

Which ONE of the following statements is TRUE? Probability calculations are used in forensic applications of DNA fingerprinting to determine if:

DNA from two different sources have matching alleles.

DNA samples were degraded before analysis.

A match between alleles of different DNA samples might have occurred by chance.

An unknown DNA sample is from a male or female.

Alleles in the suspect's DNA are different from alleles of a victim's DNA.

Which ONE of the following statements is TRUE? VNTR sequences are very useful in DNA forensic and population genetic analysis because:

Their centromeric location aids chromosomal analysis.

The high frequency of alleles in the population means that homozygotes are rare.

They can easily be detected in Northern blots.

The low frequency of alleles in the population means that homozygotes are rare.

They are rarely sex linked.

Which TWO of the following statements regarding VNTR sequences are TRUE:

The variation in repeat lengths probably arise due to unequal crossing over.

Repeated sequences in a tandem array are only ever found at a single chromosomal locus.

Southern blot hybridization of restriction enzyme digests of human DNA to VNTR probes reveals DNA "fingerprints".

VNTRs are generally of the order of 10-80kbp.

VNTRs are never found within genes.

Which ONE of the following statements regarding the human mitochondrial genome is/are TRUE:

The human mitochondrial genome has 370 genes.

The human mitochondrial genome is a single, linear chromosome.

The human mitochondrial genome encodes several tRNA genes.

The human mitochondrial genome was sequenced during the Human Genome Project.

The DNA sequence of the control region exhibits little variability between individuals.

Which ONE of the following statements is TRUE? Buffer AL contains Tris-HCl buffer. This is used in the Cheek Cell DNA isolation because:

It maintains an alkali pH to ensure RNA hydrolysis.

It maintains an acid pH to ensure depurination of the DNA.

It chelates Mg^{2+} ions that are necessary for nuclease activity.

It ensures an ionic strength necessary to avoid strand denaturation.

It maintains a neutral pH that helps nucleic acid stability.

Which ONE of the following statements is TRUE? The QiaAmp spin columns are an example of spin-column based nucleic acid purification. These columns use which of the following compounds to bind DNA under appropriate ionic conditions:

- A. Agarose.
- B. Silica.
- C. Cellulose.
- D. Polylysine.
- E. Sepharose

Which TWO of the following statements are TRUE? The AL buffer causes cellular lysis because it contains:

- A. Detergent.
- B. Lysozyme.
- C. Protease.
- D. Mg^{2+} .
- E. NaCl

The sequence of the upstream, 5' primer that you used for the pMCT118 (aka DS180) VNTR is 5'-GAAACTGGCCTCCAAACACTGCCCGCCG-3'. The sequence of the downstream, 3' primer that you used was 5'-GTCTTGTGGAGATGCACGTGCCCCTTGC-3'. On the basis of this information, which TWO of the following statements are TRUE:

The sequence primed by the 3' primer ends with the DNA sequence 5'-CGGCGGGCAGTGTGGAGGCCAGTTTC-3'

The 5' primer anneals to the sequence 5'-GAAACTGGCCTCCAAACACTGCCCCGCCG-3'.

The sequence primed by the 5' primer ends with the DNA sequence
5'-GTCTTGTTGGAGATGCACGTGCCCCTTGC-3'.

The 3' primer anneals to the sequence 5'-GTCTTGTTGGAGATGCACGTGCCCCTTGC-3'.

The 3' primer anneals to the sequence 5'-GCAAGGGGCACGTGCATCTCCAACAAGAC-3'.

For the mtDNA reaction, the thermal cycler was programmed with the following step file:

94°C for 6 minute initial denaturation and enzyme activation.

35 cycles of (94°C - 30 sec/ 51°C - 30 sec/ 72°C - 1 min)

72°C for 10 minute final extension

Link to a 4°C soak file.

Which of the following statements regarding this PCR are TRUE:

The 94°C incubation is to allow annealing of the primers to the cheek cell DNA.

The 51°C incubation is to allow annealing of the primers to the cheek cell DNA.

The 72°C incubation is to allow extension of DNA synthesis by the thermophilic polymerase.

The 35 cycles should produce 10^{35} molecules from each molecule of DNA in the starting solution.

The 4°C soak allows continued extension by the thermophilic polymerase.

Which THREE of the following statements regarding the mtDNA sequencing trace files (.ab1) are TRUE:

The size of the peaks corresponds to the intensity of fluorescence.

The first few bases are usually of higher quality than the others.

The first few bases are close to the primer annealing site.

The signal intensity of the CC_MITODNA1_A07 sequence is A=233,C=202,G=103,T=183 (file in the Test Sequences folder on DropBox).

The CC_MITODNA1_A07 chromatogram shows more than 400bp of readable sequence.

Which TWO of the following statements regarding the agarose gels of the mtDNA PCR product are TRUE:

There do not seem to be any non-specific PCR products on the gel.

The single band visible at approximately 1.7kbp corresponds to primer-dimer artefacts.

Heterozygous individuals show two bands on the gel.

The 1.7kbp band present in some individual lanes is consistent with product of the expected size.

Some lanes do not contain any bands indicating a failure to amplify the expected DNA.

Which TWO of the following statements regarding mitochondrial inheritance are TRUE:

Due to recombination, mutations in mitochondrial DNA tend not to be inherited together.

Males pass on mitochondrial DNA specifically to their daughters.

Females pass on mitochondrial DNA due to the large cytoplasm of the egg.

Mitochondrial DNA haplotypes are clusters of mutations that are inherited within specific lineages.

Mitochondrial DNA sequence variation is least amongst African populations.

Which TWO of the following components must be present in the PCR mixes that you used?

- A. E.coli DNA polymerase.
- B. NTPs (ribonucleotide triphosphates).
- C. ddNTPs (di-deoxynucleotide triphosphates).
- D. Mg^{2+}
- E. Oligonucleotide primers

Which ONE of the following statements is TRUE? Alec Jeffreys first used VNTRs (or mini-satellites as they were originally called) to block the deportation of a Ghanese boy by proving his maternity. This was due to:

- A. Sharing all of the bands in a DNA fingerprint between the mother and son.
- B. Sharing of one band in a DNA fingerprint between the mother and son.
- C. Sharing of half of the bands in a DNA fingerprint between the mother and son.
- D. Comparing DNA fingerprints of the mother and son on a Western blot.
- E. Showing that the allele frequency of the myoglobin intron VNTR was greater in the mother than in the son

Which TWO of the following reasons help indicate that the sample identified as "HVR49" is unlikely to be accurately genotyped as haplogroup H2a2a1?

The low quality of the sequence calling means that it is impossible to align the HVR1 and HVR2 sequences and assemble them.

There are mutations identified that mean that it cannot be the same as the CRS.

The sequence appears to be from a PCR artefact because the sequence doesn't appear to have aligned at all to the CRS.

If you carefully edit the sequences and genotype them at the Haplogrep server it suggests a haplotype of H1e3.

The H2a2a1 haplogroup is the most common haplogroup in the world.

Which TWO of the following statements regarding mtDNA is/are TRUE?

- A. Yeast with defective mitochondria which are unable to carry out oxidative phosphorylation caused by large deletions in their mtDNA cannot grow.
- B. Liver cells have thousands of mtDNA genomes per mitochondrion.
- C. mtDNA genomes cannot recombine during meiosis.
- D. The D loop or control region of human mitochondrial DNA is hypervariable.
- E. Mutations in mtDNA accumulate more slowly than in nuclear DNA.

Which TWO of the following statements are types of DNA sequence variation?

- A. The most common type of DNA sequence variation in the human genome are VNTRs.
- B. The most common VNTR allele frequencies are less than 0.001 in human populations.
- C. The SNP mutation rate in humans is of the order of 3×10^{-8} per base pair.
- D. Indels are always caused by insertion of new bases into a chromosome.
- E. The length of a branch in a haplotype network corresponds to the number of DNA sequence differences between two haplotypes

Which TWO of the following statements regarding Single Nucleotide Polymorphisms (SNPs) is/are TRUE?

- A. SNPs are normally inherited in a non-Mendelian fashion.
- B. A-G changes (purine to purine) are more common than A-T changes (purine to pyrimidine).
- C. SNPs occur roughly every 100kbp in the human genome.

- D. SNPs in the mitochondrial DNA control region are more common than in the nuclear genome.
- E. The International HapMap Project focused on common SNPs whereby more than 10% of the population show either allele

Which TWO of the following statements about sequencing quality (Q) scores is/are TRUE?

- A. A score of 10 denotes 90% base call accuracy.
- B. A score of 40 presents 1 in 40 chance of incorrect base call.
- C. A score of 30 presents 1 in 1000 chance of incorrect base call.
- D. A score of 10 denotes 10% base call accuracy.
- E. A score of 30 presents 1 in 100 chance of incorrect base call

Which TWO of the following statements regarding the Hardy-Weinberg equilibrium is/are TRUE?

- A. It does not apply to a gene where there are more than two alleles present in a population.
- B. Mating is not assortative with regards to MHC alleles in humans.
- C. The genotype frequencies of heterozygotes will be $2pq$ where p is the frequency of one allele and q the frequency of the other.
- D. It only applies where there is no migration of alleles into or out of the population.
- E. It does not apply to VNTR alleles and genotypes.

