Examples of pedigrees

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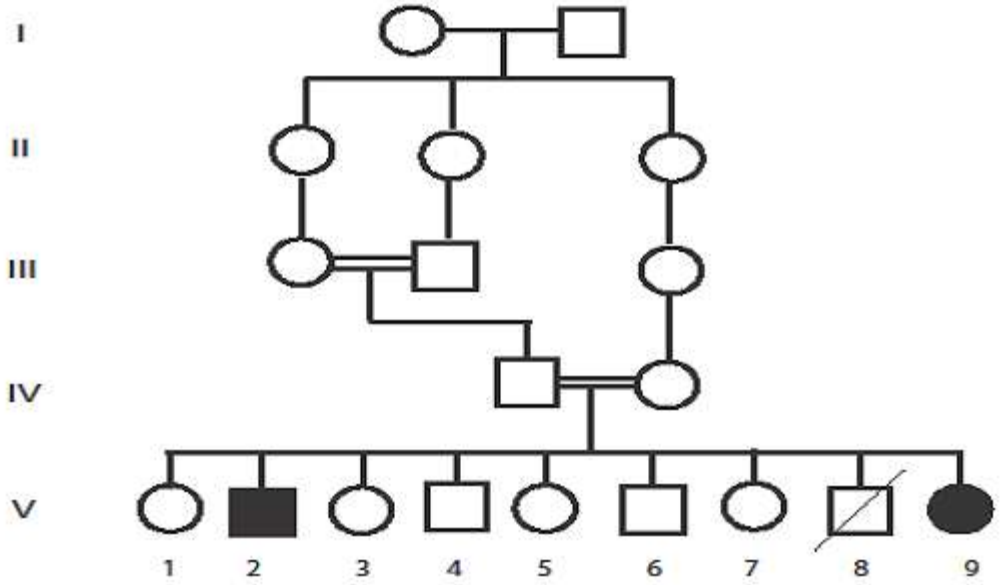
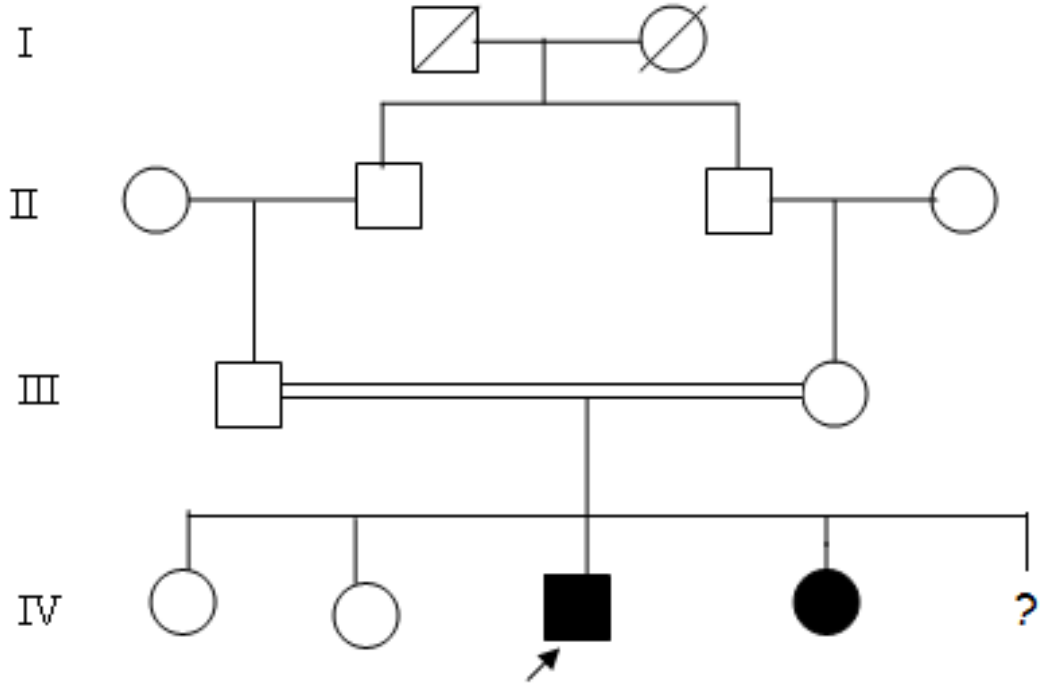
Pedigree 1
What is the most probable mode of inheritance in this pedigree?



Answer
Pedigree 1

- There are two affected siblings, a girl and a boy (black symbols)
- The parents are relatives (consanguineous) as indicated by the double line drawn between the symbols of mother and father and the pedigree structure
- The parents are not affected
- This is horizontal transmission
- Most probably the mode of inheritance is autosomal recessive
- Both parents are carriers of the abnormal gene, although do not show any of the features of the disease
- Recurrence risk, that is the risk of having another affected is 25% in each pregnancy
- To know if any of the non affected brothers and sisters is a carrier of the abnormal gene, molecular studies are indicated. The risk of being a carrier is around 66% (2/3)

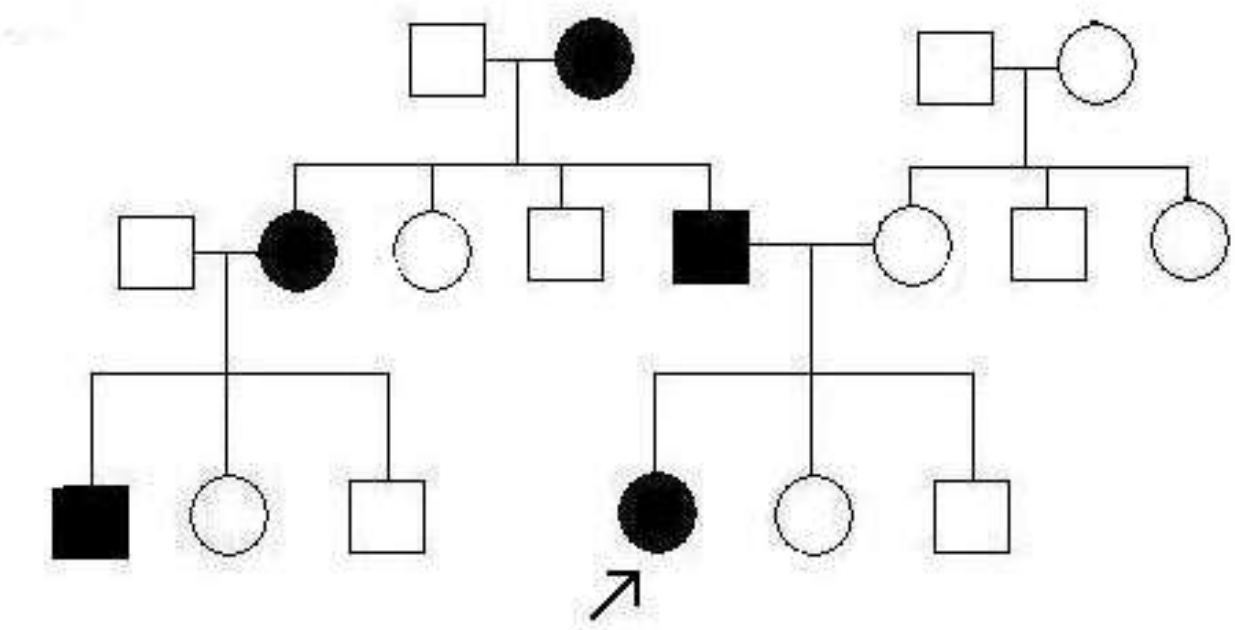
Pedigree 2:
What is the risk that the fetus is affected?
0%, 5%, 25%, 50%



Answer
Pedigree 2

- It is 25%
- Read answer to pedigree 1
- Both parents are carriers, so their genotypes are Dd And Dd where D is the normal allele and d is the abnormal allele. The probabilities for children are:
- 25% DD
- 50% Dd (carriers)
- 25% dd (affected)

Pedigree 3
What is the most probable mode of inheritance in this pedigree?



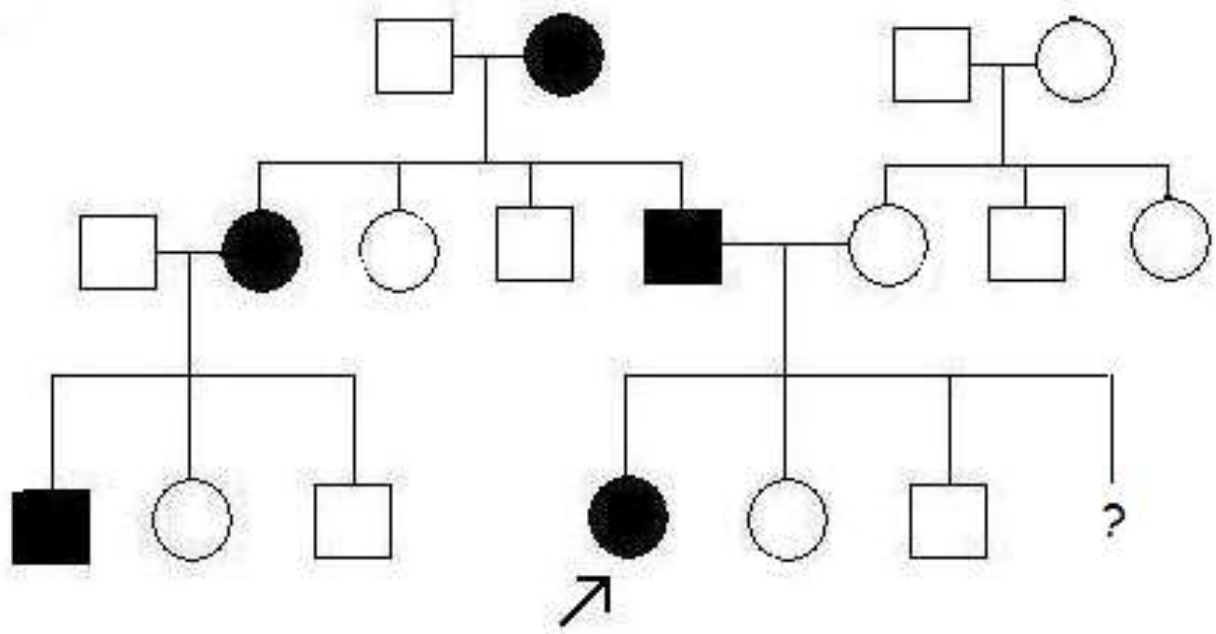
Answer
Pedigree 3

- The proband is a girl (marked by the arrow)
- Her father is affected
- Her parents are not related
- She has a paternal aunt, a cousin and her paternal grandmother who are also affected
- It is a vertical transmission
- Both males and females are affected
- Most probably this is an autosomal dominant mode of inheritance

Pedigree 4

What is the risk that the fetus is affected?

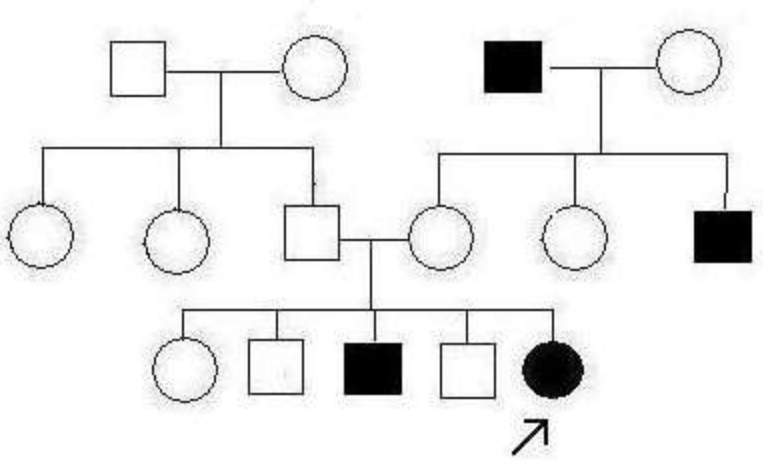
0%, 5%, 25%, 50%



Answer
Pedigree 4

- Since this is most probably an autosomal mode of inheritance and the father is affected, the risk to have an affected baby is 50%
- The father genotype is Dd where D is the abnormal allele and d is the normal allele
- The mother is dd
- In each pregnancy there is a 50% chance of having a nonaffected child dd and 50% chance to have an affected child Dd

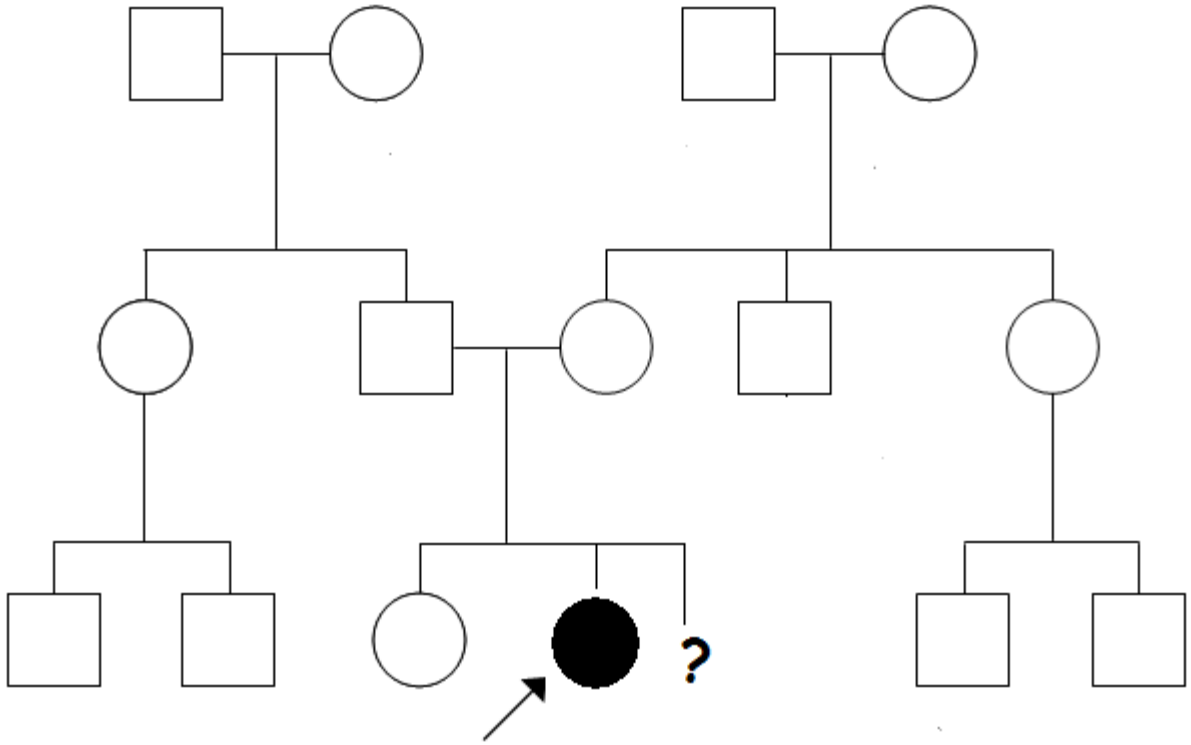
Non-penetrance in autosomal dominant conditions



- There is usually no carrier status for autosomal dominant conditions although a non affected person with an abnormal allele could be seen and this is called non-penetrance as in the mother of proband in this pedigree

Pedigree 5

The shaded symbol is a baby affected by spina bifida
(Neural tube defect)
What is the risk that the fetus is affected by the same condition?
0%, 3%, 10%, 50%



Answer pedigree 5

- Spina bifida usually has a multifactorial (MF) mode of inheritance where both environmental and genetic factors play a role.
- For MF conditions, the risk of recurrence is emperical, that is observational. If one child is affected, the risk to have another affected child is 2-4%.