Basic Genetics

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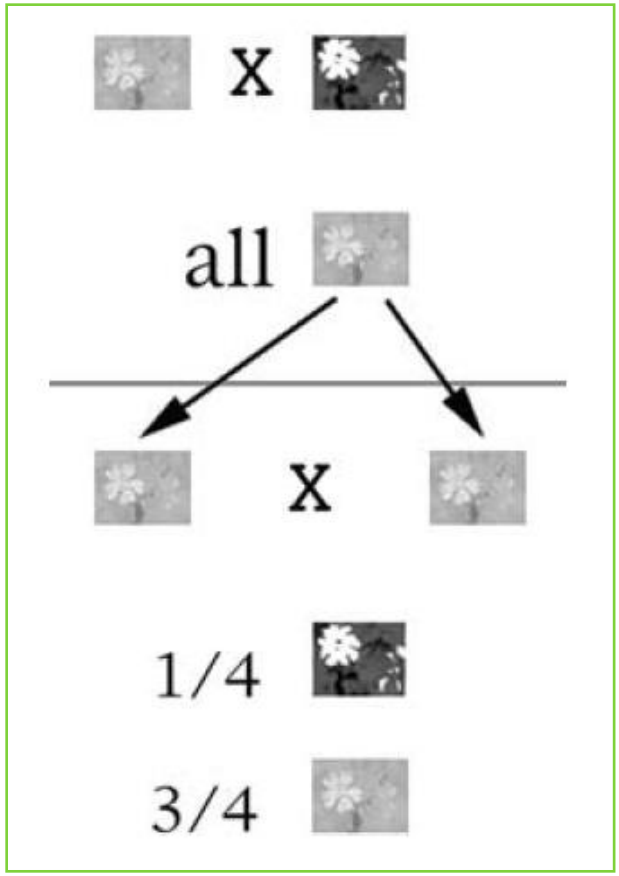
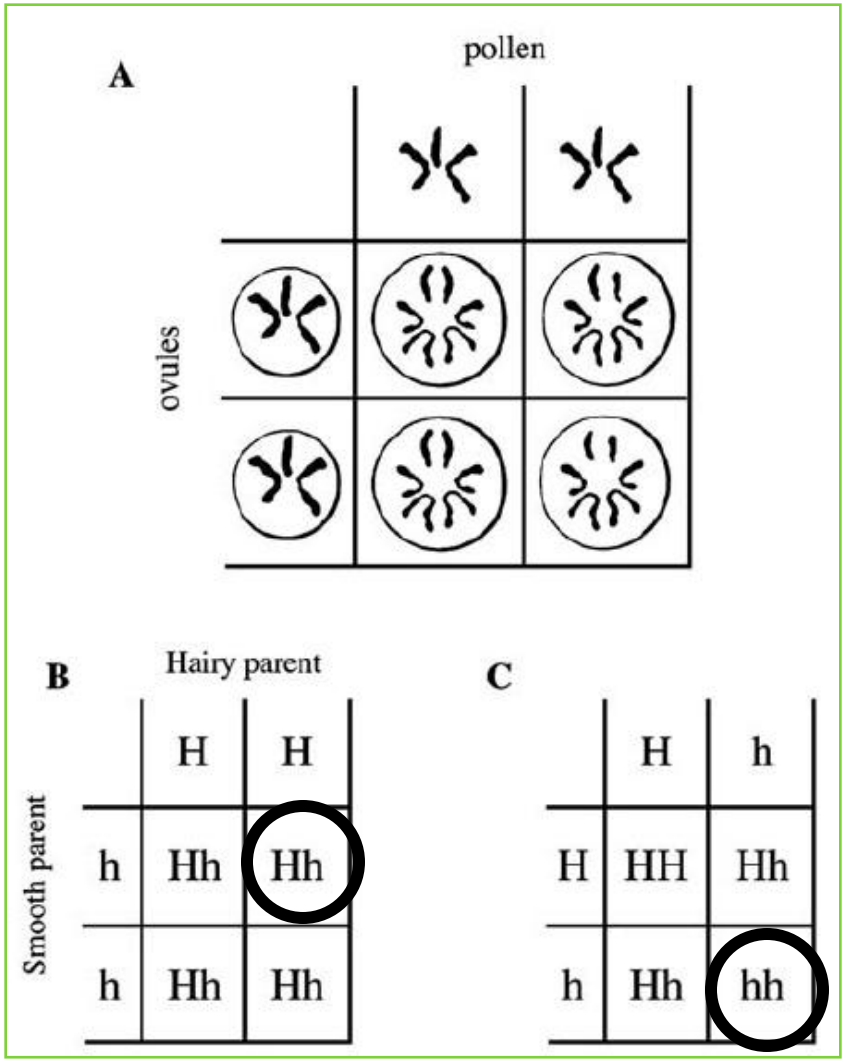
Inheritance in Single-Gene Traits



- Each parent provides equal genetic contribution
- Characteristic: dominant = visible; recessive = not visible
- In gametogenesis, these characteristic are separated/segregated → Segregation (I)
- If there are more than one traits, the genes will be separated and passed independently to offspring → Independent assortment (II)

The Punnet Square



- Plant with hairy-leafed x smooth-leafed
  - Hairy leafed (H)=dominant → determine phenotype
  - Smooth-leafed (h)=recessive
- Hh=heterozygotes
- HH/hh=homozygotes

NOTE:

- Genotype vs Phenotype
- Incomplete Dominance

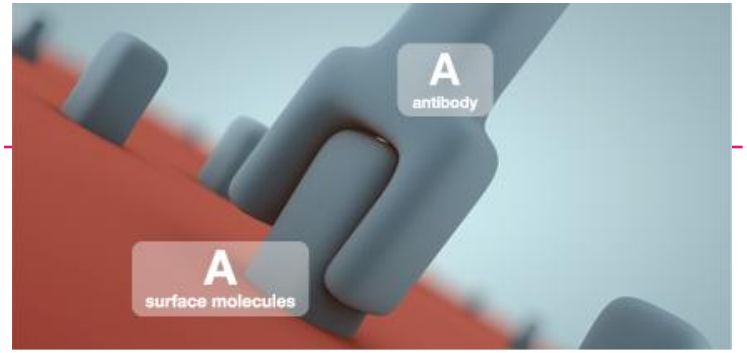
Mendelian/single gene inheritance

- Classical patterns:
  - Autosomal: dominant, recessive
  - X-linked: dominant, recessive
- Non-classical patterns

Blood Types

- Blood Types,
  - Depend on antigen/agglutinogens
- <u>Codominant</u>

Blood group	Antigen(s) present on the red blood cells	Antibodies present in the serum	Genotype(s)	Can receive blood from
A	A antigen	Anti-B	AA or AO	
B	B antigen	Anti-A	BB or BO	
AB	A antigen and B antigen	None	AB	
O	None	Anti-A and Anti-B	OO	

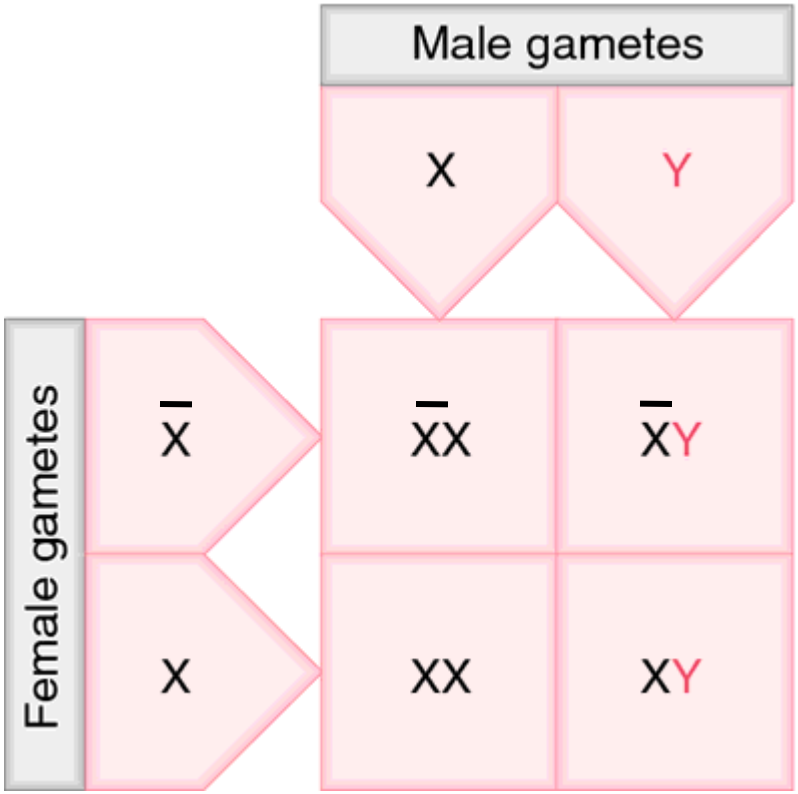


blood type	red blood cell surface molecules	plasma antibodies
type A	A only	B only
type B	B only	A only
type AB	A & B	neither
type O	neither	both

- http://learn.genetics.utah.edu/content/begin/traits/blood/

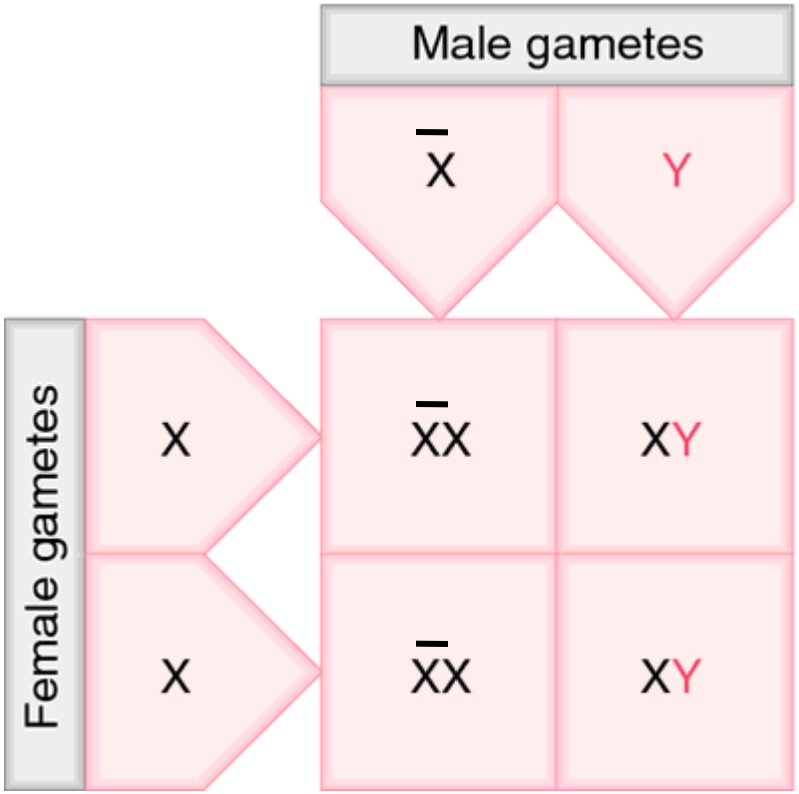
Hemophilia

- X-linked traits
- Defects in gene located on the x chromosome
- The possibilities are:
  - Male normal
  - Male affected
  - Female normal
  - Female carrier



Color Blindness

- Color Blindness: total, partial
- The possibilities are:
  - Female normal
  - Female carrier
  - Male normal
  - Male affected

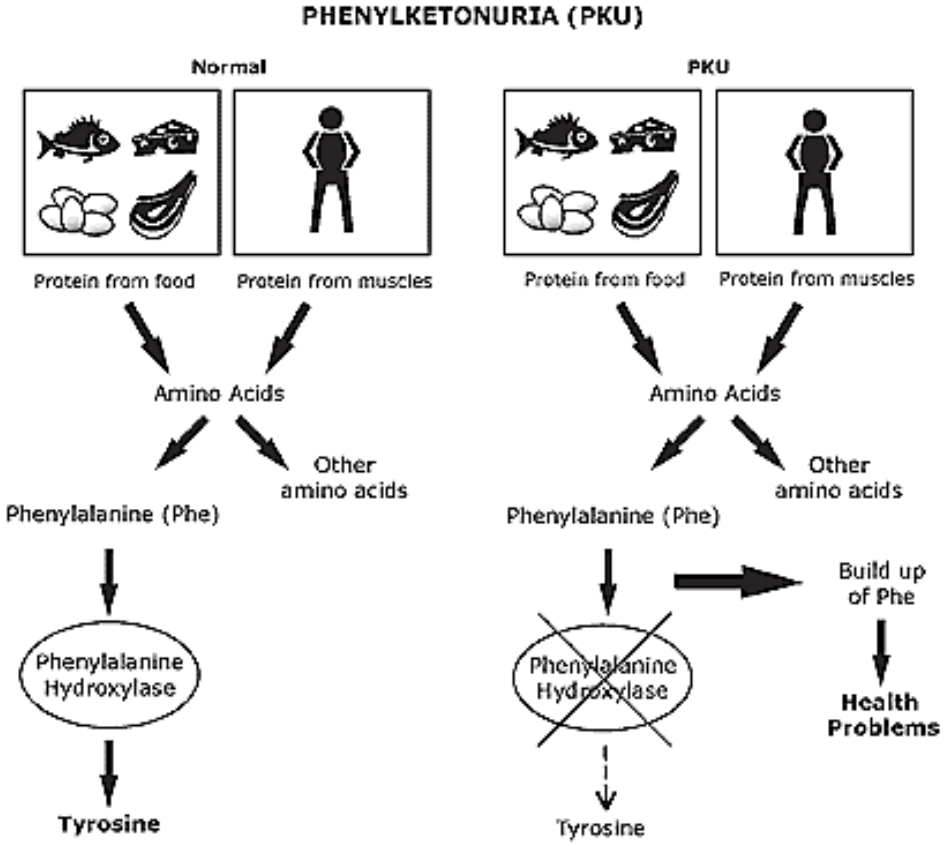


Prostate and Breast Cancer

- Cancer can caused by a single gene defect or other factors
- Prostate and breast cancer are now known to have a genetic basis
  - Prostate cancer: *HPC* gene (HPC1)
  - Breast cancer: *BRCA1* and *BRCA2* gene → the risk of getting breast ca is 92% but only about 5-10% out of all breast ca patients.

Genetic Metabolic Diseases

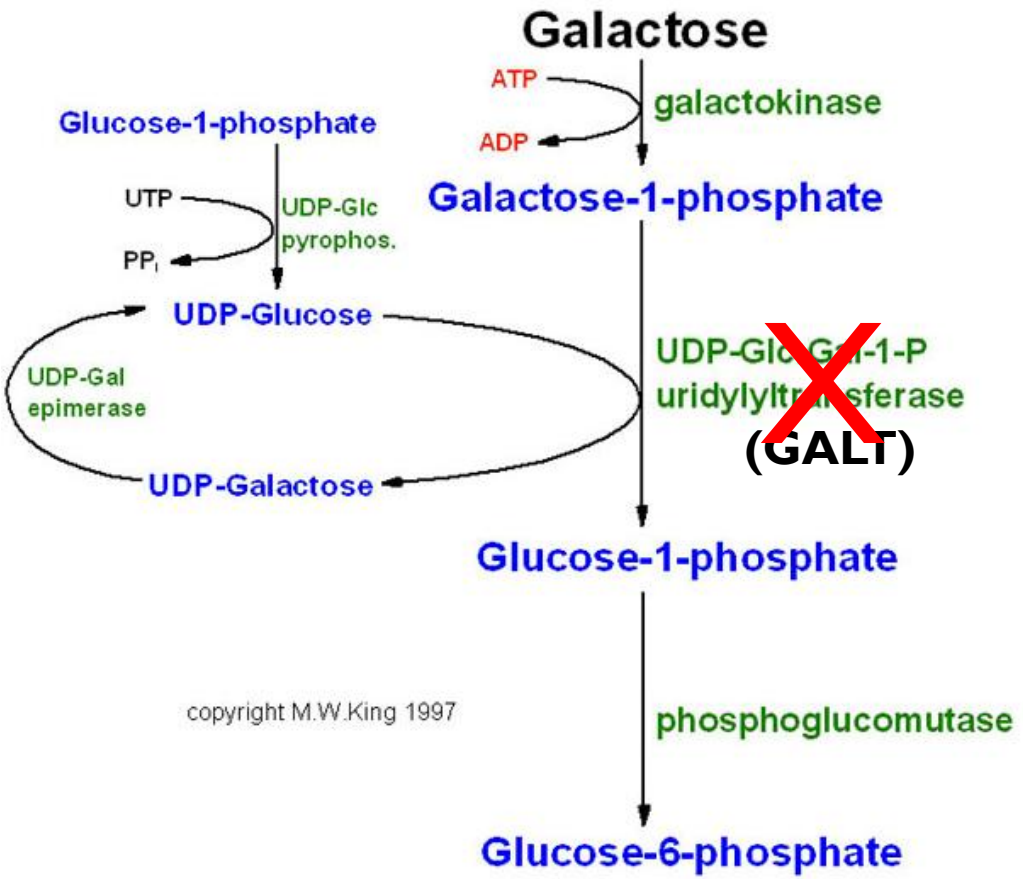
- PKU (Phenylketonuria)
- Autosomal recessive
- Can cause mental retardation
- Screening for PKU



- **Aspartame → aspartic acid and phenylalanine**

Genetic Metabolic Diseases

- Galactosemia
- Autosomal recessive
- Lactose (milk) → galactose and glucose
- Vomit and diarrhea a few weeks after birth



Disorders of Carbohydrate Metabolism - Galactosemia

- Classic galactosemia
  - Deficiency of galactose-1-phosphate uridyl transferase - - - -> accumulation of galactose-1-phosphate in many tissues
  - Failure to thrive, infantile cataracts, mental retardation, progressive hepatic failure leading to cirrhosis and death
  - Prevention – early removal of galactose from the diet
- Galactokinase-deficiency galactosemia
  - Much less frequent than classic type
  - Often marked by only infantile cataracts

Genetic counseling

- Definition:
  - a process of communication and education which addresses concerns relating to the development and/or transmission of a hereditary disorder

- For whom genetic counseling:
  - Parents with a previous child with a (possible) genetic disorder
  - One of the parents has a (possible) genetic disorder
  - Patient(s) in the family with a (possible) genetic disorder
  - Consanguinity of parents
  - Exposition to teratogenic/mutagenic drug

Cont'd…

- Consultand is provided with these following informations:
  - The medical dx, its prognosis and possible treatment
  - The mode of inheritance of the disorder and the risk of developing and/or transmitting it
  - The choices or options available for dealing with the risks

- Steps of genetic counseling:
  1. Establishing the dx
     - Taking a family history; examination
  2. Calculating and presenting the risk
  3. Discussing the options
  4. Communication and support

Cont'd

- Estimation of recurrence risk usually require :
  - The diagnosis and its **mode of inheritance**
  - Analysis of the **family pedigree**
  - The result of **test** (linkage study, clinical data)

Estimation of Risk in Mendelian Disorders

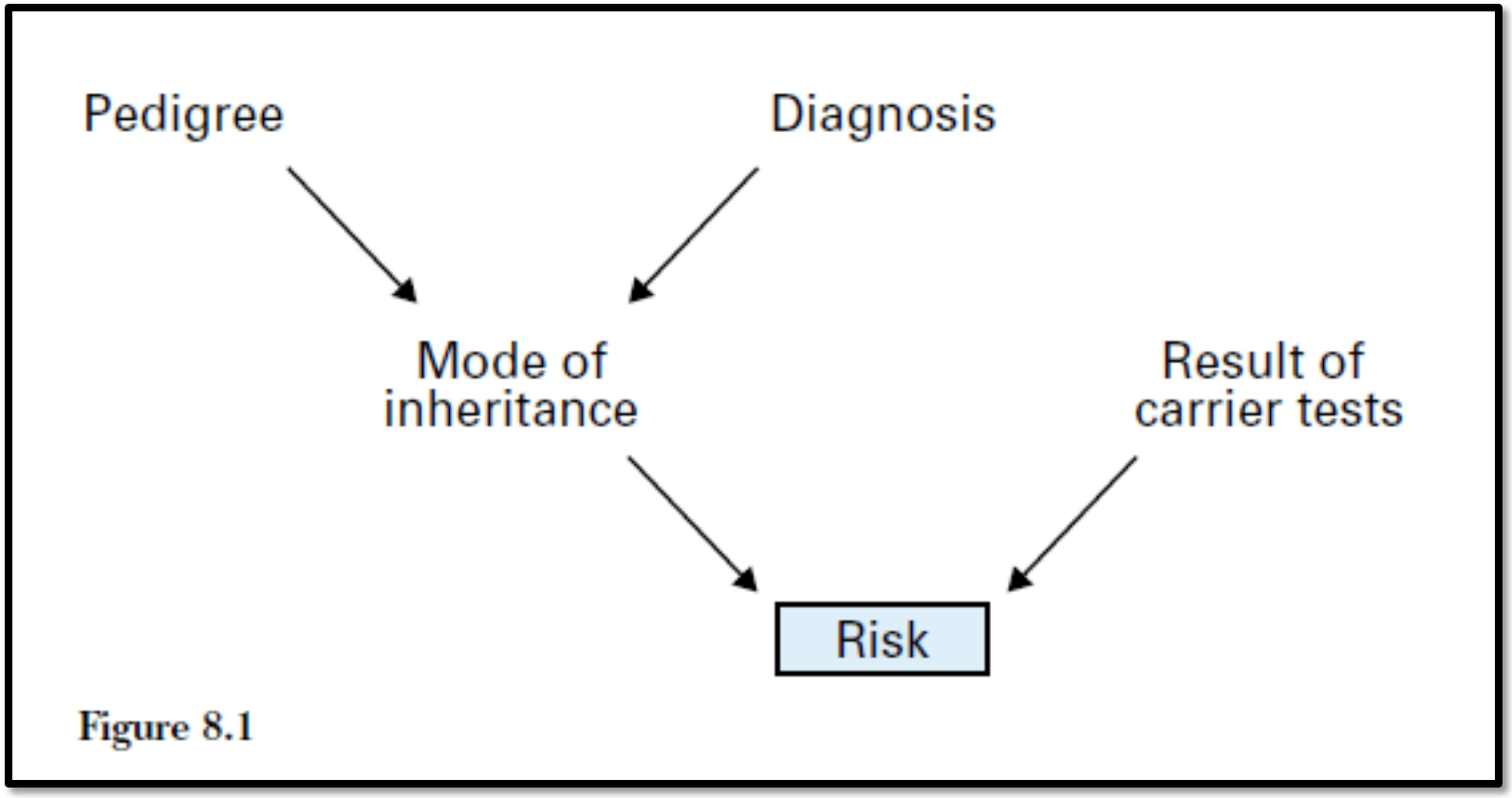
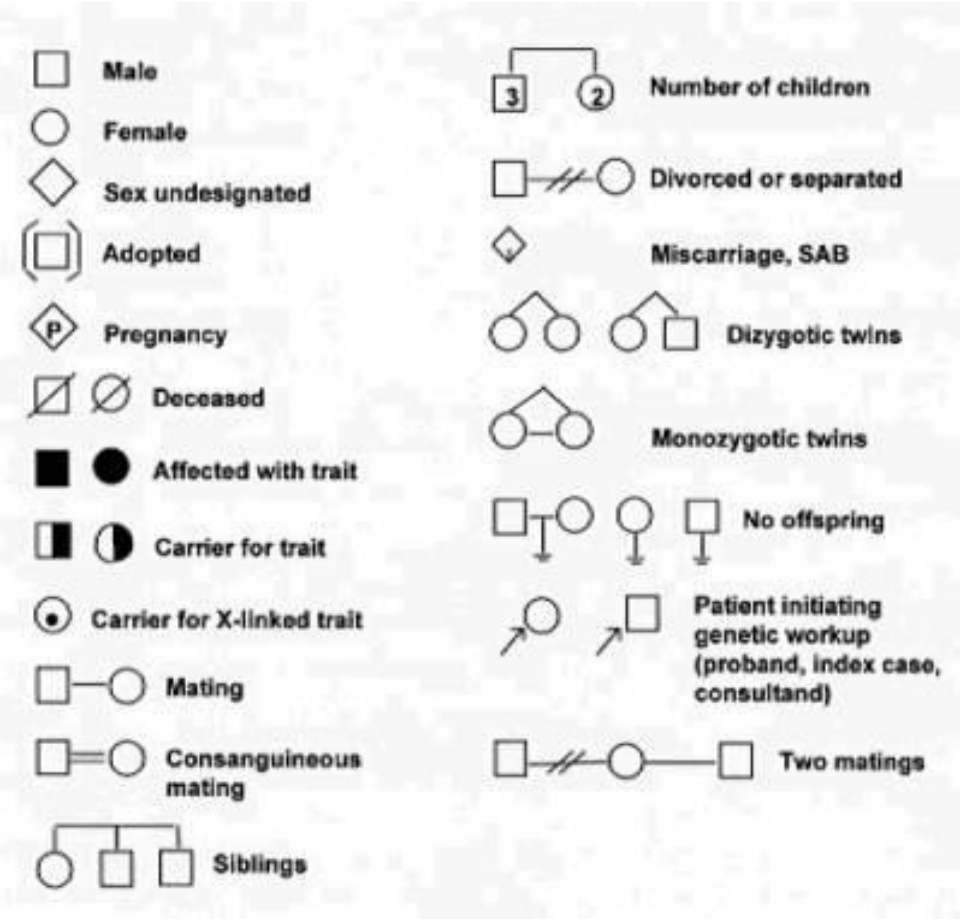


Figure 8.1

Risk calculations

- In genetic counselling, we want accurate risk assessment for families with genetic disease
- What kinds of information can be used?
  - **Pedigree**
  - **Biochemical**
  - **Karyotiping**
  - **DNA**

Drawing pedigree

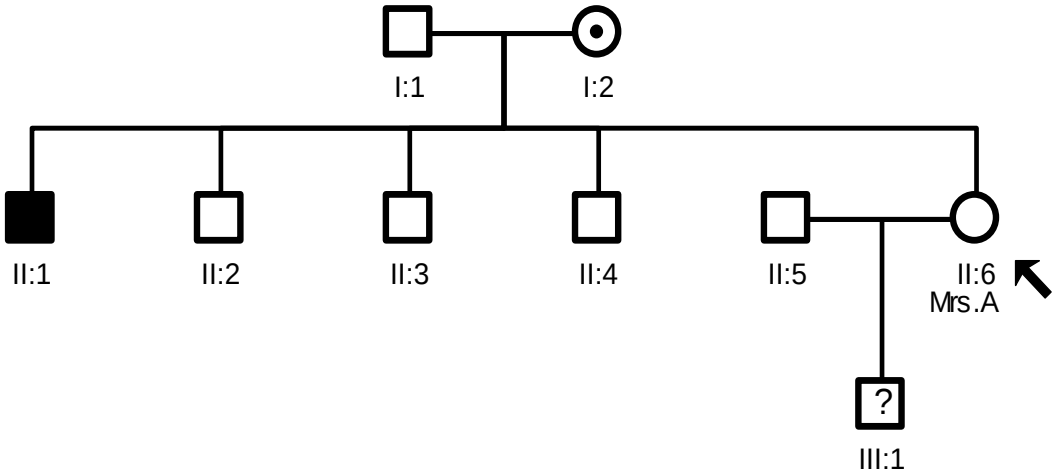


I, II, III, etc = Generations are numberered from the top of the pedigree

III_1 , III_2, III_3, etc = Individuals in each generation

Discussion

- Mrs A has a brother with a severe haemophilia A. He died due to a cerebral bleeding at the age of 8 years. She has also 3 healthy brothers. There is no other affected family member.
  - *X-linked recessive; Skipping generation and not all male affected*



  - The chance for Mrs A to be a carrier for haemophilia A: *50%*
  - The possibility of the son to affected hemophilia <u>if his mother is carrier</u> is *50%*

References:

- Omoto CK & Lurquin PL. 2004. Genes and DNA: A Beginner's Guide to Genetics and Its Applications. Columbia University Press, New York. (xviii+217 pages)