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 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 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
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 hamartoma 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
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 & hetrozygous 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