Investigative Genetic Genealogy (IGG): current methods, knowledge and practice



Genetisti Forensi Italiani

CONGRESSO NAZIONALE

LA GENETICA FORENSE: QUALE FUTURO?

10|11|12 Novembre 2022 GENOVA

Christopher Phillips

Forensic Genetics Unit, University of Santiago de Compostela, Spain

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Buckskin Girl, a 37-year MP ID case - identified 11th April 2018







DNA Doe Project is pleased to announce our first success story. We have identified Buckskin Girl, as confirmed this week by the Miami County, Ohio Sheriff's Office. While all other forensic identification methods failed, our pioneering approach using genetic genealogy succeeded.

We participated in a press conference held in Troy, Ohio this afternoon where the news was made public by Chief Deputy Stephen Lord.

- Found in a roadside ditch in Troy, Ohio, 24th April 1981
- > 2001 STR profile generated
- > 2009 mtDNA profile to CODIS
- > 2018 DNA Doe project involved
- New DNA extract made from highly degraded blood sample in storage
- Whole genome sequencing by Full Genomes Corporation
 - 28 March 2018, DNA profile uploaded to <u>GEDmatch</u> community database
- Successful match with 1st cousin-once removed (equivalent to 2nd cousin)
- Full analysis took 4 hours
- 11th April 2018 press conference identified as Marcia King, aged 21

The Golden State Killer Suspect - identified 24th April 2018





- 13 murders and 13 kidnappings linked to North California region
- Former police office so had easy access
- He possibly interfered with evidence

UNKNOWN SUSPECT

East Area Rapist/Golden State Killer California 1976 to 1986



REWARD

The FBI is offering a reward of up to \$50,000 for information leading to the arrest and conviction of the individual known as the East Area Rapist/Golden State Killer.

REMARKS

The East Area Rapist/Golden State Killer (EAR/GSK) is described as a White male, currently thought to be between the ages of 60 and 75 years old, and approximately 5'10" tall. He may have/had blond or light brown hair and an athletic build. He may have had an interest in the military, or had some military training, leaving him familiar and proficient with firearms.

The Golden State Killer Suspect - identified 24th April 2018





- 13 murders and 13 kidnappings linked to North California region
- Former police office so had easy access
- > He possibly interfered with evidence
- Four months to process rape-kit DNA

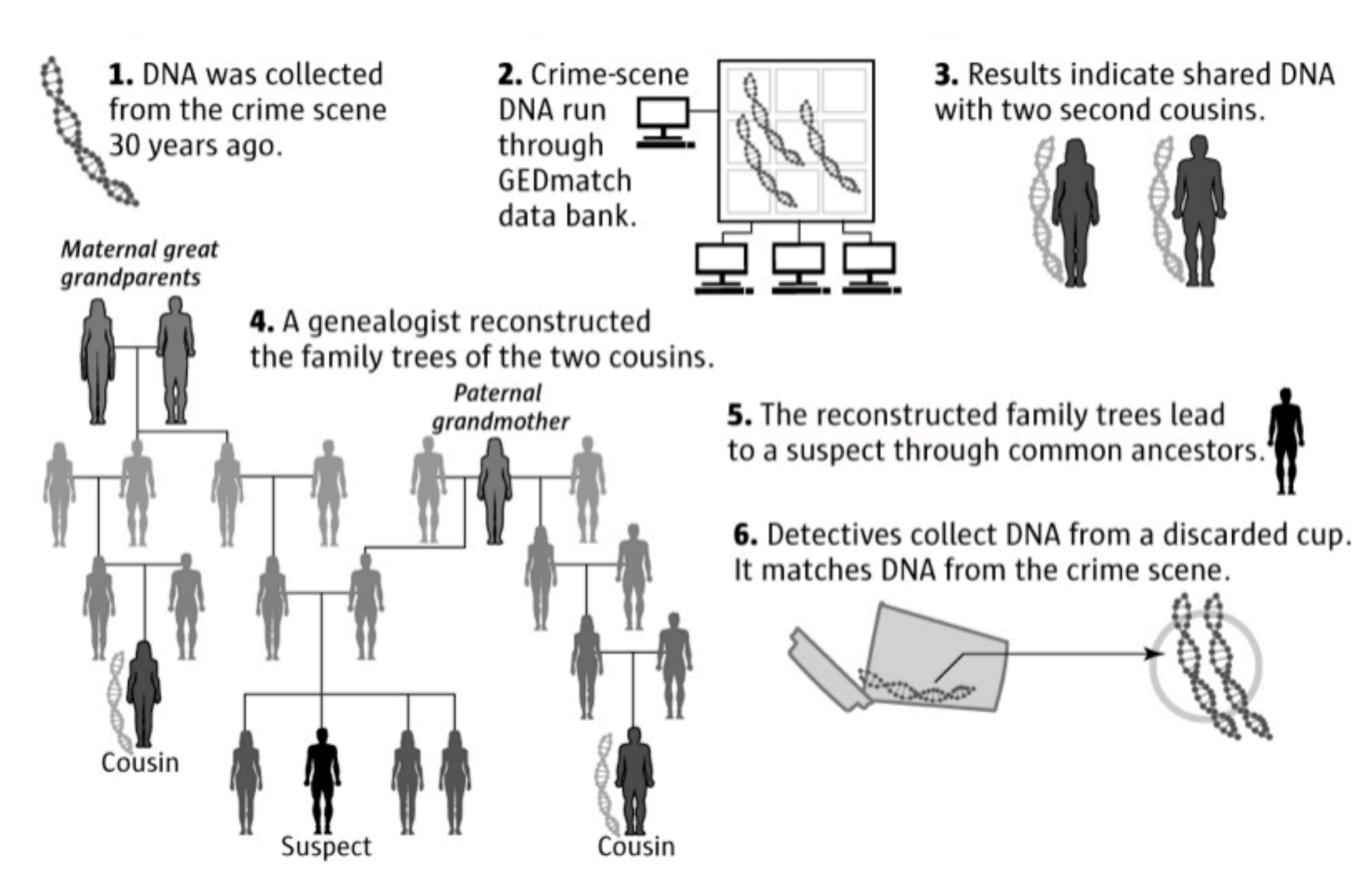


- It is likely, but not admitted, that a DTC 'buccal swab' was dosed with DNA and the SNP data uploaded to <u>GEDmatch</u>
- 10-20 relatives shared the same greatgreat-great grandparents
- Genealogist Barbara Rae Venter made multiple, extensive family trees
- Two suspects matched likely location, age, appearance, modus operandi data
- Collected DNA from cup/car door/tissue
- 24th April 2018 press conference identified as Joseph James DeAngelo, aged 72: 8 plus 4 FD murder charges

- Directly descended from Italian immigrants to the US
- Likely to have premature baldness (but probable age was ~70Y)
- Blue eyes

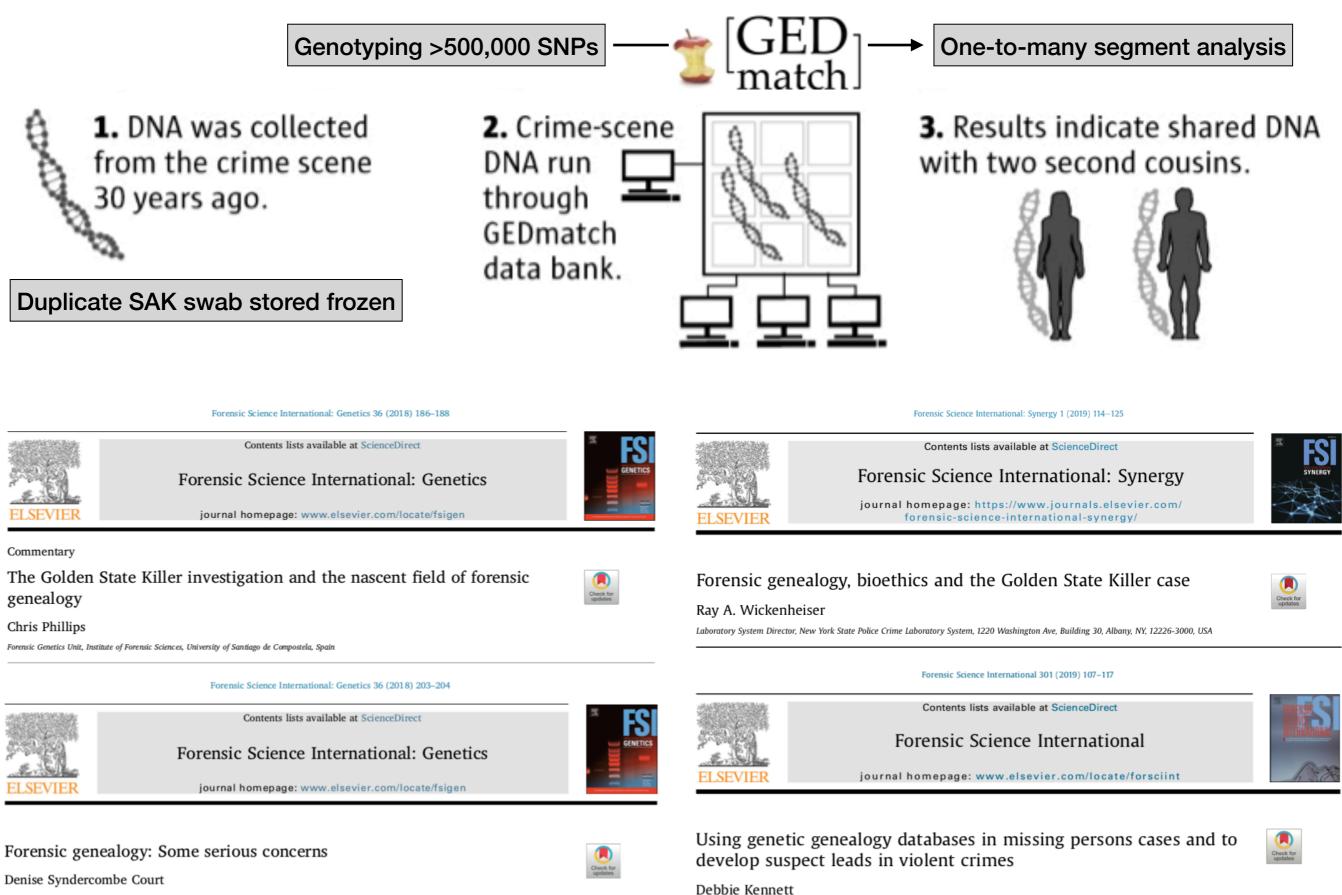
Identifying the Golden State Killer - the critical steps





Identifying the Golden State Killer - steps 2-3 were DNA analysis



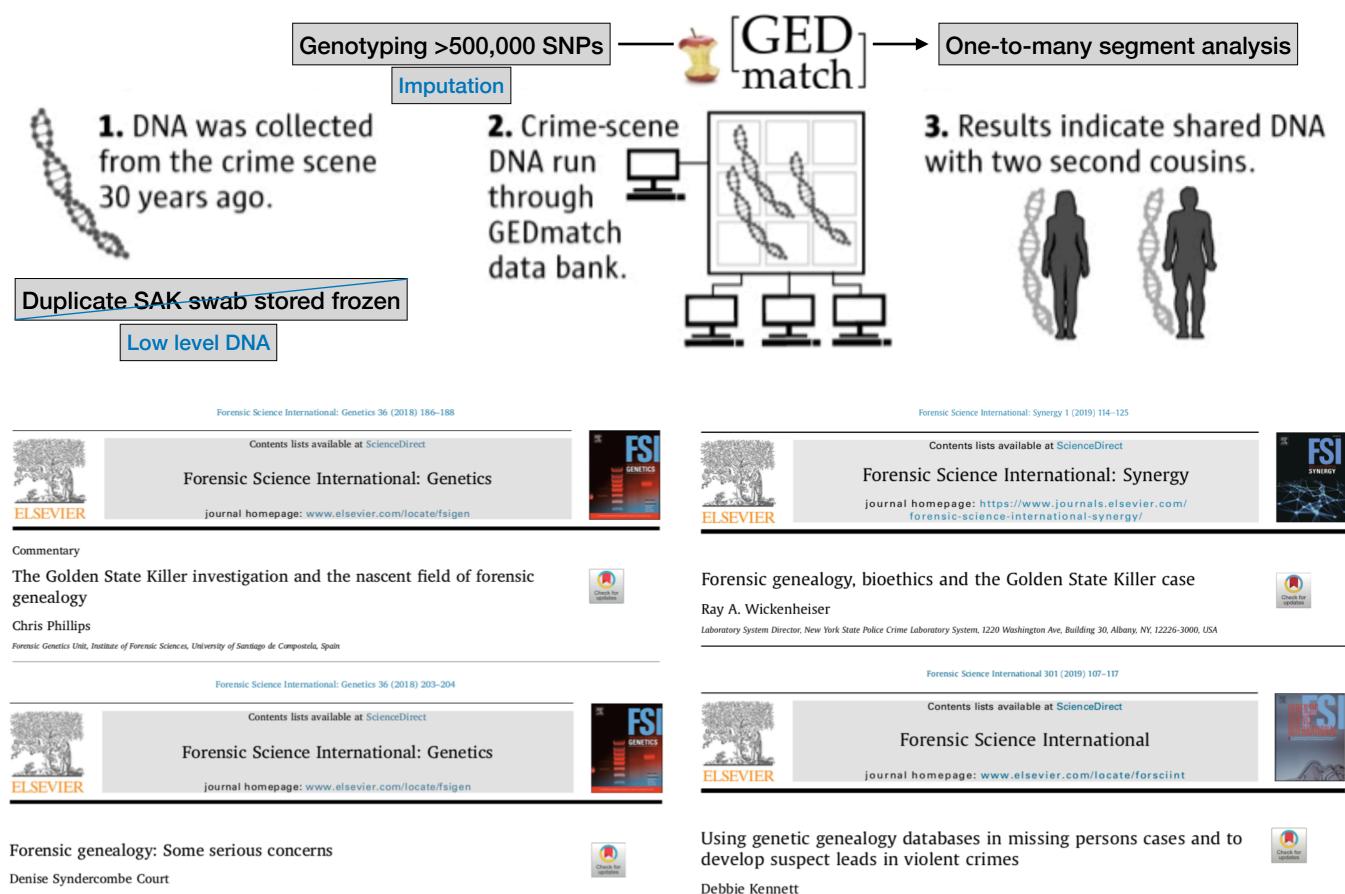


King's Forensics, Faculty of Life Sciences and Medicine, King's College, London, UK

Research Department of Genetics, Evolution and Environment, University College London, Gower Street, London WC1E 6BT, United Kingdom

Identifying the Golden State Killer - steps 2-3 were DNA analysis



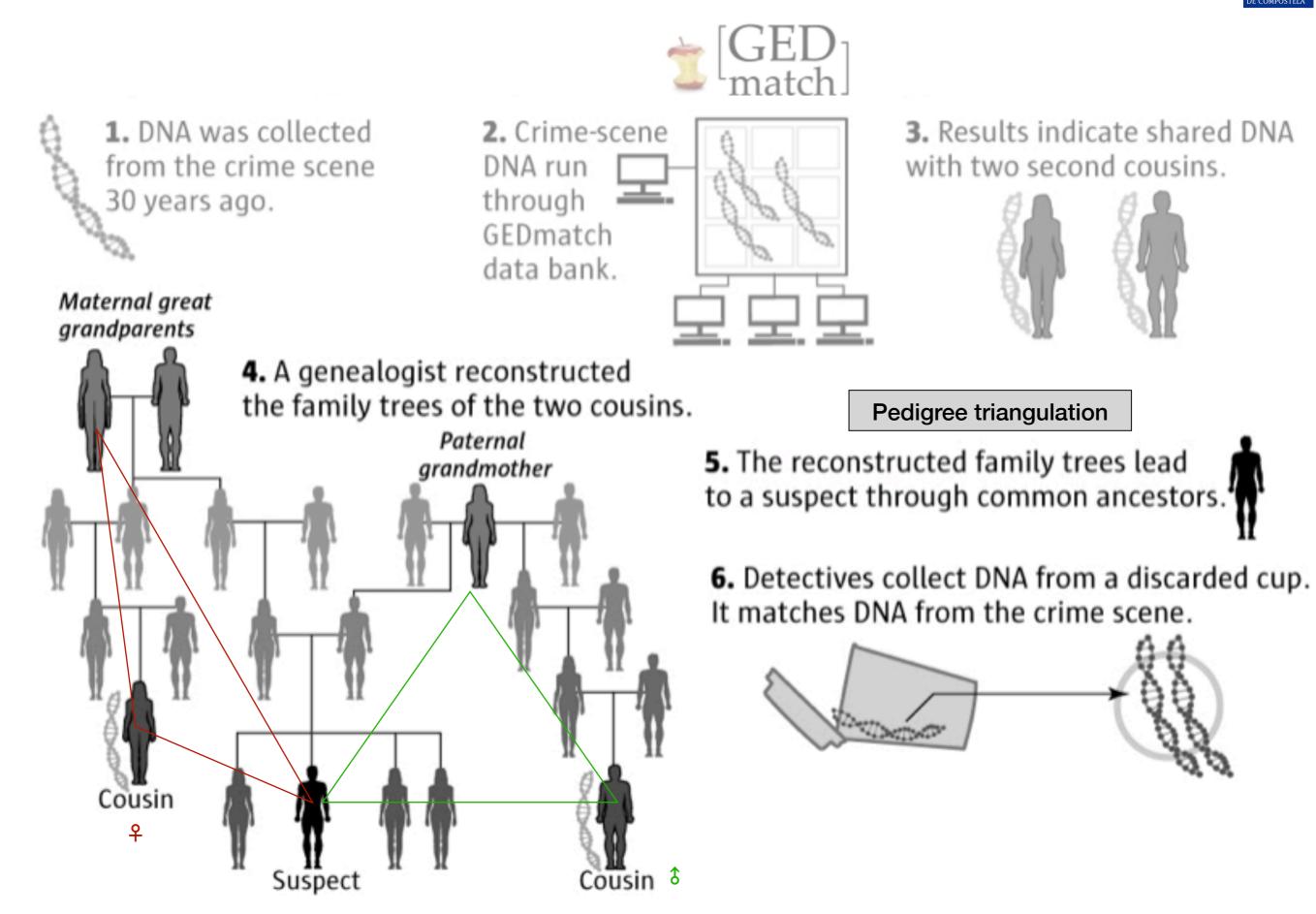


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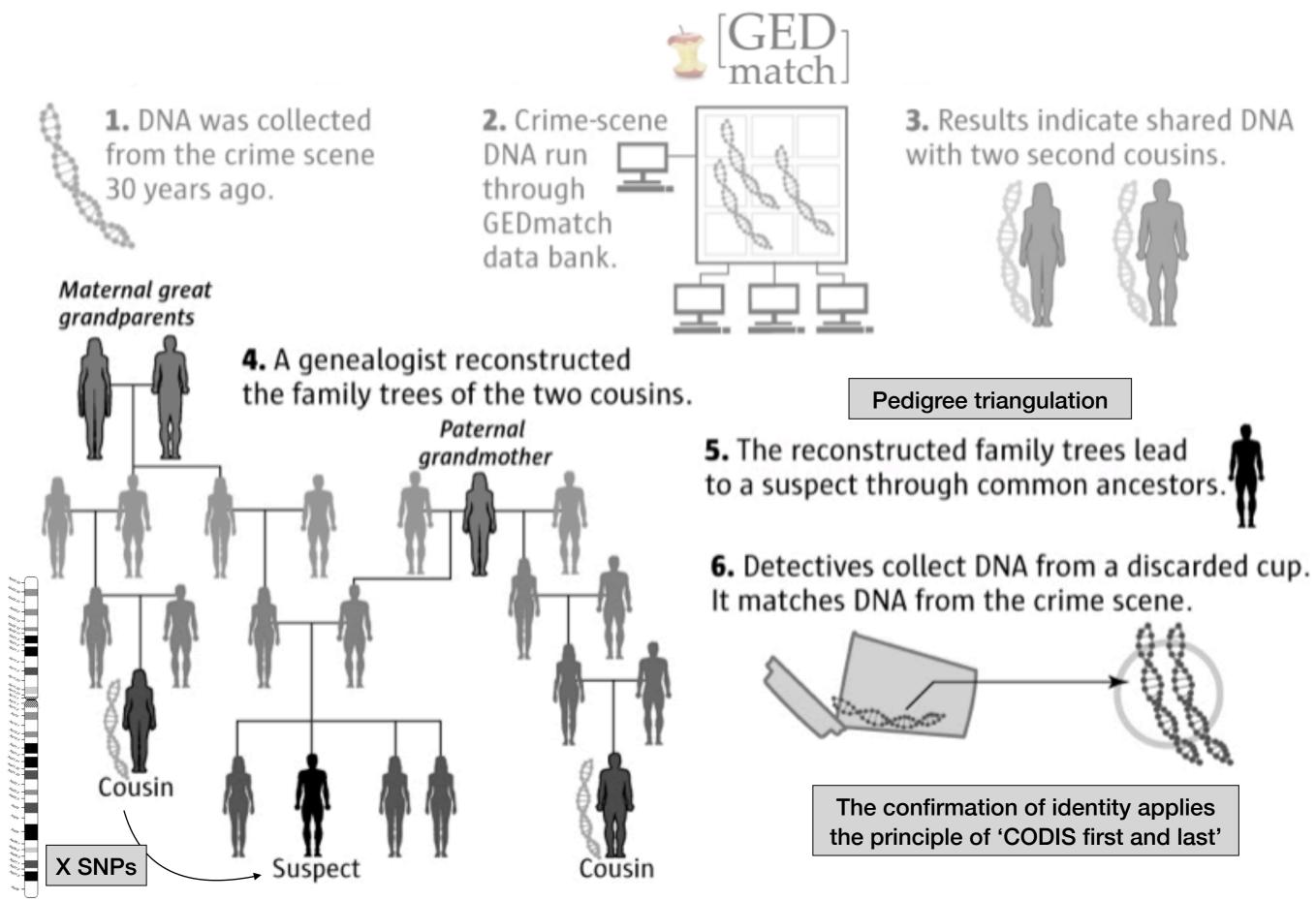
Identifying the Golden State Killer - steps 4-5 were genealogy



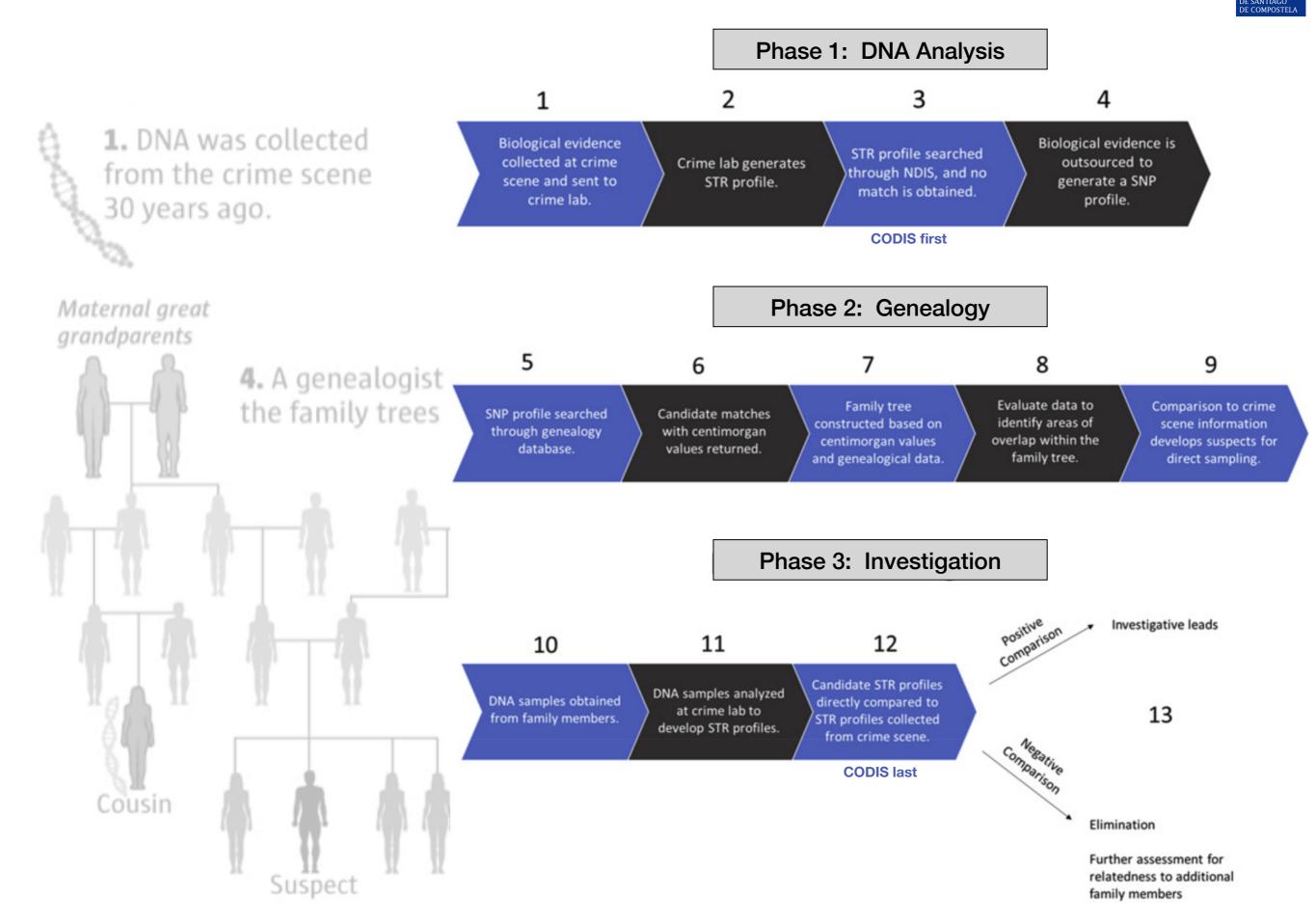


Identifying the Golden State Killer - steps 4-5 were genealogy



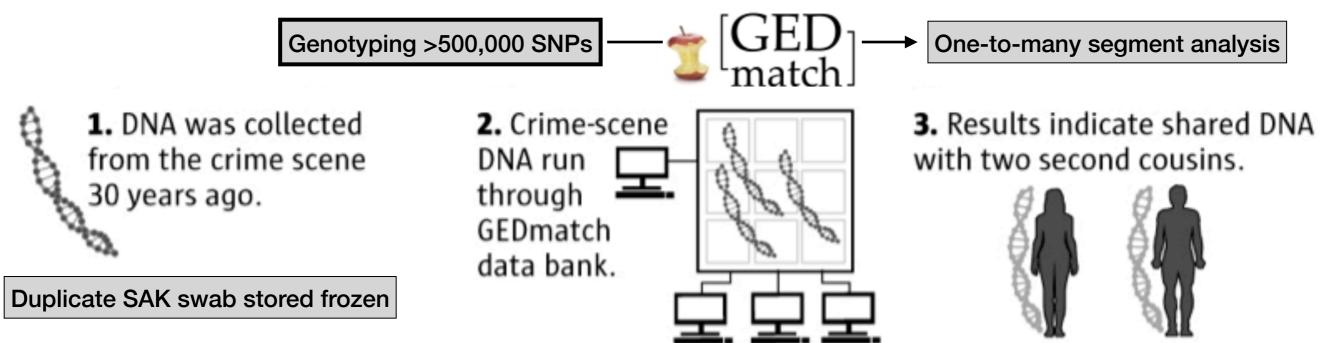


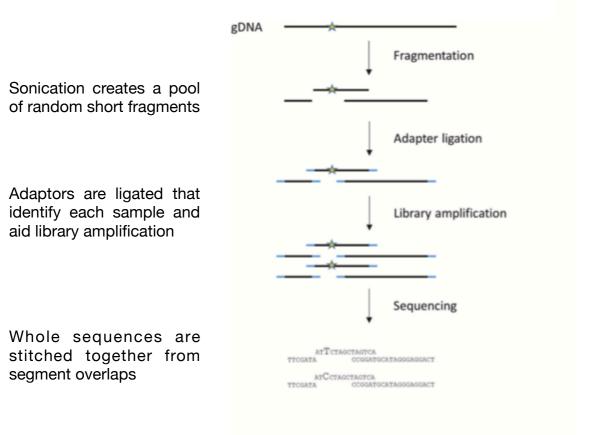
The IGG workflow in more detail



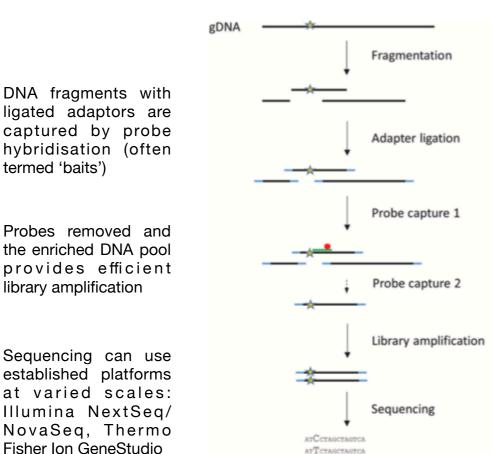
Identifying the Golden State Killer - genotyping high numbers of SNPs $\, {
m U}$



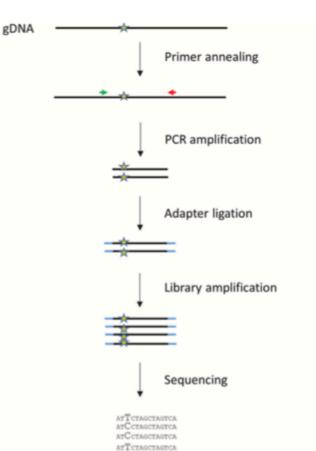




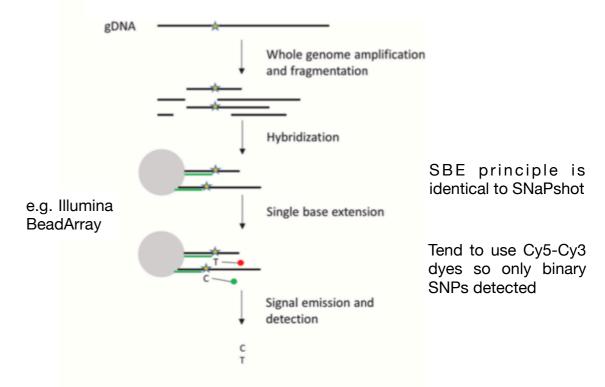
Hybridisation capture methods



MPS using conventional forensic PCR

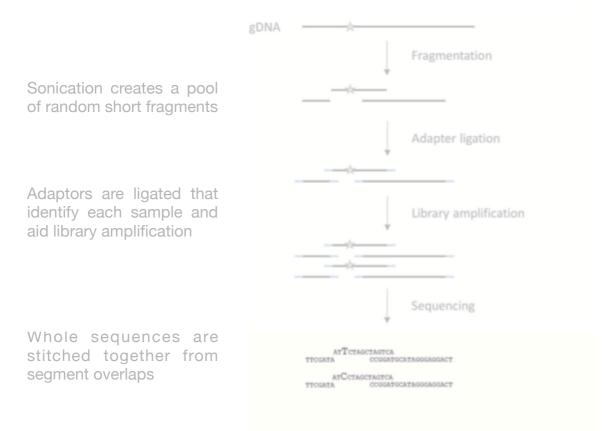


Whole-genome SNP arrays



Large-scale SNP genotyping

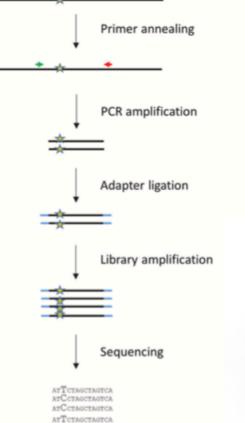
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Fisher Ion GeneStudio

MPS using conventional forensic PCR



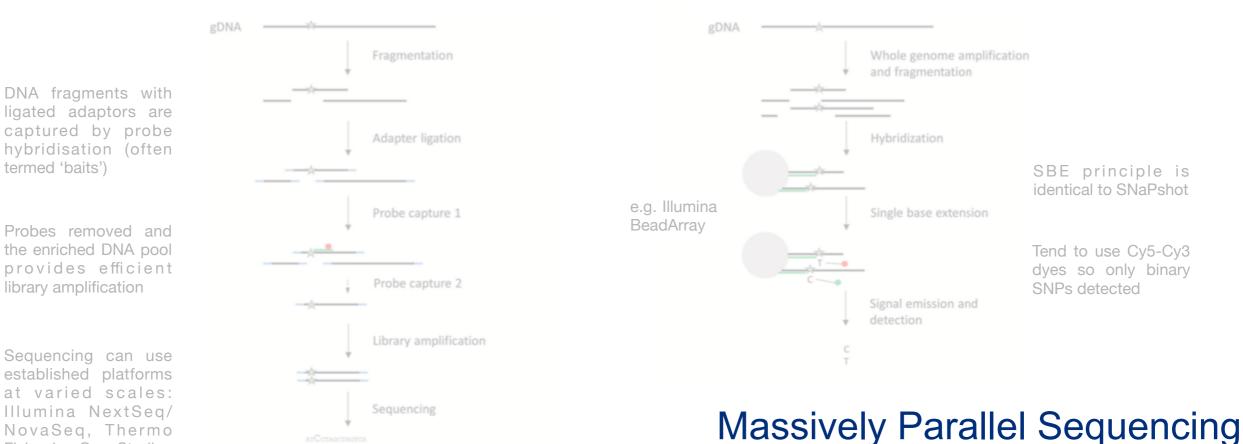


Whole-genome SNP arrays

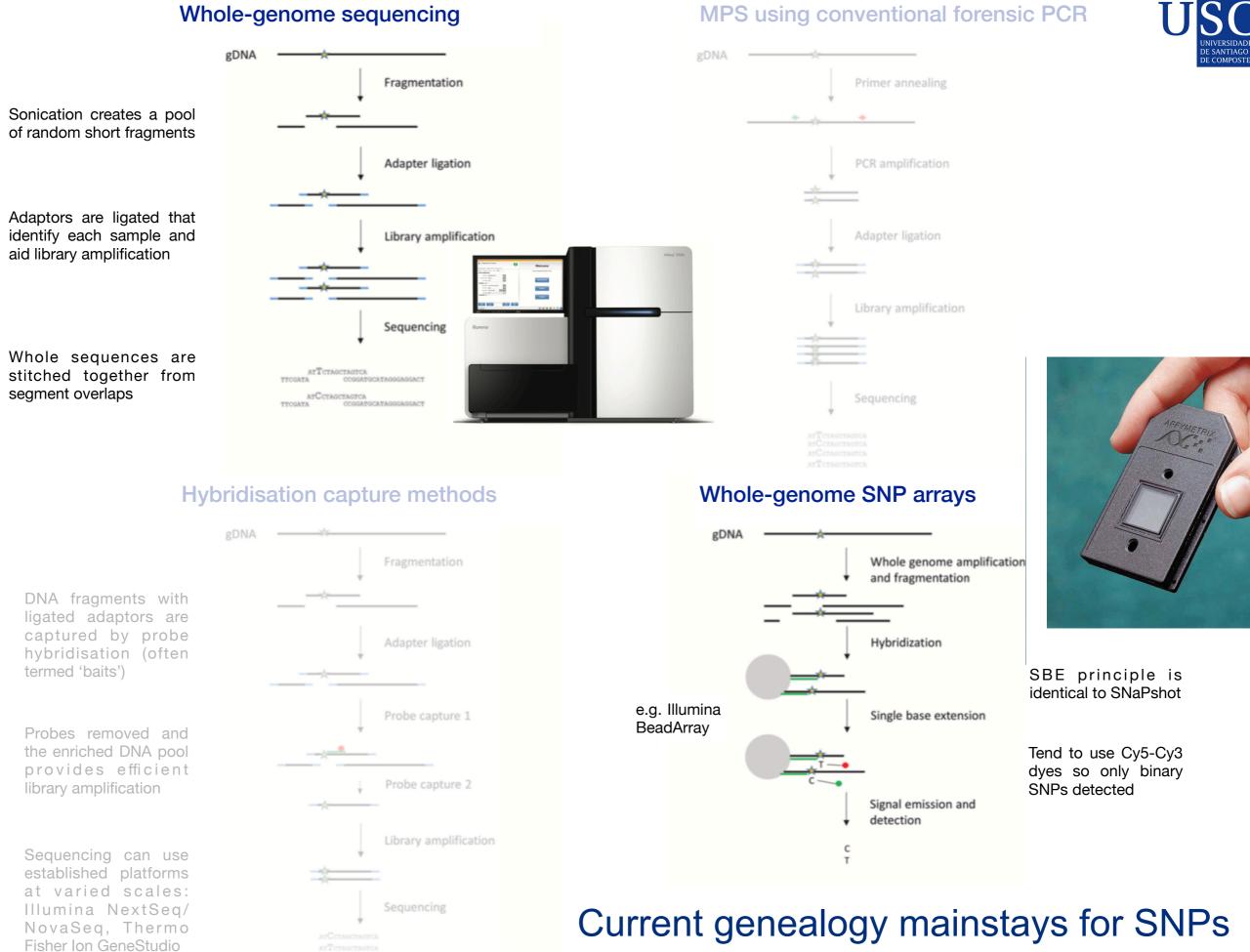


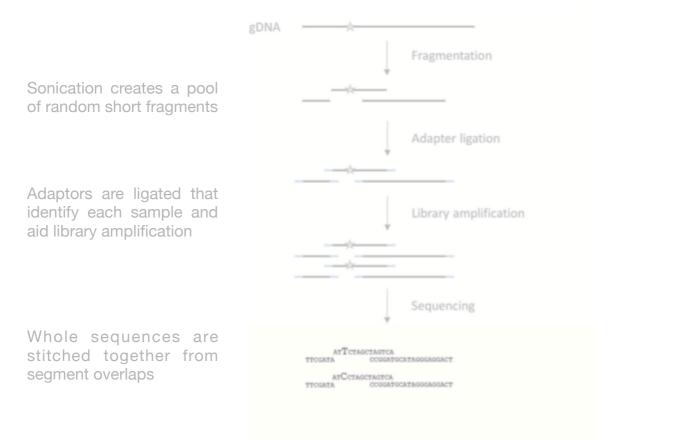


Hybridisation capture methods



gDNA





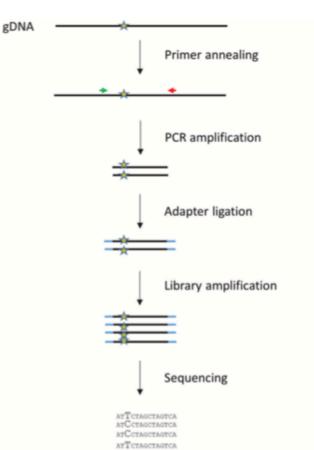
Hybridisation capture methods

atCCTAGCTAGTCA

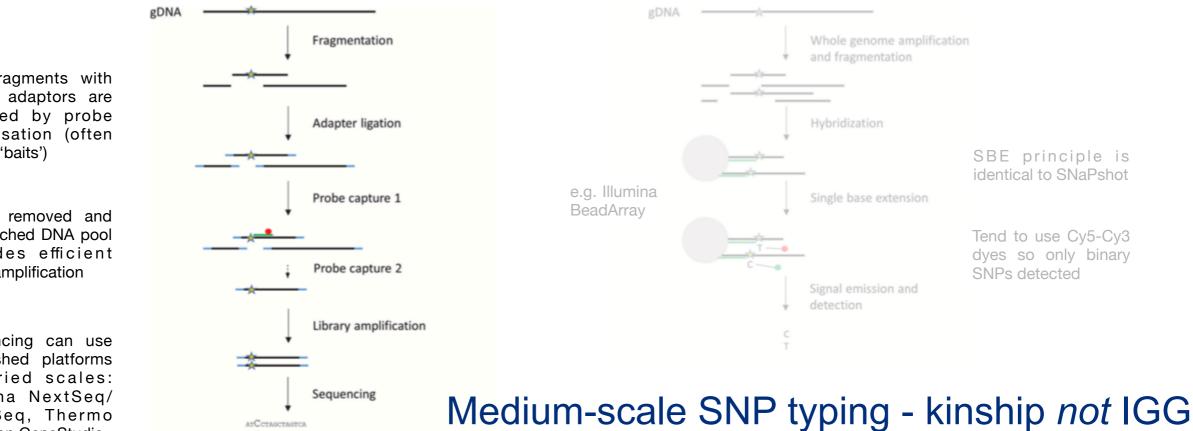
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MPS using conventional forensic PCR

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Whole-genome SNP arrays



DNA fragments with ligated adaptors are captured by probe hybridisation (often termed 'baits')

Probes removed and the enriched DNA pool provides efficient library amplification

Sequencing can use established platforms at varied scales: Illumina NextSeq/ NovaSeq, Thermo Fisher Ion GeneStudio

Case	WWII Context	Sample	Sample Type	Relationship to Service Member (DOR)	
		1	Buccal swab	Grandniece (3)	
А	USS Oklahoma	2	Buccal swab	Nephew (2)	
		3	Left femur	Self	
		4	Buccal swab	Nephew (2)	
		5	Buccal swab	Great grandnephew (4)	
В	USS Oklahoma	6	Buccal swab	Great grandniece (4)	
		7.1	Left femur	Self	
		7.2	Lettientui		
		8	Buccal swab	First cousin twice removed, male (5)	
		9	Buccal swab	Nephew (2)	
С	Austria	10	Buccal swab	Daughter (1)	
		11.1	Longhone	6.16	
		11.2	Long bone	Self	
		12	Buccal swab	Sister (1)	
		13	Buccal swab	Grandniece (3)	
D	Italy	14.1			
		14.2	Right parietal	Self	
		14.3			
		15	Buccal swab	Son (1)	
Е	Tarawa	16	Buccal swab	Daughter (1)	
Б	iaidwa	17.1	Right tibia	C-16	
		17.2	Right ubia	Self	

G C A T T A C G C C A T G C A T



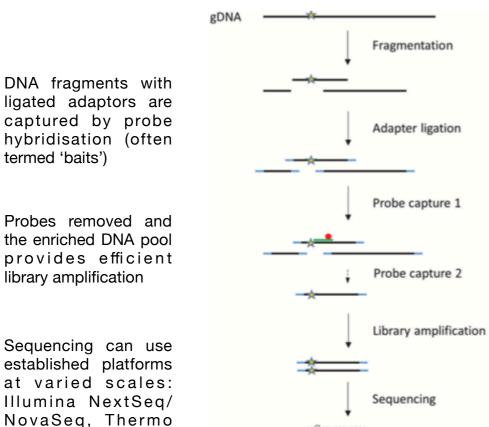
Article

The FORCE Panel: An All-in-One SNP Marker Set for Confirming Investigative Genetic Genealogy Leads and for General Forensic Applications

Andreas Tillmar ^{1,2,*}, Kimberly Sturk-Andreaggi ^{3,4,5}, Jennifer Daniels-Higginbotham ^{3,4}, Jacqueline Tyler Thomas ^{3,4} and Charla Marshall ^{3,4,6,*}

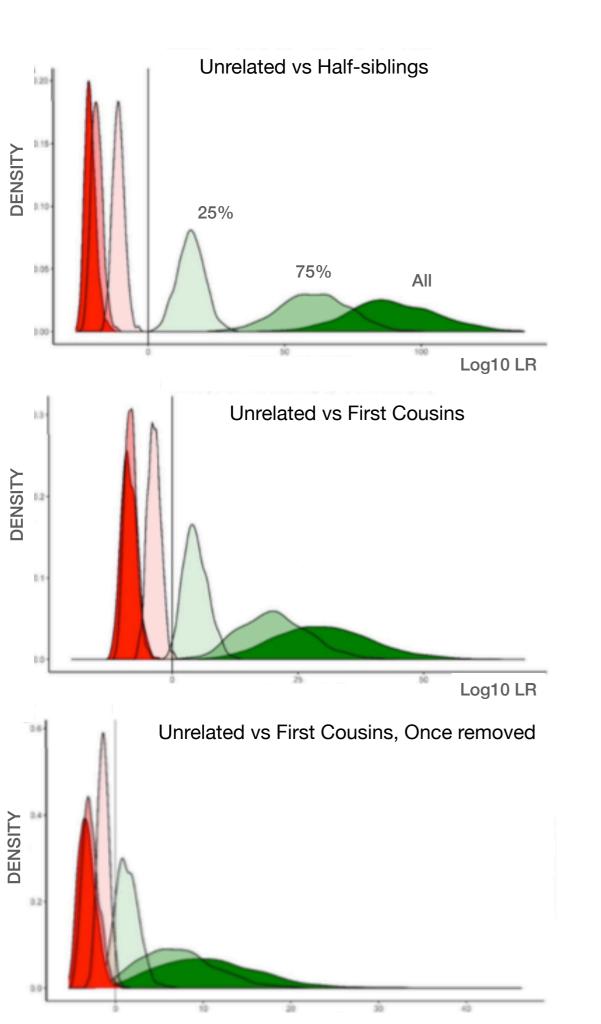
SNP Panels (# SNPs)
Infinium Global Screening Array (654,027), Infinium Omni Express (710,000), Infinium CytoSNP-850K (850,000)
ForenSeq DNA Signature Prep Kit: Primer Mix A (94)
Precision ID Identity Panel (90)
QIAseq Investigator 140 SNP panel (140)
ForenSeq DNA Signature Prep Kit: Primer Mix B (56)
Precision ID Ancestry Panel (165)
VISAGE panel (115)
ForenSeq DNA Signature Prep Kit: Primer Mix B (24)
VISAGE panel (41)
Precision ID Identity Panel (34)
AmpliSeq (884)
5422 SNPs in total Ancestry 237 4 FORCE 5,422 V-SNP 829 Y-SNP 829

Hybridisation capture methods



atCctagctagtca atTctagctagtca

Fisher Ion GeneStudio







Article

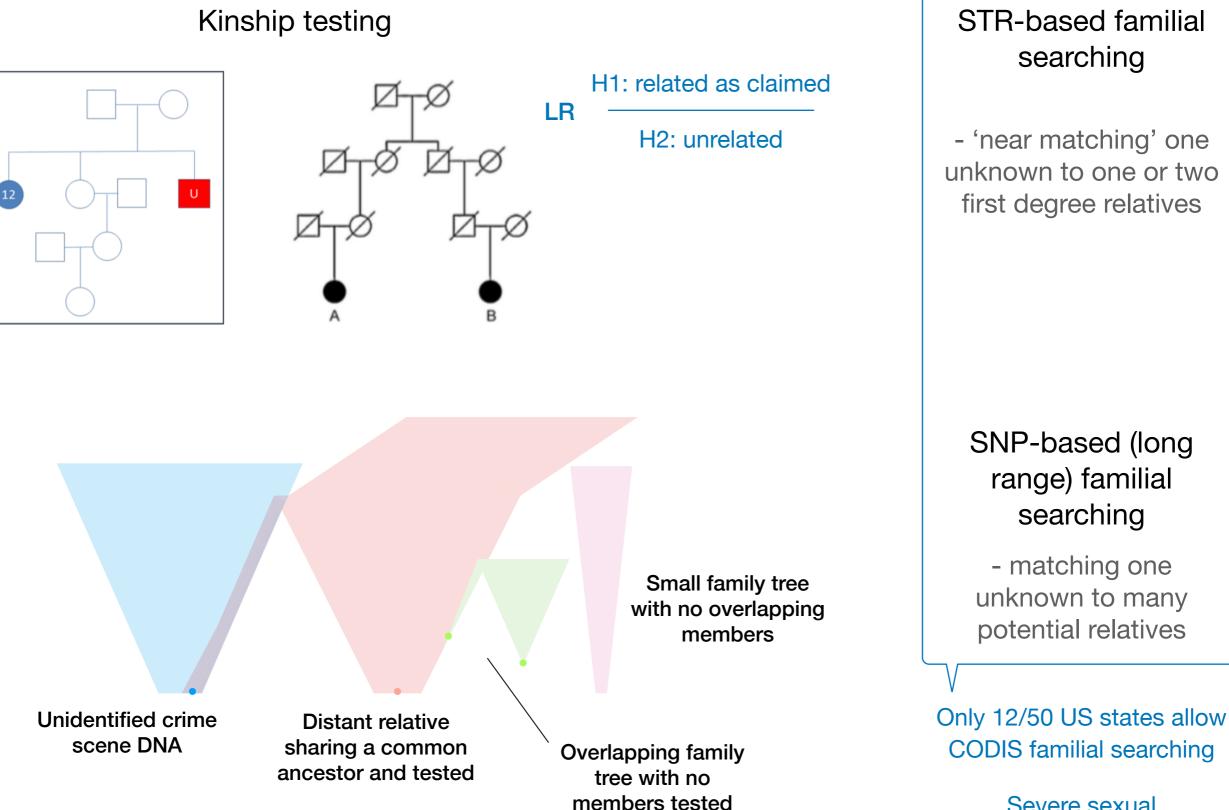
The FORCE Panel: An All-in-One SNP Marker Set for Confirming Investigative Genetic Genealogy Leads and for General Forensic Applications

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Marker Type	SNP Panels (# SNPs)					
Kinship SNP/ X-SNP						
	ForenSeq DNA Signature Prep Kit: Primer Mix A (94)					
iiSNP	Precision ID Identity Panel (90)					
	QIAseq Investigator 140 SNP panel (140)					
	ForenSeq DNA Signature Prep Kit: Primer Mix B (56)					
aiSNP	Precision ID Ancestry Panel (165)					
	VISAGE panel (115)					
	ForenSeq DNA Signature Prep Kit: Primer Mix B (24)					
piSNP	VISAGE panel (41)					
N CNTD	Precision ID Identity Panel (34)					
Y-SNP	AmpliSeq (884)					
The FORC	E panel Phenotype Contract of the second sec	entity 38 hship 929				

IGG looks beyond the pairwise comparisons of kinship tests

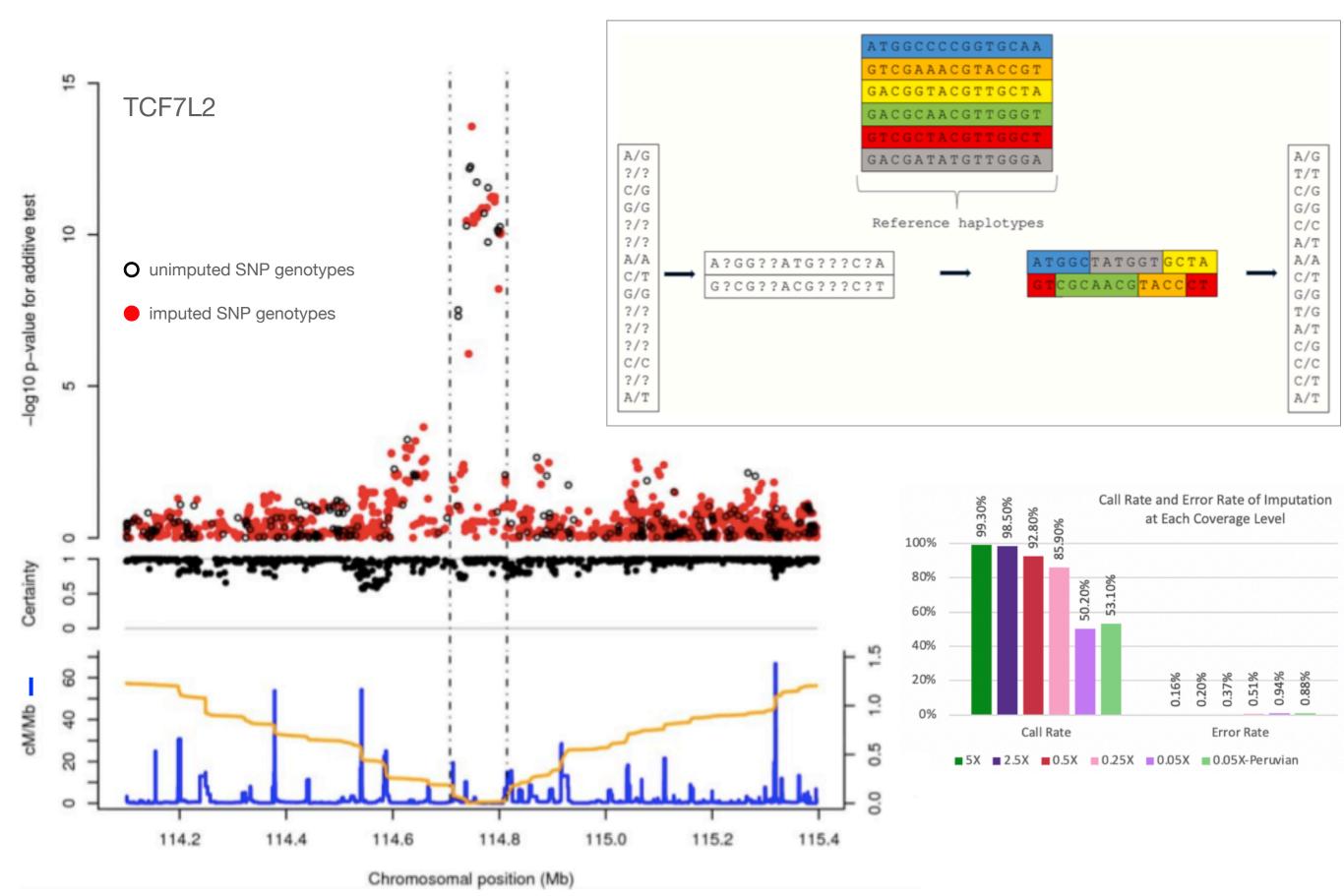




Severe sexual assault kit backlog

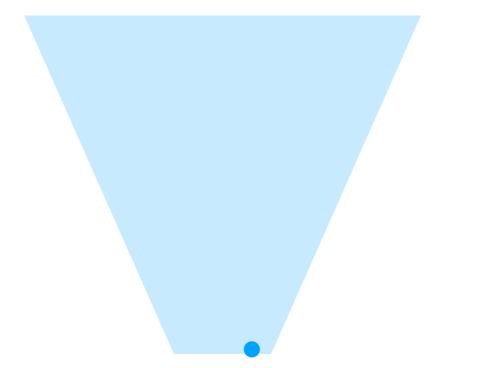
Principle of imputation assumes no recombination in closely sited SNPs - so missing genotypes are inferred from haplotype patterns





STR-based familial searches finds a group of close relatives



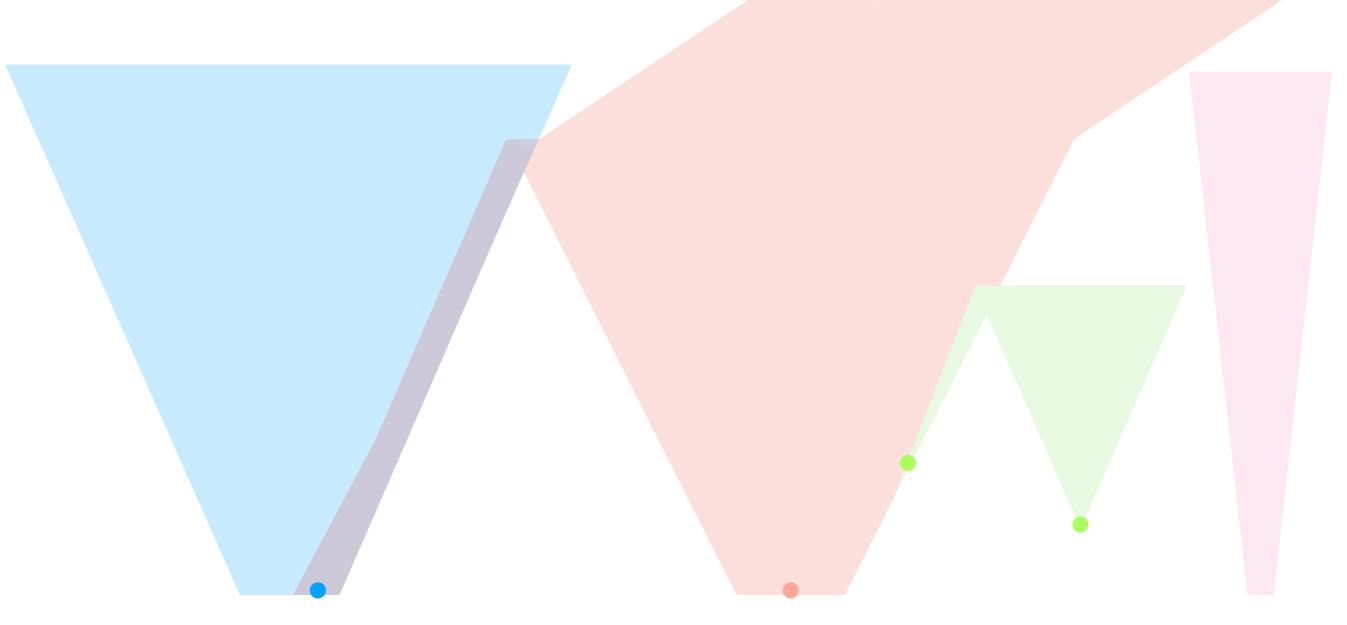


5-10 generations

Crime-scene donor (not in a DNA database)

Most pedigrees are unlinked until they go back > 10-20 generations





Crime-scene donor

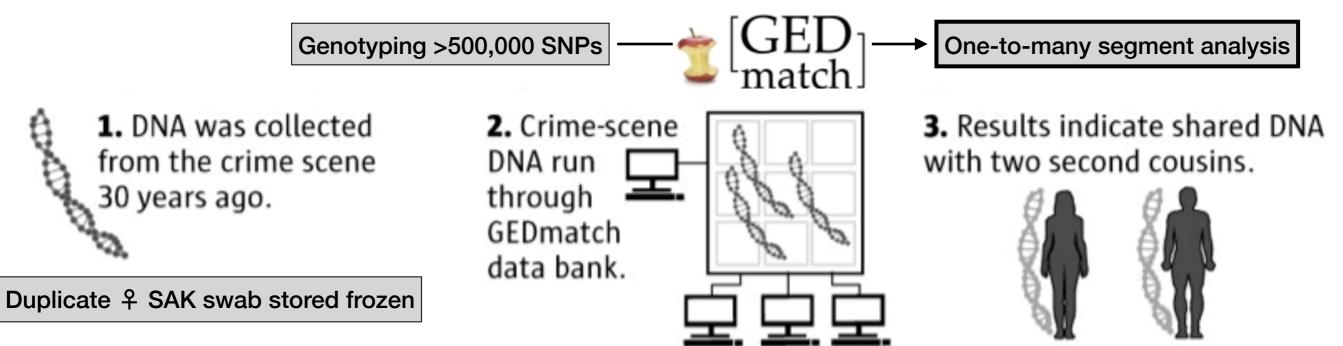
Related Individuals

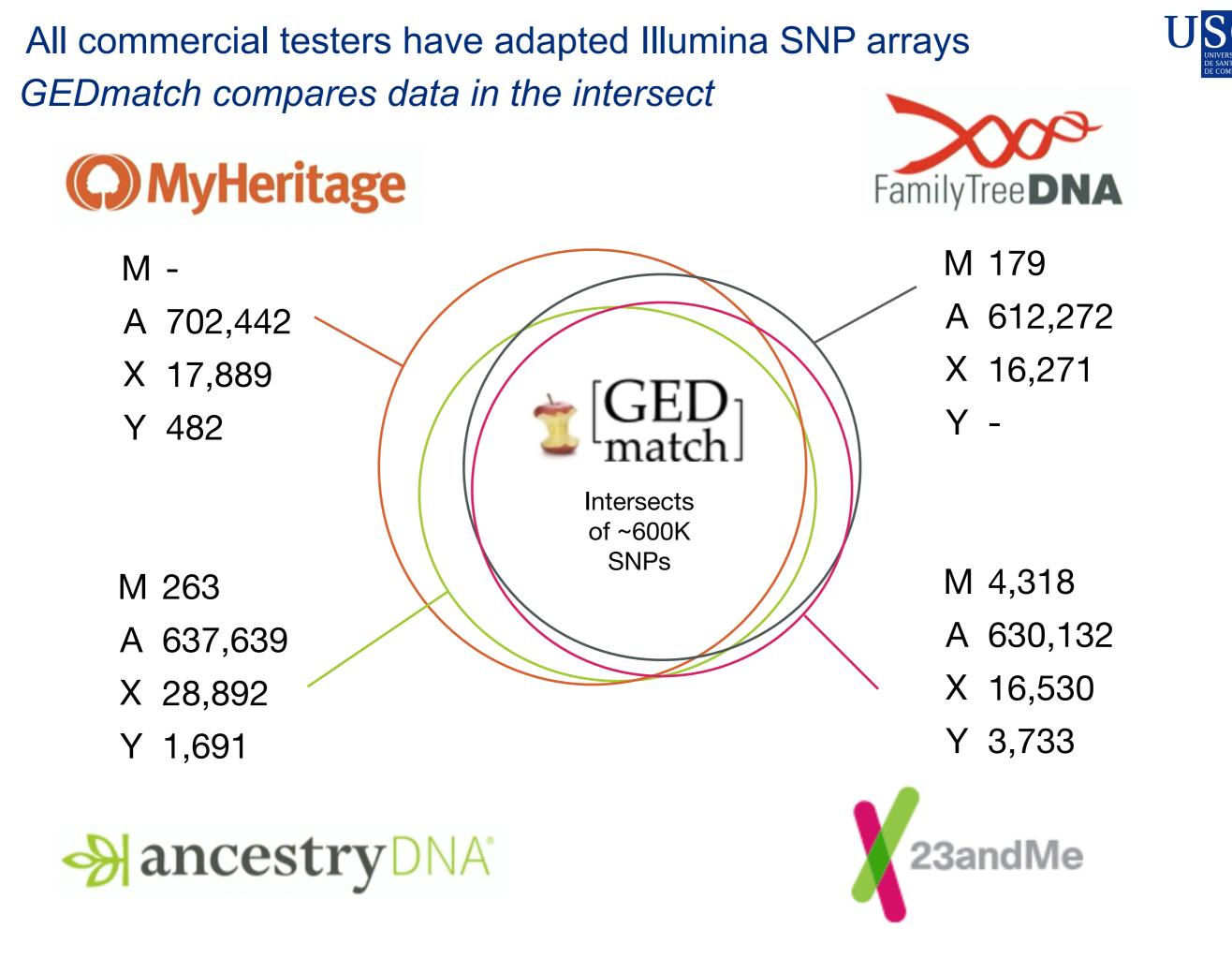
Unlinked pedigree

Individuals without offspring so pedigree ends

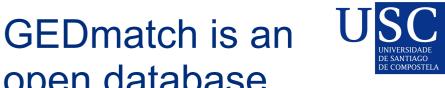
Identifying the Golden State Killer - segment analysis











		Now to use One to Many.	any WATCH VIDEO			open database
One-to-many DNA cor	nparison for: Kit es in Tier 1 One-to-many ve	Ĵ⊙ ersion.				NEW MATC Notification New feature
Filter By: Autosomal X	With this offset 0	With this limit 50 Prev Next Tips	CM size 7 Select all	Tag Groups None All One	Overlap Cutoff 45000	2021 We are pleased to announce a new feature at 0 Match Notification emails! If you are a free user, you can receive weekly e of new matches that are 100 cM or greater.
						If you are a Tier 1 user, you can receive weekly notifications, and can choose 30 cM, 70 cM, 10 as the threshold for new match notifications.



le are pleased to announce a new feature at GEDmatch: New latch Notification emails!

you are a free user, you can receive weekly email notifications f new matches that are 100 cM or greater.

you are a Tier 1 user, you can receive weekly OR daily email otifications, and can choose 30 cM, 70 cM, 100cM or 200 cM as the threshold for new match notifications.

[GED_{match}]

Upload DNA Free Tools -GEDmatch Forums Family Trees -Genealogy Comparisons / Searches 👻 Home

△ CHRISTOPHER PHILLIPS

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	A492222	*Zobiana	zobiana@hotmail.co.uk		1803	2	F			235.7	48.2 Q	2.96	0	0	Migration - F2 - A
	JQ8223445	Kate Wilson	macfearsome@gmail.com		142	2	F			51.9	12.5 Q	4.05	0	0	FTDNA
	MX9810637	Simon Eriksson	simon.frewins@gmail.com		401	2	М			49.5	25.2 Q	4.09	0	0	23andMe
	M714230	Jacqueline Gordier	Jacquigordier@hotmail.com		1744	2	F	H1c3		48.3	38.8 Q	4.11	0	0	Migration - V4 - M
	YF5534989	Margaret Stephens	stracey@traceylawfirm.com		339	2	F			48.1	10.4 Q	4.11	0	0	-
	RJ5853300	Justin Kincaid	kincaid4christ@yahoo.com		175	2	М		R-M269	43.4	13.3 Q	4.18	0	0	FTDNA
	A373001	Joe Harper	jmharper@frontiernet.net		1204	2	М	C1c	M269	38.2	26.5 Q	4.28	0	0	Migration - F2 - A

GEDmatch applies IBD chromosome segment analysis to match closest relatives

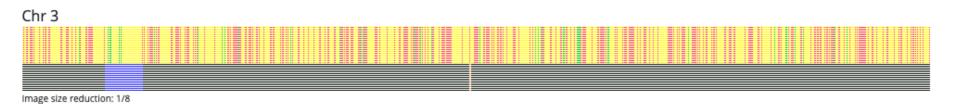


Chr	B37 Start Pos'n	B37 End Pos'n	Centimorgans (cM)	SNPs
1	182,392,412	204,081,196	19.9	772



Chr 2

Chr	B37 Start Pos'n	B37 End Pos'n	Centimorgans (cM)	SNPs
3	10,275,475	16,110,666	9	322



Chr 4

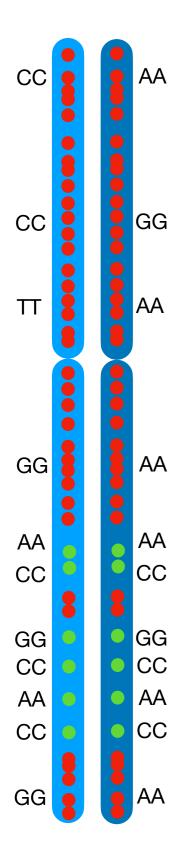
Chr	B37 Start Pos'n	B37 End Pos'n	Centimorgans (cM)	SNPs
5	10,148,758	28,028,745	20.8	636

Individual marker indications:	
Base Pairs with Full Match	
Base Pairs with Half Match	
Match with Phased data	
Base Pairs with No Match	

Validity of segments:	
Significant	
Moderate	
Low	
Insignificant	
Large gap between adjacent SNPs	
No Match	

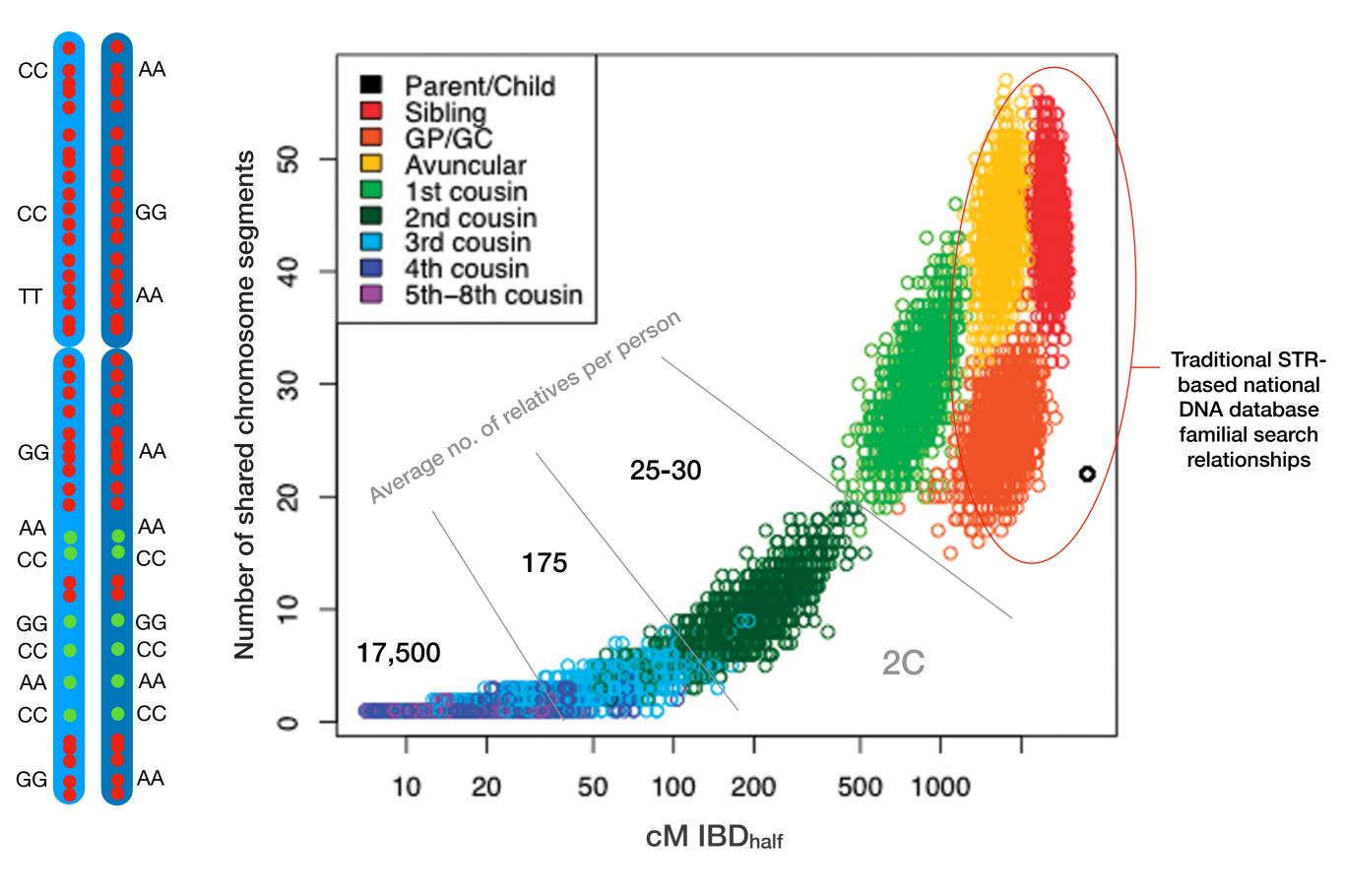
Defines IBD segments by identifying runs of homozygosity





	€	GΑ	A G	G /	A G	λÂ	G	GC	G C	Т	A C	G G	G	AC	З А	A	G A	AA	G (CC	C C	C(G G	G	G	AA	A G	С	AA	A A ⁻	Т	Forensic DNA
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IBD segments can identify 2nd to 3rd cousins with reasonably good likelihoods, less reliably beyond these relationships



The first crowdsourced genome study was the Shared cM Project



August 2017

Blaine T. Bettinger www.thegeneticgenealogist.com More about this project CC 4.0 Attribution License Interactive version by Jonny Perl at DNA Painter Last updated 7th Oct 2017 Source for this version

Filter

Enter the total number of cM for your match here:

i i

reset

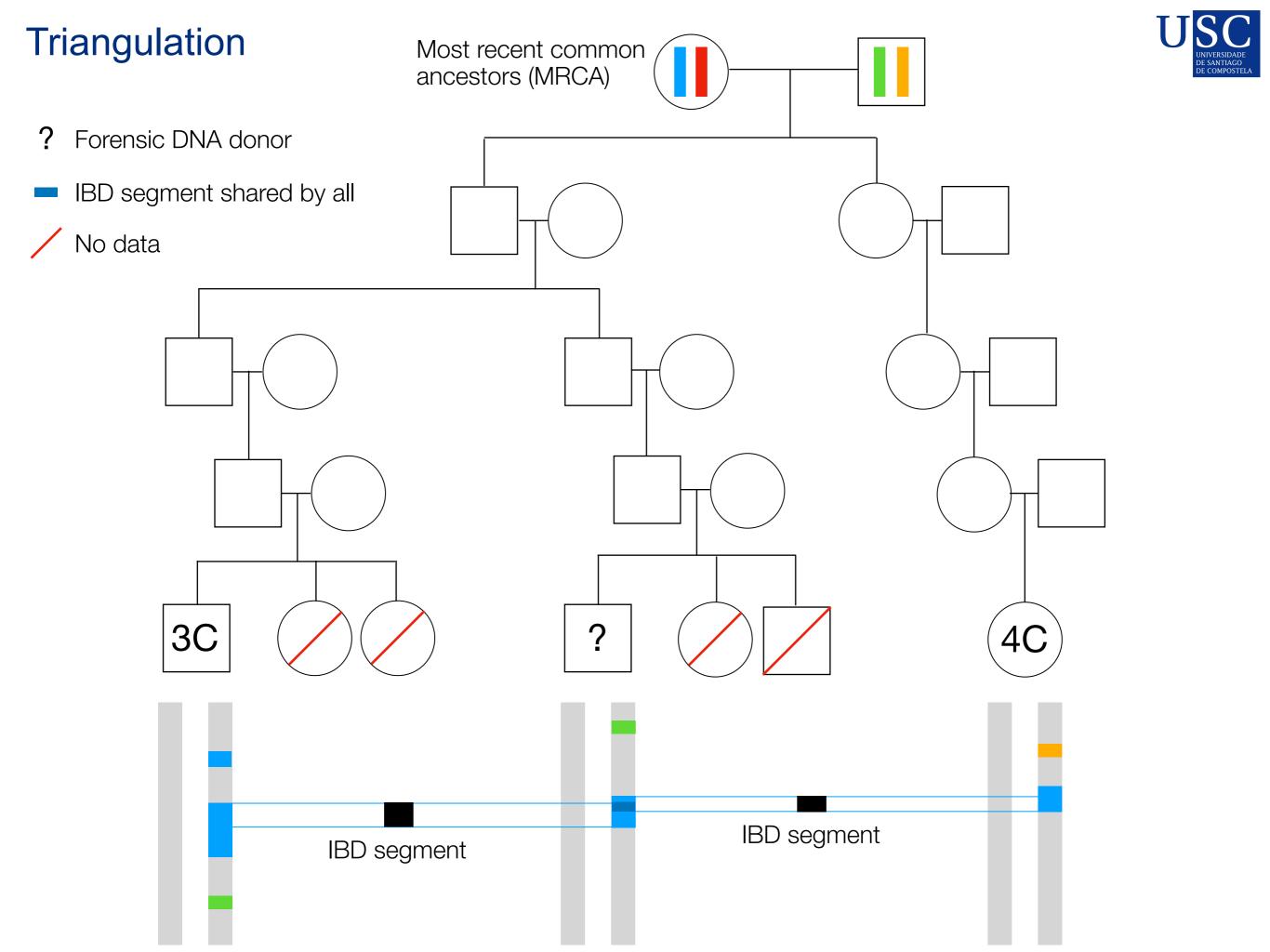
Then any relationships that fit will stand out below

Click here for a sharable link to the cM amount above

How to read this chart

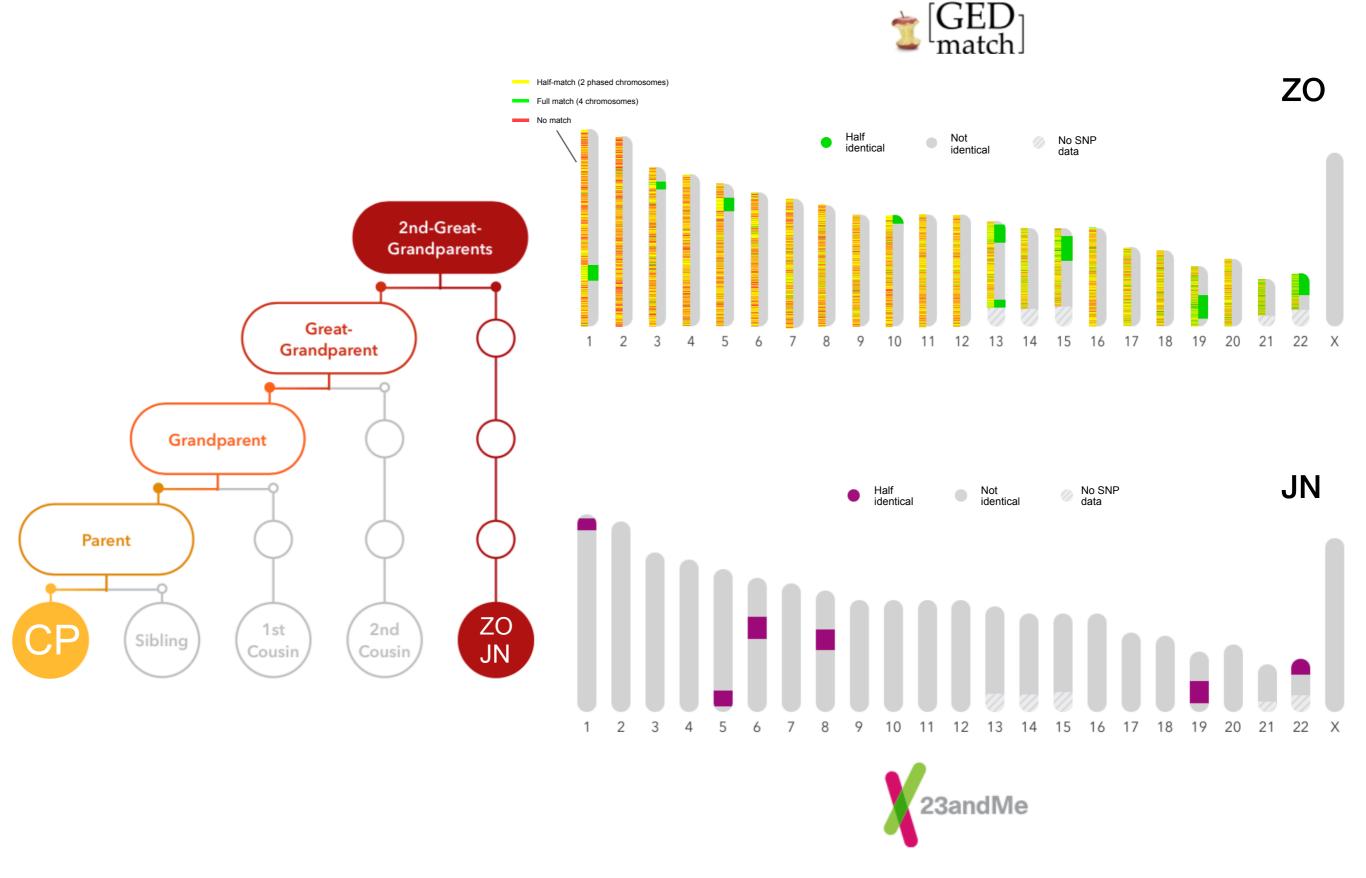
Relationship Average Range (low to high) (99th percentile)

Half GG- Aunt/Uncle 187 12 – 383			G	reat-Grandpare 881 464 – 1486	nt			Great-Great Aunt/Uncle 427 191 – 885	1C3R 123 0 – 283	2C3R 57 0 – 139	Other Relationships
Half 1C2R 145 37 – 360	Half-Great Aunt / Uncle 432 125 – 765			Grandparent 1766 1156 – 2311			Great Aunt/Uncle 914 251 – 2108	1C2R 229 43 – 531	2C2R 74 0 – 261	3C2R 35 0 – 116	6C 21 0 – 86
Half 2C1R 73 0 – 341	Half 1C1R 226 57 – 530	Half Aunt/Uncle 891 500 – 1446		Parent 3487 3330 – 3720		Aunt/Uncle 1750 1349 – 2175	1C1R 439 141 – 851	2C1R 123 0 – 316	3C1R 48 0 – 173	4C1R 28 0 – 117	6C1R 16 0 – 72
Half 3C 61 0 – 178	Half 2C 117 9 – 397	Half 1C 457 137 – 856	Half-Sibling 1783 1317 – 2312	Sibling 2629 2209 – 3384	SELF	1C 874 553 – 1225	2C 233 46 – 515	3C 74 0 – 217	4C 35 0 – 127	5C 25 0 – 94	6C2R 17 0 – 75
Half 3C1R 42 0 – 165	Half 2C1R 73 0 – 341	Half 1C1R 226 57 – 530	Half-Niece / Nephew 891 500 – 1446	Niece / Nephew 1750 1349 – 2175	Child 3487 3330 – 3720	1C1R 439 141 – 851	2C1R 123 0 – 316	3C1R 48 0 – 173	4C1R 28 0 – 117	5C1R 21 0 – 79	7C 13 0 – 57
Half 3C2R 34 0 – 96	Half 2C2R 61 0 – 353	Half 1C2R 145 37 – 360	Half Great- Niece / Nephew 432 125 – 765	Great Niece / Nephew 910 251 – 2108	Grandchild 1766 1156 – 2311	1C2R 229 43 – 531	2C2R 74 0 – 261	3C2R 35 0 – 116	4C2R 22 0 – 109	5C2R 17 0 – 43	7C1R 13 0 – 53
Half 3C3R	Half 2C3R	Half 1C3R 87 0 – 191	Half GG Niece / Nephew 187 12 – 383	Great Great Niece / Nephew 427 191 – 885	Great- Grandchild 881 464 – 1486	1C3R 123 0 – 283	2C3R 57 0 – 139	3C3R 22 0 – 69	4C3R 29 0 – 82	5C3R 11 0 – 44	8C 12 0 – 50



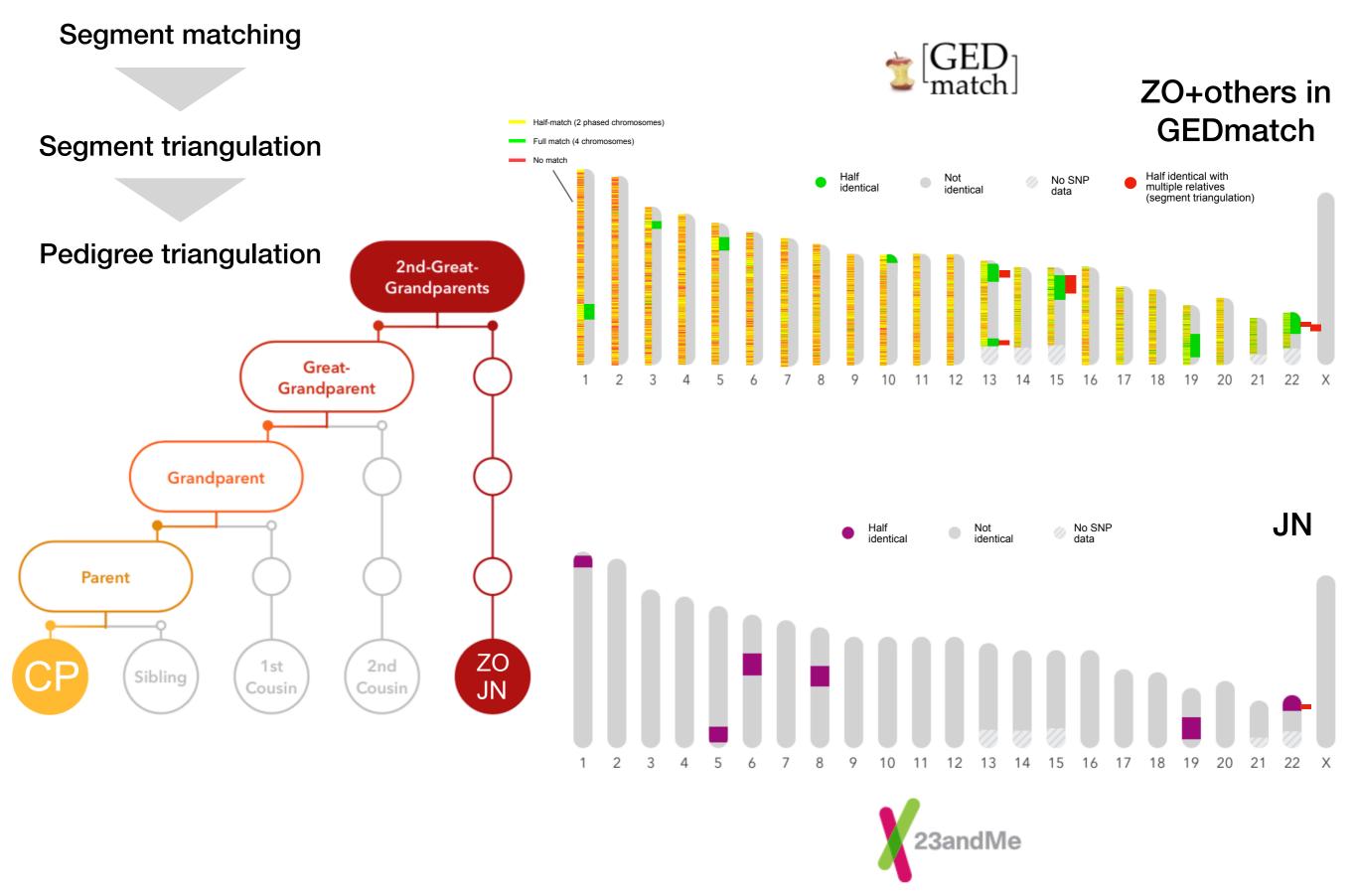
Exploring segments - two different matches in different databases can initiate triangulation studies





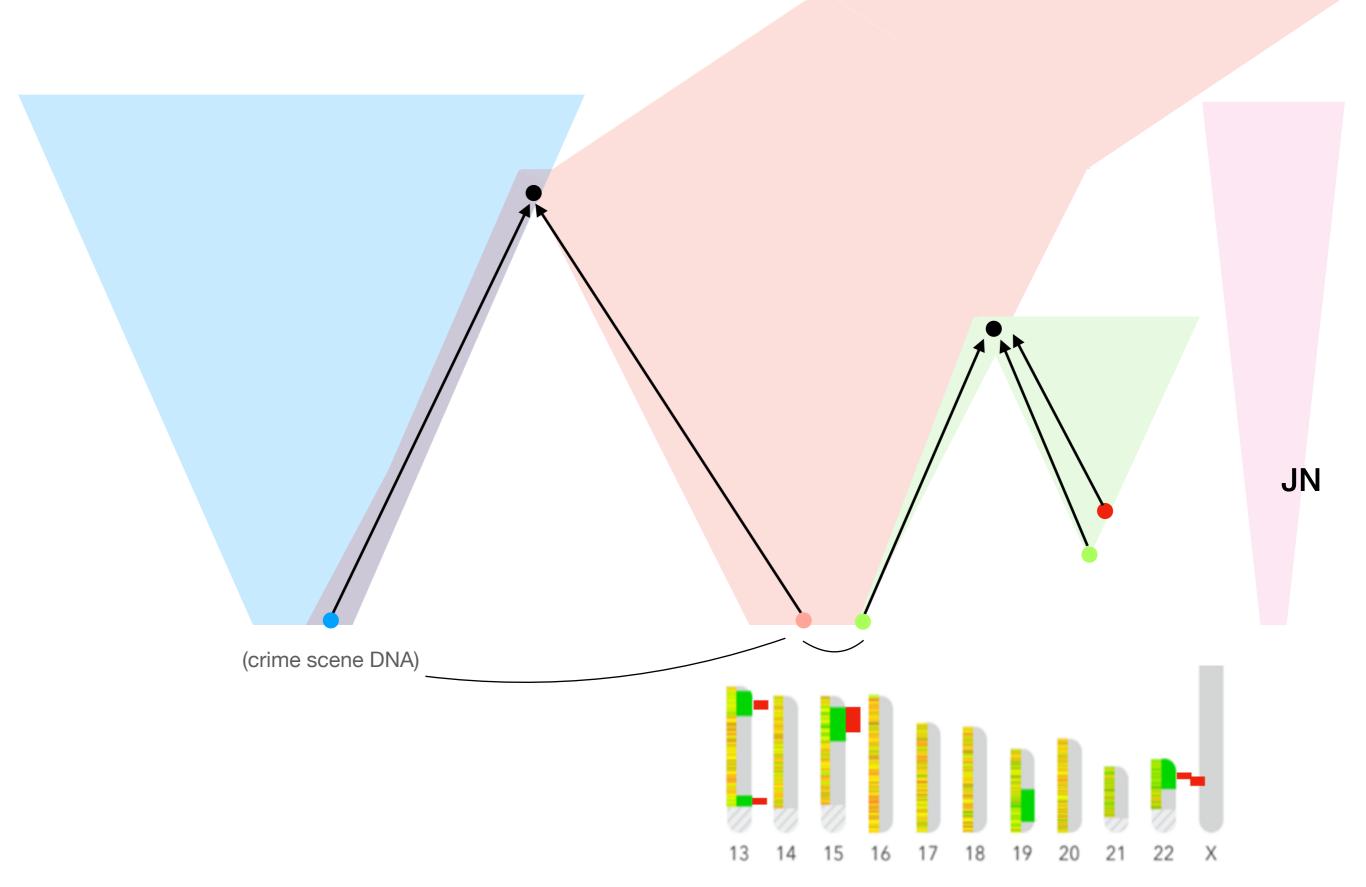
Multiple individuals with matching segments allows triangulation





Segment triangulation indicates shared ancestors amongst kinships







Forensic Science International: Genetics 46 (2020) 102233



journal homepage: www.elsevier.com/locate/fsigen

Whole-genome sequencing of human remains to enable genealogy DNA database searches – A case report

Andreas Tillmar^{a,b,*}, Peter Sjölund^c, Bo Lundqvist^d, Therese Klippmark^a, Cajsa Älgenäs^a, Henrik Green^{a,e}

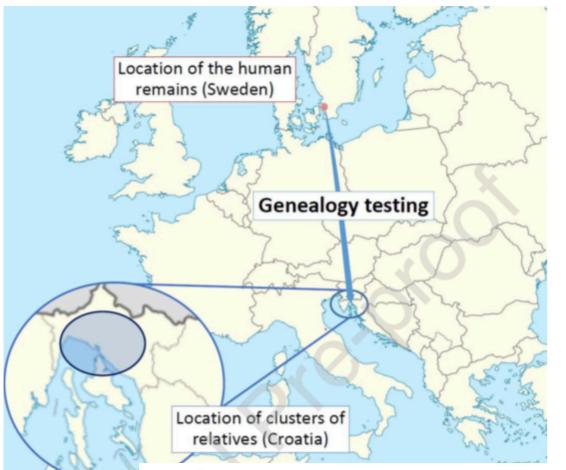


Table 1. Summary of the quality parameters and criteria used for the genotype calling

Parameter	Description	Threshold for genotype calling
Coverage	Number of individual reads that has a certain nucleotide at a given position.	≥10 (homozygote) ≥5 per allele (heterozygote)
Allelic balance*	[coverage for the most common nucleotide]/ [coverage for the most common nucleotide + coverage for the second most common nucleotide].	=1 (homozygote) 0.5≤x≤0.7 (heterozygote)
Q score	Mean quality score (nucleotide Q).	>25
Forward/Reverse- ratio	Forward and reverse read ratio for the nucleotide(s) comprising the genotype.	>0.2





- 3 x 3 ng DNA from a femur of murder victim generated 3 libraries for HiSeq^x
- Fragments of av. 400 bp size gave av. 150 bp read lengths as basis for 1,378,481 SNPs for an optimum GEDmatch intersect
- DNA quality and SNP call reliability were checked with STRs and the Qiagen 140-SNP ID panel respectively
 - SNPs using Table 1 thresholds
 - GEDmatch search made before terms and conditions changed
 - A cluster of relatives was found within a 40km radius in NW Croatia - likely from a single pair of victim's grandparents



Contents lists available at ScienceDirect

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Research paper



Getting the conclusive lead with investigative genetic genealogy – A successful case study of a 16 year old double murder in Sweden

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Table 2

Summary of the established dataset and database searches.

Sample/ library preparation	Dataset	Database	Number of genotypes ^a (approx.)	Total shared segment length for top hits
DNA library 1.1	WGS analysis	GEDmatch	155,000-269,000	Less than 30 cM
DNA library 1.1	WGS analysis and genotype imputation	GEDmatch	864,000–1026,000	Less than 30 cM
DNA library 1.1 + DNA library 1.2	WGS analysis and genotype imputation	GEDmatch	908,000–1050,000	Less than 30 cM
DNA library 2.1	WGS analysis	GEDmatch	1279,000	Less than 30 cM
	÷	FTDNA	1861,000	~350 cM, ~100 cM, ~60 cM and decreasing

Cases in National Forensic Labs now being reported



- Mother and child stabbed to death in Linköping in 2004. Mass screen of 6000 men to try to match DNA left at scene
- 3 separate whole-genome-sequence runs from 20 ng pooled DNA with 150 bp paired-end sequencing (NovaSeq 6000). Targeted 1,378,481 SNPs
- SNP genotypes were increased with imputation and a 1.3 million and 1.9 million dataset was applied to GEDmatch and FT DNA

Table 1

Summary statistics from the WGS runs.

Parameter	WGS run 1 (DNA library 1.1)	WGS run 2 (DNA library 1.2)	WGS run 3 (DNA library 2.1)
Average Coverage	13X	10X	60X
Duplication rate	~ 70%	~ 60%	< 10%
Median insert size	~ 60 bp	~ 60 bp	~ 180 bp

Investigators may need to extend to target testing to fill pedigree





 DNA was collected Results indicate shared DNA Crime-scene from the crime scene with two second cousins. DNA run 30 years ago. through GEDmatch data bank. Maternal great grandparents A genealogist reconstructed the family trees of the two cousins. Pedigree triangulation Paternal The reconstructed family trees lead grandmother to a suspect through common ancestors. 6. Detectives collect DNA from a discarded cup. It matches DNA from the crime scene. Cousin Target testing has been used in US with surreptitious sample collection major privacy infringement issues Suspect Cousin

Concluding remarks



The application of IGG is most actively pursued in the US, but has been successfully used elsewhere - notably Sweden

	Country	Percentage
1	United States	65%
2	United Kingdom	9%
3	Canada	6%
4	Australia	4%
5	France	2%
6	Germany	1%
7	Sweden	1%
8	Ireland	1%
9	New Zealand	1%
10	Netherlands	1%

Uploads to GEDmatch

Concluding remarks



The application of IGG is most actively pursued in the US, but has been successfully used elsewhere - notably Sweden

Ethical considerations about individual privacy have been ignored in some cases and this has caused concern

Should we be making use of genetic genealogy to assist in solving crime?

A report on the feasibility of such methods in the UK

September 2020

UK and Sweden currently consider ethical/ privacy concerns outweigh operational benefits of IGG and have not advocated its use - but such policies also relate to the success of STR-based familial searching in the UK compared to the US

Concluding remarks



The application of IGG is most actively pursued in the US, but has been successfully used elsewhere - notably Sweden



Ethical considerations about individual privacy have been ignored in some cases and this has caused concern



A move towards much larger MPS SNP multiplexes will open up IGG to more labs - but extending familial search to 2nd-3rd cousin - e.g. Kintelligence (10,230 SNPs) + GEDmatch Pro



VEROGEN

ForenSeq Kintelligence Kit

The only forensic genetic genealogy assay that targets the relevant markers in degraded and low-input DNA samples



Thank you



Genetisti Forensi Italiani

CONGRESSO NAZIONALE

LA GENETICA FORENSE: QUALE FUTURO?

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Forensic Science International: Genetics

journal homepage: www.elsevier.com/locate/fsigen

Review article

Investigative genetic genealogy: Current methods, knowledge and practice



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