GENODERMATOSES

Definition

Genetically determined skin disorders with a little alteration by environmental factors

Familial

Any condition more prevalent in relatives of any affected individual

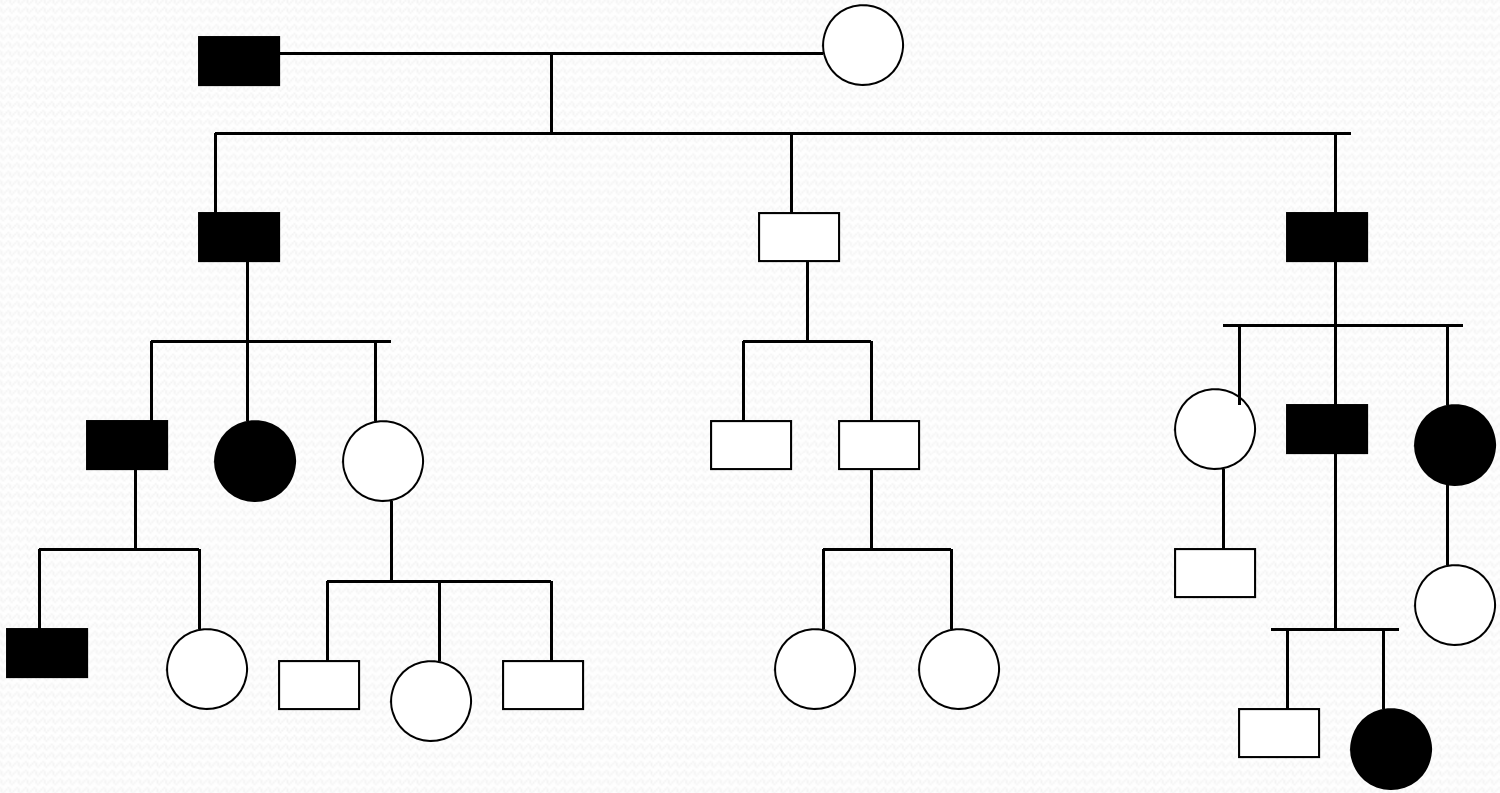
Hereditary

Disorder transmitted from one generation to other

Congenital

Disorder present at or before birth

Autosomal dominant inheritance



AD disorders affect both male /females

Affected persons are heterozygous for abnormal allele

Every affected individual will have an affected paren

Affected parent transmit disorder to half their sons/ half daughters

Ex. Incontinentia Pigmenti,NF,Tuberous sclerosis

-50% of children of an affected parent will be affected

- Age of onset m/b variable
- Severity of the disease varies among individual in a family
- AD disorders may show lack of penetrance.

Ex. NF, Tuberous selerosus

NEUROFIBROMATOSES :

(VON RECKLING HAUSEN DISEASE)

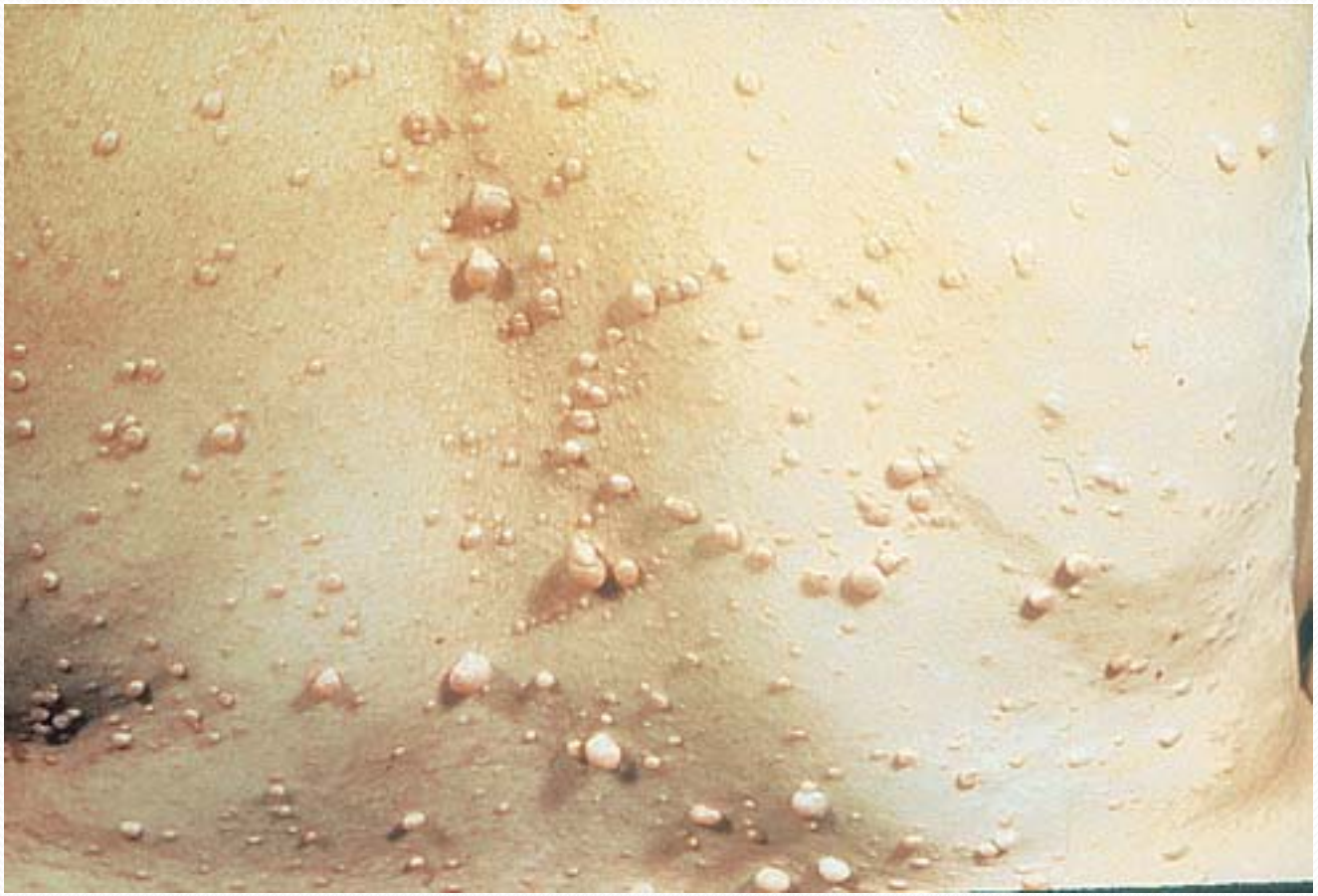
FAMILIAL AD INHERITED DISORDER

i.e. NF – I

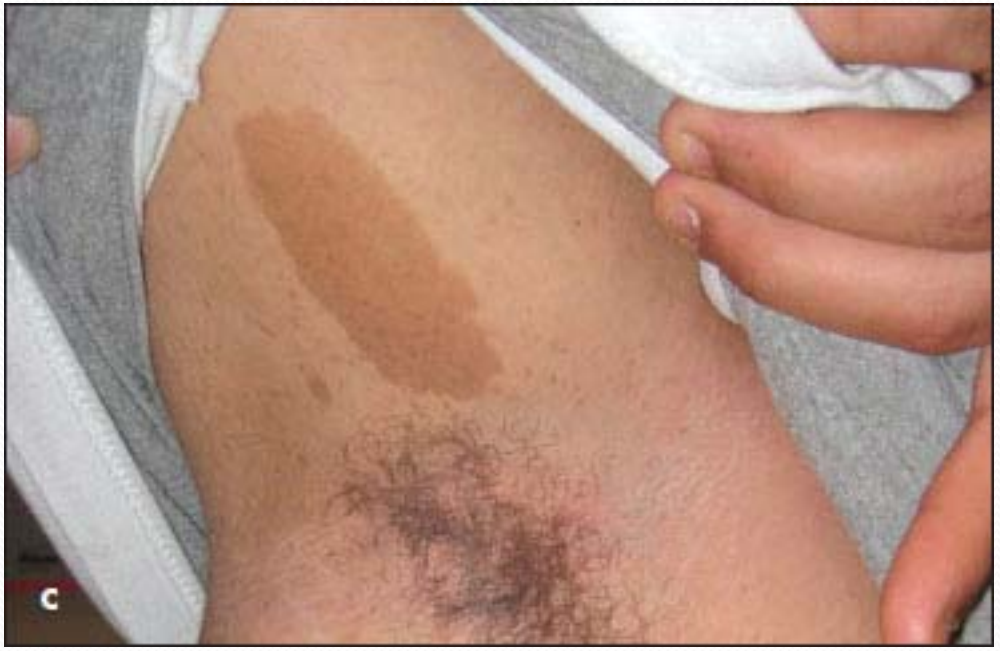
NF – II

NF-I

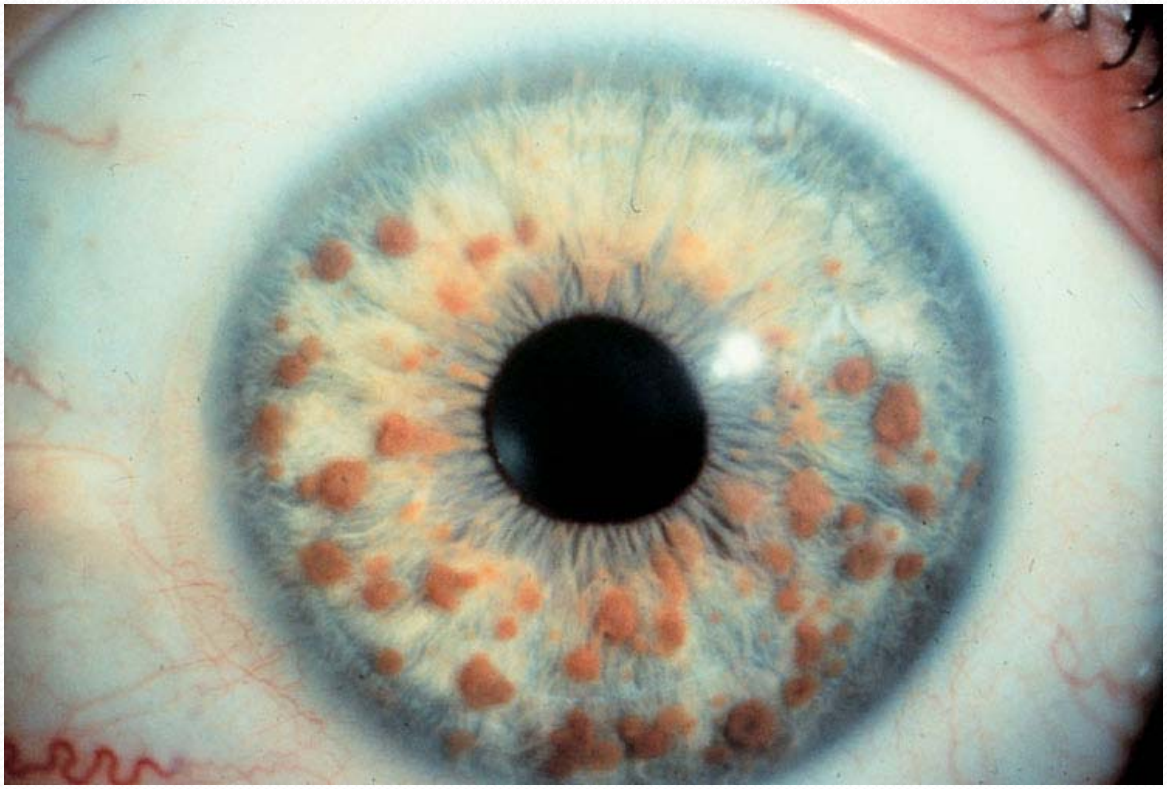
Cutaneous neurofibromas



Café au lait macules



Lisch nodules



NF – II

Located on chr. 22

-B/L acoustic schwanomas

- CNS tumours of meningeal and glial

- Café au lait spots

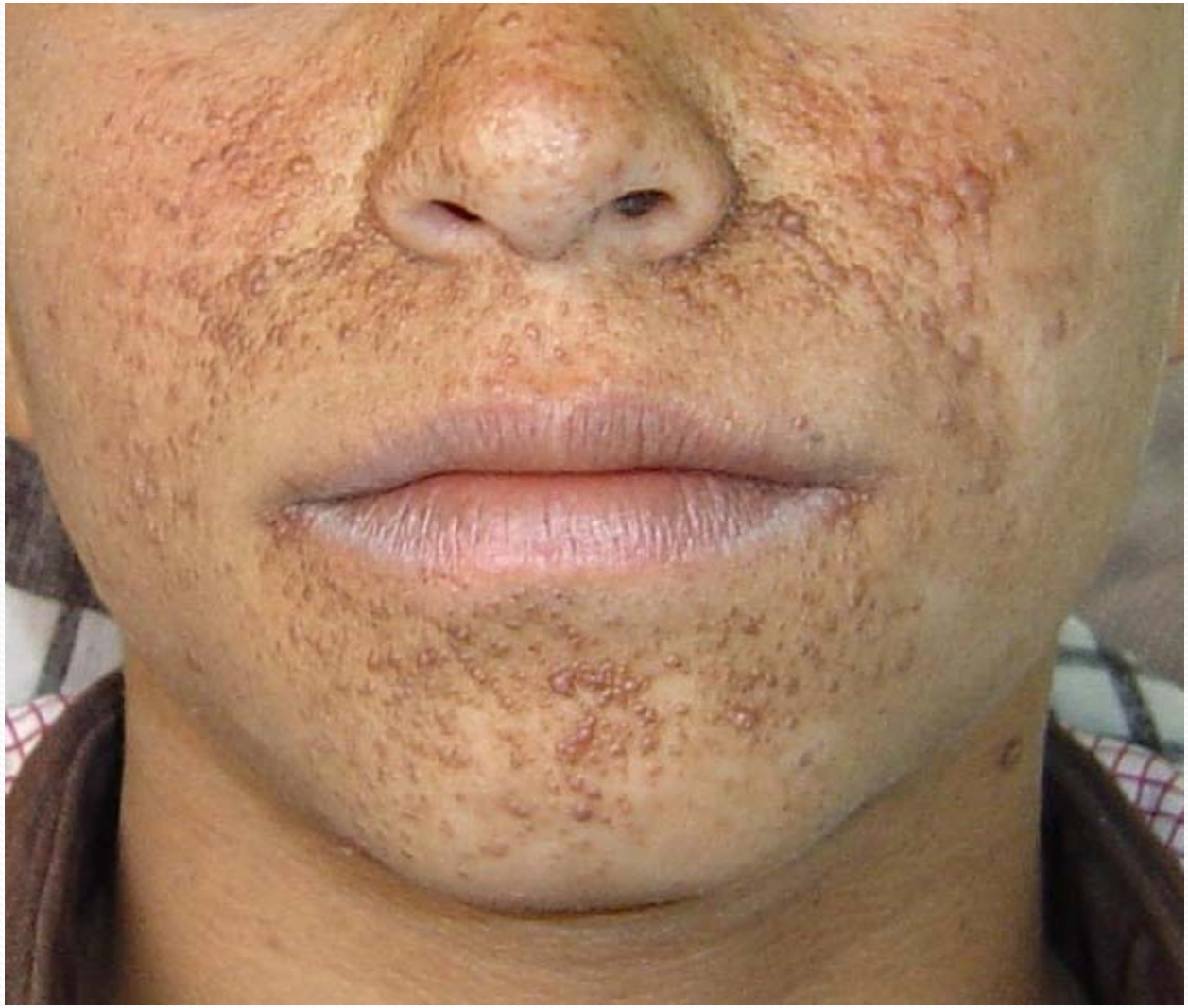
- Cut. NF

TUBEROUS SCLEROSIS (EPILOIA, Bourneville's disease)

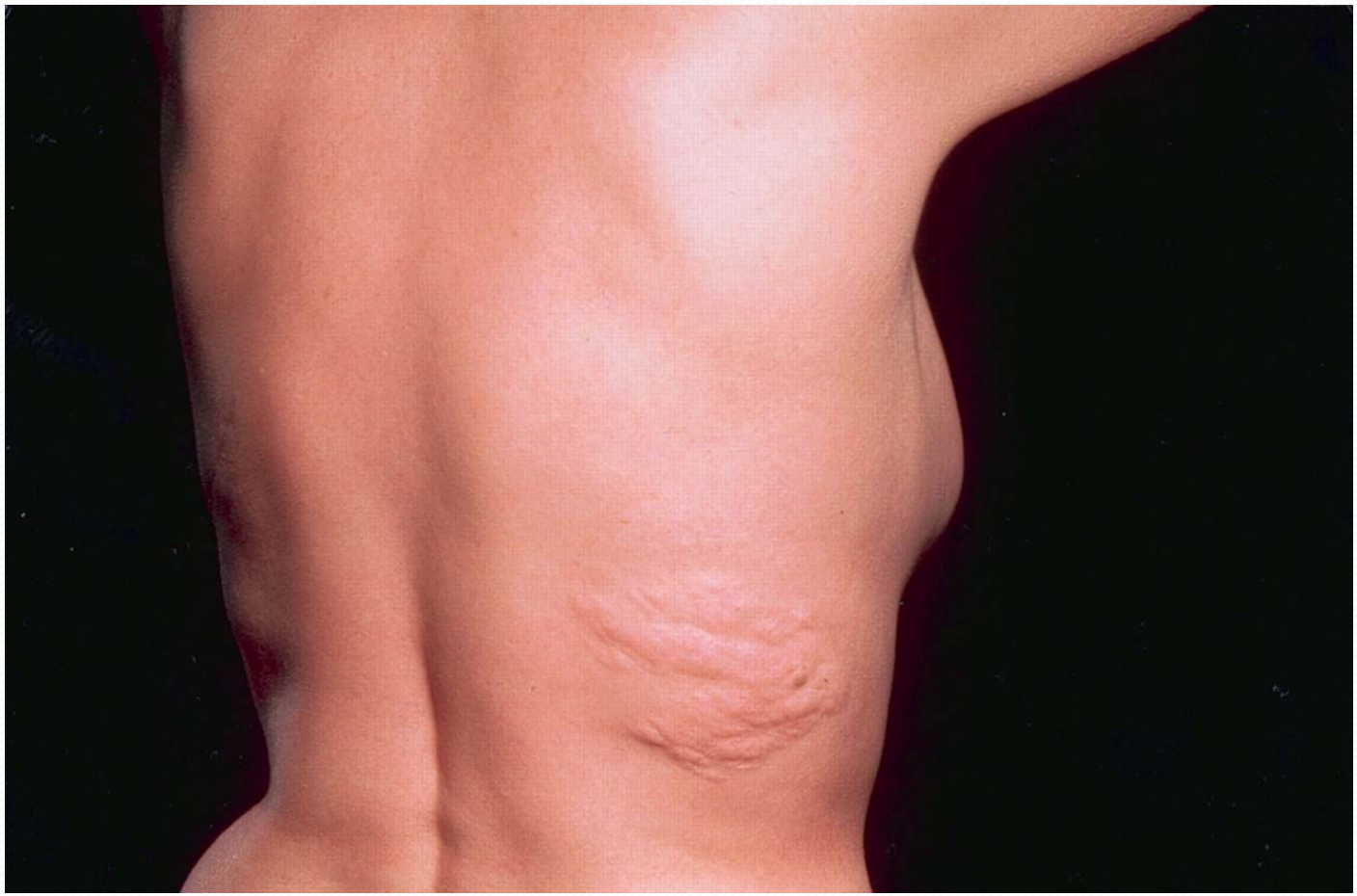
- AD

Characterized by hamortoma formation in skin, brain, eye, kidney, heart

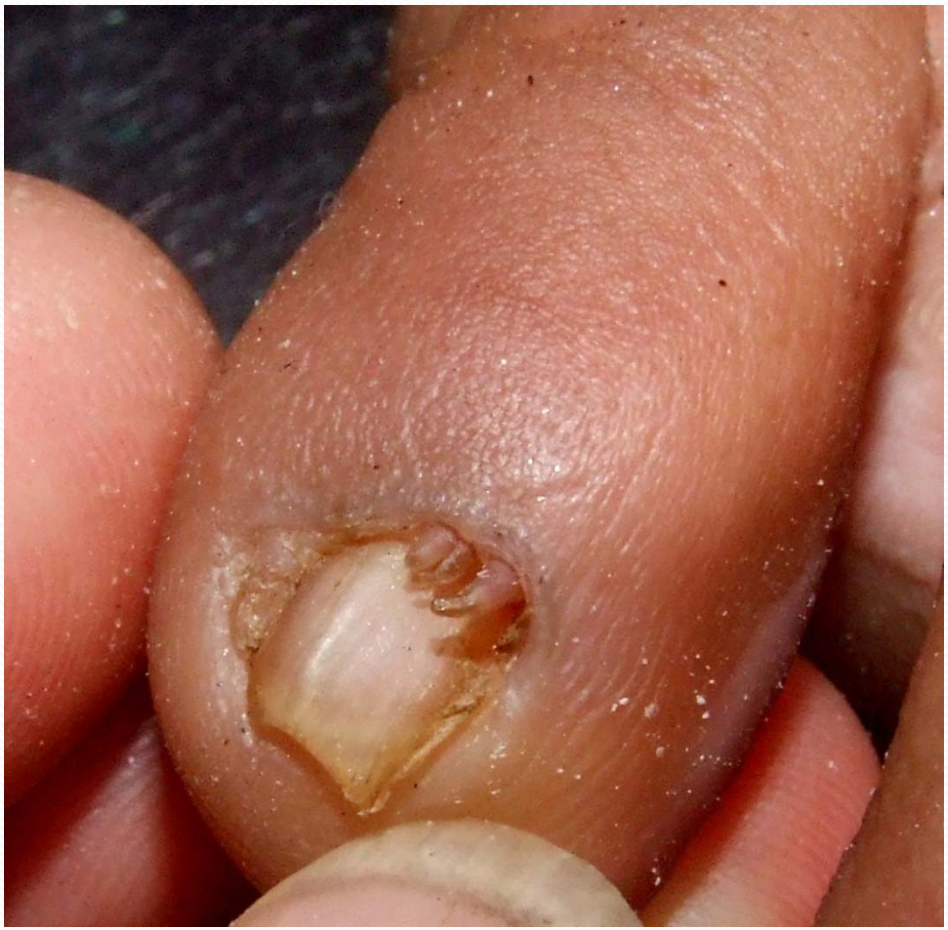
Skin lesions: Angiofibromas



Shagreen patch

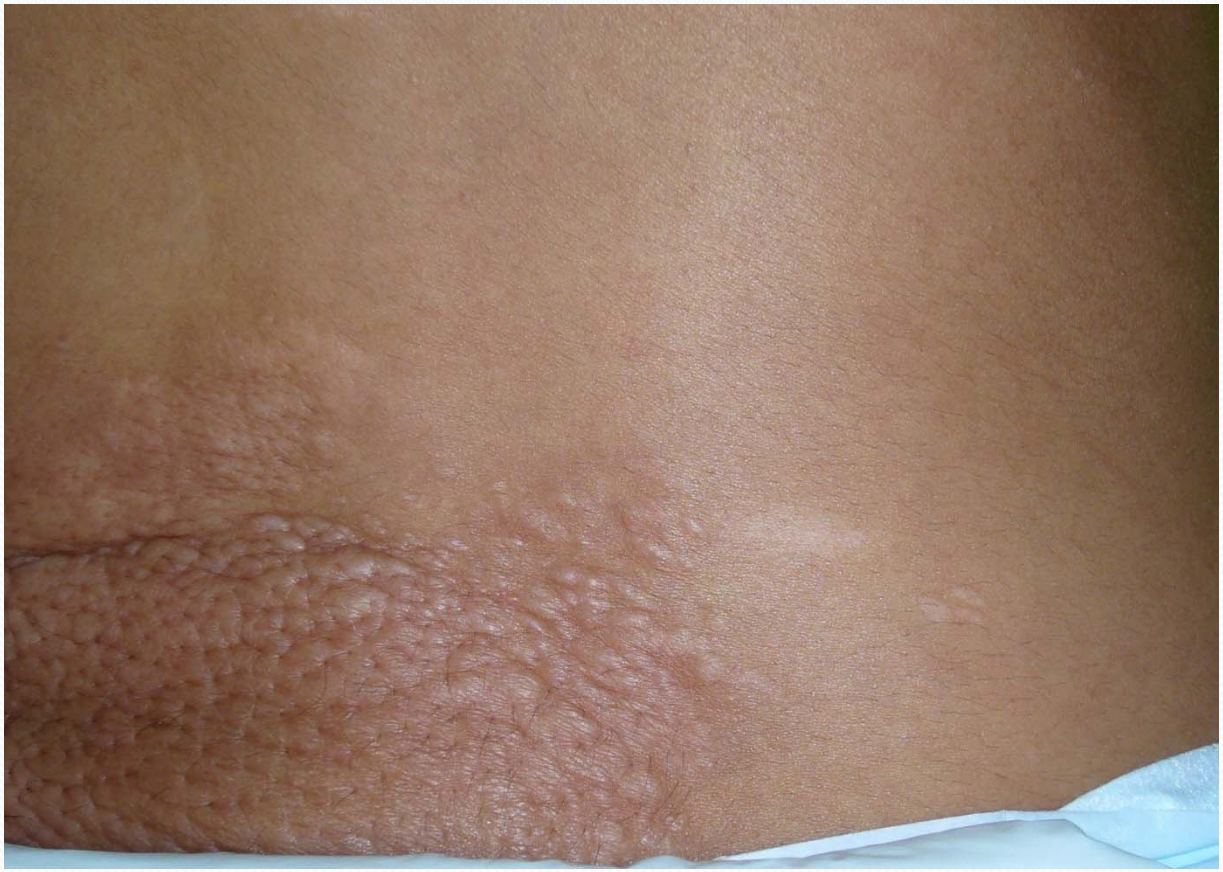


Periungual fibromas



Other Features

Ash leaf macules



Other features:

Ocular – Retinal phacomata

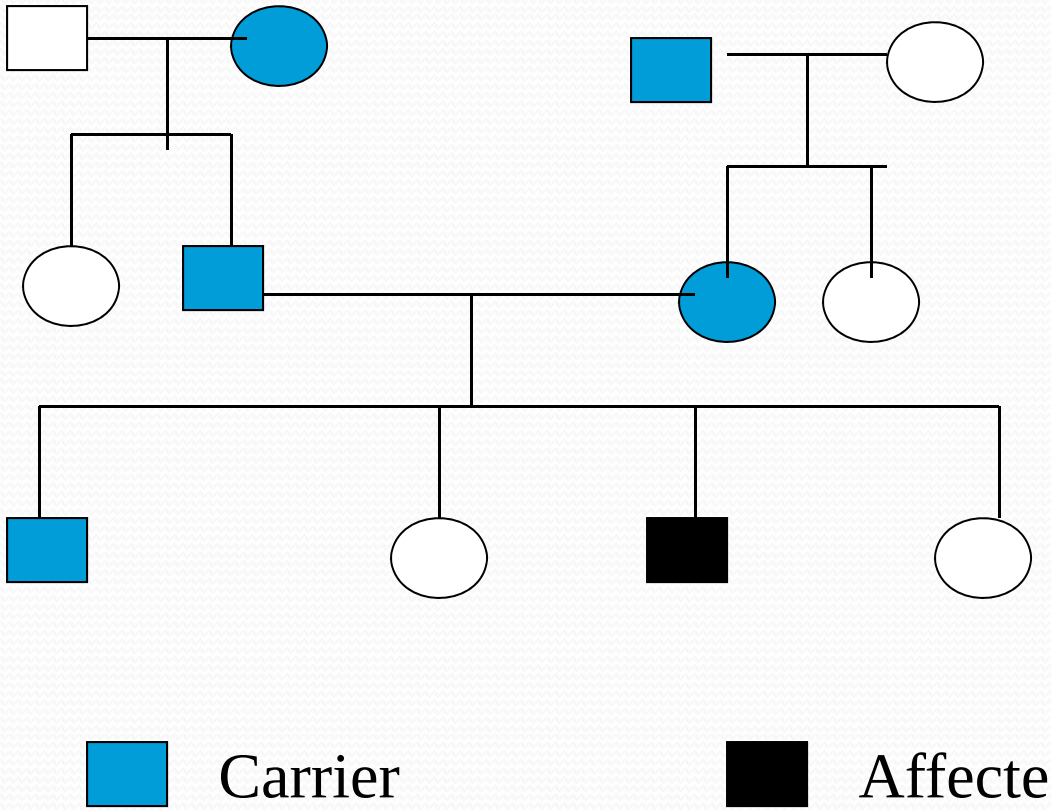
Pigmentary abnormalities

Cardiac/renal tumours

Rhabdomyomas

GIT tumours

AUTOSOMAL RECESSIVE INHERITANCE



a) AR disorders affect both male & female

b) Affected individuals are homozygous

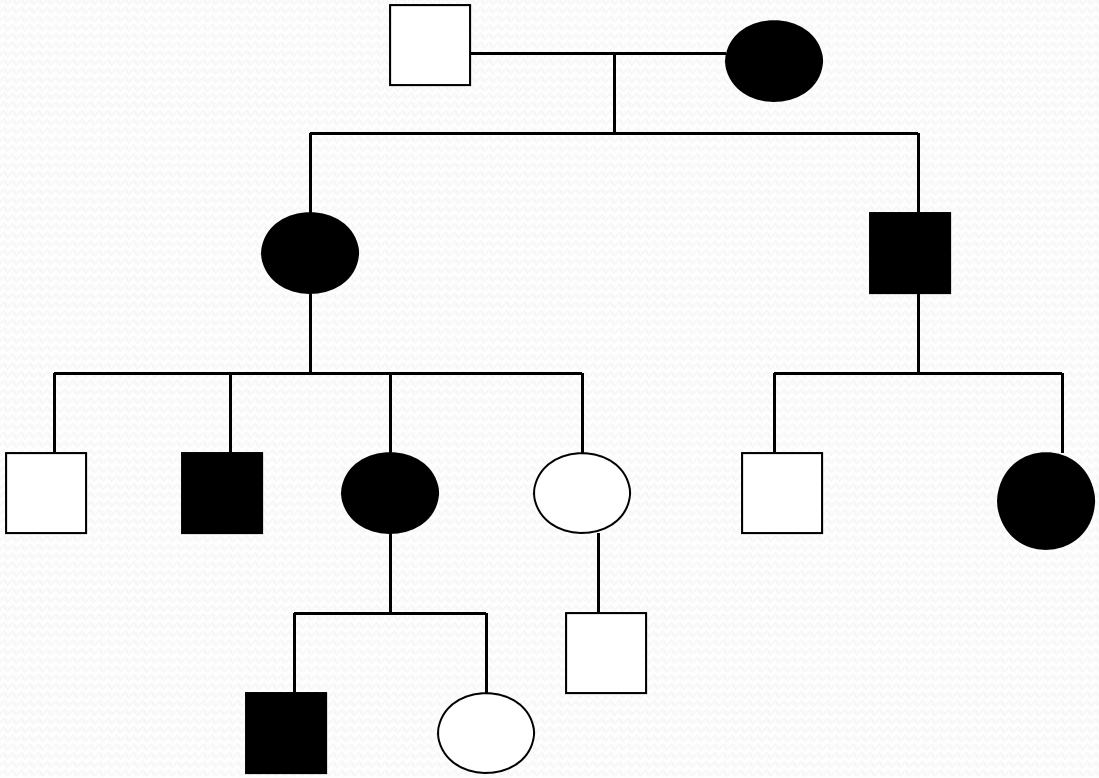
c) AR in indiv. -where both parents are carriers

d) 1:4 children affected in heterozygous parents

e) Consanguity increases risk of AR disease

f) Severe presentation as in disease of metabolism

X-LINKED RECESSIVE INHERITANCE



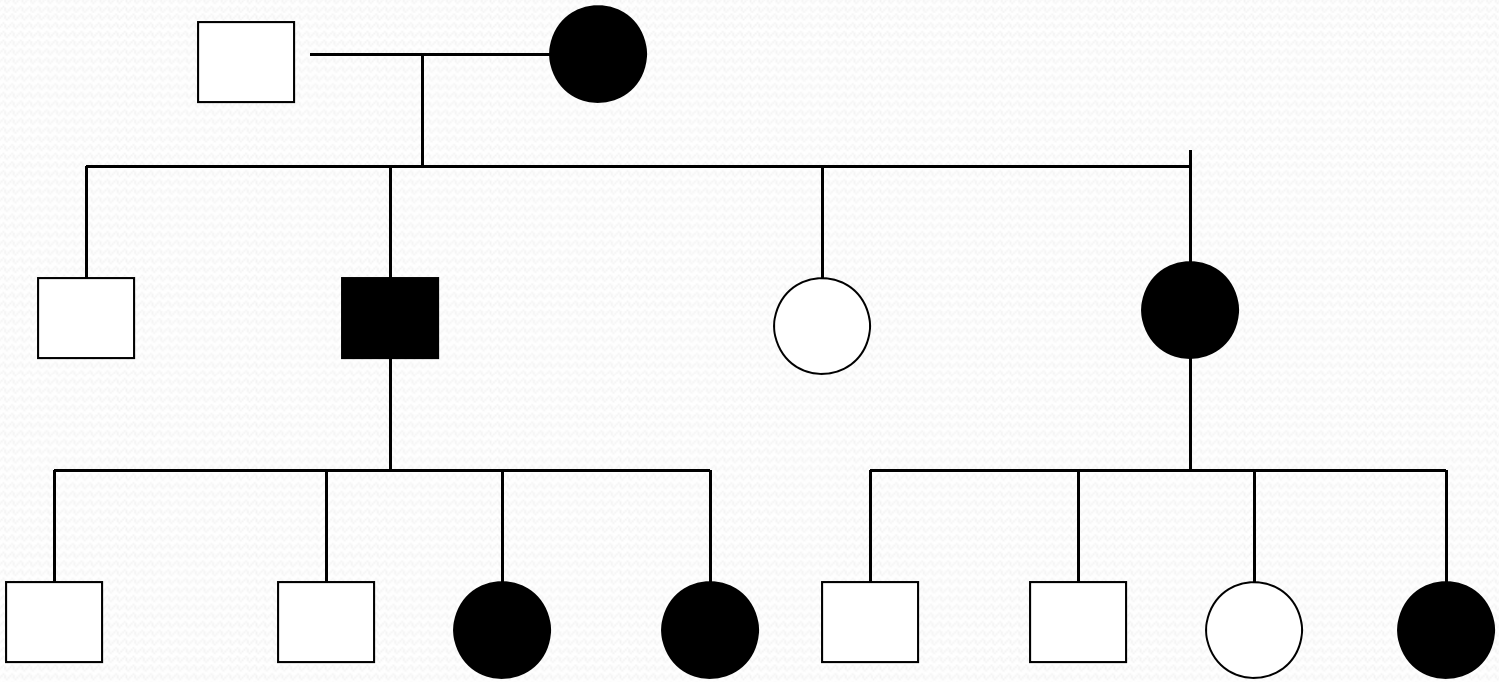
i) Only males are affected

ii) Transmitted thro healthy female carriers

iii) Female carrier transmit disorder to half her sons, not from father to son.

iv) Ex: Ocular albinism

X- LINKED DOMINANT INHERITANCE



i) Occurs in both hemizygous & hetrozygousfemale

ii) Affected males transmit disease to their daughters but not to sons

iii) Affected females transmit disease to half her sons half her daughters.

Ex. Incontinentia pigmenti

Syndromes associated with DNA instability:

Xeroderma pigmentosum:

- AR
- Photosensitivity
- Pigmentary changes
- Premature skin ageing

Others : Neoplasia

Abnormal DNA repair

neurological complication

Disorders of keratinization:

Icthyosis

Fish scale

AD – Vulgaris

i.e. progressive, persistent

non inflammatory scaling