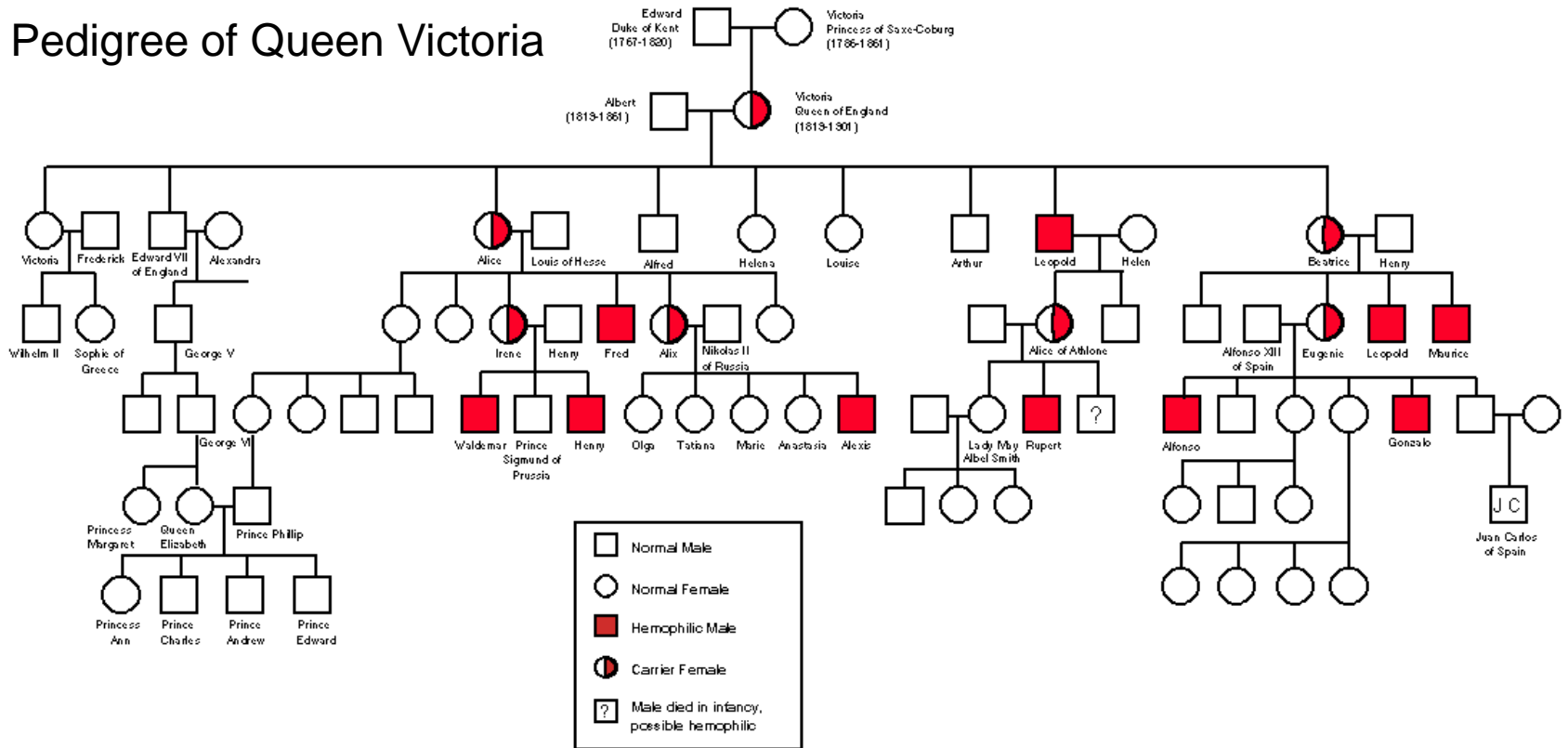


Pedigree Analysis

Pedigree of Queen Victoria



What is a pedigree?

- Shows a pattern of inheritance in a family for a specific trait (phenotype)
- Genotypes can usually be determined
 - Why would we want to use a pedigree in genetics?
- Track the occurrence of diseases such as:
 - *Huntington's* – simple dominant – lethal allele – causes breakdown of the brain
 - *Cystic fibrosis* – 1/2500 – mucus accumulates (white North Amer.)
 - *Tay-Sachs disease* – lipids accumulate in CNS (Jewish)
 - *Phenylketonuria* – missing enzyme causes problems in CNS (Nordic/Swedish)

The Symbols used:



Unaffected male



Affected male



Unaffected female



Affected female



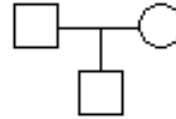
Person whose sex is not known



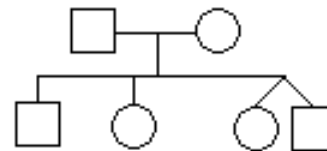
Marriage (mating)



Consanguinous marriage



vertical line = offspring
(in this case, son)



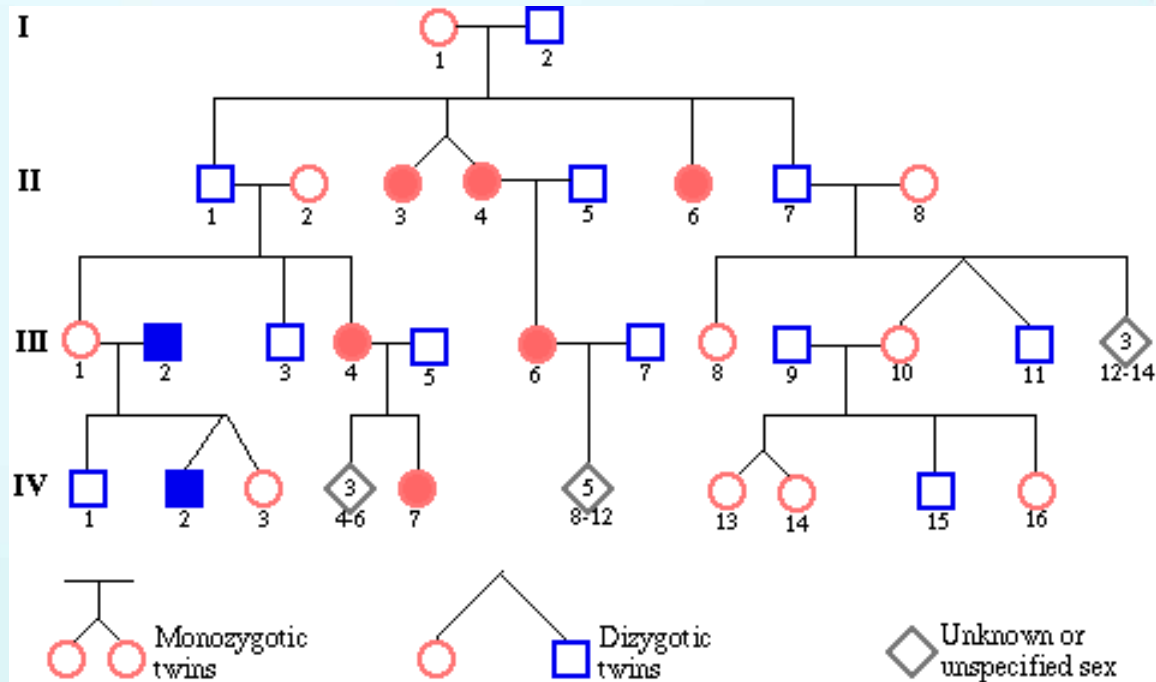
A family of four brothers and sisters. The last two are non-identical twins



Identical twins

Sample pedigree:

- generations are numbered with Roman Numerals
- oldest offspring are on the left



How many males are present? How many females?

How many females show the trait being studied?

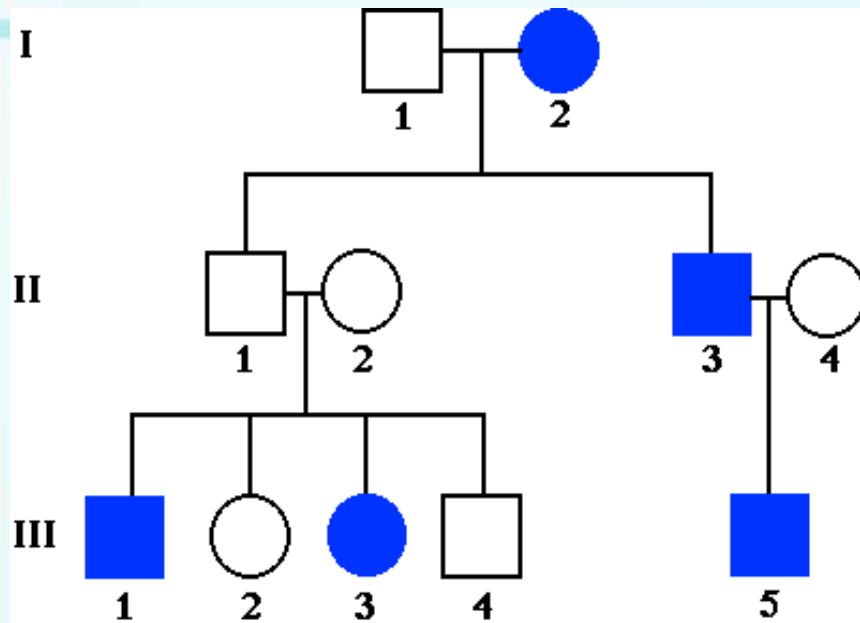
What is the sex of offspring III-9?

How many offspring did the generation I parents have?

What is the difference between the II-3 & 4 and IV-2 & 3?

Inheritance patterns:

- **Autosomal dominant:**



Pedigree 2. An idealized pedigree demonstrating the effects of incomplete penetrance.

The disease is passed from the father (II-3) to the son (III-5), this never happens with X-linked traits.

The disease occurs in three consecutive generations, this never happens with recessive traits.

Males and females are affected, with roughly the same probability.

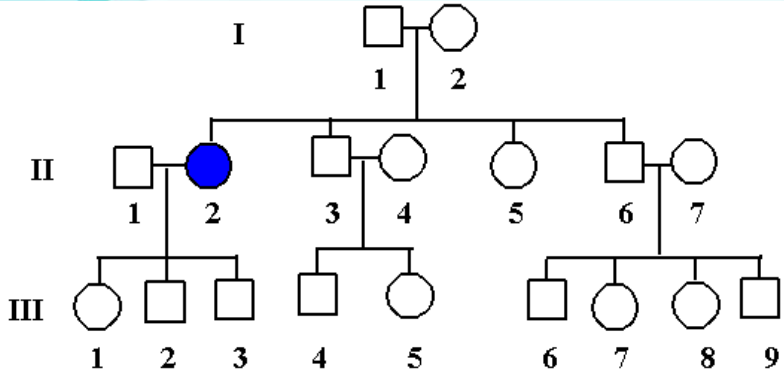
–Examples: Polydactyly

–Huntington's disease

Inheritance patterns:

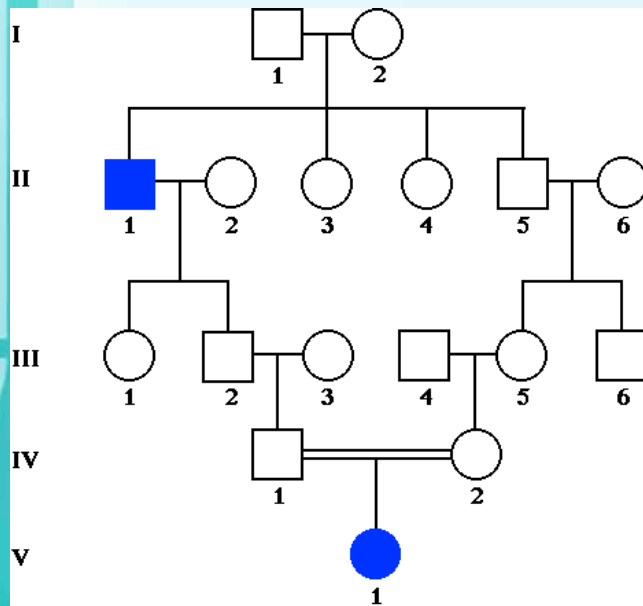
• Autosomal recessive

		Father's Gametes	
		D	d
Mother's Gametes	D	DD	Dd
	d	Dd	dd



Males and females are equally likely to be affected.

The recurrence risk to the unborn sibling of an affected individual is 1/4.



The trait is characteristically found in siblings, not parents of affected or the offspring of affected.

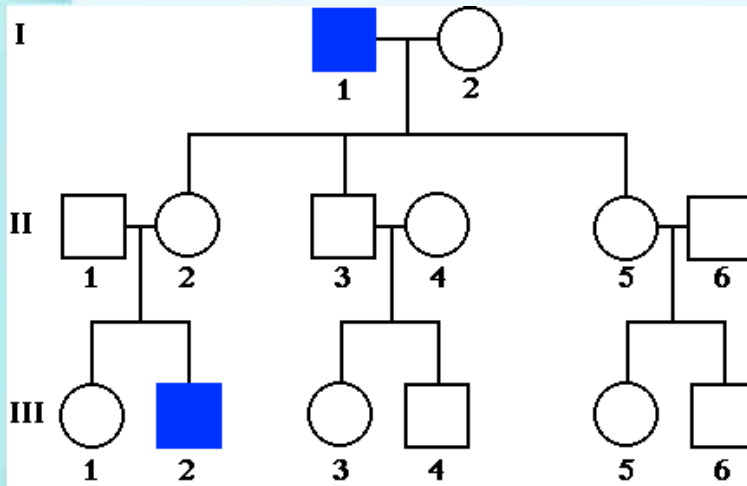
Parents of affected children may be related. The rarer the trait in the general population, the more likely a consanguineous mating is involved.

–Cystic fibrosis

–Tay-Sach's disease

Inheritance patterns:

- **Sex-linked recessive conditions**



Pedigree 7. X-linked recessive inheritance.

The disease is **never passed from father to son.**

Males are much more likely to be affected than females.

• **All affected males in a family are related through their mothers.**

Trait or disease is typically passed from an affected grandfather, through his carrier daughters, to half of his grandsons.

– **Examples:**

– **Colour-blindness**

– **Duchenne Muscular Dystrophy**