

Genetika

Gangguan dan Abnormalitas

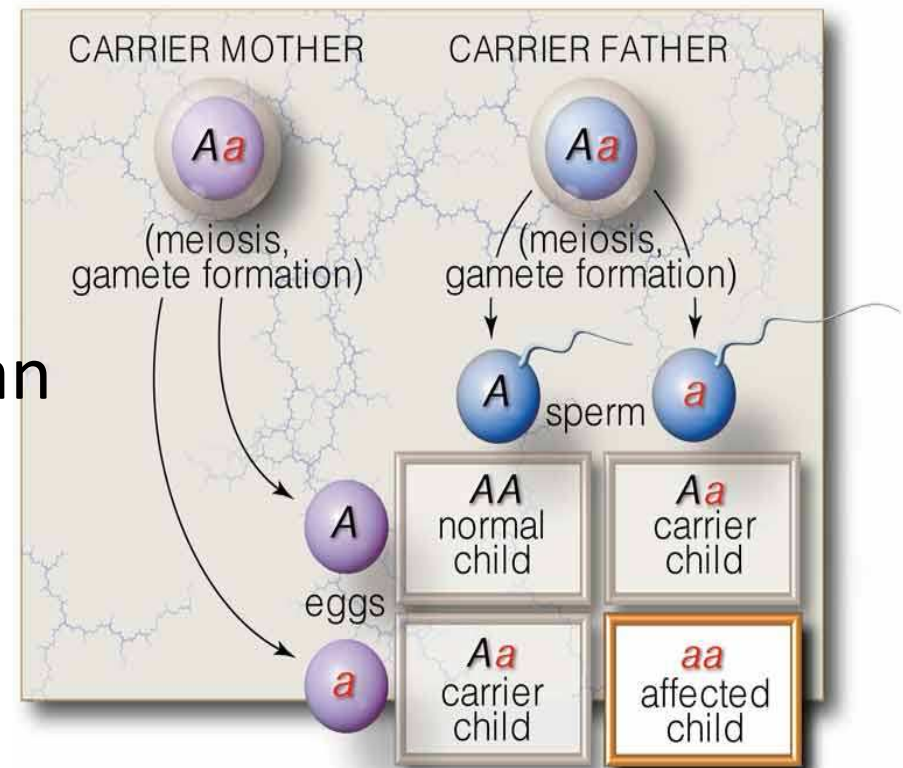
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Genetic Disorder

- Inherited conditions that cause mild to severe medical problems (Syndrome)
- May be...
 - Recessive
 - Dominant
 - Codominant or Incompletely dominant

Autosomal Recessive Inheritance Patterns

- Terletak pada autosom
- Penyebab dari sebagian besar gangguan dan kelainan
- Carriers (heterozigot)
- Akan selalu berada dalam gen

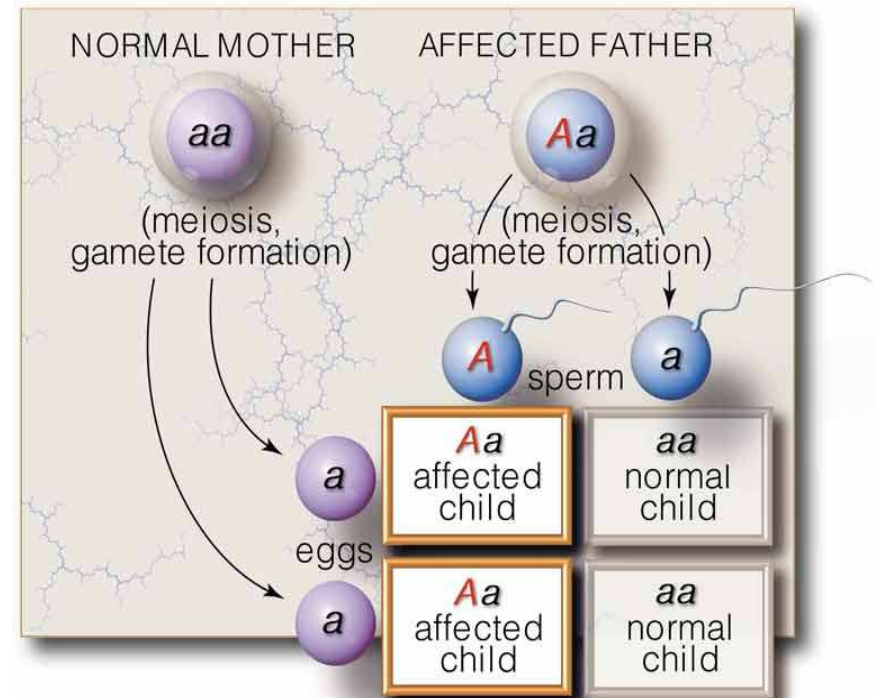


Autosomal Recessive Inheritance

- Galactosemia
- Cystic Fibrosis
- Albinism
- Phenylketonuria
- Hitchhiker's thumb
- Unpigmented eyes (Blue eyes)

Autosomal Dominant Inheritance

- Typically will appear each generation
- Lethal Dominant alleles
 - age matters
- Dominant abnormalities



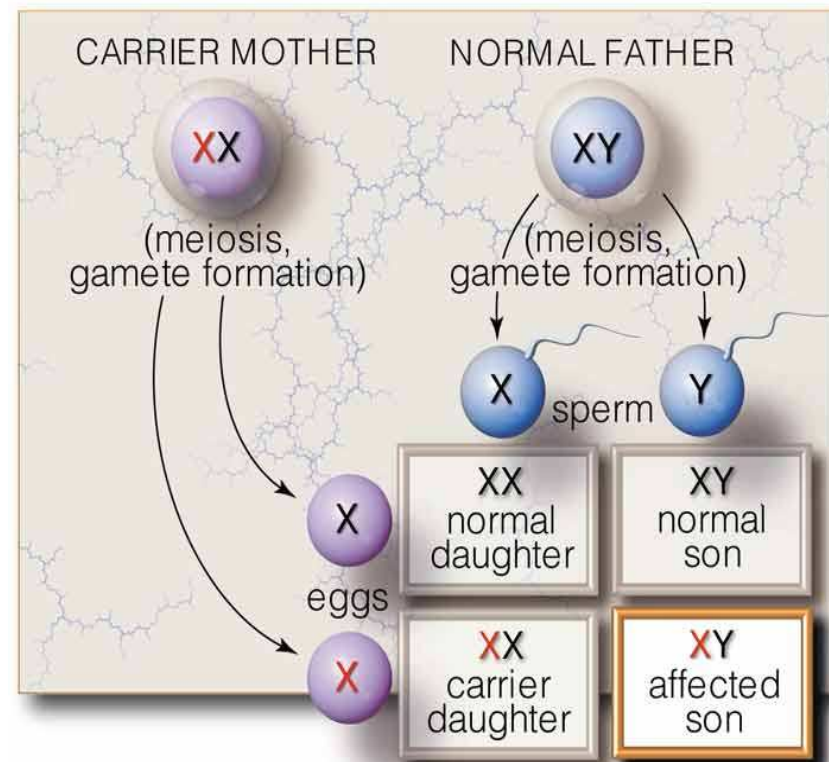


Autosomal Dominant Inheritance

- Achondroplasia
- Huntington's Disease
- Polydactyly
- Progeria
- Marfan syndrome
- Extra fingers and toes

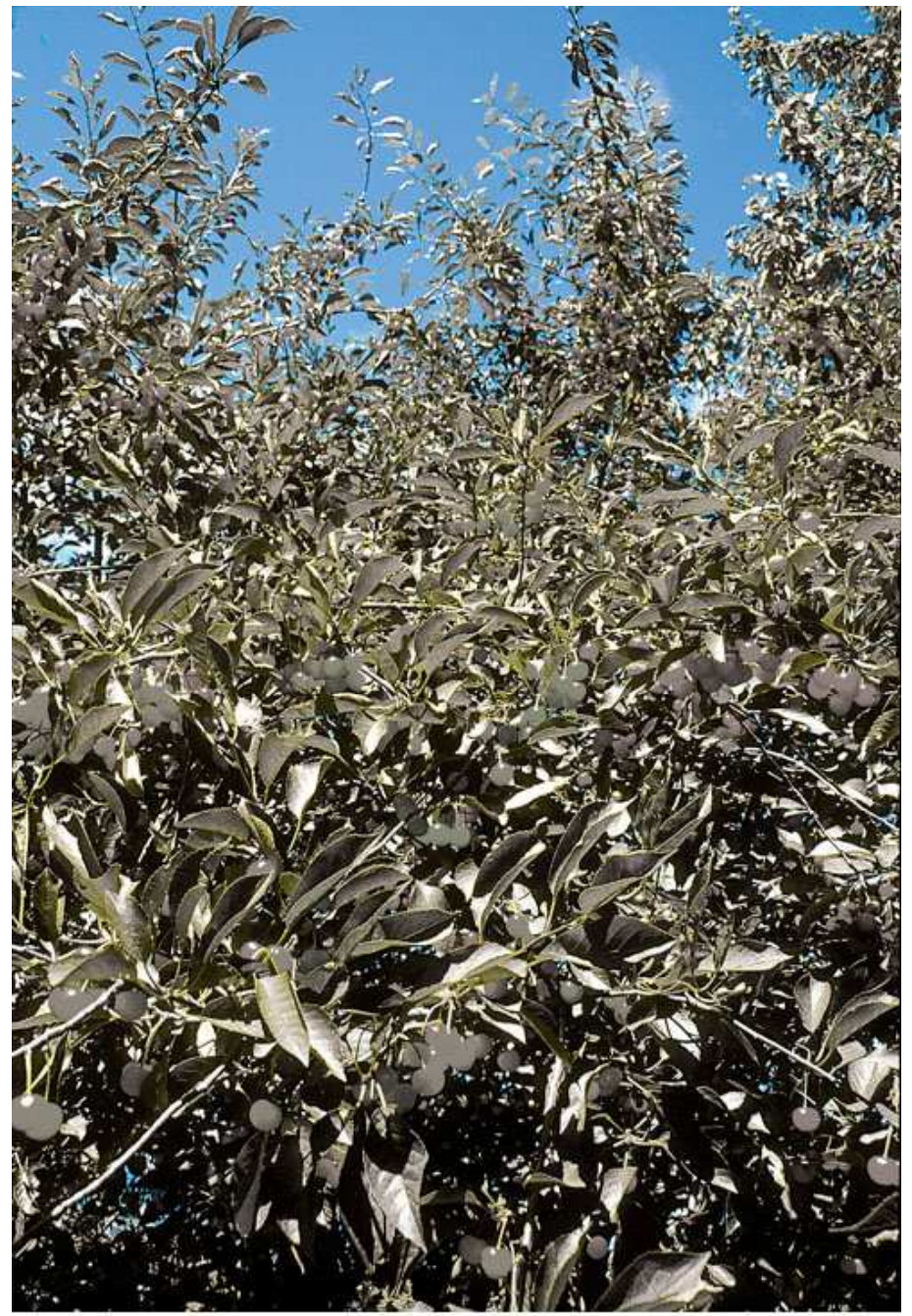
X-Linked Recessive Inheritance

- Terletak pada kromosom X
- Lebih sering pada laki-laki
- Anak mewarisi dari ibu



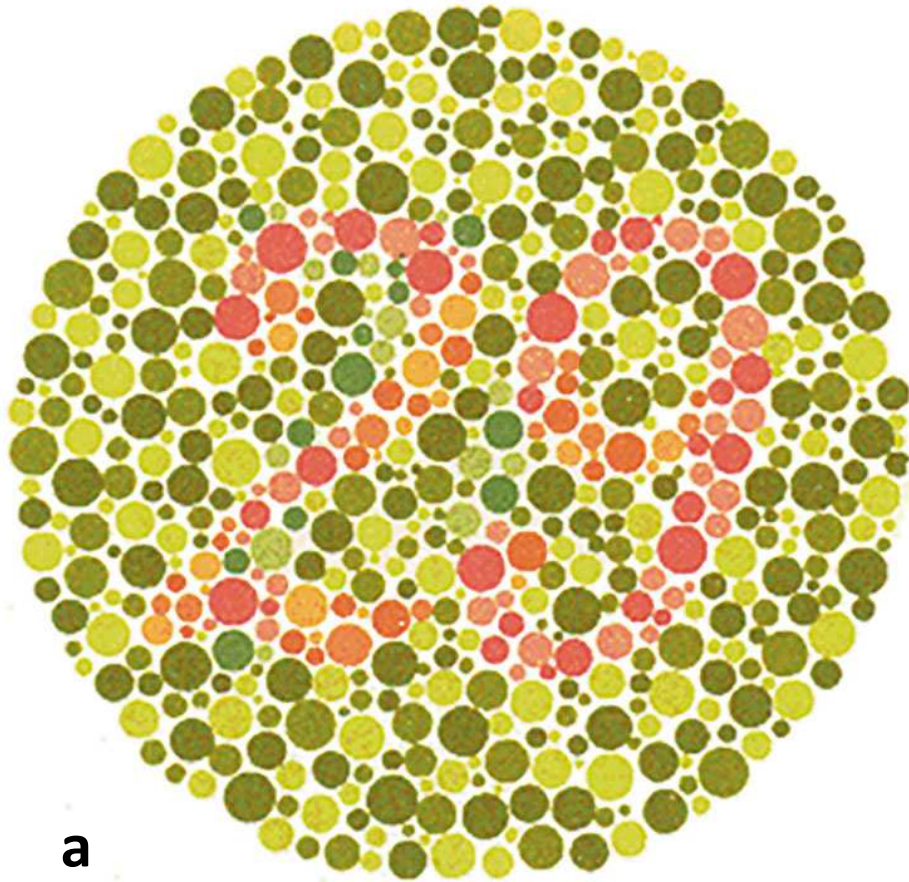


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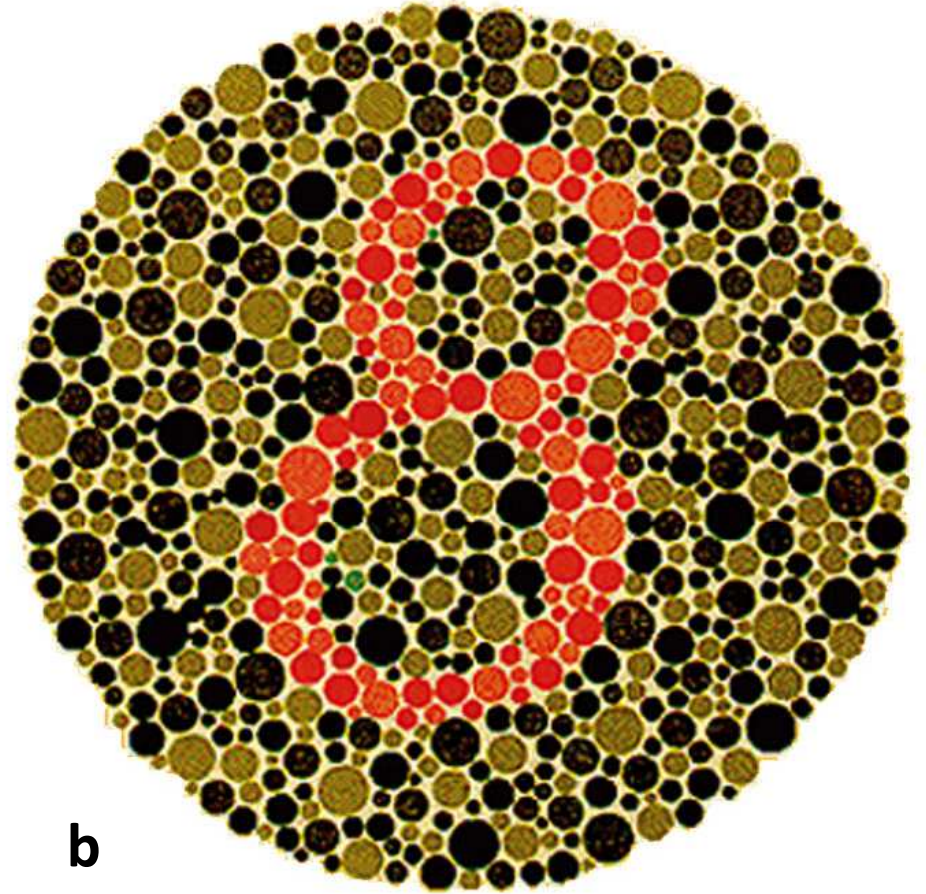
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Fig. 8-27, p.128



a

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b

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Fig. 8-27, p.128

X-Linked Inheritance

- Colorblindness
- Hemophilia
- Pattern Baldness
- Muscular dystrophies

Pedigree

- Jejak yang fenotipe selama beberapa generasi
- Mengidentifikasi dominasi atau resesif

Pedigree Symbols



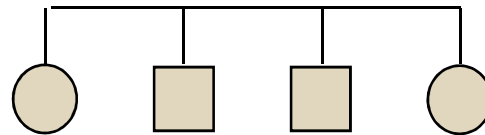
male



female



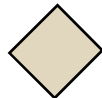
marriage/mating



offspring in order of birth,
from left to right



Individual showing trait being
studied

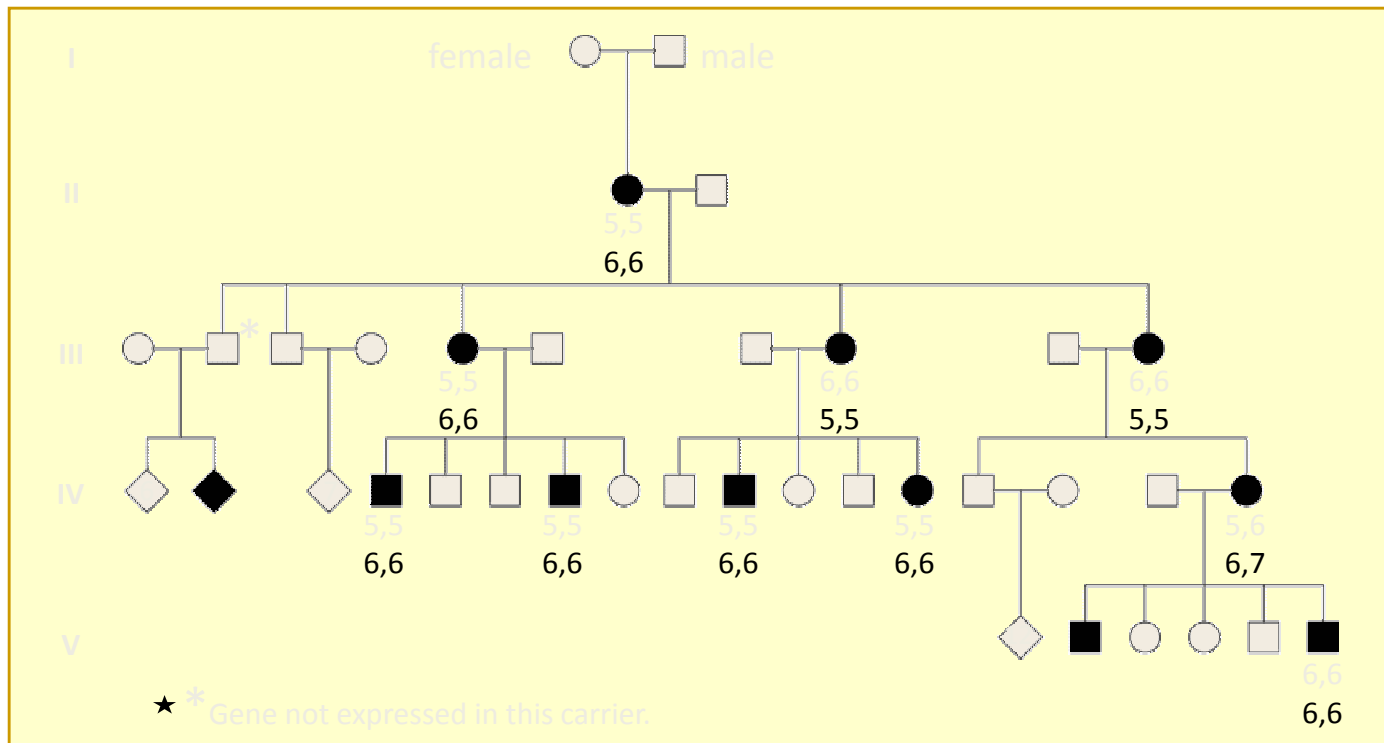


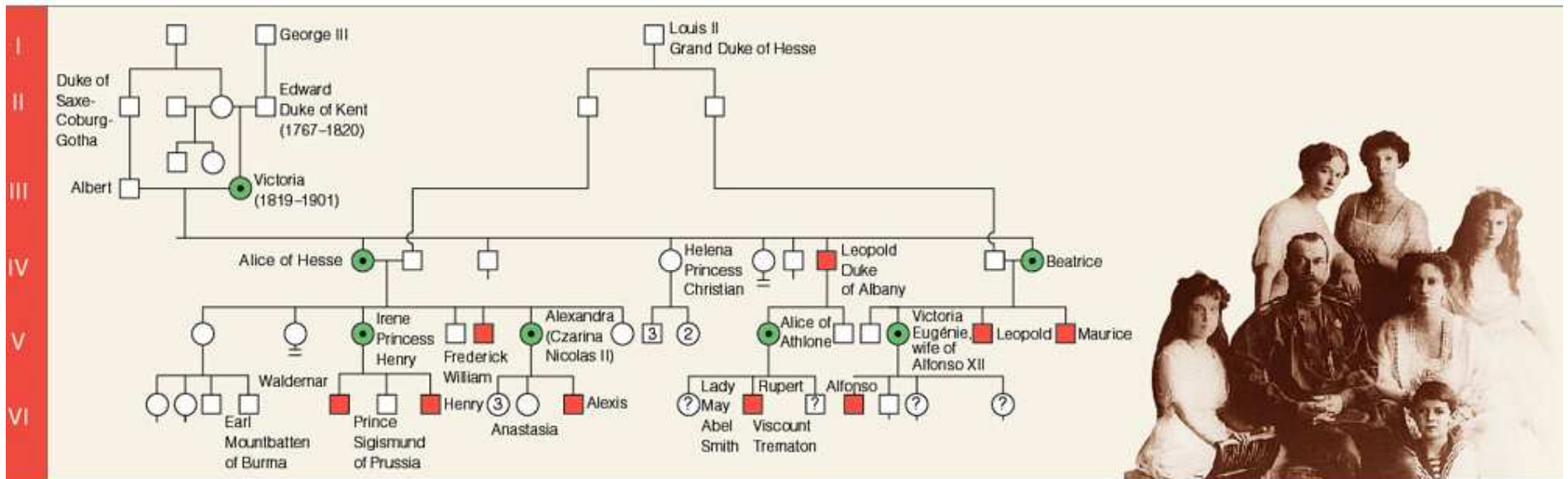
sex not specified

I, II, III, IV...

generation

Pedigree for Polydactyly





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Fig. 8-28, p.129