Bioinformatics Study Guide 2

Genes, Genomes, and Genetic Variation

Review the basic structure of genes (see previous study guide) since an understanding of genome structure and genetic variation depends on understanding gene structure.

What is a genome?

Genome is complete set of genes or genetic material in organism

What two distinct genomes are present in human cells? [hint: nucleus and mitochondria both have DNA) What three genomes would be present in a plant cell?

Human cells: haploid and diploid. (nuclear, mitochondrial)

Plant cells: nucleus, mitochondria, and plastids/chloroplast. Haploid, diploid, tetraploid.

Note that viruses and bacteria also have genomes.

Which kind of genomes consist of DNA? Which kind of genomes may be either DNA or RNA?

Cellular genome is DNA, but some viruses have RNA or DNA.

When was the first viral genome sequenced? When was the first draft of the human genome completed?

Virus: first completed in 1977.

Human: 2000 for first draft, 2003 completed

What differences are seen in the genome of human as compared to yeast or bacteria in terms of organization (packing and presence of genome wide repeats)?

In yeast, genes are tightly packed, but they aren’t in humans. Also, humans have a lot more genome wide repeats. A lot.

What kinds of repeated and unique sequences are present in the human genome?

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What fraction of the human genome actually encodes proteins?

3%!!!!!

What experimental method was first used to sequence genomes, including the human genome?

Sanger method

How can the results of DNA sequencing be used to identify mutations?

Sequence electropherogram

Note the new methods being used to sequence DNA: pyrosequencing, DNA chips, and nano pore sequencing.

What are the steps of genome assembly?

Genome assembly: fragment DNA sequences, find overlaps between reads, assemble overlaps into contigs, assemble contigs into scaffolds.

What are the two basic strategies for genome sequencing?

Bottom up (shred all books, piece books together one at a time, put books back on shelf) and top down (take one book at a time from a known location and shred it, piece it back together, then place it back on the shelf in the library)

What are the definitions of genotype and phenotype?

Genotype: genetic constitution

Phenotype: the set of observable characteristics of an individual resulting from the interaction of the genotype with the environment

What other factors modify how a genotype is manifested in terms of phenotype?

The environment!!

What is a genetic locus?

Specific location or position of a specific gene

What is genetic variation?

Diversity in gene frequency, either in individuals or in species.

What is the origin of genetic variation?

Mutation is the ultimate source of genetic variation.

How is a lesion different than a mutation?

Lesion: damage to a specific part of a DNA sequence

Mutation: a section of DNA replaced by a differently coded section

What are the various types of mutations?

Missense

Nonsense

What are alleles?

Alternative form of a gene that can arise due to mutation. Found at same place on chromosome.

How many alleles are possible in an individual diploid organism?

2 alleles

How many alleles are possible for a genetic locus in a population of individuals?

What is a genetic polymorphism?

What are single nucleotide polymorphisms?

What are wild-type alleles?

Allele that codes for most common phenotype

What a mutant alleles?

Mutated form of a gene

What do we mean by the terms homozygous and heterozygous?

Homozygous: two of the same allele

Heterozygous: two different alleles

How can a loss of heterozygosity take place?

What are dominant versus recessive states?

Understand the mutations involved in human sickle cell anemia and beta thalassemia.

Human sickle cell anemia: glu 6 to valine mutation

Beta thalassemia: mutations in the HBB gene