Forensic DNA science

Professor
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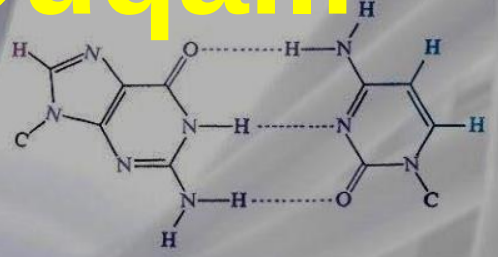

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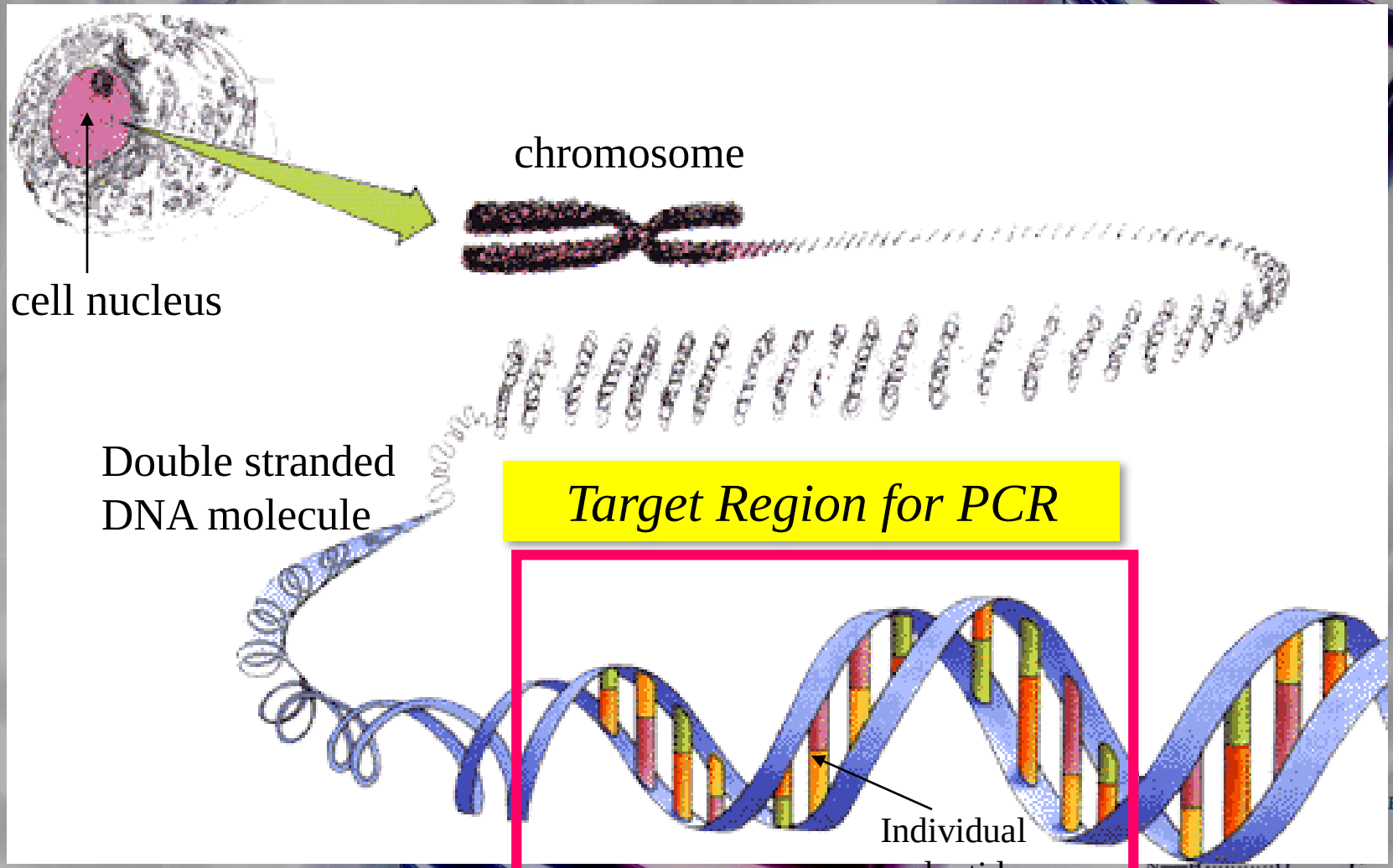
DNA in the Cell



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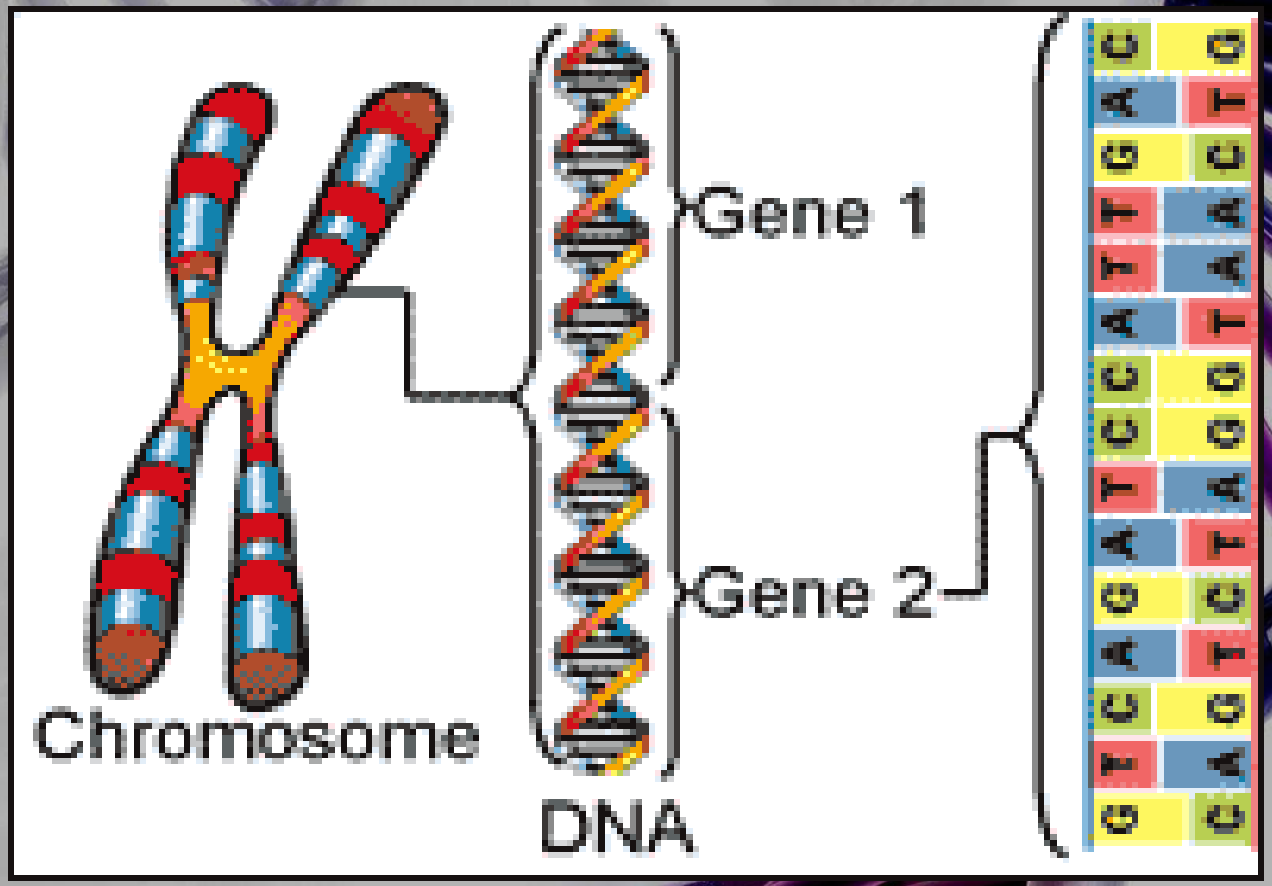
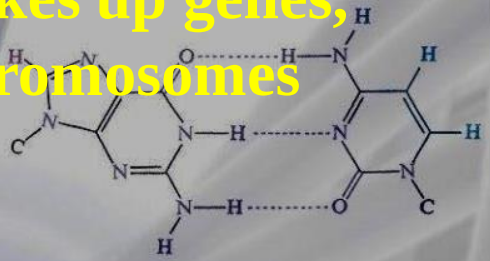


Nucleotides make up DNA. DNA makes up genes, and genes are small segments of chromosomes



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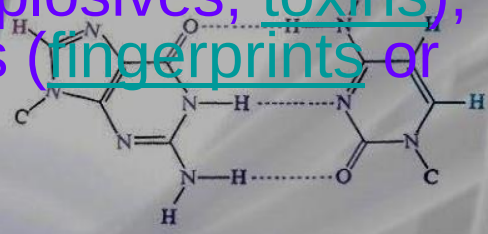
WHAT IS FORENSIC SCIENCE?

- Forensic science is a multidisciplinary subject used for examining crime scenes and gathering evidence to be used in prosecution of offenders in a court of law. Forensic science techniques are also used to examine compliance with international agreements regarding weapons of mass destruction.

- The main areas used in forensic science are biology, chemistry, and medicine, in addition to physics, computer science, geology, and psychology. Forensic scientists examine objects, substances (including blood or drug samples), chemicals (paints, explosives, toxins), tissue traces (hair, skin), or impressions (fingerprints or tidemarks) left at the crime scene.



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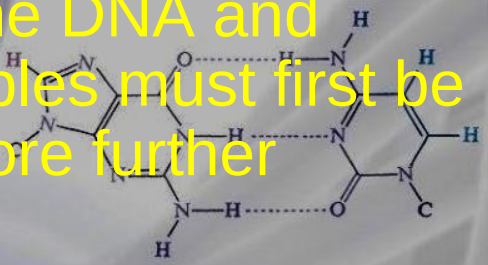
- DNA is the main method of identifying people. Victims of crashes or fires are often **unrecognizable**, but adequate DNA can be isolated and a person can be positively identified if a sample of their DNA or their family's

- DNA is taken for comparison. Such methods are being used in the identification of the remains in Yugoslav war victims, the World Trade Center terrorist attack victims, and the 2002 Bali bombing victims.

- Biological traces, investigated by forensic scientists come from bloodstains, saliva samples (from cigarette buts or chewing gum) and tissue samples, such as skin, nails, or hair. Samples are processed to isolate the DNA and establish the origin of the samples. Samples must first be identified as human, animal, or plant before further investigation proceeds.



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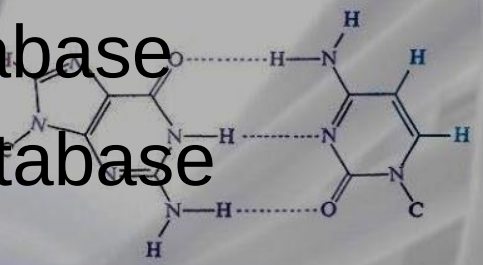
Brief History of Forensic DNA Typing

- 1980 - Ray White describes first polymorphic RFLP marker
- 1985 - Alec Jeffreys discovers multilocus VNTR probes
- 1985 - first paper on PCR
- 1988 - FBI starts DNA casework
- 1991 - first STR paper
- 1995 - FSS starts UK DNA database
- 1998 - FBI launches CODIS database



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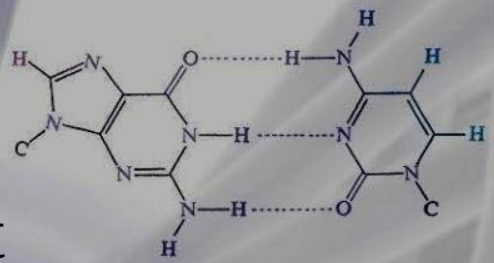
DNA Use in Forensic Cases

- Most are rape cases (>2 out of 3)
- Looking for match between evidence and suspect
- Must compare victim's DNA profile

Challenges

- Mixtures must be resolved
- DNA is often degraded
- Inhibitors to PCR are often present



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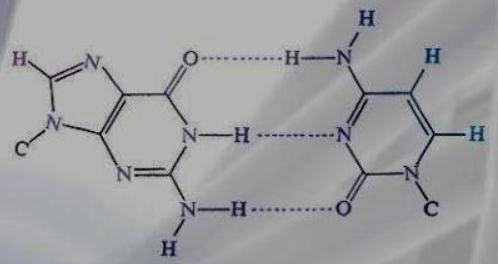
Human Identity Testing

- Forensic cases -- **matching suspect with evidence**
- Paternity testing -- **identifying father**
- Historical investigations
- Missing persons investigations
- Mass disasters -- **putting pieces back together**



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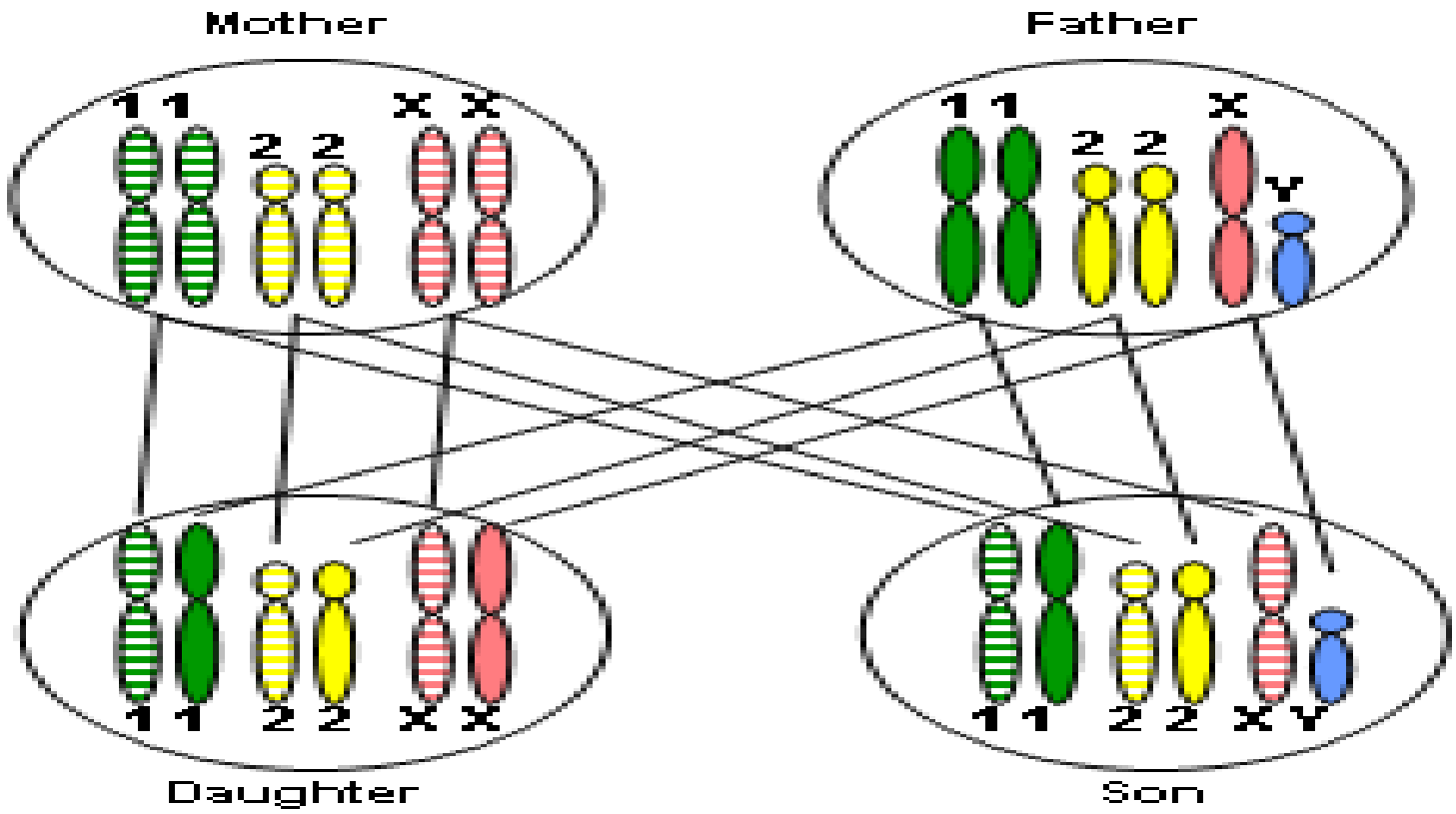
An Example of How Chromosomes Are Passed from Parents to Children



The figure to the right shows the transmission of chromosomes (and therefore genes) from parents to children. In this figure, three pairs of chromosomes are shown: pair #1 (green); pair #2 (yellow); pair #3 - sex chromosomes (pink and blue).

Steps in DNA Sample Processing

Sample Obtained from Crime Scene or Paternity Investigation

Biology

DNA Extraction → DNA Quantitation → PCR Amplification of Multiple STR markers

Technology

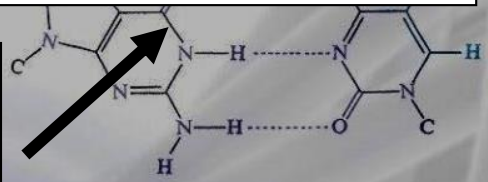
Separation and Detection of PCR Products (STR Alleles) → Sample Genotype Determination

Genetics

Comparison of Sample Genotype to Other Sample Results

If match occurs, comparison of DNA profile to population databases

Generation of Case Report with Probability of Random Match



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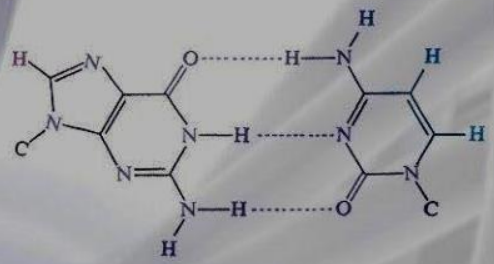
Sources of Biological Evidence

- Blood
- Semen
- Saliva
- Urine
- Hair
- Teeth
- Bone
- Tissue



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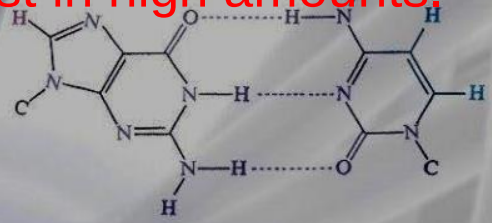
Mitochondrial DNA (mtDNA)

- ***Unlike nuclear DNA, which is passed from both mother and father to the offspring, mitochondrial DNA (mtDNA) is maternally inherited.***

- At fertilization, the mtDNA, contained in the tail end of the spermatozoa, is never allowed to enter the ovum, deleting the male mtDNA information from the offspring's genome.

- Mitochondrial DNA is useful for forensic purposes because of two properties. First, part of the mitochondrial genome is highly polymorphic, making it useful for human identification. The two most variable regions known as HV1 and HV2 are usually amplified and sequenced to compare the difference between the evidence and reference samples.Secondly, although mtDNA comprises less than 1% of the total DNA within a cell, its genes exist in high amounts.



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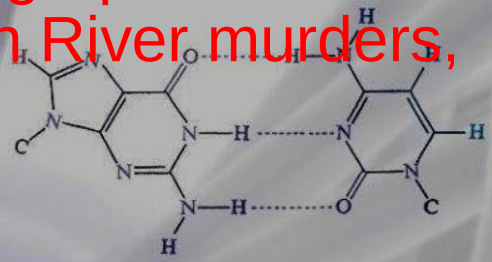
Mitochondrial DNA (mtDNA)

- Because mtDNA is present in high copy number, it is very useful when analyzing degraded samples or samples that lack nuclear DNA. One example is hair, which is a common item of evidence, especially when there is little or no root present to test.

- The examination of mtDNA in evidence such as bone and teeth, which may contain degraded DNA, can produce a satisfactory profile because of the high copy number of mitochondrial sequences. Mitochondrial DNA testing was performed in a number of high-profile cases such as the Boston Strangler, the Green River murders, and the Laci Peterson homicide.



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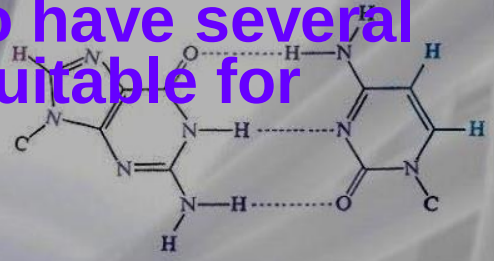
Short Tandem Repeats (STRs)

- The human genome is full of repeated DNA sequences. These repeated sequences come in various sizes and are classified according to the length of the core repeat units, the number of contiguous repeat units, and/or the overall length of the repeat region. DNA regions with short repeat units (usually 2-6 bp in length) are called **Short Tandem Repeats (STR).**

- STRs are found surrounding the chromosomal centromere (the structural center of the chromosomes). STRs have proven to have several benefits that make them especially suitable for human identification.



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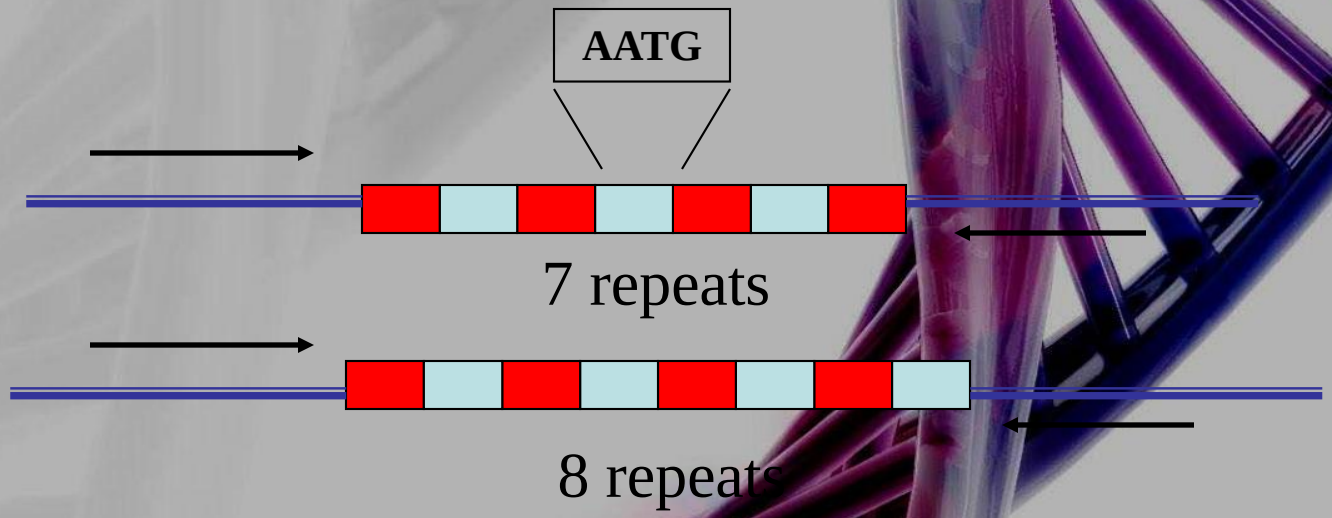
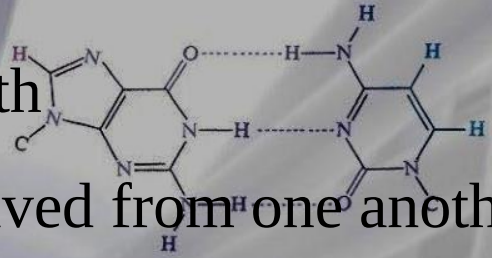
Short Tandem Repeats (STRs)



the repeat region is variable between samples while the flanking regions where PCR primers bind are constant

Homozygote = both alleles are the same length

Heterozygote = alleles differ and can be resolved from one another



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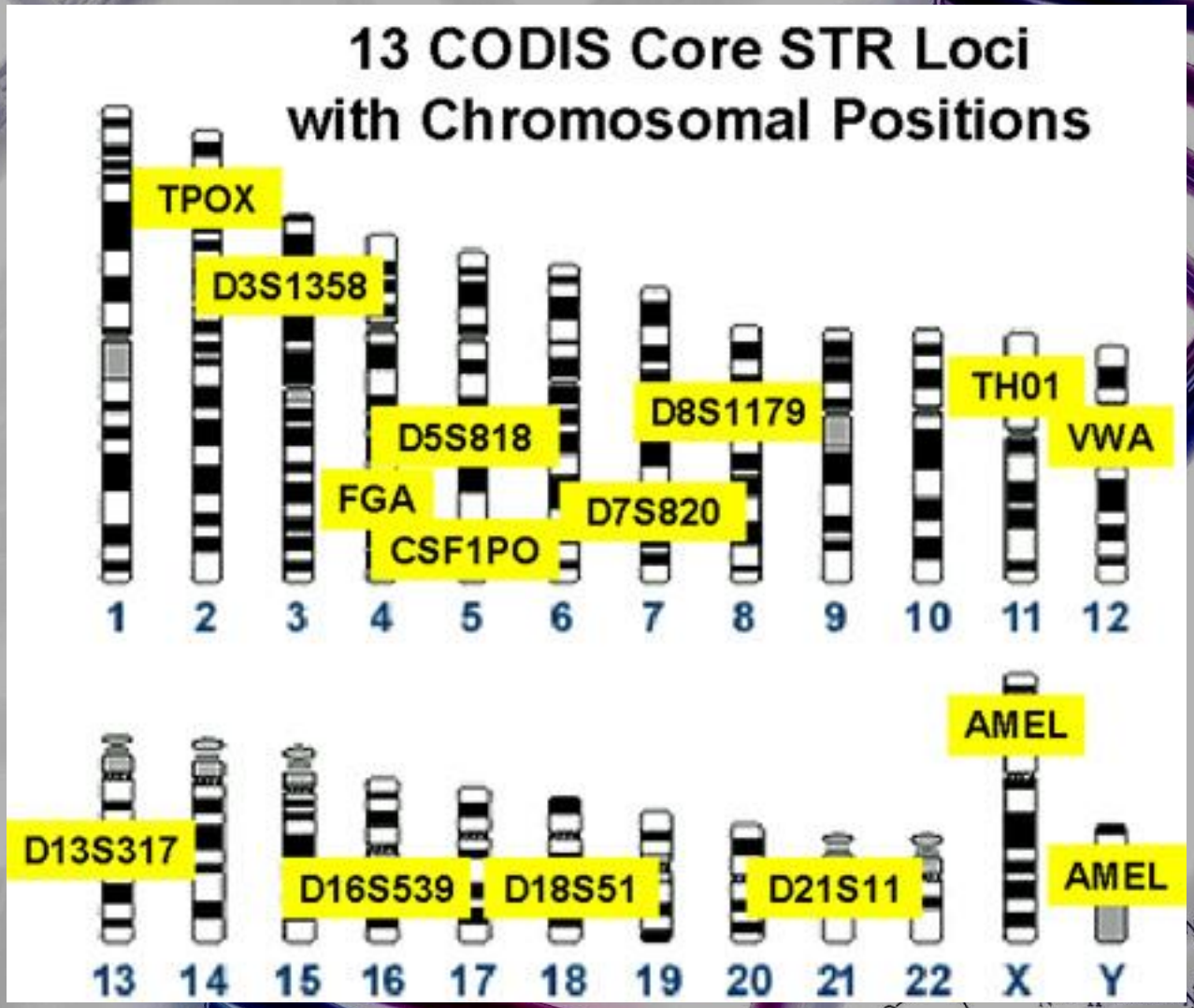

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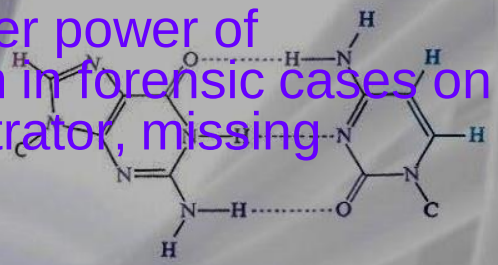
Why STR chosen for human identification?

- For human identification purposes, it is important to have DNA markers that exhibit the highest possible variation in order to discriminate between samples.
- The smaller size of STR alleles make STR markers better candidates for use in forensic applications, in which degraded DNA is common. PCR amplification of degraded DNA samples can be better accomplished with smaller target product sizes.
- Because of their smaller size, STR alleles can also be separated from other chromosomal locations more easily to ensure closely linked loci are not chosen. Closely linked loci do not follow the predictable pattern of random distribution in the population, making statistical analysis difficult.
- Because of these characteristics, STRs with higher power of discrimination are chosen for human identification in forensic cases on a regular basis. It is used to identify victim, perpetrator, missing persons, and others.



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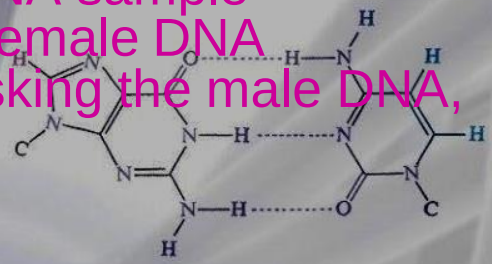
Y-STR

- Y- STRs are Short Tandem Repeats (STRs, refer to the found on the male-specific Y Chromosome. The coding genes, mostly found on the short arm of the Y Chromosome, are vital to male sex determination, spermatogenesis and other male related functions. The Y-STRs are polymorphic among unrelated males and are inherited through the paternal line with little change through generations.

- Y-STRs have been used by forensic laboratories to examine sexual assault evidence. In a sexual assault case, evidence such as vaginal swabs will contain both female and male DNA. Differential extraction is often used to separate the male component from the female component. More often, however, the male and female components cannot be separated completely. As a result, the female component could exist prominently even in the male component after separation. When the "male DNA sample" undergoes the PCR amplification process, the female DNA component is amplified as well, sometimes masking the male DNA, which makes analysis difficult.



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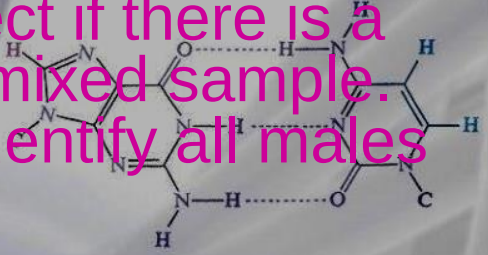
Y-STR

- Masking does not occur when Y-STRs are examined. Since there is no Y-STR in the female evidence, the only contribution of Y-STR can only come from the assailant(s) in a sexual assault case. The male component will be easily detected, since only this part of DNA will be amplified. The Y-STR system is especially helpful when there are more than one assailant. The mixed pattern in the evidence can help to identify those males responsible for the assault.

- Y-STR is also used for non-sexual assault cases where mixed samples are collected from evidence. Sometimes, regular STR will cause the masking effect if there is a very small quantity of male DNA in the mixed sample. Performing Y-STR testing can help to identify all males who have contributed to the evidence



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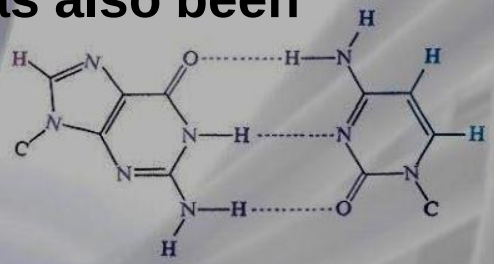
Y-STR Paternal Ancestry DNA Test

- The Y-STR (paternal ancestry) DNA test can confirm your relationship to long-lost relatives and paternal ancestors. This ancestry DNA test is also often used to provide additional evidence. They would need to ask a biological male relative, such as a father, brother, paternal uncle, or paternal grandfather to contribute a sample for comparison testing with her potential paternal relative or ancestor.
-
- The genealogy DNA test is based on the fact that the Y chromosome is passed from father to son relatively unchanged through many generations. The illustration to the right shows a typical inheritance pattern for the Y chromosome. Because the Y chromosome follows the same father-to-son pattern much like surnames in Western culture, the test has also been referred to as a "Surname Test."



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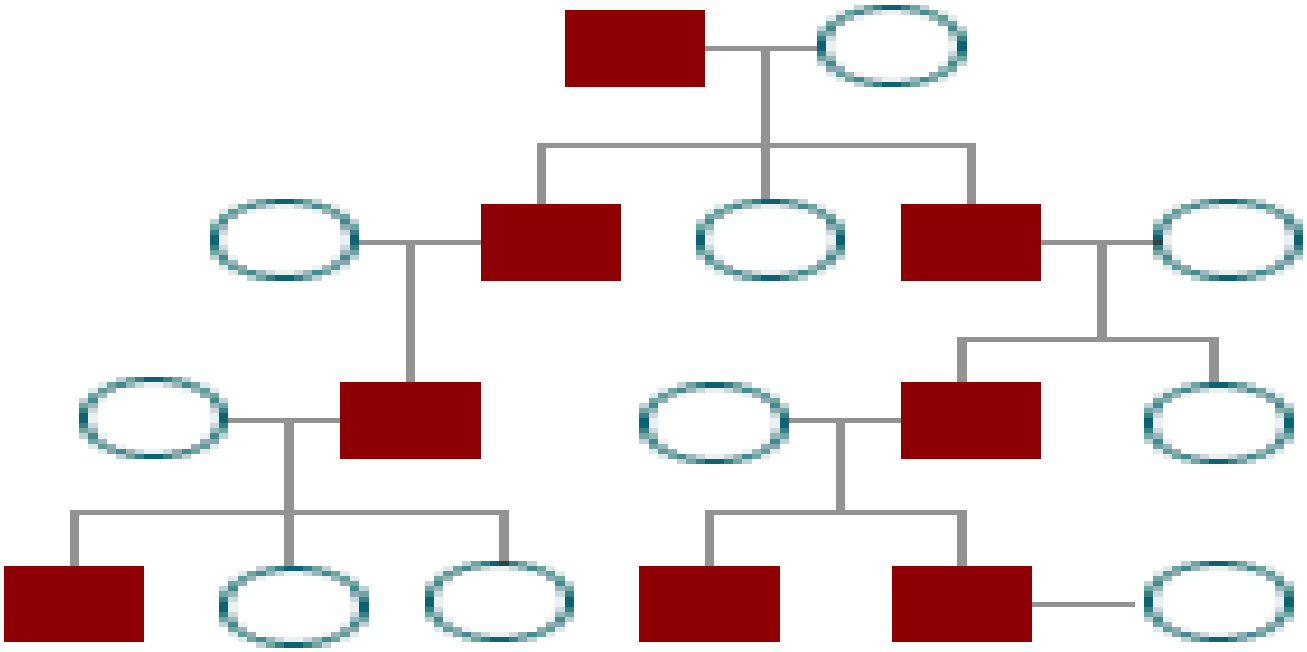
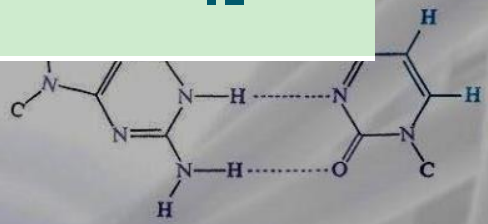


Male Female

Red indicates paternal lineage



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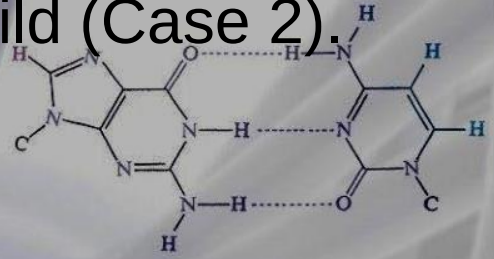
HOW DNA TYPING IS USED IN DETERMINING PATERNITY

- This analysis is based on the fact that a person possesses two DNA copies originating from the mother (maternal copy) and the father (paternal). The DNA from the mother, the alleged father and the child are analyzed to evaluate paternity. The DNA copy contributed by the biological father should be present in the alleged father with the Probability of Paternity >95% (Case 1). If the child possesses DNA that did not originate from the mother or the alleged father, then the alleged father cannot be the biological father of the child (Case 2).



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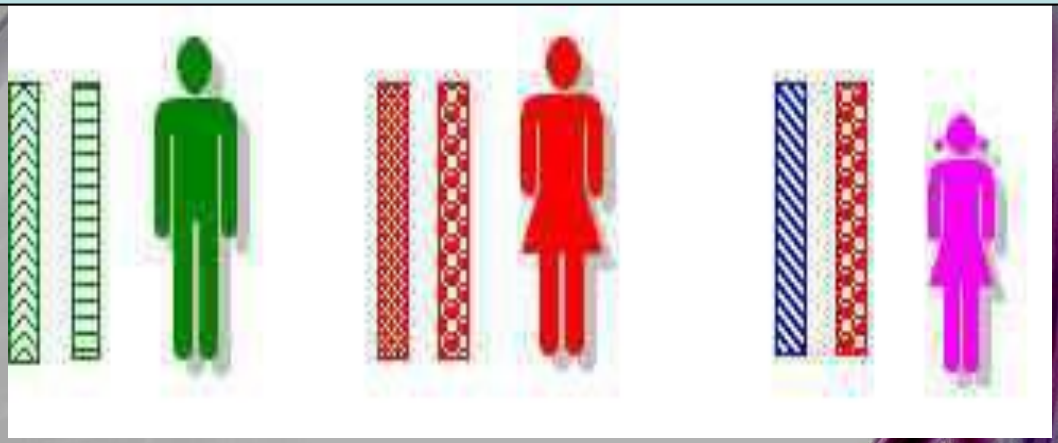
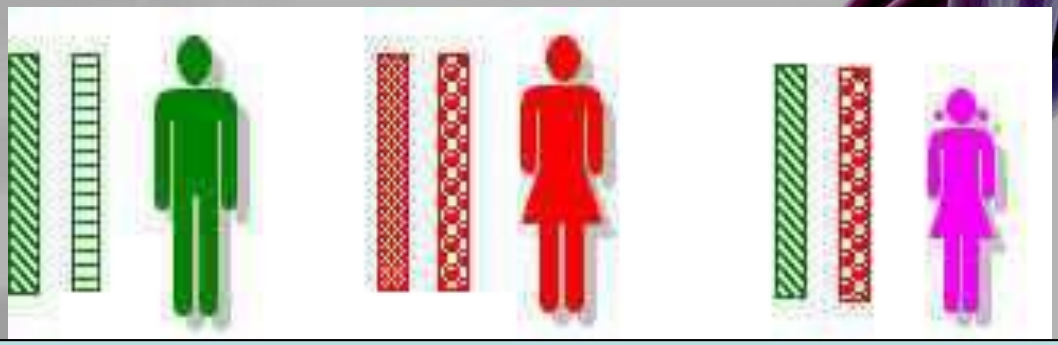
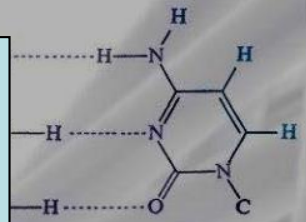
The two possibilities are diagrammed briefly below:

CASE 1: Exclusion of Paternity





CASE 2: Possibility of Paternity



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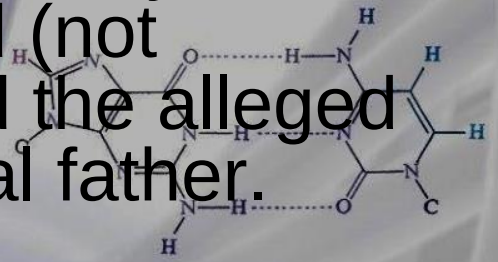
• *Y-STR Paternal Ancestry DNA Test*

- in a Y-STR DNA test, specific locations on the Y chromosome are examined to generate a Y-STR profile for each male tested. Males who are related through their fathers will tend to have the same or similar Y-STR profiles, and males who are not related will likely have different Y-STR profiles.

- An example, if a male child and his alleged uncle (alleged father's full brother) are tested, their Y-STR profiles must match. If they do not, then the alleged uncle is excluded (not considered a biological uncle) and the alleged father is probably not the biological father.



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