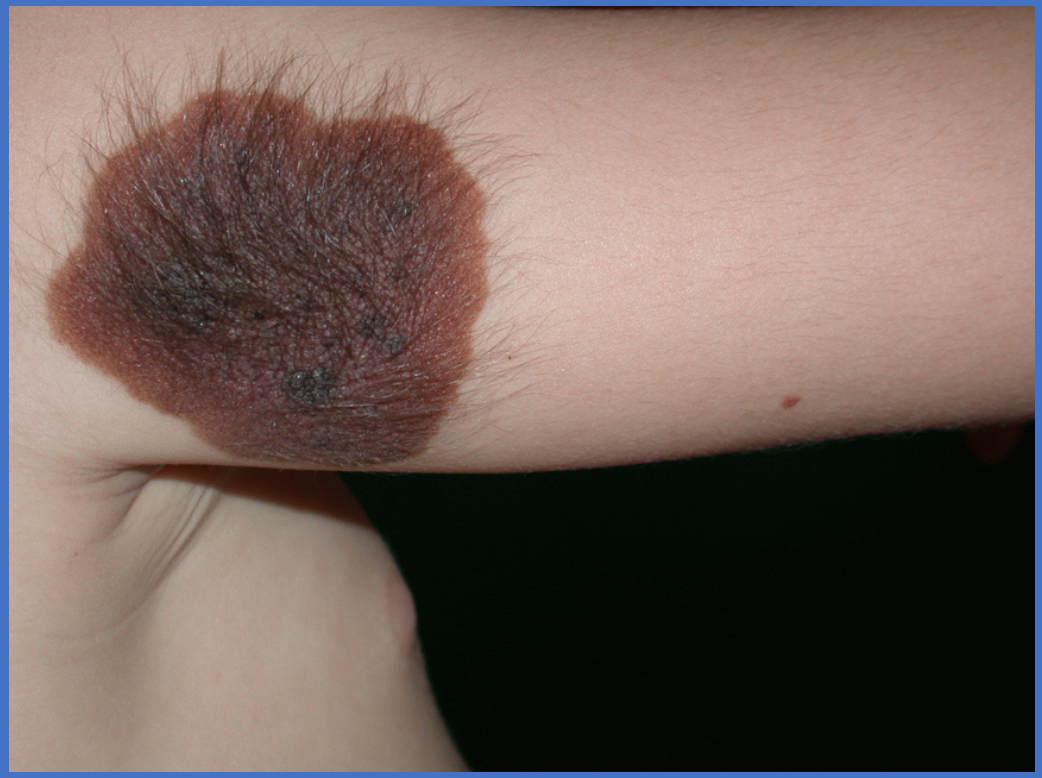
- Multiply the diameter at infancy by:
- 1.7 for head lesions
- 2.8 for Hands, feet, torso, forearms, arms, hips
- 3.4 for thigh lesions
- 3.3 for leg lesions

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.

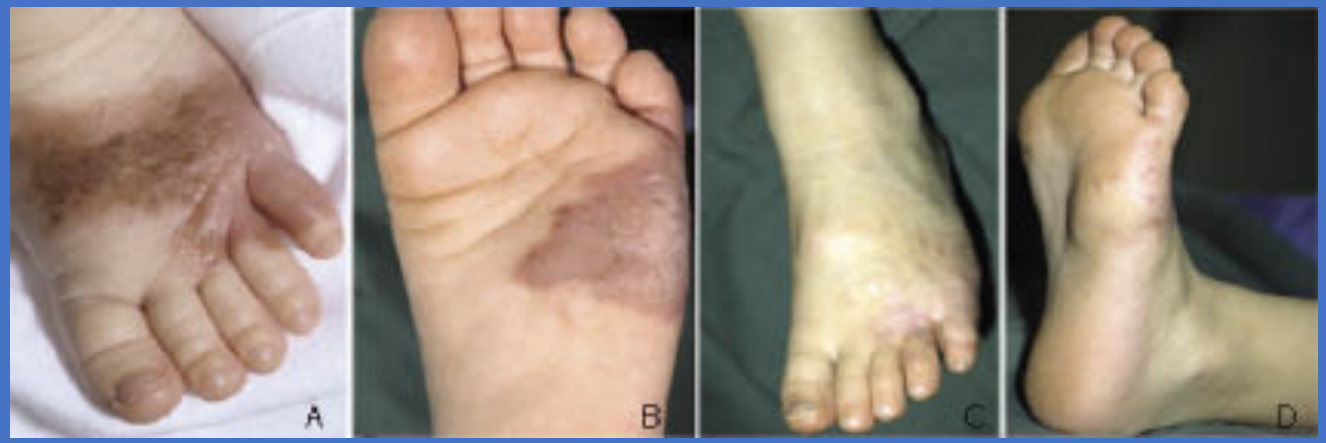
Grow coarse dark hair



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

Get raised, darkened with pebbly, verrucous surface

Spontaneous lightening



Acta Derm Venereol. 2012 Nov;92(6):605-6. doi: 10.2340/00015555-1413.

Proliferative nodules

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

Can it turn to skin cancer?

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

Management
Small/medium CMN

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

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 ??

Congenital melanocytic nevi : when to 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

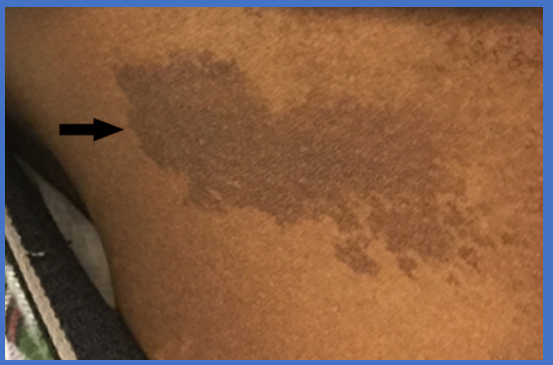
Café au lait

- 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

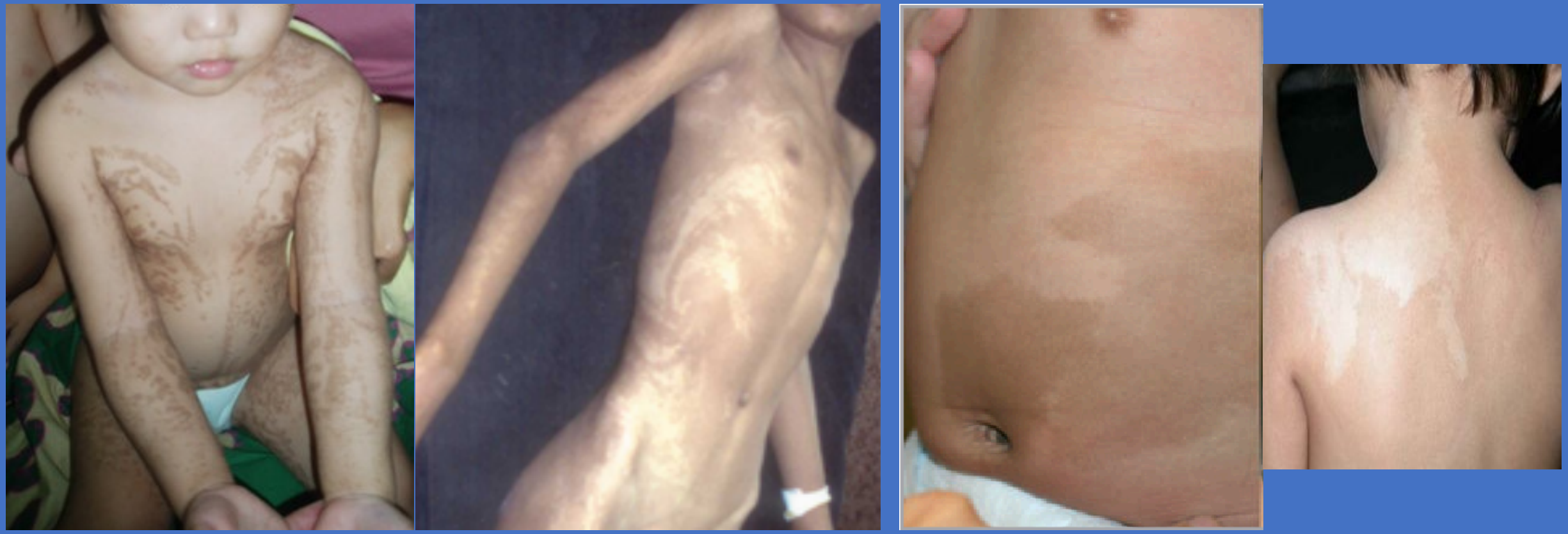
CALMs which need dermatology referral?

- 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



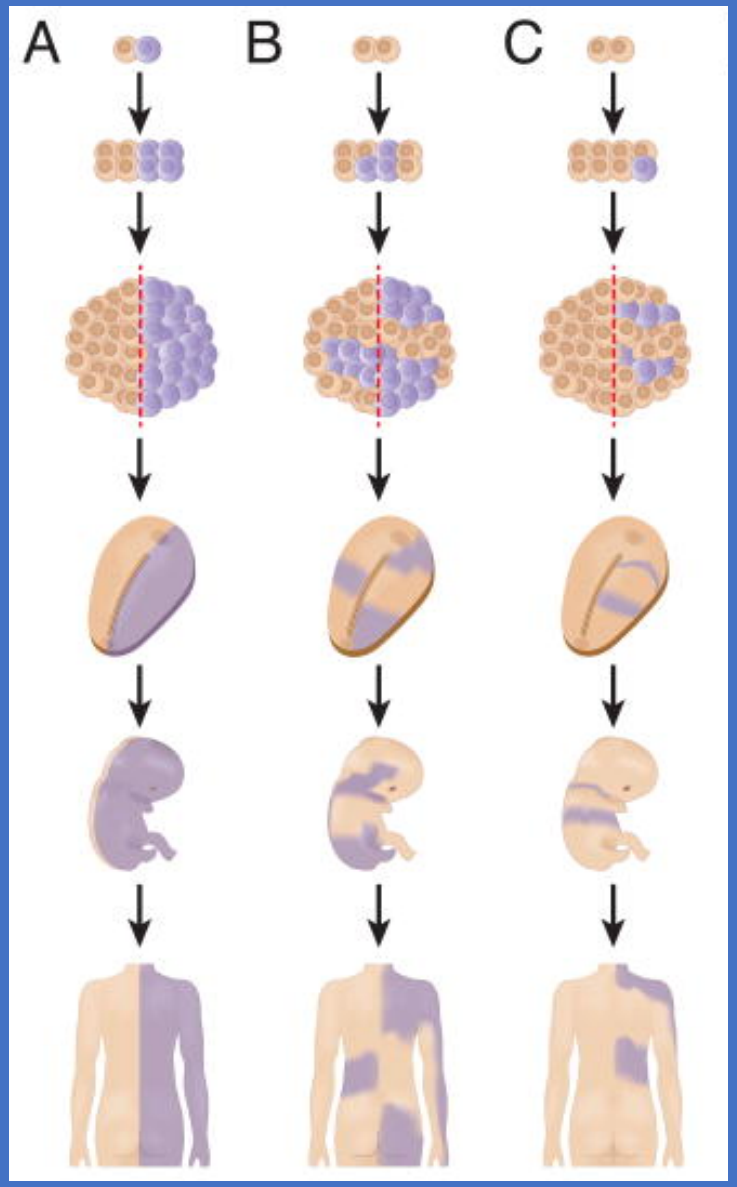
Indian Pediatrics 2004;401

https://d3i71xaburhd42.cloudfront.net/178ed593eee4ef675a5ee0173569f4b8651f8b47/3-Figure2-1.png

Egyptian dermatology online journal vol 4 No.2;7 Dermatology 2008

Pigmentary mosaicism

- Patterned Hypopigmentation/ hyperpigmentation
- No single entity but a cutaneous sign of mosaicism
- Reflects clone of cells with decreased / increased pigment production potential
- Genetic analysis of affected skin compared to unaffected skin may help to pinpoint the genetic alteration



https://www-ncbi-nlm-nih-gov.proxy3.library.mcgill.ca/pmc/articles/PMC4490042/bin/nihms686045f1.jpg

Pigmentary mosaicism

- Any systemic association?
- The incidence of systemic involvement is 4-30%
- The more widespread a pigmentary anomaly is, the more 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 American-African 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 refer?

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

Take home messages

- Birthmarks are mostly caused by mosaicism
- They can herald an associated abnormality
- A complete history and physical exam is important
- Although most birthmarks are benign ,some need early referral


THE SLIDE MAN – QUESTION MARK