

MCBD II Core Lecture - Mar 13 2025


From Forensic Genetics to Forensic Genomics








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 1993 Mag. rer. nat (Biology, Uni Innsbruck)

 1999 Dr. rer. nat (Biology, Uni Innsbruck)



 2001 Venia Docendi (Legal Medicine)

506 original peer-reviewed publications
 >28k citations, h-index 95, i10-index 353 (Google Scholar)

Elected memberships (excerpt)

- 1998 European DNA Profiling Group
- 1999 European Network of Forensic Science Institutes
- 2001 International Society of Forensic Genetics
- 2012 Executive Board Member
- 2015-2019 President
- 2020 Vice President
- 2009 Nationale Akademie der Wissenschaften Leopoldina
- 2023 Mitglied der Spurenkommision








2


CSI laboratory (ISO17025)
 Austrian Central DNA laboratory
 EDNAP, ENFSI, Interpol
 Int. reference lab for forensic DNA testing

DVI laboratory (ISO17025)
 e.g. Tsunami (Sri Lanka, 2004), Chile (1973 regime victims), Missing Mexican students (2014)

Forensic molecular research laboratory
 Mitochondrial DNA databasing (EMPOP)
 Population genetics (mito, Y)
 Forensic DNA Phenotyping (appearance, ancestry, age)
 New Technologies (Massively Parallel Sequencing)




3

Locard's exchange principle

A perpetrator of a crime will bring something into the crime scene and leave with something from it

Both can be used as **forensic evidence**

A C G T

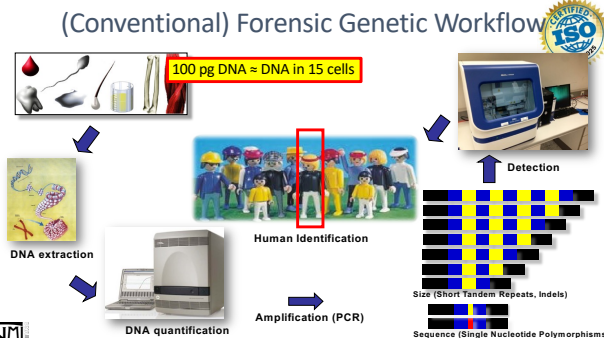


Edmond Locard
(1877-1966)

W. de Gruyter, 1916

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(Conventional) Forensic Genetic Workflow



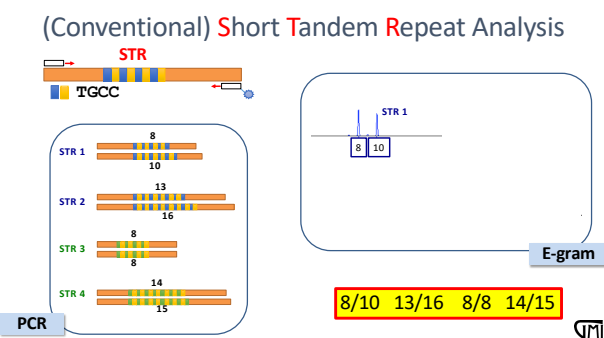
100 pg DNA ≈ DNA in 15 cells

DNA extraction → **DNA quantification** → **Amplification (PCR)** → **Detection** → **Human Identification**

Size (Short Tandem Repeats, Indels)
Sequence (Single Nucleotide Polymorphisms)

8

(Conventional) Short Tandem Repeat Analysis



STR

TGCC

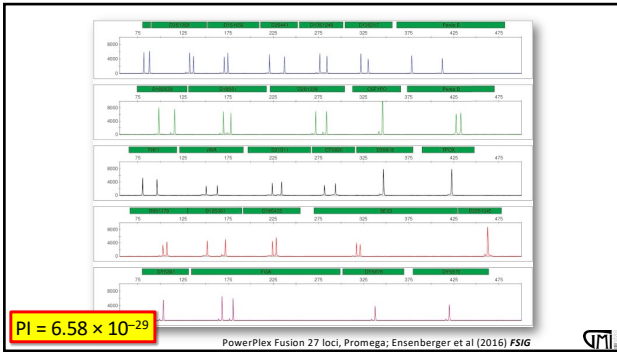
PCR

STR 1

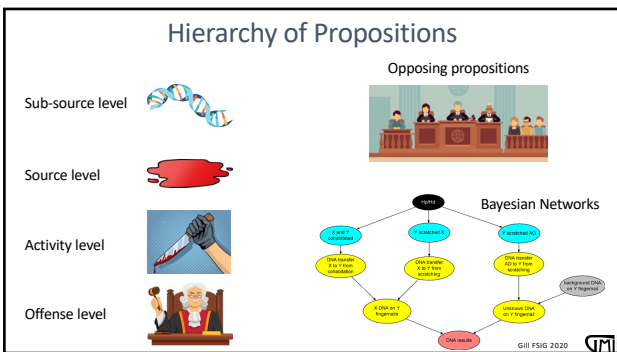
E-gram

8/10 13/16 8/8 14/15

9



10




11




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The Royal family was held under house arrest by the Bolsheviks in the Ipatiev House, Ekaterinburg (April - July 1918)



Nikolay Sokolov (1919)
Judge and Chief Investigator

From the Sokolov collection (Harvard)



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Basement room of the Ipatiev House where the Russian Imperial family was murdered July 16-17 1918 by members by Bolsheviks



From the Sokolov collection (Harvard)




17



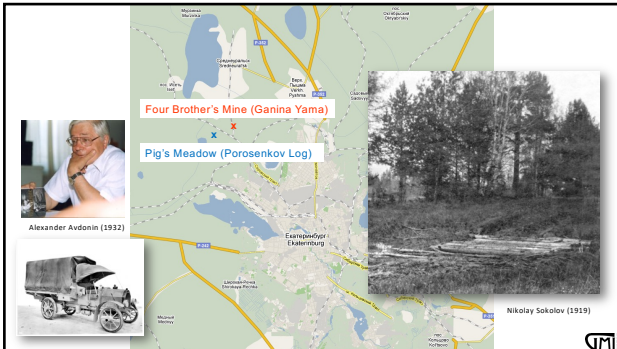
Four Brother's Mine (Ganina Yama)

Ekaterinburg

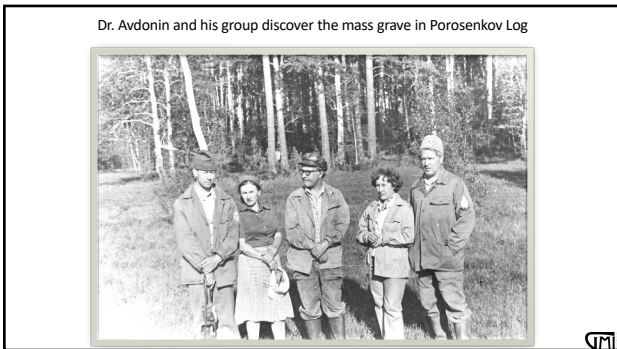
Nikolay Sokolov (1919)



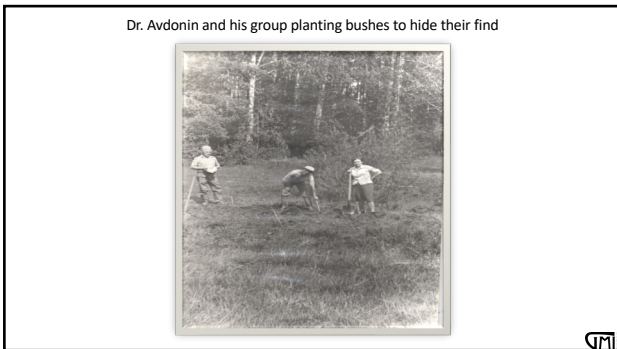
18



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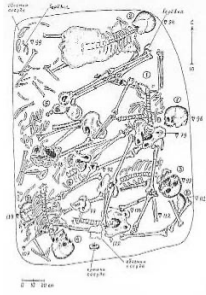


20



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1991 – Official discovery of the mass grave and excavation of the remains



SMI

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Identification of the remains of the Romanov family by DNA analysis

Peter Gill¹, Pavel L. Ivanov², Colin Kimpton¹, Romelle Piercy¹, Nicola Benson¹, Gillian Tully¹, Ian Evett¹, Erika Hagelberg³ & Kevin Sullivan¹

Nature Genetics – Feb. 1994

SMI

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Anna Demidova



Alexei Trupp



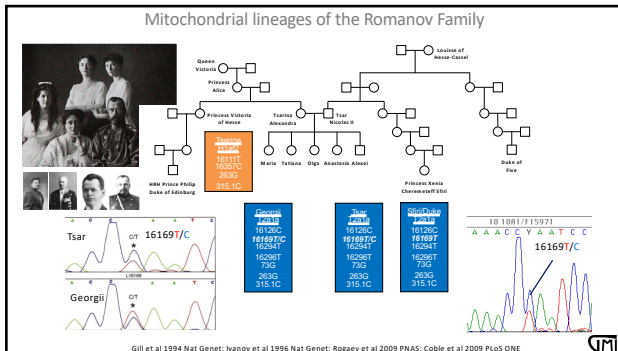
Ivan Kharitinov



Dr. Eugene Botkin

SMI

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Table 2 Summary of mtDNA differences compared to the Anderson¹⁰ reference sequence

Origin of sample	DNA source	Length sequenced (bp)	Positions within hypervariable regions (HVR) of mitochondrial DNA																
			HVR 1							HVR 2									
			16111	16158	16281	16284	16289	16290	16296	16304	16311	16327	73	146	195	308	309.1	309.2	315.1
			C	T	C	C	C	A	C	C	T	T	T	A	T	T	A	C	C
Servant 1 (?)	Femur skeleton 1	760	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Servant 2 (?)	Femur skeleton 6	742	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Servant 3 (?)	Femur skeleton 9	650	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Royal Physician - Dr. Botkin (?)	Femur skeleton 2	736	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Daughter 1 of Tsar/Tsarina (?)	Femur skeleton 3	755	T	-	-	-	-	-	-	-	-	-	C	-	-	-	-	-	-
Daughter 2 of Tsar/Tsarina (?)	Femur skeleton 5	634	T	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Daughter 3 of Tsar/Tsarina (?)	Femur skeleton 6	760	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Tsarina Alexandra (?)	Femur skeleton 7	744	T	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Duke of Edinburgh (Grand-nephew of Tsarina)	Blood sample	760	T	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Tsar Nicholas II (?)	Femur skeleton 4	782	-	C	Y	-	-	-	-	-	-	-	T	-	-	-	-	-	-
Gr. Gr. grandson of Louise of Hesse-Cassel	Blood sample	781	-	C	T	-	-	-	-	-	-	-	T	-	-	-	-	-	-
Gr. Gr. Gr. granddaughter of Louise of Hesse-Cassel	Blood sample	782	-	C	T	-	-	-	-	-	-	-	T	-	-	-	-	-	-

... Sequence unchanged from reference sequence; -, No nucleotide assignment; *, Nucleotide absent from reference sequence; Y, CT heteroplasmy.

700 < LR < 8.4x10⁵

Gill et al 1994 Nat Genet

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Prince Alexei, aged 8 or 9, in a detail from an official Russian royal-family photograph taken in 1913. His sister Grand Duchess Anastasia's hand drapes over his shoulder.

Remains of Czar Nicholas II's Son May Have Been Found

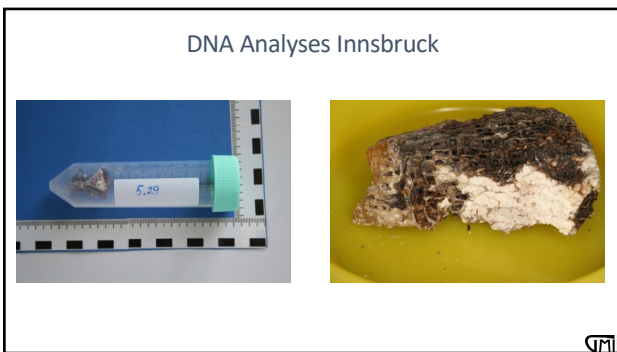
- FRIDAY, AUGUST 24, 2007
- MOSCOW — The remains of the last czar's hemophiliac son and heir to the Russian throne, missing since the royal family was gunned down nine decades ago by Bolsheviks in a basement room, may have been found, an archaeologist said Thursday.

<http://www.foxnews.com/story/0,2933,294360,00.html>

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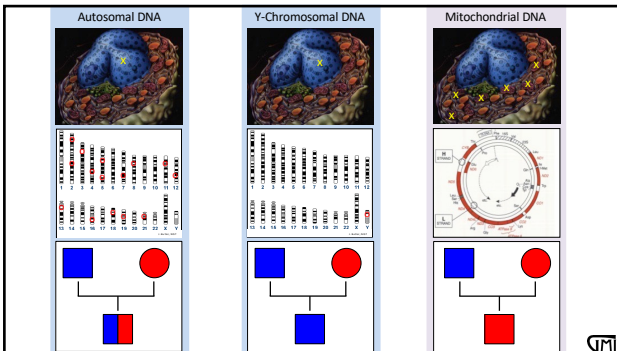
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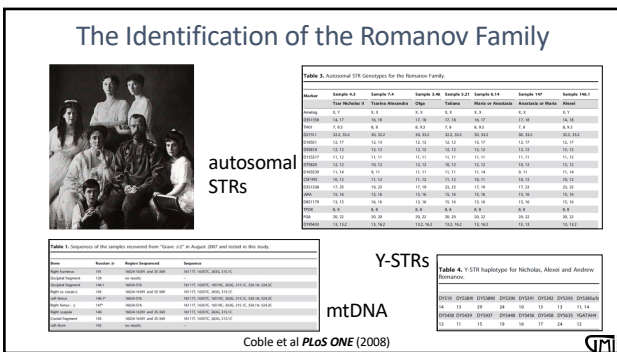
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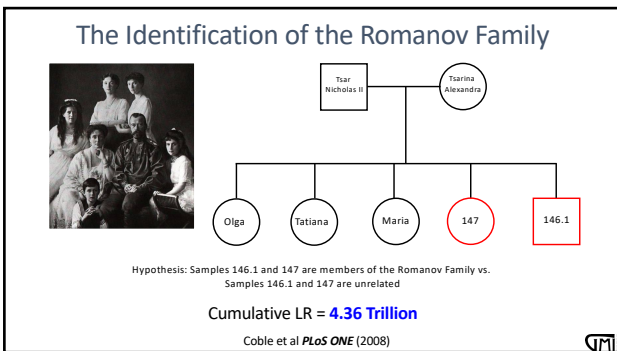
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Confirmation of the Tsar's STR profile



Nagasaki, 1891



blood stain on shirt

Full matches in Y-Chromosomal and autosomal STR profiles

Coble et al *PLoS ONE* (2008)



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The Imposters

Establishing the identity of Anna Anderson Manahan
 Peter Gill, Colin Kimpton, Rosemary Aliston-Greiner, Kevin Sullivan, Mark Stoneking, Terry Melton, Julian Nott, Suzanne Barrit, Rhonda Roby, Mitchell Holland & LTC Victor Weedn
Nature Genetics 9, 9–10 (1994)



Anna Anderson Manahan

Origin of sample	DNA source	Length sequenced (bp)	Position within the non-coding region (ref. 11)					
			16111	16128	16261	16292	16302	16307
Reference sequence	-	-	C	T	C	C	T	T
Duke of Edinburgh (Great nephew of Tsarina)	Blood sample	403	T	-	-	-	-	C
Anna Anderson	Infantina sample	403	C	T	T	T	C	-
Anna Anderson	Hair sample (3 hairs)	344-352	C	T	T	C	-	-
C. Maucher (Great nephew of Schandorowicz)	Blood sample	380	C	T	T	C	-	-

... Sequence unchanged from reference sequence.



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"They never answered the other forensic evidence — the mountain of forensic evidence — in her favor," says a still-believing Robert Crouch, shown last month placing flowers near the memorial marker of the late Anna Manahan.



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The Imposters

Total number of "Russian imperial children" claimants since 1918:

	Olga: 28 claimants	
	Tatiana: 33 claimants	
	Maria: 53 claimants	
	Anastasia: 33 claimants	
	Alexei: 81 claimants	



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PLOS ONE

OPEN ACCESS PEER REVIEWED

RESEARCH ARTICLE

Mystery Solved: The Identification of the Two Missing Romanov Children Using DNA Analysis


Michael D. Coble, Odile M. Loretto, Mark J. Wadhams, Suni M. Edson, Kerry Maynard, Camia E. Meyer, Harald Niederstätter, Cordula Berger, Burkhard Berger, Anthony B. Falsetti, Peter Gill, Walther Parson, Louis N. Finkel

Published: March 11, 2009 • <https://doi.org/10.1371/journal.pone.0004838>



241 Save	120 Citation
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Truth of Anastasia Romanov





Öffentliche Gruppe · 704 Mitglieder

Gruppe beitreten



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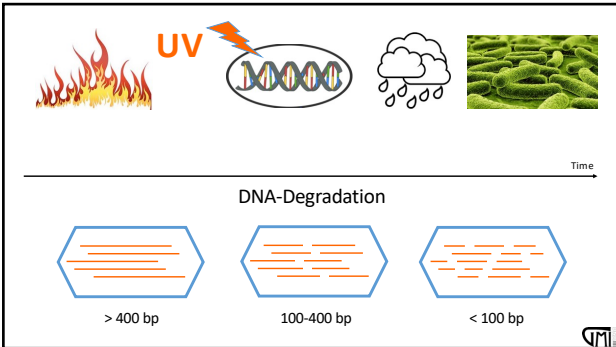
Celebrity Genetics

				
				
Wolfgang A. Mozart (+1791) Parson 2006	Friedrich v. Schiller (+1805) Parson 2008	King Richard III (+1485) King et al 2014	Kaspar Hauser (+1883) Parson et al in 2024	Schliederhannes (+1803) Parson et al under review

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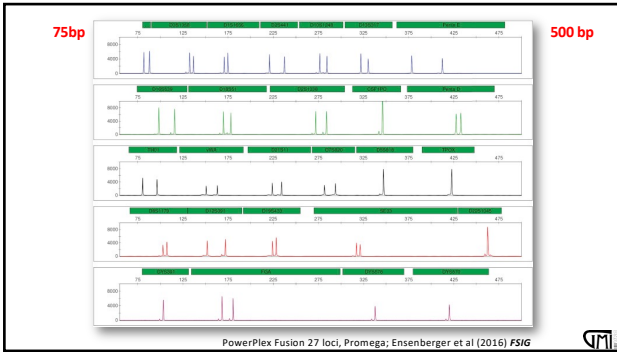


DNA-Degradation

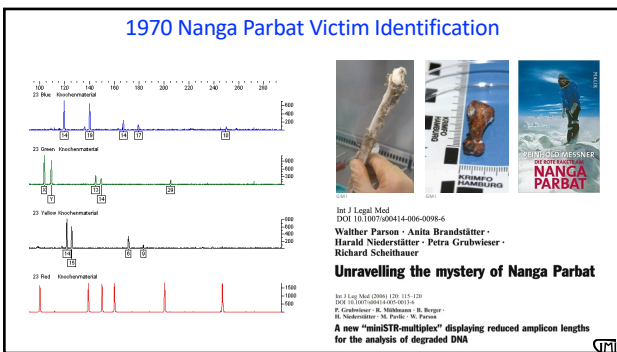
Time →

> 400 bp 100-400 bp < 100 bp

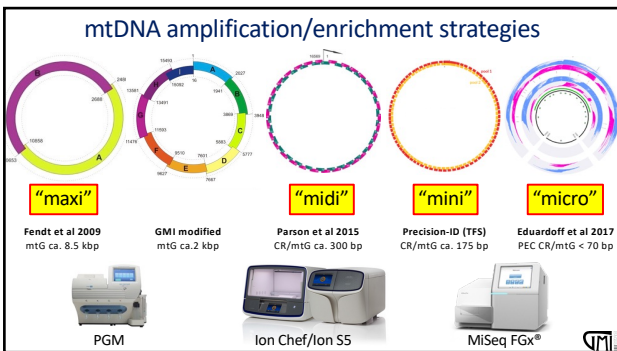
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Primer Extension Capture mtDNA CR MPS

Briggs et al Science (2009)

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SEPTEMBER 26, 2014, IGUALA, MEXICO

43 male students from the Ayotzinapa Rural Teachers College went missing

Bag was found in a **water** with putative human remains

We received **17 severely burnt fragmented** remains

REUTERS

Austrian forensic experts may shed light on Mexico massacre

BY SHADIA NASRALLA
PUBLISHED 11:50 AM GMT+2 Sep 26, 2014

Austrian forensic experts with 43 samples of DNA from remains were found inside the mass graves in Iguala, near Toluca in the southwestern state of Guerrero, September 26, 2014.

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Conventional DNA analysis

17 samples cleaned and DNA extracted (Phenol/Chloroform)

qPCR nDNA (Niederstatter et al 2007) brought promising quants in only **1 sample**

This sample gave a **full aSTR profile** matching **1** of the 43 families

qPCR mtDNA (Niederstatter et al 2007) brought **no quants** in the remaining 16 samples (143 bp)

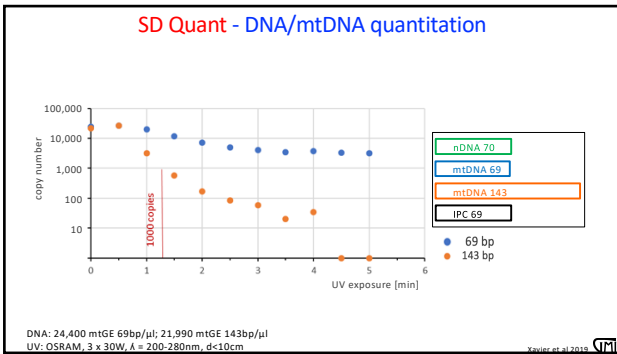
AMERICAS

Remains of Student in Mexico Identified

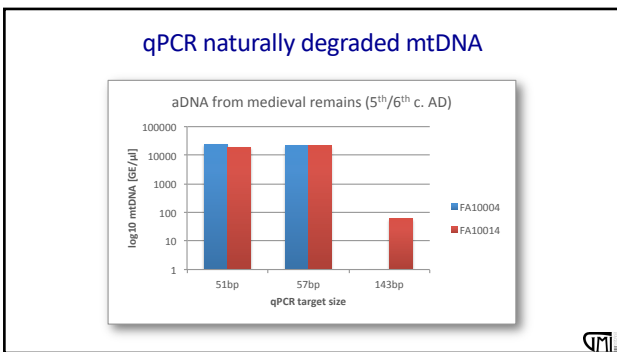
By RABBIT C. ABRONIA and PAULINA YELLAGER OCT 2, 2014

Cristian Mena, father of Alexander Mena, whose remains were said to have been identified, mourns at his home in Iguala, Mexico, on Sunday.

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9 of 16 “hopeless” samples brought PEC results

NEWS

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Latin America & Caribbean

Remains of second Mexican student identified

© 17 September 2015 Latin America & Caribbean

The relatives of the 42 missing students have held regular protests and marches for the past year.

genes

MDPI

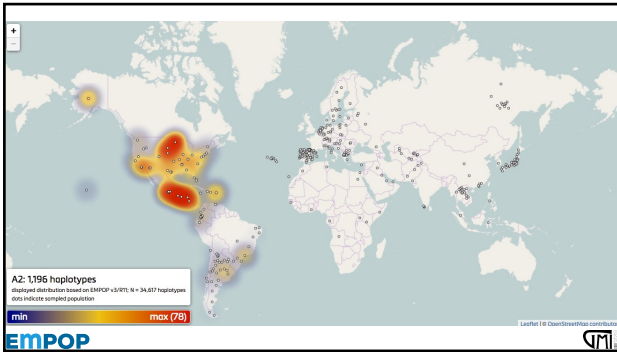
Optimized mtDNA Control Region Primer Extension Capture Analysis for Forensically Relevant Samples and Highly Compromised mtDNA of Different Age and Origin

Mayra Edvardoff ^{1,*}, Catarina Xavier ¹, Chistina Strobl ¹, Andrea Casan-Vargas ² and Wálter Paron ^{1,3,†}

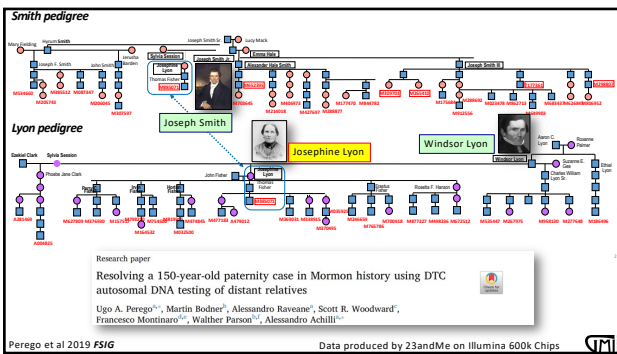
<https://www.genesjournals.com>

JMI

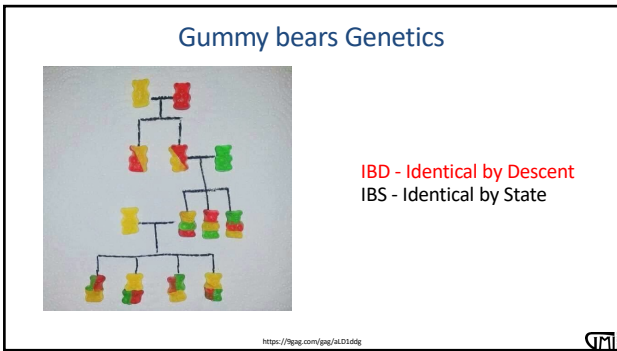
51



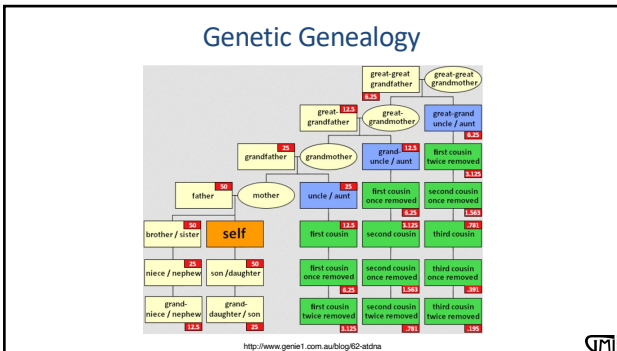
52



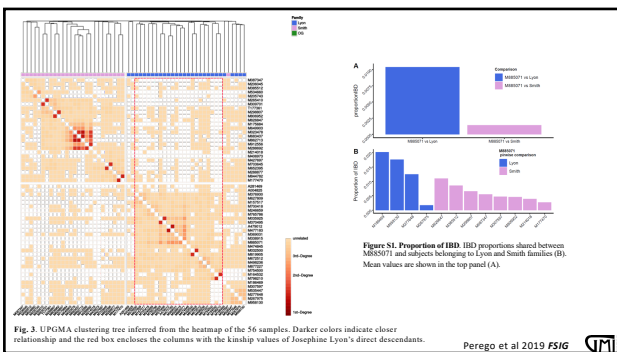
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The Case of the „Golden State Killer“ 1974 - 1986

- 13+ murdered, 50+ raped, 120+ burglarized

?

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**The Case of the „Golden State Killer“
1974 - 1986**

- The investigation never stopped ...
 - Numerous suspects had been identified and excluded
- The investigator
 - Paul Holes (Contra Costa County district attorney's office) – close to retirement
- The DNA sample
 - An unused frozen rape kit from 1980 was analyzed, compared to the CODIS profile from the unknown rapist for authentication
 - Submitted to commercial provider for genome-wide SNP analysis, then uploaded to public GEDmatch database
- The genealogist
 - Barbara Rae-Venter, retired patent attorney working in genealogy to help adoptees find their parents



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**The Case of the „Golden State Killer“
1974 - 1986**

GEDmatch Tools for DNA and Genealogy Research

GEDmatch provides DNA and genealogical analysis tools for amateur and professional researchers and genealogists. Most tools are free, but we do provide some premium tools for users who wish to help support us with contributions. You will need to upload DNA and / or genealogical (GEDCOM) data to make use of the tools here. Registration requires your name, email and a password of your choice. Click [HERE](#) to register.

- Two parental family trees established – of Italian and British origin
- The British side was informative leading to hundreds of potential relatives, connecting a 3rd cousin with a putative great-great-grandfather
- Further searches involved birth records, newspaper clippings, social media profiles and family tree data
- One suspect providing a voluntary sample was excluded

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**The Case of the „Golden State Killer“
1974 - 1986**

- 13+ murdered, 50+ raped, 120+ burglarized



- Joseph James DeAngelo, arrested April 24, 2018 in Sacramento, CA
 - Born in 1945, Vietnam veteran, 1971 BSc in criminal justice, police officer 1973-1980 in Sacramento and Exeter, married 1973 & divorced 1991, 3 daughters, living since then in Citrus Heights, Sacramento

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The future of Forensic Genetics

1985-1995	Exploration
1995-2005	Harmonization
2005-2015	Growth
2015-	Sophistication

adapted from Butler JM *Phil. Trans. R. Soc. B* (2015)

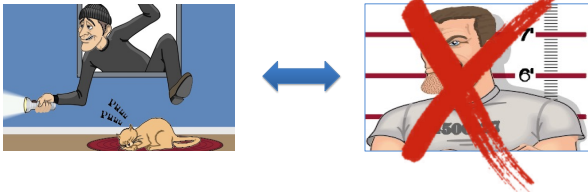
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Paradigm Shift in Forensic Genetics


Identification - **Prediction**
Genetics - **Genomics**

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Cases without suspects




Provide investigative leads through DNA




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the invisible gorilla



Ca. 1 min movie of two teams playing basketball
Q: How often do the teams pass a ball
A lady in a gorilla costume walks through
228 test persons watching the movie

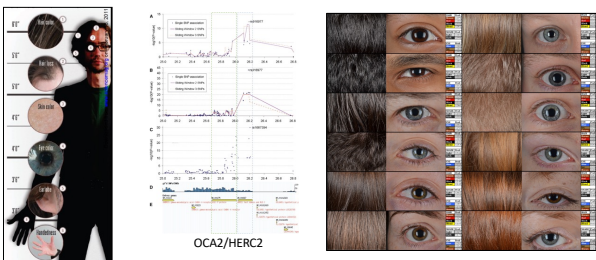
And Other Ways Our Intellions Deceive Us



Results
194 counted the passes correctly
In most groups ca. 50% of the participants did not report seeing the gorilla
Success **higher** in participants who counted the black team
= **Inattentual blindness**

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Forensic DNA Phenotyping



OCA2/HERC2

Kayser et al *AHG* 2008

Kayser et al *FSIG* 2015



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Ancestry Informative DNA Markers

The Madrid bomb investigation

Slide kindly provided by C. Phillips, USC Spain

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Age estimation

Heidegger et al 2022

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The Vaastra Case

Traces from perpetrator's blood and semen found at the crime scene
 No suspect/database matches
 Blame on close-by asylum seekers refuge database
 Case went cold
 Case re-opened in 2012
 Y-chromosomal analyses led to framer Jasper S. living close by

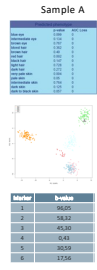
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Forensic Genomics (Wahlfach) VU1 (June 2025)

Appearance
Hirraples-S
Erasmus MC Genetic Identification
Indiana Purdue University Walsh Lab Genetic Tools

Ancestry
University Santiago Compostela (Spain), PCA, SNIPPER
STRUCTURE/CLUMPAC, mtDNA

Age
Somatic panel and model for blood



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