

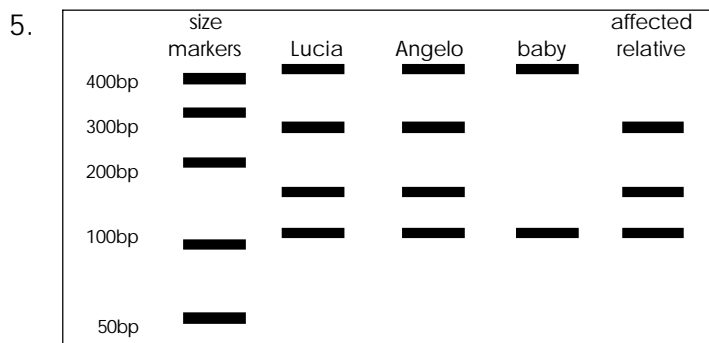
Will My Baby Be Ill?

Learning Objectives

- Polymerase Chain Reaction.
- Restriction Enzymes.
- Gel Electrophoresis.
- Fetal tissue sampling methods.

Answers

1. A nonsense mutation is a change in base sequence which introduces a stop codon, resulting in premature termination of translation.
2. Mael, because it recognises a sequence that is present in one allele (ie the $\beta 39$ allele) but not in the other (ie normal β -globin allele). This means that digestion of the region of DNA in which this sequence lies, with the Mael enzyme, will produce different sized DNA fragments which can be visualised using gel electrophoresis.
3. The essential ingredients of PCR are:
 - DNA
The DNA sample which you wish to amplify.
 - DNA polymerase
This is an enzyme found in all cells which replicates DNA. The DNA polymerase used in PCR is derived from a bacterium which lives in thermal springs, called *Thermus aquaticus*, or Taq for short. Because of the temperature of its natural environment, Taq polymerase is resistant to high temperatures, unlike DNA polymerase from humans and other organisms. This resistance to high temperature is useful because during the denaturation step of PCR, the reaction solution is subjected to very high temperatures of about 95°C.
 - Primers
Short single-stranded pieces of DNA which are complementary to regions of DNA on either side of the region you wish to amplify. After the DNA sample is "melted" - that is, separated into two strands - the primers anneal (bind) to the sample, and give the DNA polymerase a starting point to start building more DNA.
 - Nucleotides
Free nucleotides of adenine, thymine, cytosine and guanine (A, T, C & G) are added to the solution from which the new strands of DNA are built.
 - Buffer
The ingredients are mixed in a solution containing salts and other chemicals which help the polymerase to function.
4. Cells could be obtained from the fetus at this stage of pregnancy by chorionic villus sampling (CVS). This is the most common method of obtaining fetal cells at 10-12 weeks of pregnancy. CVS involves passing a very fine needle through the mother's skin into the uterus, to the placenta. A finer needle (inside the first needle) and a syringe are used to draw some tissue from the placenta. Placental tissue has the same genetic make-up as the fetus, but is not a part of the fetus. Amniocentesis can also be used to obtain fetal cells, by taking a sample of the fluid surrounding the fetus, but this is usually performed later in pregnancy, at 14-18 weeks.



Resources

Reference Sheets D and E

©2000 MCRI, QCGS and Gene CR