# Don't Forget mtDNA

FamilyTreeAZ.com

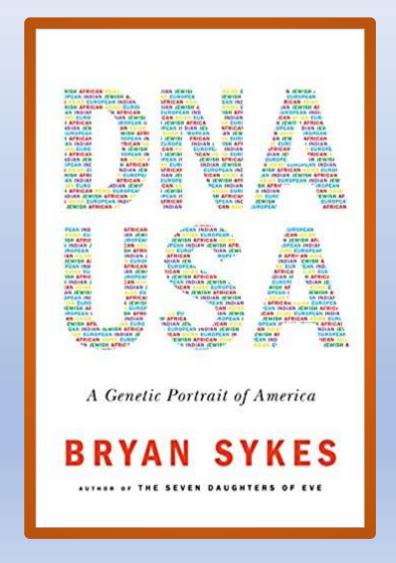


Ken Waters

Jan 7, 2021



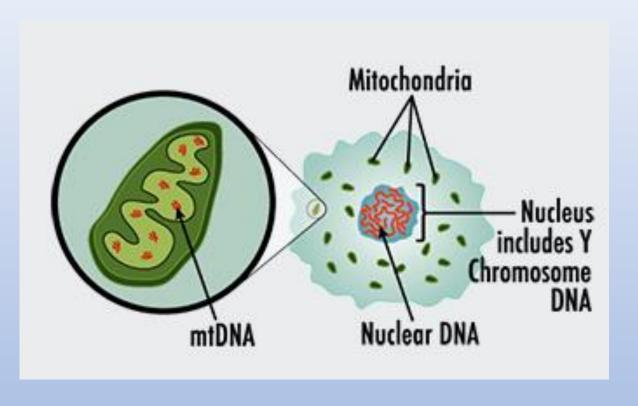
# Two Inspirations



My Matrilineal Line – first 4 generations

### What is mtDNA?

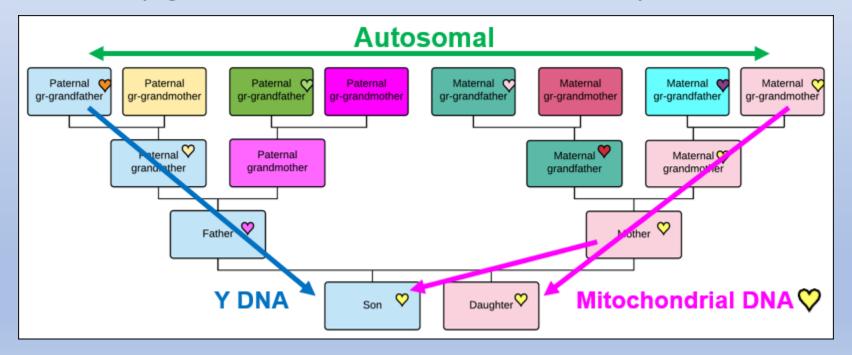
- mtDNA = Mitochondrial DNA
- Part of every cell in our body but is DNA that is NOT part of the nucleus (unlike autosomal and Y-DNA)
- Mutates about every 2,500 years so it is retained through generations



Source: https://projects.nfstc.org/fse/01/01-08.html

# What is Different about mtDNA for Genealogy?

- Matrilineal inheritance passed only through females
- Passed for many generations over thousands of years



Courtesy: Roberta Estes at https://dna-explained.com/wp-content/uploads/2021/01/Y-mtdna-autosomal-pedigree.png

### Who Should Take a mtDNA Test?

- Anyone (male or female) interested in solving a specific matrilineal line
- To complement other DNA tests that might have been done like autosomal and Y-DNA
- Anyone just curious to see what happens

Please be aware --- this is very different from the autosomal tests that many of us have taken

Similar to a Y-DNA test you may receive a limited number of matches, unlike the autosomal test

However, you might have matches with common ancestors many generations back

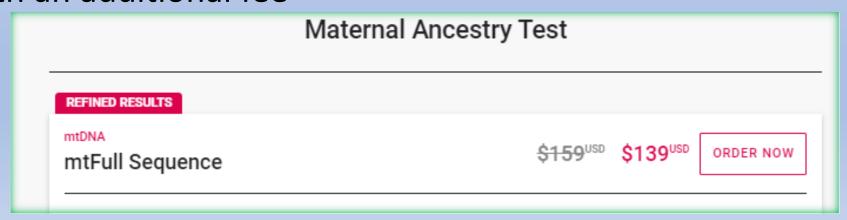
Some people report disappointment with their results, so Your Mileage May Vary!

### How to Test for mtDNA

- Only 3 of the 5 testing companies currently test for mtDNA
  - Family Tree DNA
  - Living DNA (haplogroup information only)
  - 23andMe (haplogroup only)
- Only Family Tree DNA has matching

## mtDNA Testing on Family Tree DNA

- Was on sale over the holidays for \$139 Normal price is \$159
- Recommend only purchasing the "mtFull" test to get all 16,569 markers
  - NOTE: They used to provide a more limited test but I don't see it available anymore
- If you already have a test with FTDNA then you can reuse the sample with an additional fee

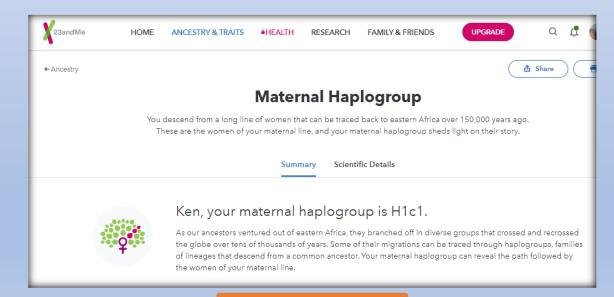


# What About Haplogroups?

- We have paternal (Y-DNA) and maternal haplogroups
- Indicate deep ancestry ( > 500 years to thousands of years)



FTDNA



Your haplogroup is H1c Learn more about your results -> You're viewing your coverage map which shows you where people who share your haplogroup live today. Click on "Migration Map" below to see the journey your ancestors took to get where you live today. See where in the world your DNA is commonly found Tuareg (Fezzan) 61.0% Basque 27.0% 25.0% Portugal Cantabria 23.5% Andalusia 24.0%

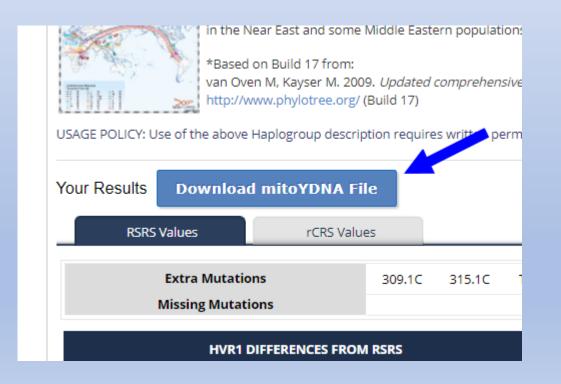
Maternal (mtDNA) Coverage Map

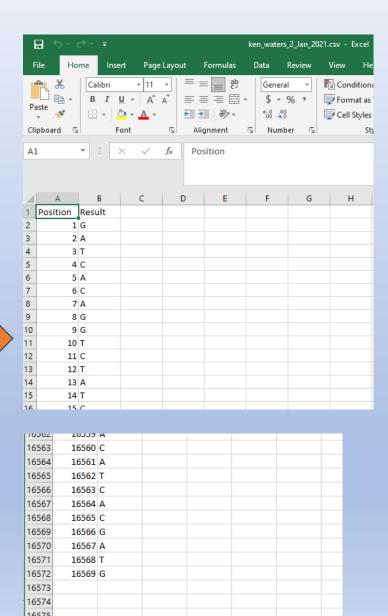
23andMe

LivingDNA

## Downloading Raw mtDNA Results from FTDNA

Results in a CSV file ~ 120 KB with 16,569 base markers (NOTE: \*not\* base pairs as this DNA only comes from mother)





# Downloading FASTA Data

B11096-FASTA, fasta - Notepad

 Upload FASTA files to https://dna.jameslick.com/mthap/FASTA.html

 The FTDNA web code does not properly download the FASTA file but I was able to dig out the proper direct link and with that I could download the file cation and linking it to your name, nloading your raw data, you assume the

Download FASTA File: FASTA

### FASTA Result

#### FASTA format

From Wikipedia, the free encyclopedia

In bioinformatics and biochemistry, the FASTA format is a text-based format for representing either nucleotide sequences or amino acid (protein) sequences, in which nucleotides or amino acids are represented using single-letter codes. The format also allows for sequence names and comments to precede the sequences. The format originates from the FASTA software package, but has now become a near universal standard in the field of bioinformatics. [4]

The simplicity of FASTA format makes it easy to manipulate and parse sequences using text-processing tools and scripting languages like the R programming language, Python, Ruby, and Perl.

#### Contents [hide]

- 1 Original format & overview
- 2 Description line
  - 2.1 NCBI identifiers
- 3 Sequence representation
- 4 FASTA file
  - 4.1 Filename extension
  - 4.2 Compression

#### **FASTA** format

Filename .fasta, .fna, .ffn, extensions .faa, .frn Internet text/x-fasta media type Developed by David J. Lipman William R. Pearson<sup>[1][2]</sup> Initial release 1985 Bioinformatics Extended from ASCII for FASTA Extended to FASTQ format[3] Website www.ncbi.nlm.nih.gov /BLAST/fasta.shtml ₽

#### (BACK)

1/4/2021

mthap version 0.19b (2015-05-11); haplogroup data version PhyloTree Build 17 (2016-02-18) +mods

B11096-FASTA-mtDNA Haplogroup Analysis Report

raw data source B11096-FASTA.fasta (16KB)

FASTA format was uploaded. Based on the markers found, assuming the following regions were completely sequenced: HVR1 (16001~16569) HVR2 (1~574) CR (575~16000).

Found 16569 markers at 16569 positions covering 100.0% of mtDNA.

#### Markers found (shown as differences to rCRS):

HVR2: 263G (309.1C) (315.1C) 318C 477C

CR: 750G 1438G 3010A 4769G 8860G 9150G 13707A 15326G

**HVR1:** 16193T 16263C (16519C)

#### **Best mtDNA Haplogroup Matches:**

#### 1) H1c1

**Defining Markers for haplogroup H1c1:** 

HVR2: 263G 477C

CR: 750G 1438G 3010A 4769G 8860G 9150G 15326G

HVR1: 16263C

Marker path from rCRS to haplogroup H1c1 (plus extra markers):

 $H2a2a1(rCRS) \Rightarrow 263G \Rightarrow H2a2a \Rightarrow 8860G 15326G \Rightarrow H2a2 \Rightarrow 750G \Rightarrow H2a$ 

 $\Rightarrow$  4769G  $\Rightarrow$  H2  $\Rightarrow$  1438G  $\Rightarrow$  H  $\Rightarrow$  3010A  $\Rightarrow$  H1  $\Rightarrow$  477C  $\Rightarrow$  H1c  $\Rightarrow$  9150G 16263C

 $\Rightarrow$  H1c1  $\Rightarrow$  (309.1C) (315.1C) 318C 13707A 16193T (16519C)

### mitoYDNA

- http://mitoydna.org
- A free site to upload Y-DNA as well as mtDNA (the CSV file shown a few slides back)



Home About Privacy FAQ/Help Kits Tools

#### News and Information

If you are shopping and want to help support free and accessible Y and mtDNA data? Check out our store, mitoYDNA Gear or use mitoYDNA AmazonSmile when making your dayto day purchases, mitoYDNA AmazonSmile campaign.

We have new Help Videos available on our mitoYDNA YouTube Channel





A Y and mitochondrial DNA Database - Crowdsourced, Free and Accessible

Get the mitoYDNA Chrome Extension required for downloading your mtDNA file for uploading to mitoYDNA.



Want to join the discussion? Join us on Facebook mitoYDNA User Group Like Share 915 people like this. Be the first of your friends. Twitter @mitoYDNA

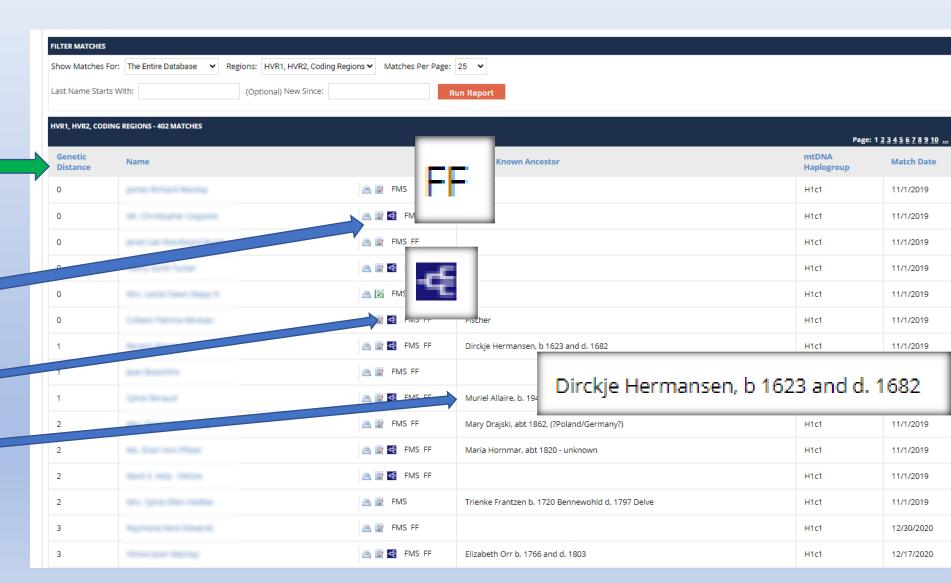
#### Read the mitoYDNA BLOG

#### **Create New Kit**

mitoYDNA ID	First Name	Last Name	Biological Gender	EKA First Name	EKA Last Name	EKA Birth Place	Haplogroup		User Notes
T10183	Ken	Waters	Male	John	Waters	Ireland	M-269	Edit   YDNA   Delete	
T14169	Ken	Waters	Male	Sophia	Armagh	Tewskbury, New Jersey	H1c1	Edit   mtDNA   Delete	

# My Match Results

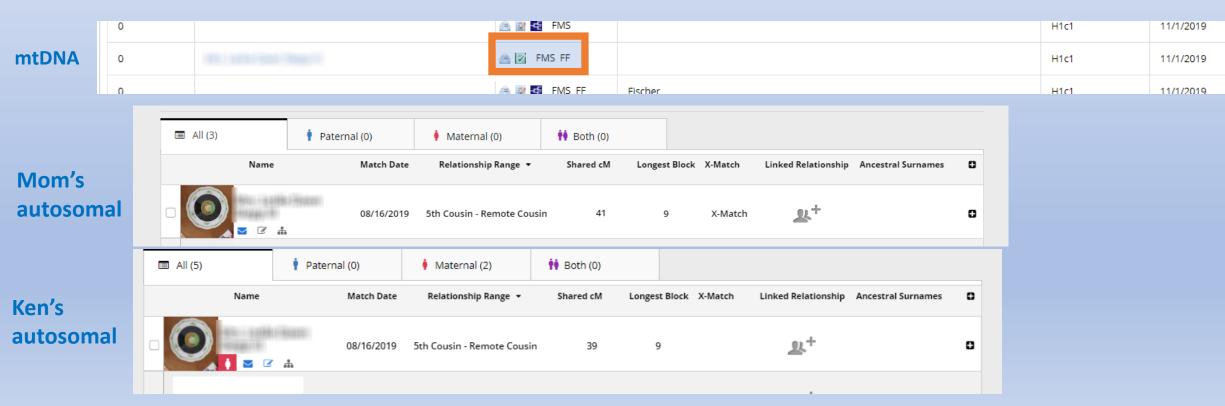
- Closest matches at top, measured by "Genetic Distance"
- Useful:
  - Indicates if they have FF (autosomal test)
  - Tree indicator if exists
  - Earliest
     matrilineal 
     person in tree



### 

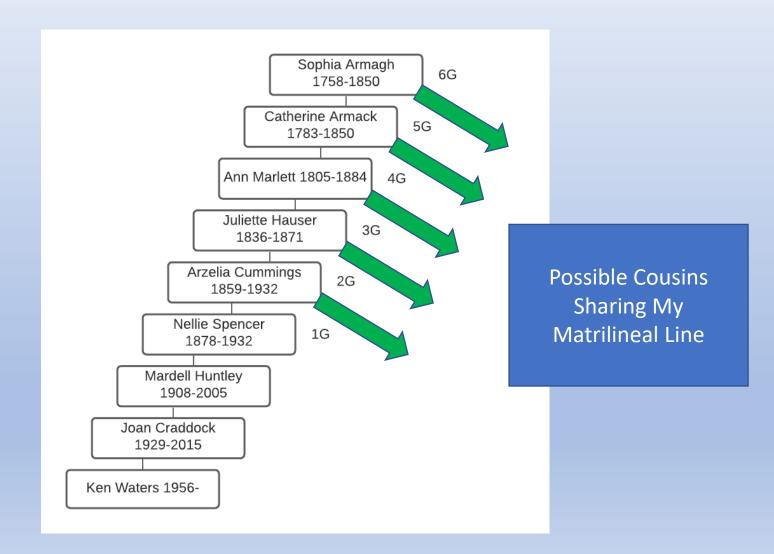


 Using my Mom's uploaded kit from Ancestry I found a 41 cM autosomal (FF) match who also matched my mtDNA matches at a Genetic Distance of 0; also matched me at 39 cMs



# Primary Goal – Compare My Matrilineal Line to Others – Find that Common Ancestor

My known matrilineal line



# My Project Goal

- Build Q&D (Quick and Dirty) trees in Ancestry (private-unsearchable) for each of the 0 Genetic Distance mtDNA matches
  - Only follow matrilineal line
- Start by downloading Matches (.csv)



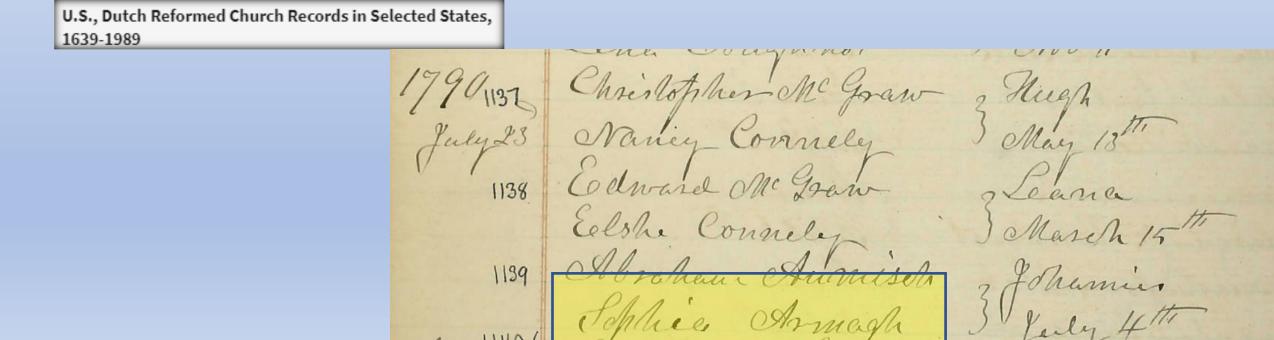
• I'm starting with the 6 matches with 0 Genetic Distance (GD)

### One Clue Researching My Line – Likely Dutch Heritage

My earliest known matrilineal ancestor was Sophia Armagh:

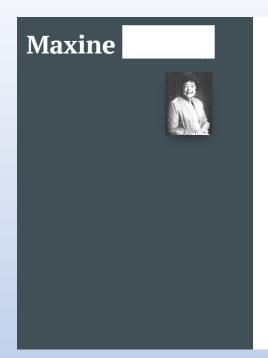


Found a great research source for her—baptism of her son:



### **Build the Trees!**

- For each name I use the email address for a search in BeenVerified to get the birthdate and possible mother and then search them in Ancestry, Google, FB, obits
- First one I was able to find an obit for the test-taker's mother. YES!
- Unfortunately I hit a dead end on his 4<sup>th</sup> GGM. So, on to the next one



83, of Mesa, Arizona passed away on March 22, 2012. Maxine was born on May 21, 1928 in Okemah, Oklahoma where she spent her childhood days. She married in 1949 in Anchorage, Alaska. Being a dedicated airline pilot's wife, she lived in numerous states including Alaska, Wisconsin, Michigan and Minnesota while raising their six children. She was an active member of the Republic Airline Pilot's Wives group for over thirty years. In 1992 she moved to Mesa, Arizona and has been an active member of the community. She was the Chairman of Conventions and Director of Correspondence for the Second Air Division Association-Eighth Air Force; an active member of the Assistance League of East Valley serving as Chairman of the 2011 & 2012 Annual Fashion Show Fundraiser; regularly attended her Red Hat luncheons and served in various ministries at St. Bridget Catholic Church. Maxine is survived by her husband Oak of 62 years, their children, Randall, Susan, Doug, Robert, James & Anne; five sons and daughters in-law, seven grandchildren and eleven great-grandchildren. Services will be held at 10:00am on Thursday March 29 at St Bridget Catholic Church, Lindsay & McDowell in Mesa; Reception immediately following in the church hall. Arrangements were made by Meldrum Mortuary. Memorial donations can be made to Assistance League of East Valley, Attn: I Parnell, 1950 N. Arizona Ave. Ste 3, Chandler AZ 85225 in lieu of flowers.

Share

Send Flower



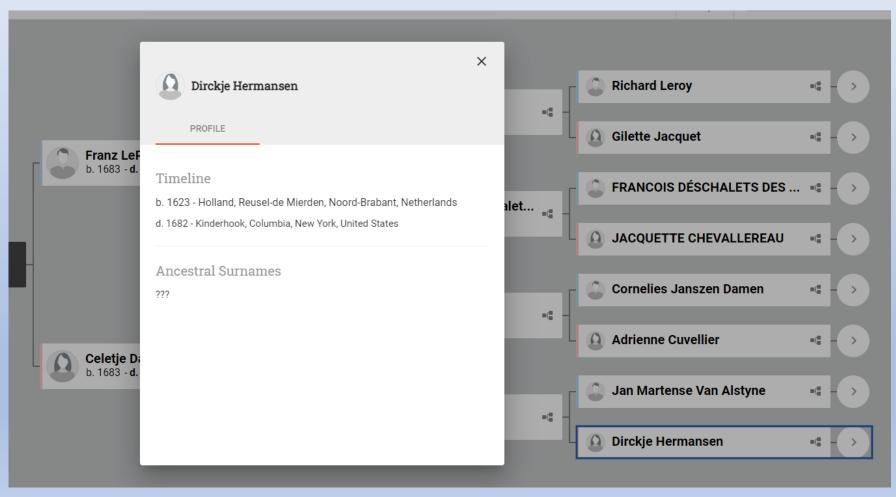
# The Results---after about 4 hours of tedious research

Surname matches!

Match # **Match Birth Autosomal Earliest** 1960 Susan Devore 1809 KY 1 No 2 1982 Yes, no match Sarah Devore 1798 VA 3 1948 Yes, no match None, couldn't determine her mother 4 1948 No Only back to mother Helen Evelyn James 1914 PA 5 1958 Yes, 41 cM match I couldn't build her tree but she claims Dirckje Hermansen, b 1623 and d. 1682 from Holland as a matrilineal ancestor this is same as another of my mtDNA close matches 6 1943 Yes, no match Shows GGM Louisa Fahrenkamp born in 1852 in Germany

### One Other Glimmer of Good Hope

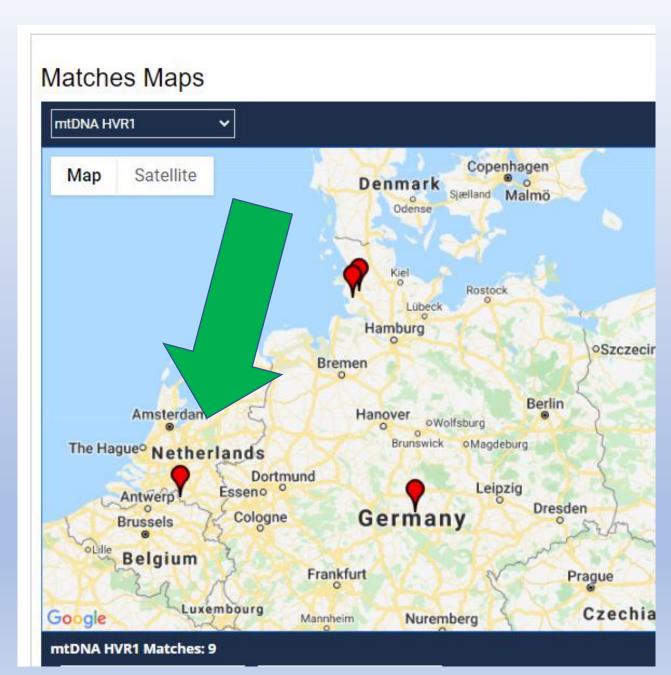
- GD of 1, this match has an ancestor from Holland who settled in the Hudson Valley where my earliest matrilineal ancestor was
- And, Dirckje was also in my Match #5's tree



## Matches Map

- Be sure to look at the Matches Map on FTDNA
- Can often provide some clues to your matrilineal line
- In my case it supports what I've found to be Holland roots





## In Summary

mtDNA could be useful to you --- or may not be; YMMV

- I did get something out of it:
  - Confirmed matrilineal heritage back to Holland
  - Found one close match that also matched on autosomal and was able to communicate with her about an ancestor in Holland
  - Traced likely linkage of my 6GGM in Hudson Valley NY to others in the small New Holland community of Kinderhook
  - Two of closest matches I was able to get down to the same matrilineal surname – unfortunately not a familiar surname to my tree

# Well, Was it Worth the \$159 for Me?

Waste of Money; broke a brick wall

**Your Mileage May Vary!!!** 

## Lastly, Some Common Acronyms

- CRS: Cambridge Reference Sequence (mitochondrial full sequence of one European woman from the 1970s)
- rCRS: Revised Cambridge Reference Sequence
- RSRS: Recombined Sapiens Reference Sequence (2012, reference to "mitochondrial Eve")
- HVR1, HVR2: Hypervariable Regions (more common to change/mutate)
- Note: taking the full mtDNA test returns all bases (1-16,569); HVR1 only returns 16,024 through 16,383; HVR2 only returns 57 through 372.

### Good Resource for More Info



I-dna/

https://dna-explained.com/mitochondrial-dna/

# "My Journey" Video from FTDNA

"My Journey" Video

### FamilyTreeAZ.com



Ken Waters
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familytreeaz.gedmatch@gmail.com

Presentations: http://www.familytreeaz.com/Presentations/

#### **Genealogy Presentations**

Closure -- Solving an Adoption Mystery(Nov 2020)

Using GEDMatch's New MRCA Tool (Sep 2020)

Identifying Those DNA Matches (Mar 2020)
My Top 10 Tips for Genetic Genealogy
Just Tested -- Now What? (February 2020)
Comparison of DNA Testing Sites (January 2020)
DNA Testing Options (January 2020)
DNAPainter (January 2020)
Ancestry Tag Groups (December 2019)
ThruLines (November 2019)
How Am I Related? (October 2019) Handout
Intro to DNA Genetic Genealogy (September 2019)