DEPARTMENT OF BIOTECHNLOGY
FACULTY OF ENGINEERING & TECHNOLOGY

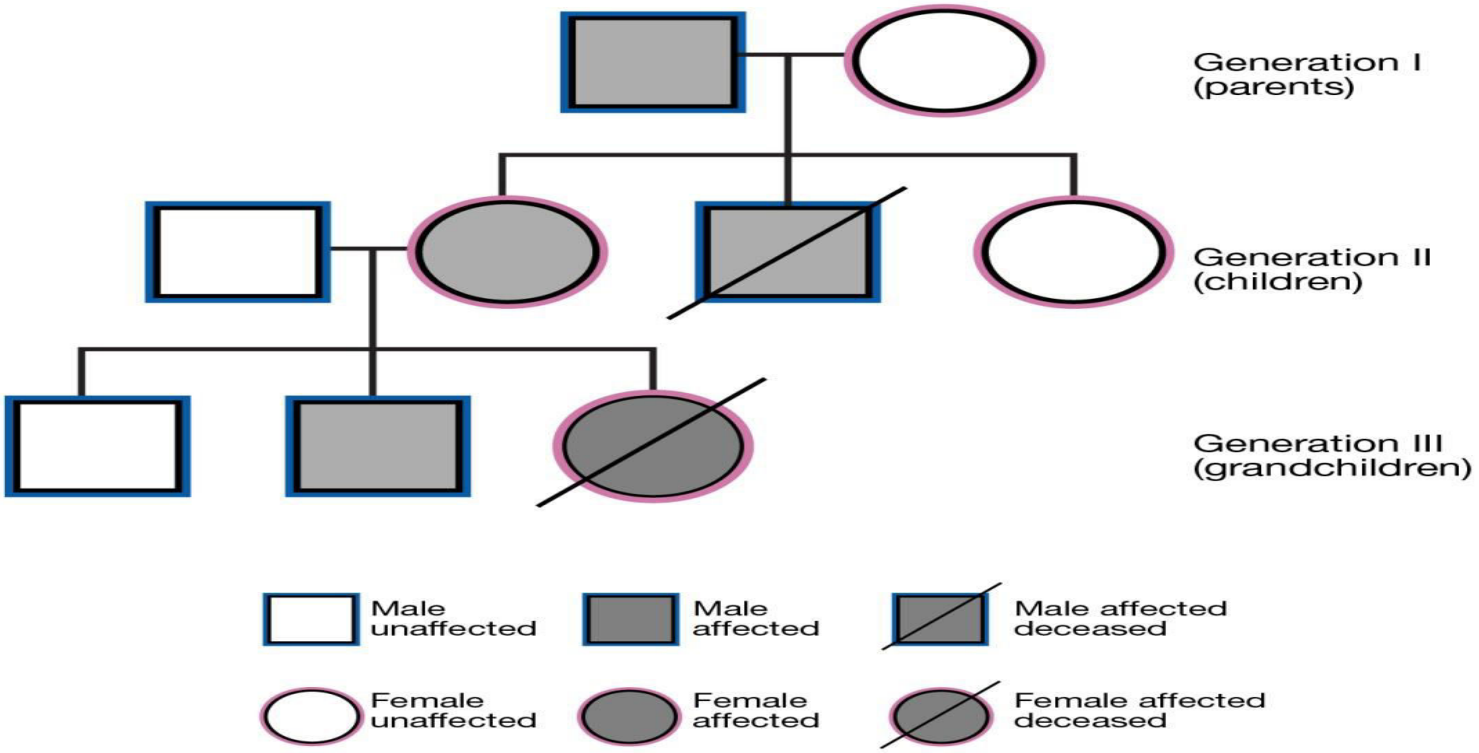
LT 26. Pedigree Varietal

Content outline
Pedigree Varietal

What is pedigree?

Pedigree: A pedigree is a genetic representation of a family tree that diagrams the inheritance of a trait or disease though several generations. The pedigree shows the relationships between family members and indicates which individuals express or silently carry the trait in question. Varietal means variation or being a variety in distinction from an individual or species.

➢Pedigree simply refers to characteristics and varietal refers to diversity



•A pedigree is a representation of our family tree. It shows how individuals within a family are related to each other. We can also indicate which individuals have a particular trait or genetic condition. If we take a pedigree, which we usually try to include at least three generations, we might be able to determine how a particular trait is inherited. Using that information, we might be able to tell the chance that a given individual will have the trait themselves or could pass it on to their children. There are standard ways to draw pedigrees so that we can all look at a pedigree and understand it. We use squares to represent males and circles to represent females. We then can number our generations with roman numerals, so the top generation would be generation one, or Roman numeral I. Along this line, we'd indicate males and females. We would indicate marriages between individuals with a horizontal line connecting the two individuals.

•If an individual has a genetic trait, we would blacken those individuals in or shade them so that it would be understood that they had a particular trait. We would then draw a line, a vertical line, off the horizontal line where we would indicate any of their children that they had, and we would then indicate if any of their children were infected

•And we can do this for as many generations as we have. It's important when we draw pedigree that we try to put in as much information as possible. So for example, if there have been children that died in early infancy or were stillborn, we also want to include those individuals. And those are typically shown as very small blackened-in symbols to indicate there was a loss of a child, either in pregnancy or early in life.

Application of RFLP in determining pedigree varietal

•Restriction enzymes cut DNA at precise points producing a collection of DNA fragments of precisely defined length.

•These can be separated by electrophoresis, with the smaller fragments migrating farther than the larger fragments.

•One or more of the fragments can be visualized with a "probe" — a molecule of single-stranded DNA that is

- •complementary to a run of nucleotides in one or more of the restriction fragments and is
- •radioactive (or fluorescent).

•If probes encounter a complementary sequence of nucleotides in a test sample of DNA, they bind to it by Watson-Crick base pairing and thus identify it.

•Polymorphisms are inherited differences found among the individuals in a population.

•RFLPs have provided valuable information in many areas of biology, including:

•screening human DNA for the presence of potentially deleterious genes ("Case 1");

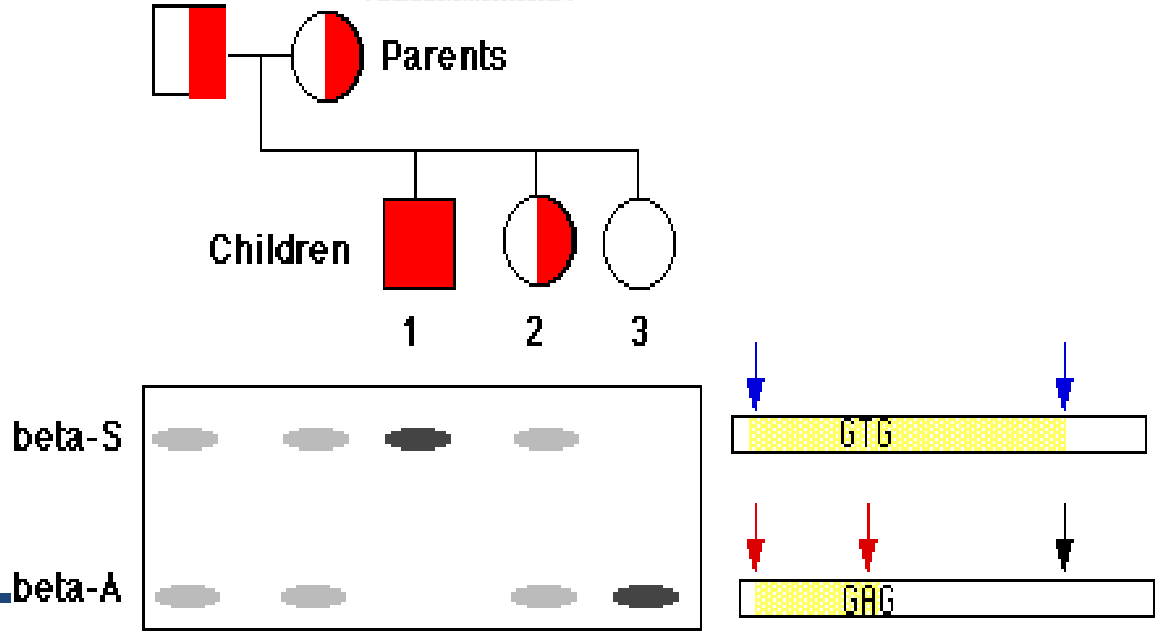
•providing evidence to establish the innocence of, or a probability of the guilt of, a crime suspect by DNA "fingerprinting" ("Case 3").

Case 1: Screening for the sickle-cell gene

•Sickle-cell disease is a genetic disorder in which both genes in the patient encode the amino acid valine (Val) in the sixth position of the beta chain ($beta^S$) of the hemoglobin molecule. "Normal" beta chains ($beta^A$) have glutamic acid at this position.

•The only difference between the two genes is the substitution of a T for an A in the middle position of codon 6.

•This converts a GAG codon (for Glu) to a GTG codon for Val and abolishes a sequence (CTGAGG, which spans codons 5, 6, and 7) recognized and cut by one of the restriction enzymes.



•When the **normal** gene ($beta^A$) is digested with the enzyme and the fragments separated by electrophoresis, the probe binds to a **short** fragment (between the red arrows).

•However, the enzyme cannot cut the **sickle-cell gene** at this site, so the probe attaches to a much larger fragment (between the blue arrows).

•The figure (from data provided by S. E. Antonarakis) shows the pedigree of a family whose only son has sickle-cell disease. Both his father and mother were heterozygous (semifilled box and circle respectively) as they had to be to produce an afflicted child (solid box). The electrophoresis patterns for each member of the family are placed directly beneath them. Note that the two homozygous children (1 and 3) have only a single band, but these are more intense because there is twice as much DNA in them.

•In this example, a change of a single nucleotide produced the RFLP. This is a very common cause of RFLPs and now such polymorphisms are often referred to as **single nucleotide polymorphisms** or **SNPs**. (However, not all RFLPs arise from SNPs.

References & Further reading