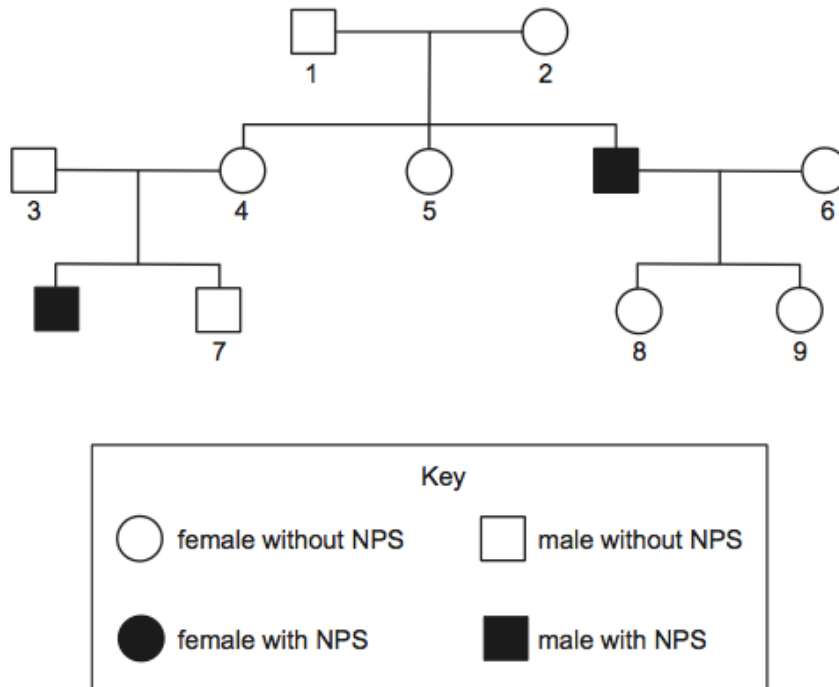


- 17 The below diagram shows a family tree of a condition known as nail patella syndrome (NPS).



Which of the following pairs of individuals **must** be heterozygous for NPS?

- A 1 and 5
- B 2 and 6
- C 3 and 7
- D 4 and 8
- E 5 and 9
- F 6 and 7

This is obviously a recessive condition, as neither 1 nor 2 are affected, but they have offspring who is. Incidentally, in Cambridge biochemistry papers, they sometimes ask the question “What type of inheritance is this? Explain your answer”. If it’s autosomal dominant (autosomal means not-on-a-sex-chromosome), then the reasoning is usually “every affected individual has an affected parent” – so if this isn’t the case, like in the pedigree above, it’s likely to be recessive.

Anyway, the easiest way to go about this is to go through the options and eliminate, rather than working out who must be heterozygous and then looking at the options.

A – 1, yes because if he were homozygous dominant (AA), none of the offspring would be affected. As a general rule, the parents of an individual with a recessive disease (aa) must both

be carriers (Aa). 5- not necessarily, because if both parents are heterozygous, then there's a 25% chance that the offspring is homozygous dominant (AA).

B – 2, yes because of the reason stated above. 6: not necessarily because none of the offspring is affected.

C – 3, yes because of the same reason as the previous two. 7, not necessarily because if both parents are Aa, then there's a 25% chance that 7 is AA.

D – 4 must be heterozygous because she's the parent of an affected child. 8 must also be heterozygous because she's the daughter of an affected parent (aa), so regardless of the mother's genotype, the daughter will have a recessive allele from the father. That makes this the correct answer, and we don't need to bother looking at the others. However, for the sake of completeness, let's do so anyway.

E – 5 doesn't have to be heterozygous as we explained earlier, even though 9 does (For the same reason that 8 does).

F – Neither have to be heterozygous, and reasons for both have already been said.