

Genetic testing- analysis of the DNA to observe genes

Results can lead to pre symptomatic treatments

Hypertrophic cardiomyopathy

- Genetic tests available for that
- Genetic test for Huntington's disease(get extra repeats)- can confirm the diagnosis=
- The DNA sequences with other people are largely identical- any nucleotides
- Genetic variation can change proteins which lead to changes in genotypes
- A and T are two different variants.
- Variant 1- hemoglobin gene a- no sickle cell anemia, not resistant to malaria
- Variant 2- t- sickle cell anemia- resistant
- 3 major types of genetic variations
- SNP- change in one DNA nucleotide
- Insertion- insertion of one or more DNA nucleotide in a genome
- Deletion- deletion of one or more DNA nucleotides=

PCR- amplify a short sketch of dna, 1st make copies, heated-denature, decreased, bind polymers, copied, higher temp.. add nucleotides to extend, make additional copies, after 3 cycles, target sequence accumulates, primer binds right outside of the sequence

Gel electrophoresis- dna molecules divided by the size and charge

Insertions and deletions detected by pcr

Single nucleotide polymorphisms -

Relp analysis- restriction enzymes(recognizes sequence) cut dna, run it on the gel...two pieces of shorter dna . ex: sickle cell hemoglobin- longer for restriction enzymes to cut

Snps and insertions/deletions can be identified by "conventional" Dna sequencing (very expensive)

10/2/13

Genome sequencing- make copies then reassemble it, the price has fallen too, advantages- can tell hereditary genes

Pcr and electrophoresis- deletions

Relp- a single nucleotide

10/9/13

Dominant or recessive- use family pedigrees...homo (both alleles r the same) hetro (genes are different)

Sex- linked disorders- more males or females are affected

y-linked- all the mens r affected, fathers will pass it to all of their sons

