#### Western Kentucky University

#### **TopSCHOLAR®**

**DLPS Faculty Publications** 

Library Public Services

9-18-2014

#### Genetic Genealogy: What Every Librarian Should Know

Rosemary L. Meszaros

Katherine Pennavaria

Follow this and additional works at: https://digitalcommons.wku.edu/dlps\_fac\_pub

Part of the Genetics Commons, Higher Education Commons, and the Library and Information Science

#### Commons

This Presentation is brought to you for free and open access by TopSCHOLAR®. It has been accepted for inclusion in DLPS Faculty Publications by an authorized administrator of TopSCHOLAR®. For more information, please contact topscholar@wku.edu.

#### Western Kentucky University

From the SelectedWorks of Rosemary L. Meszaros

September 18, 2014

## Genetic Genealogy: What Every Librarian Should Know

Katherine A. Pennavaria Rosemary L. Meszaros



# GENETIC GENEALOGY: WHAT LIBRARIANS NEED TO KNOW ABOUT DNA TESTING

**Rosemary Meszaros** 

**Government Documents & Law** 

**Katherine Pennavaria** 

**Visual & Performing Arts Library** 

**WKU Libraries** 

September 18, 2014



## Henrietta Lacks

## "HeLa" cell line

One tissue sample  $\rightarrow$  immortality (sort of)

### **DNA IS HERE TO STAY**



### **MEDICAL RESEARCH**



## PATERNITY QUESTIONS



### **CRIME SCENES**



## **GENEALOGY**

#### FIND OUT:

- 1. your ethnicity
- 2. your relatives in the database
- 3. your haplogroup
- 4. your percentage of Neanderthal DNA

#### **FACTS**



#### **DNA Test Report**

For Personal Knowledge Only

DDC8888810008

RN: 76206

DDC is accredited/certified by AABB, CAP, ACLASS for ISO/IEC 17025, CLIA, NYSDOH & ASCLD/LAB-International.

Case: 456789 Name		MOTHER Jane 1/1/2010 456789-10		CHILD Jenny 1/1/2010 456789-20		Alleged FATHER John 1/1/2010 456789-30		
								Locus
D8S1179 D21S11	1.55	10 27	14	13 29 8	(14) (30) (10)	11 29	13 30	
D7S820	1.17		10		10	10	11	
CSF1PO	1.65	8	12	111	100	8	12	
D3S1358	1.88	14	17	15	17	14	15	
TH01	2.62	6	9.3	7	9.3	7	9	
D13S317	3.43	13	0.0	11	13	11	3	
D16S539	3.32	9	12	11	12	11		
D2S1338	4.33	19	20	20	24	23	24	
D19S433	2.23	13		13		13	14	
vWA	3.62	14		14	17	17		
TPOX	1.86	11		8	11	8		
D18S51	3.06	15	17	14	17	13	14	
D5S818	1.35	12	13	11	12	11	12	
FGA	3.55	21	22	21	24	21	24	
Amelogenin		X	7-3	X		X	Y	

Interpretation:

Combined Paternity Index: 323,769

Probability of Paternity: 99.9996%

The alleged father is not excluded as the biological father of the tested child. Based on testing results obtained from analyses of the DNA loci listed, the probability of paternity is 99.9996%. This probability of paternity is calculated by comparing to an untested, unrelated, random individual of the Caucasian population (assumes prior probability equals 0.50).

This DNA test was performed using the DDC DualProcess<sup>®</sup> utilizing the AmpFISTR<sup>®</sup> Identifiler PCR Amplification and an ABI 3730 DNA analyzer. DDC PlatinumPlex<sup>®</sup> used as applicable.

Note: Since the samples were not collected under a strict chain of custody by a neutral party and the Laboratory cannot verify the origin of the samples, this test result may not be defensible in a court of law for the establishment of patemity and other legally related issues. The tested parties' names that may appear on this report have been provided by the client and cannot be verified. The laboratory assumes no responsibility for incorrect or misspelled client information. Based on the samples received from tested parties whose identities cannot be independently verified. I, the undersigned Laboratory Director, declare that the genetic data is correct as reported on 1/19/2010.

Signature

Michael L. Baird, Ph.D. Keen A. Wilson, Ph.D. L. Farris Hanna, Ph.D. Thomas M. Reid, Ph.D. Pierig Lepont, Ph.D. Marco Scarpetta, Ph.D.

## DNA sequencing is a complicated

science.

The raw results require

interpretation.

ATGACCAACAAGTGTCTCCTCCAAATTGCTCTCTGTTGTGCTTCTCCACTACAG
CTCTTTCCATGAGCTACAACTTGCTTGGATTCCTACAAAGAAGCAGCAATTTCA
GTGTCAGAAGCTCCTGTGGCAATTGAATGGGAGGCTTGAATACTGCCTCAAGCAC
AGGATGAACTTTGACATCCCTGAGGAGATTAAGCAGCTGCAGCAGTTCCAGAAGG
AGGACGCCGCATTGACCATCTATGAGATGCTCCAGAACATCTTTGCTATTTTCAG
ACAAGATTCATCTAGCACTGGCTGGAATGAGACATCTTGTTGAGAACCTCCTGGCT
AATGTCTATCATCAGAATAAACCATCTGAAGACAGTCCTGGAAGAAAAACTGGAGA
AAGAAGATTTCACCAGGGGAAAACTCATGAGCAGTCTGCACCTGAAAAGATATTA
TGGGAGGATTCTGCATTACCTGAAGGCCAAGGAGTACAGTCACTGTGCCTGGACC
ATAGTCAGAGTGGAAATCCTAAGGAACTTTTACTTCATTAACAGACTTACAGGTT
ACCTCCGAAAC, and

ATGAGCTACAACTTGCTTGGATTCCTACAAAGAAGCAGCAATTTTCAGTGTCAGA
AGCTCCTGTGGCAATTGAATGGGAGGCTTGAATACTGCCTCAAGCACAGGATGAA
CTTTGACATCCCTGAGGAGATTAAGCAGCTGCAGCAGTTCCAGAAGGAGGACGCC
GCATTGACCATCTATGAGATGCTCCAGAACATCTTTGCTATTTTCAGACAAGATT
CATCTAGCACTGGCTGGAATGAGACTATTGTTGAGAACCTCCTGGCTAATGTCTA
TCATCAGATAAACCATCTGAAGACAGTCCTGGAAGAAAACTGGAGAAAAGAT
TTCACCAGGGGAAAACTCATGAGCAGTCTGCACCTGAAAAGATATTATGGGAGGA
TTCTGCATTACCTGAAGGCCAAGGAGTACAGTCACTGTGCCTGGACCATAGTCAG
AGTGGAAATCCTAAGGAACTTTTACTTCATTAACAGACTTACAGGTTACCTCCGA
AAC.

#### **FACTS**

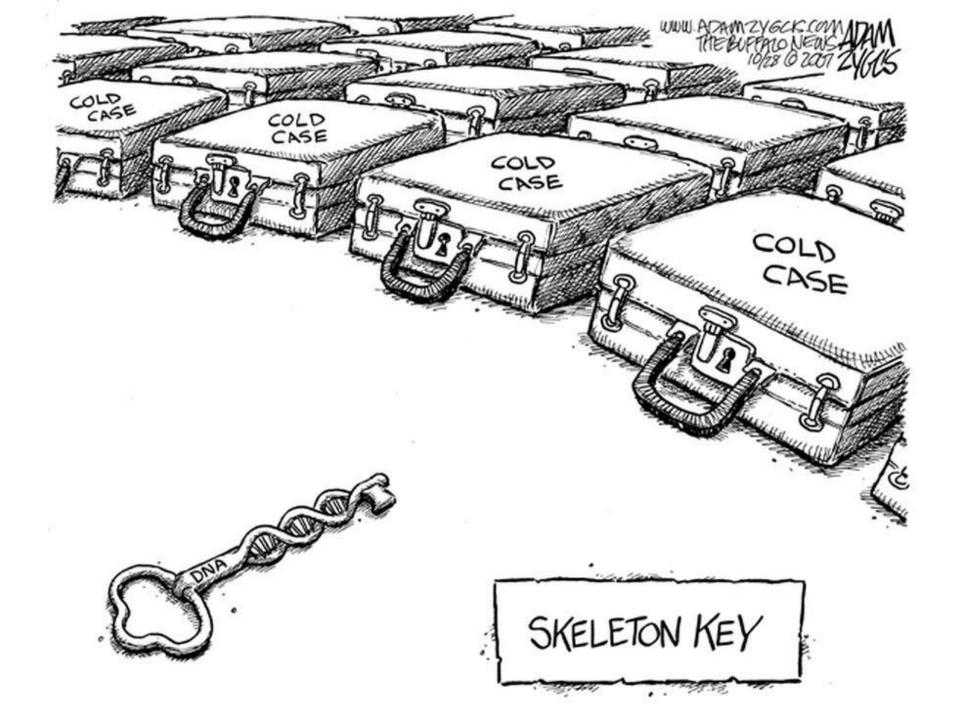


It cannot be used to recreate scary dinosaurs. (so far)

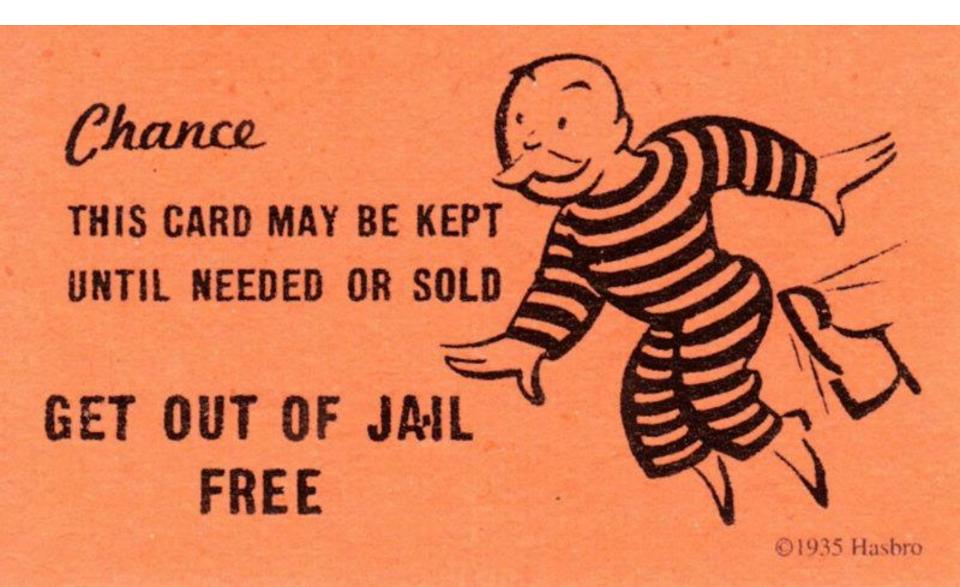
## WHATDNA SEQUENCING HAS GIVENUS



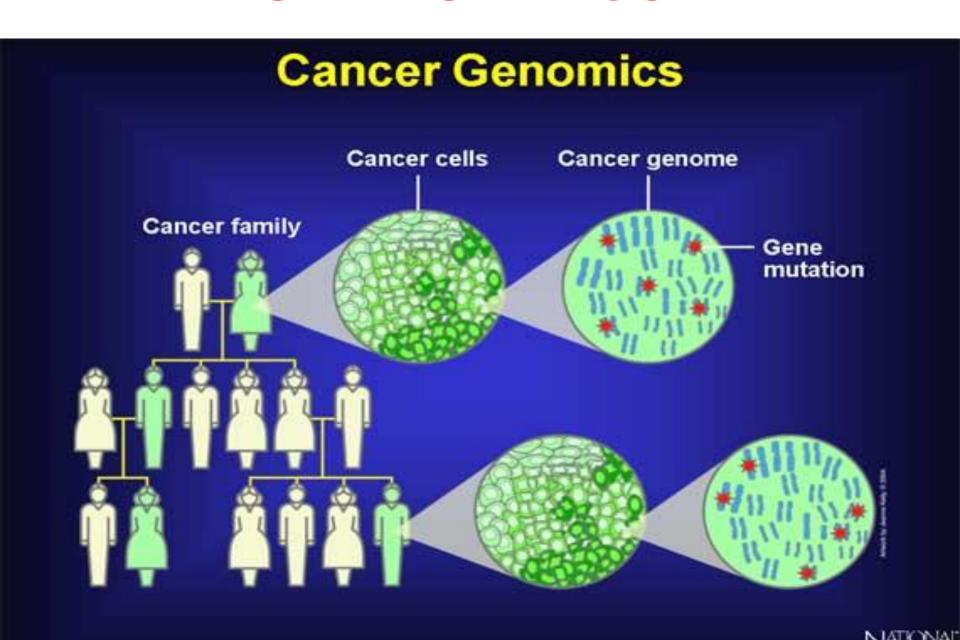
## BAD GUYS CAUGHT



#### **INNOCENT PRISONERS RELEASED**



## **HOPE FOR A CURE**



## ADVERTISING

Family Tree DNA - Genetic Testing for Ancestry, Family ...
https://www.familytreedna.com/ ▼ Family Tree DNA ▼

Establishing **DNA** fingerprints for the purpose of determining ancestral pedigrees.

Testing is available to both amateur and professional genealogists.

Login - Y-DNA - Family Finder - DNA Tests

List of DNA testing companies - ISOGG Wiki

www.isogg.org/wiki/List\_of\_DNA\_testing\_companies ▼
May 15, 2014 - This is a list of DNA testing companies offering DNA tests for genealogy, deep ancestry and ethnicity. This list is provided for information only.

Ancestry **DNA Tests** for **Genealogists** - Information genealogy.about.com > ... > Genealogy > Genetics, DNA & Health History ▼

Easy to understand guide to **DNA** and **genealogy** includes an introduction to the differences between Y-line and mtDNA **testing**, a guide to home **DNA** genetics ...

23andMe - Genetic kit for ancestry | DNA Service

https://www.23andme.com/ ▼ 23andMe ▼

23andMe is a saliva-based DNA service. ... Contact your DNA relatives across continents or across the street; Build your **family tree** ... 99. 23andMe **DNA Test** Kit ...

How to Use **DNA Testing** for **Genealogy** Research - People people.howstuffworks.com/.../genealogy/use-dna-testing-genealogy-rese... ▼

Using **DNA** for **genealogy** research is becoming more commonplace. Visit

Discover Your Ancestry with **DNA**.

Find Ethnic and Geographic Origins.

Genographic Project Kit
www.nationalgeographic.com/ ▼
Take the Revolutionary DNA Test

Take the Revolutionary **DNA Test** By Nat Geo & Discover Your Ancestry!

Free DNA Testing Kit
www.dnatesting.net/Free-DNA-Testing-Kit

(866) 633-9127

Fast, Accurate DNA Testing Results.

Free, No-Obligation **Test** Kit Today.

\$79 for Complete DNA Test www.dnafamilycheck.com/ ▼
Private and Legal Testing Options.

Results in 3 to 5 days

Best DNA Ancestry Test
www.about.com/Best+DNA+Ancestry+Test

Search for Best **DNA** Ancestry **Test**.

Find Expert Advice on About.com.

Free **DNA Test** Ancestry

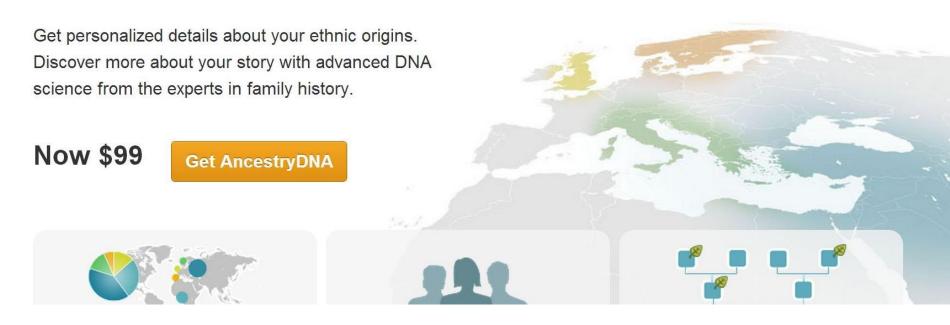
Additional 5129 Laboratory Fee Required lest henred **IDENTIGENE®** PATERNITY TEST COLLECTION KIT Additional \$129 For Alleged Father, Mother and Child

Results Typically Reported in 3 Days

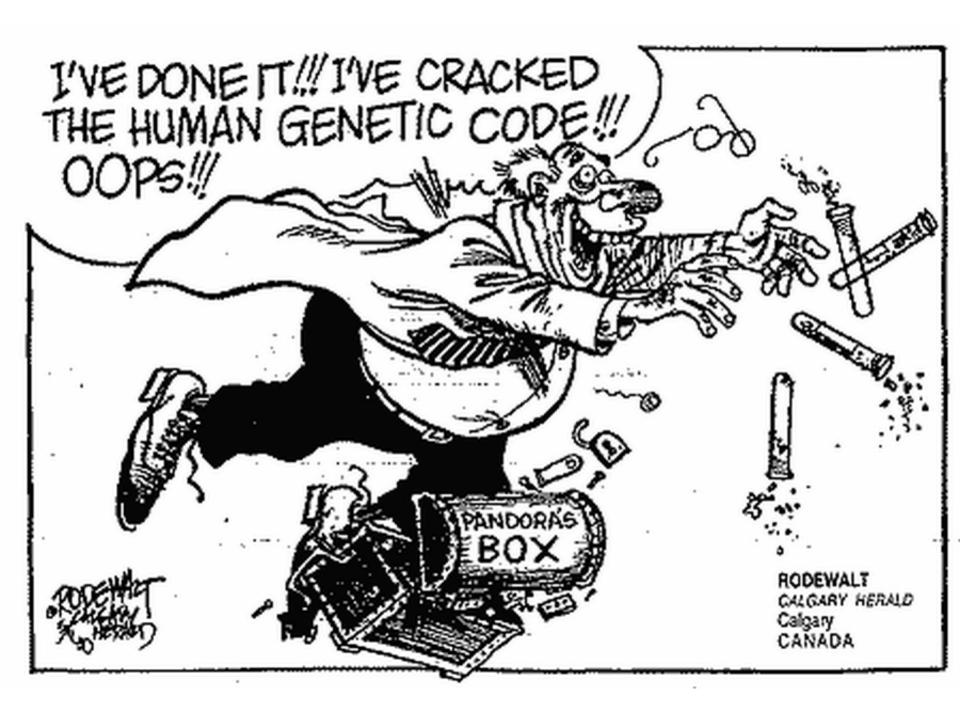




#### Family history is in our DNA. What's in yours?



## CONCERNS





"I'm afraid we found annoying personality markers in your DNA.

I can't hire you."

#### IS PRIVACY COMPROMISED?

#### YES & NO

✓ WHERE IS DNA DATA STORED?

**✓** RESTRICTIONS FOR USE?

✓ DO WE CONTROL USE OF STORED DATA?

#### FUZZY DATA

FBI: CODIS (COMBINED DNA INDEX SYSTEM)

STATE: NUMEROUS METHODS OF STORING OR DESTROYING DATA

HOSPITAL: NEWBORN DNA SAMPLES TAKEN WITHOUT PARENTAL CONSENT (NBS)

NIH: REMOVED THEIR DNA DATA FROM PUBLIC ACCESS ON WEB

#### KINSHIP SEARCHING

#### **JOYS & SORROWS OF KINFOLK:**

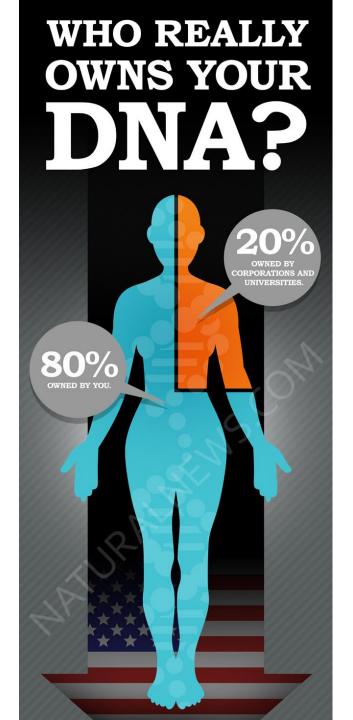
MULTIPLE COUSINS OF WHATEVER DEGREE

FIND FAMILY & CONNECTEDNESS

FAMILY TREE "BRANCHES"

But: be careful what you wish for...





# THREE TESTS ON ONE SAMPLE

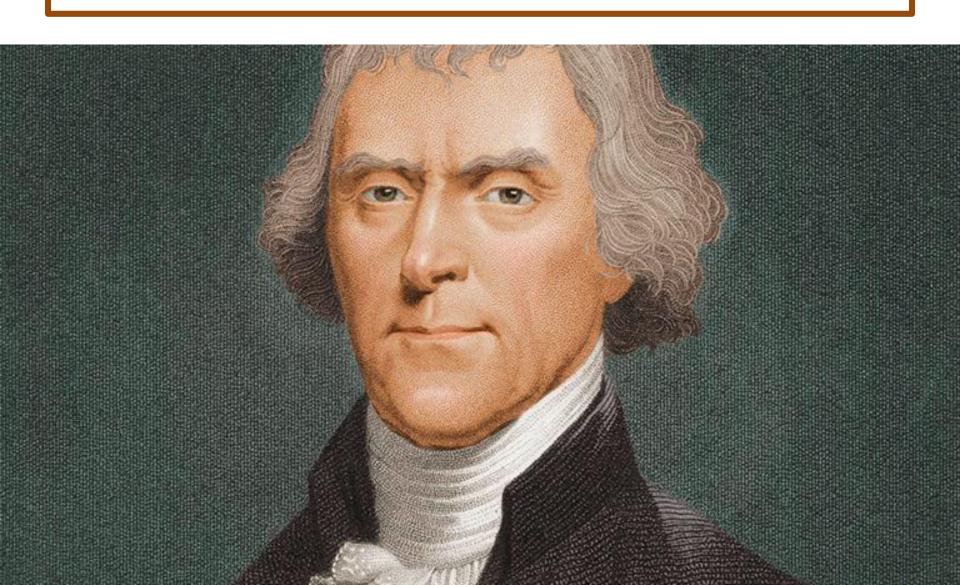
# DNA = deoxyribonucleic acid

## Y-DNA

mtDNA

atDNA

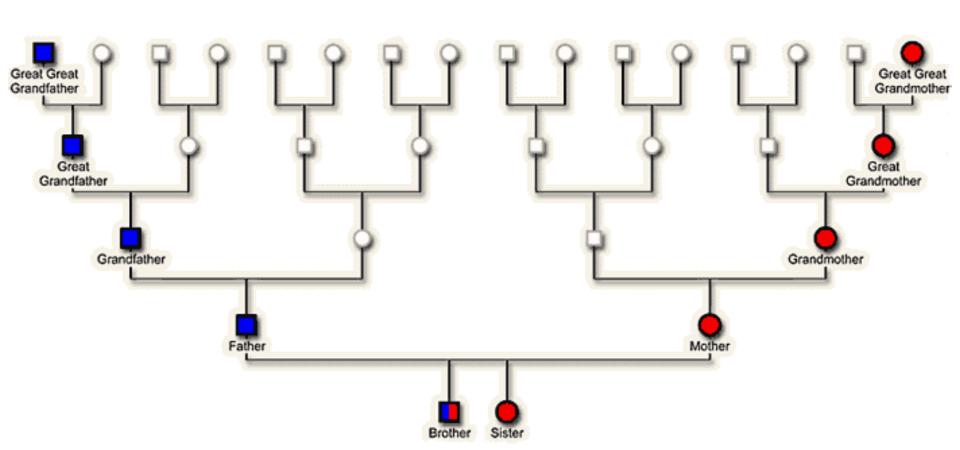
## Y chromosome (Y-DNA)

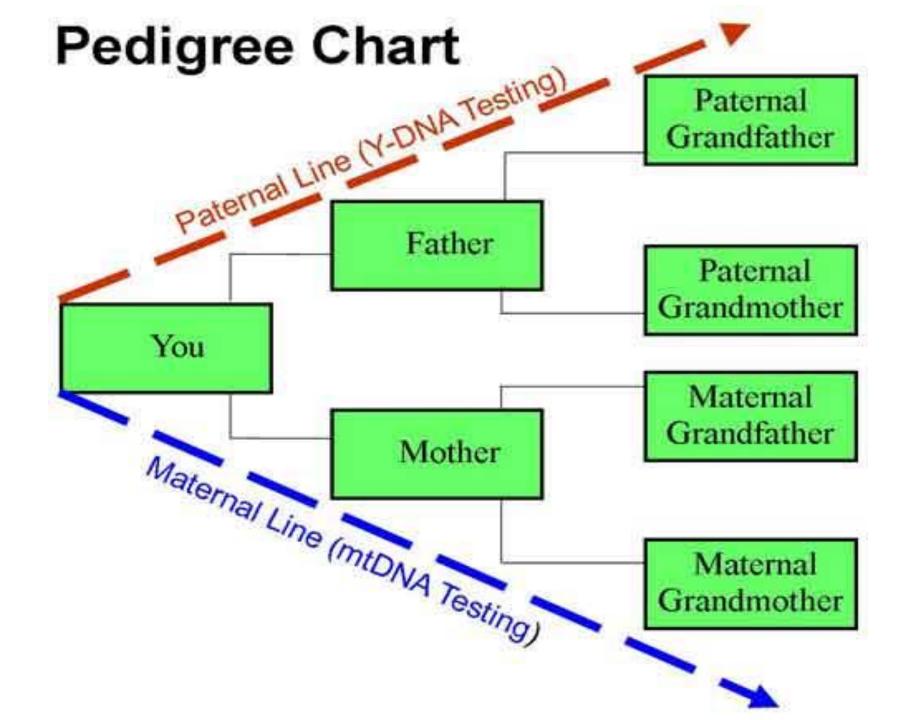


## Mitochondrial DNA (mtDNA)



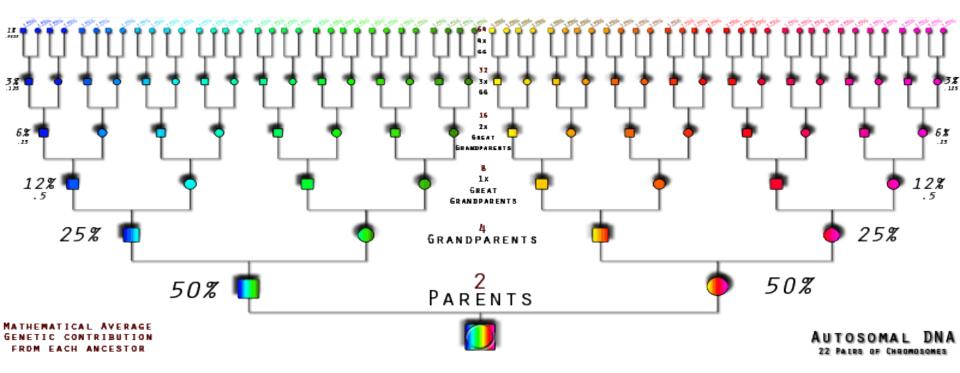
## TRACKING THE PATERNAL MATERNAL AND LINES





## Autosomal DNA (atDNA)

Our Autosomal DNA is (theoretically) representative of our whole ancestry.



## YOUR PART



## BYS (before you spit)





Not as easy as it looks.



## EXEMPT HUMAN SPECIMEN

# THEN:



# WHAT YOU FIND OUT

## Kind of weird to think about.

Sent: Thursday, August 28, 2014 4:02 AM

To: Pennavaria, Katherine

## Your sample has reached the lab.

We've received the saliva sample registered to:

Katherine Pennavaria - 19401401035494

Our technicians are inspecting your sample and preparing it for analysis. You'll receive an email when your results are ready in approximately 3-4 weeks.

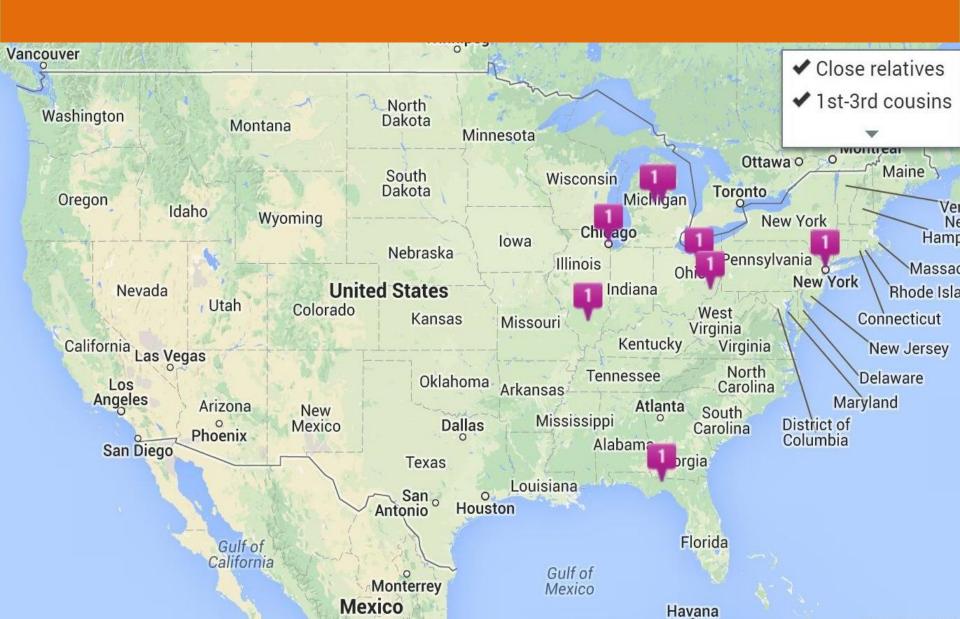
99.3%	European			
	Southern European			
21.0%	Balkan			
9.5%	Italian			
0.6%	Iberian			
0.0%	Sardinian			
18.4%	Broadly Southern European			
	Northern European			
11.6%	French & German			
7.5%	British & Irish			
1.3%	Scandinavian			
0.0%	Finnish			
7.7%	Broadly Northern European			
5.2%	Eastern European     Ashkenazi     Broadly European			
0.0%				
16.4%				
0.7%	East Asian & Native American			
	East Asian			
0.2%	Yakut			
0.0%	Japanese			
0.0%	Korean			
0.0%	Mongolian			
0.0%	Chinese			
0.4%	Broadly East Asian			
0.1%	Native American			
0.0%	Southeast Asian			
< 0.1%	Broadly East Asian & Native American			

# Yourself in numbers

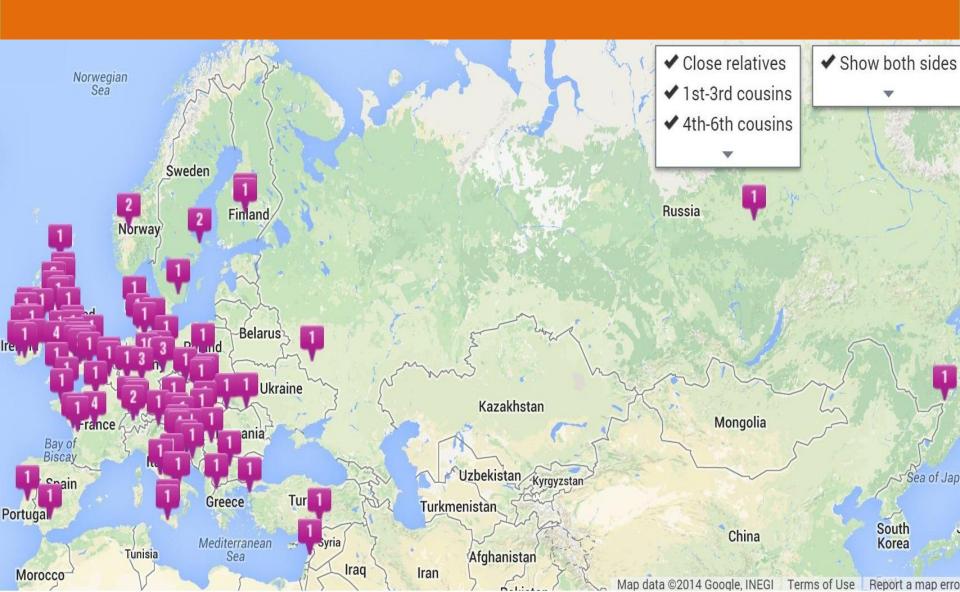
## **CONNECTIONS**

Q searc	h matches Show:	both sides - Sort: re	elationship 25 per page 7	((   1 - 25 of 991
2	Katherine Pennavaria Female	You	Chicago, IL Pennavaria Nasca LaCerra 12 more	UPDATE YOUR PROFILE
1	eric braun Male, b. 1969	2nd Cousin 5.11% shared, 13 segments	V E1b1a8a	Public Match Send a Message
1	Marsha McVey Female	2nd to 4th Cousin 1.48% shared, 4 segments	United States Mt. Pleasant, MI Wooster, OH  Lewisville, OH 3 more Northern Europe McVey  Ruble Bartenslag Stallsmith V	Public Match Send a Message
1	Peggy LaCerra Female, b. 1954	3rd to 4th Cousin 0.69% shared, 3 segments	United States Northern Europe H1a	Public Match Send a Message
1	Female	3rd to 5th Cousin 0.48% shared, 3 segments	U5b1	Send an Introduction
1	Male	3rd to 5th Cousin 0.47% shared, 3 segments	X2b R1b1b2a1a2f*	Send an Introduction
1	Isaac Larson Male	3rd to 5th Cousin 0.39% shared, 3 segments	United States Los Angeles, Monterey, Chicag  Eastern Europe Bower Larson Cohen K1a1b1a I1*	Public Match Send a Message View Family Tree

## FIRST THROUGH THIRD COUSINS



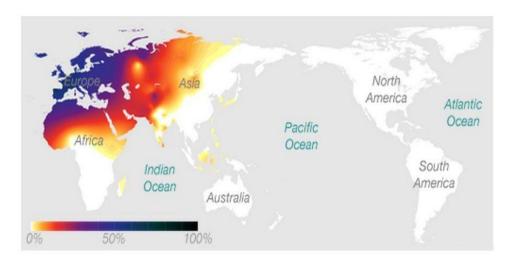
## **FOURTH THROUGH SIXTH COUSINS**



### **HAPLOGROUPS**

#### HV4a is a subgroup of RO

Locations of haplogroup R0 circa 500 years ago, before the era of intercontinental travel.



R0 originated more than 35,000 years ago in the Near East, at a time when humans were moving outward from the region into other parts of Asia and Europe. The haplogroup spawned several daughter branches that were central in those migrations, notably haplogroups H and V, both of which are most commonly found in Europe.

Maternal haplogroups are families of mitochondrial DNA types that all trace back to a single mutation at a specific place and time. By looking at the geographic distribution of mtDNA types, we learn how our ancient female ancestors migrated throughout the world.

Haplogroup: R0, a subgroup of R

Age: 35,000 years

Region: Near East, northern Africa, Western Eurasia

Example Populations: Saudi Arabs, Yemeni Jews,

Bedouin

**Highlight:** R0 includes HV and its subgroups, H, V, and a scattering of rare lineages across the Near East and Europe.

#### Haplogroups of You and Your Connections

HV4a Katherine Pennavaria

Haplogroups of Example Profiles

## Who knew?

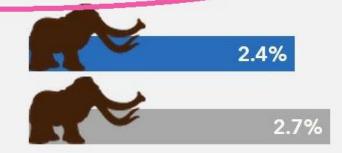
This lab estimates your genome-wide percentage of Neanderthal ancestry

#### Got Neanderthal DNA?

An estimated 2.4% of your DNA is from Neanderthals.

Katherine Pennavaria (you)

Average 23andMe user



14th percentile

For better comparison with your ethnic group, please complete your profile.

#### **MODERN HUMANS**

Higher brow Narrower shoulders Slightly taller



#### **NEANDERTHALS**

Heavy eyebrow ridge
Long, low, bigger skull
Prominent nose with developed

## In case you need to share that.

#### Neanderthal Shirts Gifts



38 results



2.8% Neanderthal Shirt \$25.95



3.1% Neanderthal Shirt \$25.95



3% Neanderthal Shirt \$25.95

### **SURVEYS**

Click on a survey to get started or to view your results



Where Are You From?

START SURVEY



**Female Fertility** 

About this survey | Published: November 2011

START SURVEY



Urinary Incontinence and Pelvic Floor Disorders

About this survey | Published: November 2011

START SURVEY



Skin Aging

About this survey | Published: September 2010

START SURVEY



**Antidepressant Efficacy and Side Effects** 

START SURVEY

## **RESEARCH QUESTIONS**

Have you ever sprained your ankle?

Which best describes your employment status?

Compared to your friends, how much do you like sweets or other sugary food?

Do you cry easily?

Have you ever had allergic rhinitis (stuffed or dripping nose caused by allergies)?

Have you ever been diagnosed with mania (been told by a medical professional that you were having a manic episode)?

Do you have an os trigonum (extra bone behind your ankle bone)?

What best describes your ancestry/ethnicity? Please check all that apply.

How many times have you been told by a doctor that you had strep throat?

Have any of your grandparents, parents, brothers, sisters, aunts, or uncles ever been

## HOW TO DECIDE

## ISOGG.ORG





← → C www.isogg.org



HOME

RESOURCES

**FOR NEWBIES** 

FOR ADMINS

FAMOUS DNA

YSNP TREE

SUCCESS STORIES

SPEAKERS LIST

MEETINGS/EVENTS

#### INTERNATIONAL SOCIETY OF GENETIC GENEALOGY

The first society founded to promote the use of DNA testing in genealogy

JOIN ISOGG

OUR MISSION | ABOUT ISOGG | Yahoo! FORUM

ISOGG PRESS

ISOGG and Southern California Genealogical Society co-sponsor

"Family History and DNA: Genetic Genealogy in 2014

Thursday, June 5, 2014 at the Los Angeles Marriott Burbank Hotel

To keep up with the latest news on both Jamboree and DNA Day see the Jamboree blog.

2014 International Genetic Genealogy Conference

Organized by the Institute for Genetic Genealogy

August 15-17, 2014 in Washington, DC at the National 4-H Youth Conference Center

See www.i4qq.orq for details.

#### Welcome to ISOGG!

Our mission also includes educating others about the use of genetics in genealogy. This can be accomplished through workshops, this website, speaker's bureau, forums, and meetings.

Meetings are important, as they allow for the dissemination of information and to maintain the momentum. While we are geographically spread out, we will do what is possible to hold meetings.

#### WHAT'S NEW?

ISOGG Sitemap

DNA Consultants/Services

Newsletter

Wiki

# HEALTH DATA NO LONGER AVAILABLE

# Status of our health-related genetic reports.

We no longer offer our health-related genetic reports to new customers to comply with the U.S. Food and Drug Administration's directive to discontinue new consumer access during our regulatory review process

At this time, we do not know the timeline as to which health reports might be available in the future or when they might be available.

## **QUESTIONS TO ASK**

What is the size of their DNA database?

Does the company allow downloading of results?

How much does it cost?

Which results are you most interested in?

### **GENEALOGY**



A Practical Guide for Librarians

KATHERINE PENNAVARIA

PRACTICAL GUIDES FOR LIBRARIANS, NO. 13

## **THANK YOU!**