

# **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**

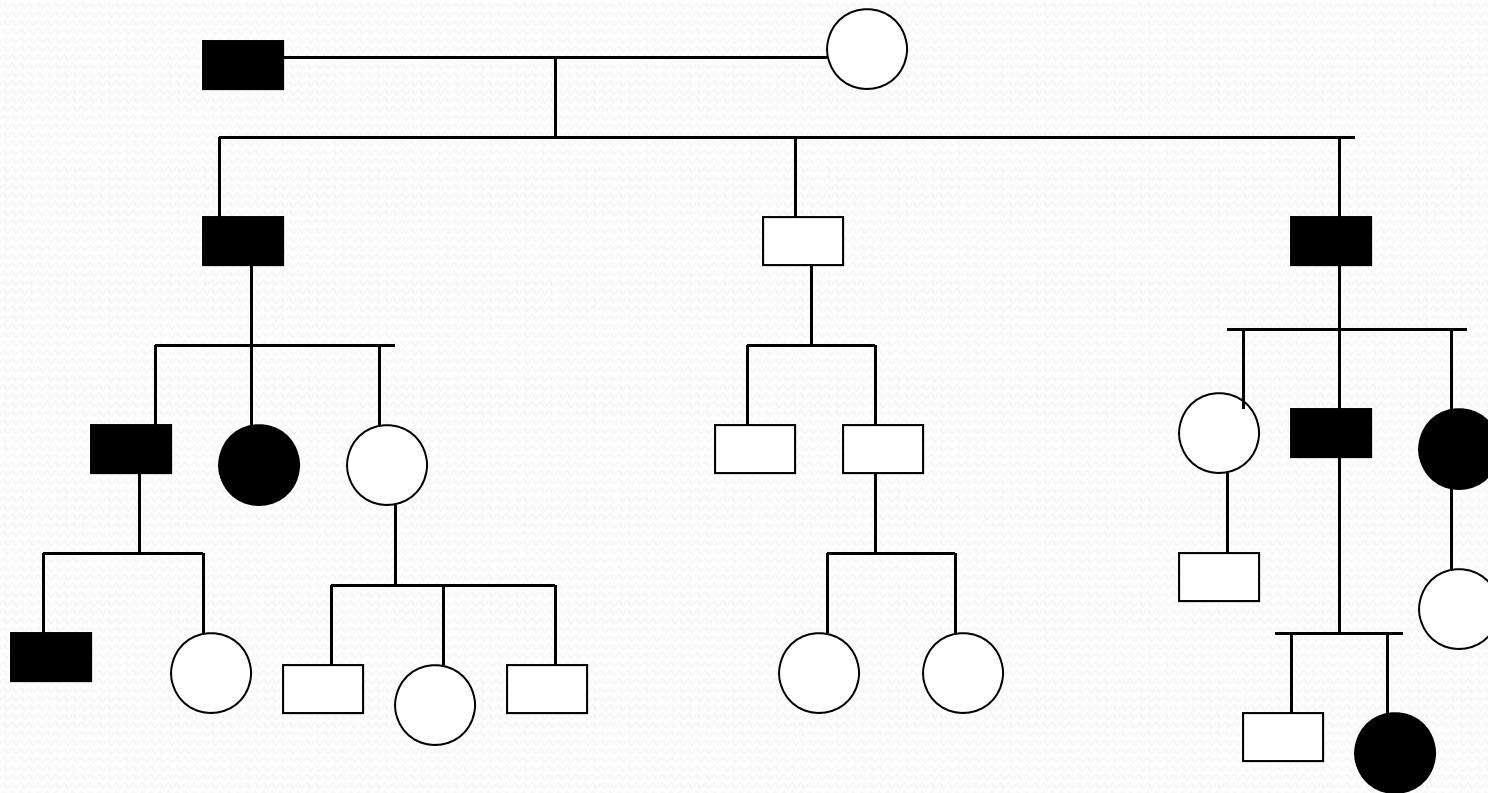
Disorder transmitted from one generation to other

## **Congenital**

Disorder present at or before birth

# GENETIC PRINCIPLES

## Autosomal dominant inheritance





**AD disorders affect both male /females**

**Affected persons are heterozygous for abnormal allele**

**Every affected individual will have an affected parent**

**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 sclerosis**

# NEUROFIBROMATOSES :

(VON RECKLING HAUSEN DISEASE)

FAMILIAL AD INHERITED DISORDER

i.e. NF – I

NF – II

NF-1

## Cutaneous neurofibromas

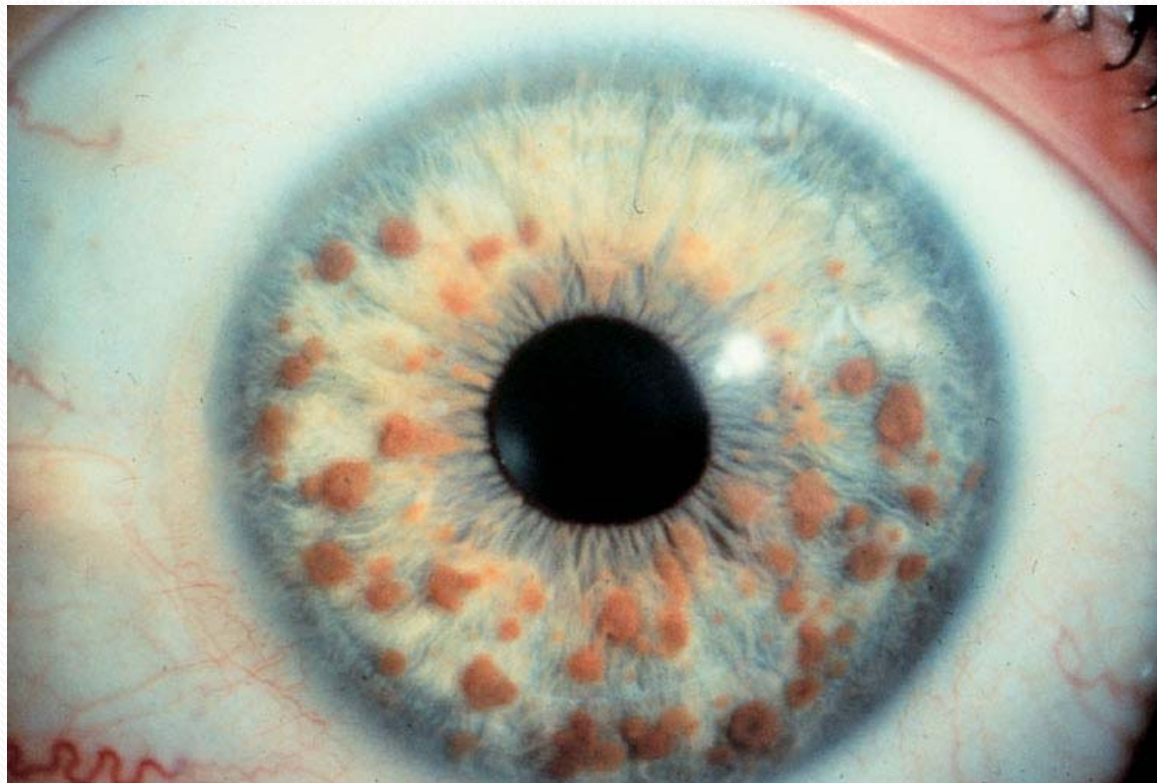


# Café au lait macules





# Lisch nodules



## NF – II

**Located on chr. 22**

- B/L acoustic schwannomas**
- 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

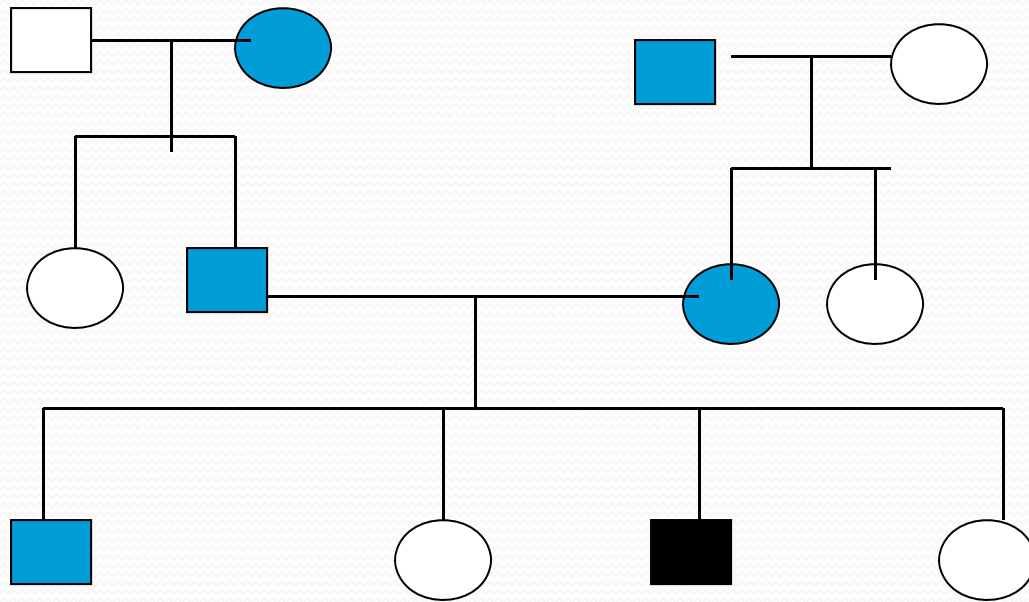
Cardiac/renal tumours

Rhabdomyomas

GIT tumours




# AUTOSOMAL RECESSIVE INHERITANCE



Carrier



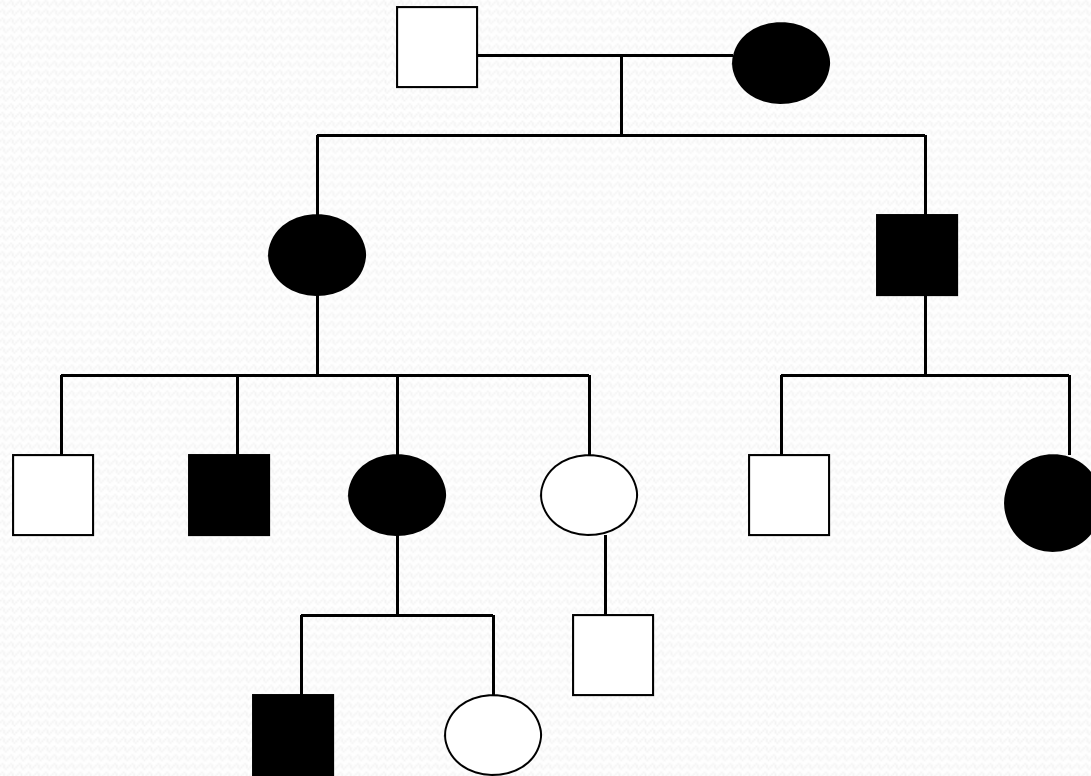
Affected


- 
- 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) Consanguinity increases risk of  
AR disease

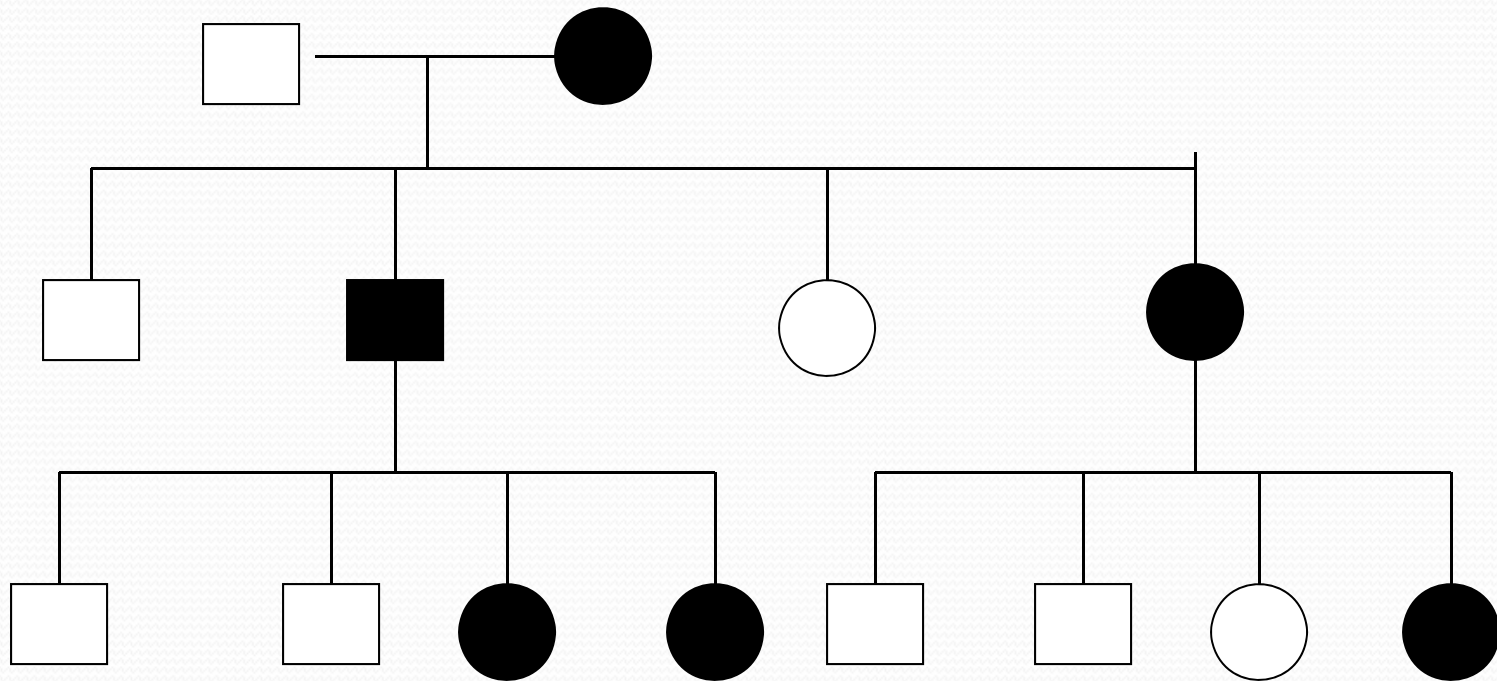
f) Severe presentation as in disease  
of metabolism

# X-LINKED RECESSIVE INHERITANCE



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- 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 & heterozygous female**

**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