



"Buccal Up – Everything You Need to Know About Genetic Testing"

DNA Testing-Forensic & Paternity Applications

Melanie S. Trapani, Ph.D., F-ABC
04Oct2017



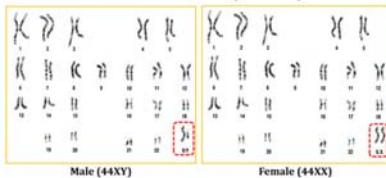
Deoxyribonucleic acid (DNA)



- >Our 'genetic blue print'
- >Unique (except identical twins)
- >Residing in the nucleus of almost every cell in our body
- >Inherited (generally half each from the mother & father)
- >46 chromosomes (23 pairs of chromosomes)



HUMAN KARYOTYPE (NORMAL)



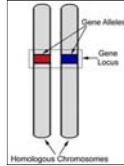
Male (44XY)

Female (44XX)



Polymorphism Definitions

- > Locus (loci = plural) – Chromosomal location or location of a gene or DNA marker.
- > Allele – Alternate possibilities for locus.
- > Heterozygous – Two alleles on homologous chromosomes are different.
- > Homozygous – Alleles on homologous chromosomes are the same.



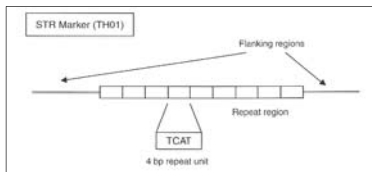
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Evolution of DNA Analysis

- > RFLP analysis – 1987 to mid 90s
- > Autosomal STR – mid 1990s
- > Mitochondrial DNA – late 1990s
- > Y-STR – early 2000s
- > Familial DNA Searching – 2008 (USA)

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Short Tandem Repeats (STRs)



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STR Length Polymorphism



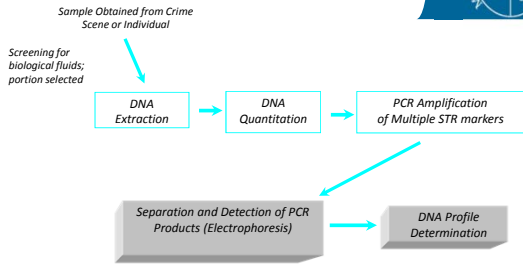
- ▶ AG-TAG-T Allele A
- ▶ AG-TAG-TAG-T Allele B
- ▶ AG-TAG-TAG-TAG-T Allele C
- ▶ AG-TAG-TAG-TAG-TAG-T Allele D
- ▶ AG-TAG-TAG-TAG-TAG-TAG-T Allele E
- ▶ AG-TAG-TAG-TAG-TAG-TAG-TAG-T Allele F

The number of repeat unit defines the allele/ typing.

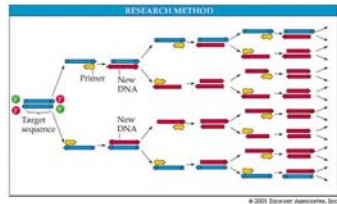
Allele F is 5 repeats longer than allele A




Steps in STR Testing

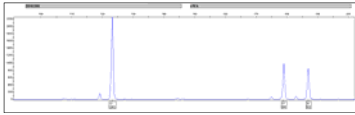


Polymerase Chain Reaction (PCR)




Electropherogram






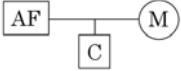
RFU

Size (basepairs)




Paternity Testing




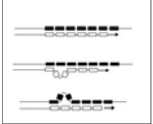


	D3S1358	FGA	D8S1179	D21S11	D18S51
Mother	18	24	13,14	30	19
Child	18	24,25	14,16	29,30	17,19
Alleged Father	15,18	22,25	13,16	29,33.2	17,21
Paternity Index	3.3529	5.9155	16.9490	2.3412	4.27




What is an STR mutation?





	D3S1358	FGA	D8S1179	D21S11	D18S51	D7S820
Mother	18	24	13,14	30	19	9,10
Child	18	24,25	14,16	29,30	17,19	9,12
Alleged Father	15,18	22,25	13,16	29,33.2	17,21	10,13
Paternity Index	3.3529	5.9155	16.9490	2.3412	4.27	0.0023



Combined DNA Index System (CODIS)



- NDIS, SDIS and LDIS
- All 50 states, the District of Columbia, the federal government, the U.S. Army Criminal Investigation Laboratory, and Puerto Rico participate in NDIS.
- NDIS upload requires DNA profile to include 20 markers and have originated (mostly) from single individual



Forensic Testing – Biological Fluids

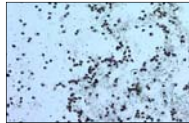


Blood
Presumptive Assays: KM, LMG, Luminol
Confirmatory: Hematrace, Hemastix



Saliva
Presumptive: RSID Assay

Seminal Fluid
Presumptive Assays: AP, p30
Confirmatory Assay: Microscopy



PLEASE NOTE: Obligatory
gruesome crime scene photo
to follow



Case #1: Autosomal STR- Blood



Description:

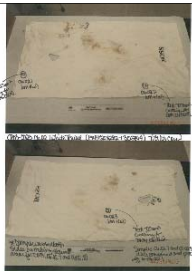
- >28 year-old woman found stabbed and strangled in bathtub with water running, with body being weighed down by household items (VCR, etc)
- >Cold case with no arrests >30 years

Evidence Received for Analysis:

- >Victim's bloody clothes
- >Two knives from victim's chest
- >Bloody towels
- >Swabs of blood from toilet seat
- >Garbage bag



Case #1: Autosomal STR- Blood



DNA Testing Results

- >All but one item produced DNA profiles consistent with victim
- >Of 14 stains tested on white towel, 12 matched DNA profile of victim.
- >Two stains from white towel originated from unknown male.

Subsequent Results

- >CODIS hit to ex-husband of roommate
- >Testing of new sample confirms match
- >2016 - Ex-husband of roommate convicted of murder



Case #2: Autosomal STR- Saliva

Description:

- >58 Year old female victim
- >Multiple lacerations to head from blunt-force trauma
- >Multiple stab wounds
- >"Leash" around neck



Evidence Received:

- >Swabs of blood from multiple locations
- >Folding knife
- >Rope used as leash
- >Victim fingernail samples
- >Swabs from two soda cans, Budweiser can



Case #2: Autosomal STR- Saliva



DNA Testing Results

- >All blood samples, produced DNA profiles consistent with victim
- >Leash and fingernail samples matched victim.
- >Swabs of drink containers = unknown male.



Subsequent Results

- >Suspect on run
- >Suspect's girlfriend provided his toothbrush and razors for DNA comparison = match
- >Suspect apprehended several states away
- >2009 Sentenced to death



Case #3: Autosomal STR- Seminal Fluid



Background

- >Sexual assault occurs in US every ~98 seconds
- >Approximately 1/3 of assaults reported
- >Estimated hundreds of thousands untested rape evidence kits in US



Source: <http://www.endthebacklog.org/>

Case Scenario

- >1984 Sexual assault of 20 year old mother of two young children
- >No leads, no arrests >30 years
- >Rape evidence kit submitted for DNA analysis in 2015



Case #3: Autosomal STR- Seminal Fluid



Evidence Received:

- >Vaginal, cervical and rectal Swabs
- >Victim fingernail scrapings
- >Victim pubic hair combings






Source: <http://www.greeleytribune.com>

DNA Testing Results

- >Sperm fraction of vaginal sample- CODIS hit to one man
- >Sperm fraction of rectal sample- CODIS hit to second man
- >Convictions in May and August 2017



Case #4: Autosomal STR: Sweat/Skin Cells






Description:


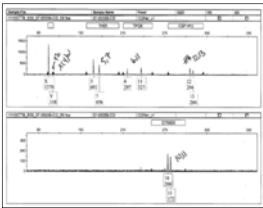

- > Interrupted home robbery
- > 86 Year old victim
- > Ankles and hands duct-taped
- > Victim beaten with piano leg
- > Jewelry pawned within 4 hours

Evidence Received for Analysis:

- > Victim's bloody clothes
- > Victim's broken dentures
- > Victim's fingernails
- > Duct tape cut from victim



Case #4: Autosomal STR: Sweat/Skin Cells


Autosomal STRs vs YSTRs

Autosomal STRs

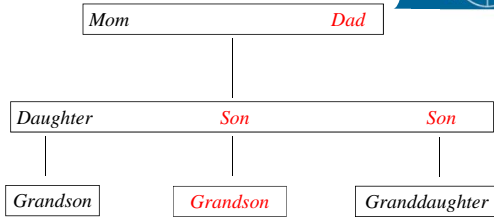
- > Located on autosomal chromosomes
- > Males and females
- > Inherited half from biological mom and half from biological father

YSTRs

- > Located on Y chromosome
- > Males only
- > Paternal inheritance



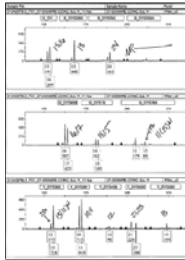
Y-Chromosome/ YSTR Inheritance



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Case #4: YSTR: Sweat/Skin Cells

- Mixed DNA Profile
- DNA profile consistent with mixture of victim DNA and DNA of boyfriend of woman whose profile is on gloves
- 50 year sentence



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Case #5: Familial DNA Searching

- From 1985-87, Seven prostitutes and/or drug users killed in South Los Angeles; same assailant attempted to murder an 8th. All bodies nude and dumped in alleys or dumpsters.
- Jan 2007: Homeless man collecting can from dumpster found nude body of 25-yr old woman; DNA collected matched that from two other slayings in 2002 and 2003. Also matched that from several other murder cases in late 1980's.
- Jan 2007: A common DNA profile linked the murders. Killer given name the "Grim Sleeper" in 2008 due to gap between murders.

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Case #5: Familial DNA Search



- > In 2010, close match to 28-yr old man obtained using Familial Search.
- > Man's father, Lonnie Franklin, Jr., became the prime suspect.
- > Investigators initiated surveillance of Lonnie Franklin to collect DNA sample for comparison with DNA evidence.



Source: <http://www.dailymail.co.uk/>



Case #5: Familial DNA Search



- > Detective "busboy" cleared trash from Franklin's table at restaurant. Video shows "busboy" wearing gloves! DNA from pizza, cup, and napkins matched saliva from victim's bodies
- > Franklin had worked with Los Angeles sanitation department: familiar with the alleys and dumpsters



Case #5: Familial DNA Search



- > July 7, 2010- Franklin arrested, home included murder weapon (gun), photos of known victims shot and bleeding, plus photos & video of 100's of other unidentified women.
- > 2016- Franklin convicted on murder of 10 murders and one attempted murder, sentenced to death

Source: <http://www.dailymail.co.uk/>



Mitochondrial DNA

CELL

Nucleus

Cytoplasm

Mitochondria

Control Region

Mitochondrial DNA

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Nuclear DNA vs. Mitochondrial DNA

- > **Nuclear DNA**
 - > Half inherited from each parent
 - > Giant spiral molecule
 - > Only two copies in cell
 - > Delicate molecule
- > **Mitochondrial DNA**
 - > Inherited only from mother
 - > Small, circular molecule
 - > Thousands of copies per cell
 - > Durable; can be recovered from limited, old and degraded tissues
 - > Common samples tested: Bone, hair (shaft only), teeth

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Mitochondrial DNA Inheritance

Mom Dad

Daughter Daughter Son

Granddaughter Grandson Grandson

Great granddaughter Great granddaughter

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Case# 6: Mitochondrial DNA



- Multiple skeletons found in Boston tidal basin
- Bullet hole in skulls, and teeth removed
- mtDNA testing of bones and hair identified three of victims as FBI informants missing at least 25 years
- 2013- Whitey Bulger convicted of murders



Thank you for your attention!

Questions?

Melanie S. Trapani, Ph.D., F-ABC
Melanie.Trapani@labcorp.com