3.02 Genetics in Primary Care - Cystic fibrosis

Naomi and James are hoping to try for their second baby. Their first child, Adam is 5 years old and has cystic fibrosis. They are worried about a future child developing the condition.

Provide: You've come today to discuss the chances of your next child having cystic fibrosis.

Elicit: Do you know much about how the condition is inherited?

Naomi: I know that it runs in the genes in some people. But I want to know what the chances are if we get pregnant again that we'll have another CF baby. It's been hard with Adam.

Provide: Cystic fibrosis is only present when a baby gets two faulty copies of the Cystic fibrosis gene. One from each parent. Because each of you do not have cystic fibrosis yourselves but your other child has cystic fibrosis each of you must have one faulty Cystic fibrosis gene each.

Elicit: with me so far?

James: Yes, so we must both be carriers?

Provide: Yes that's right James. When you have a child only one child in every 4, on average, will get 2 faulty cystic fibrosis genes. Let me show you this with a picture. As you can see there is a 3 in 4 chance that your baby won't have Cystic fibrosis. But half of your children may have one faulty Cystic fibrosis gene. 1 in 200 people in the general population have one faulty gene for cystic fibrosis.

Elicit: does that make sense?

Naomi: Yes. How will we know for sure?

Provide: There are some tests that the obstetric doctors can do to find out if your baby is likely to have cystic fibrosis. The tests aren't perfect and each carries about a 1% risk of causing a miscarriage.