

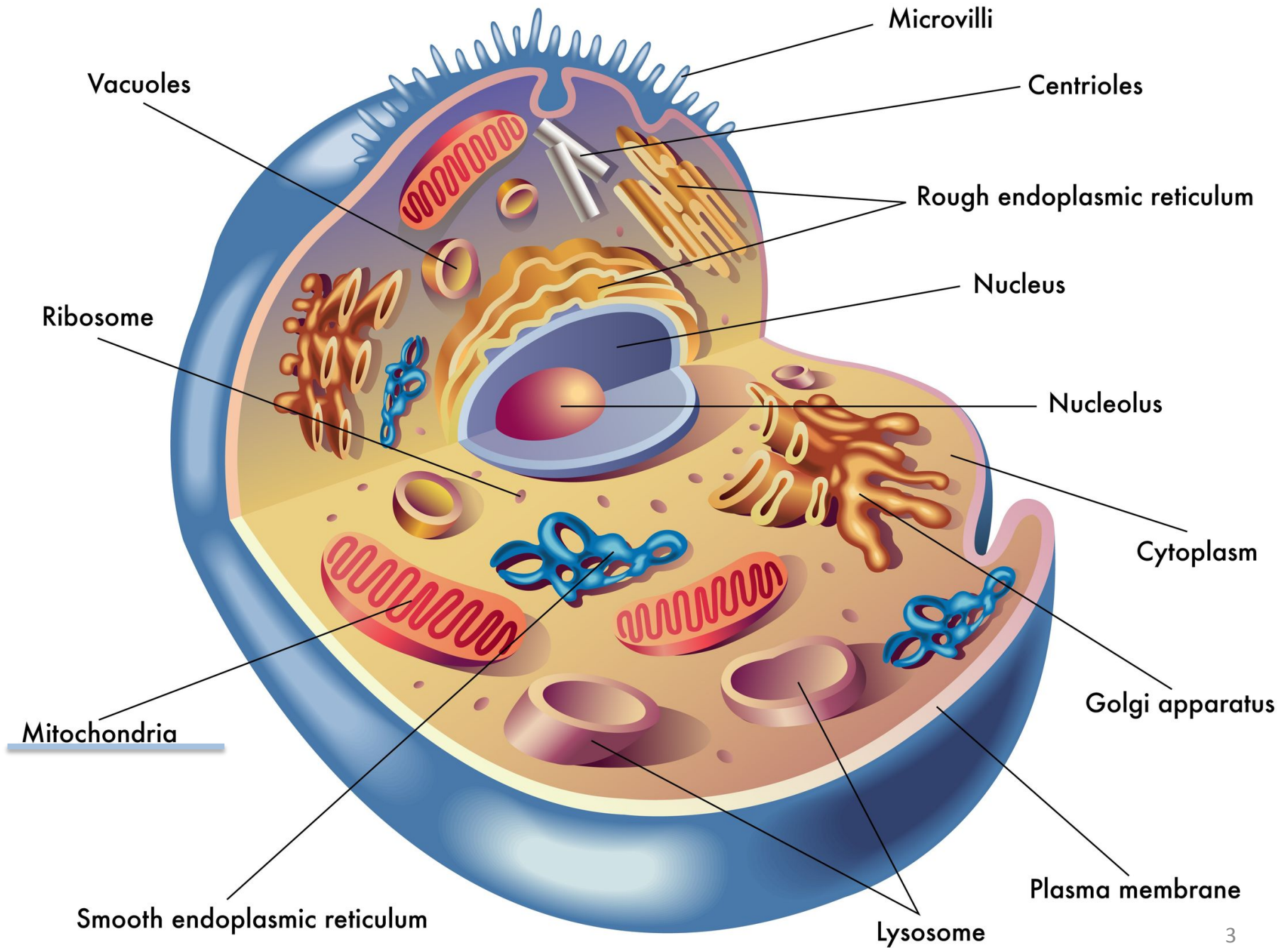
Mitochondrial DNA (mtDNA)

JGSGO

June 5, 2018

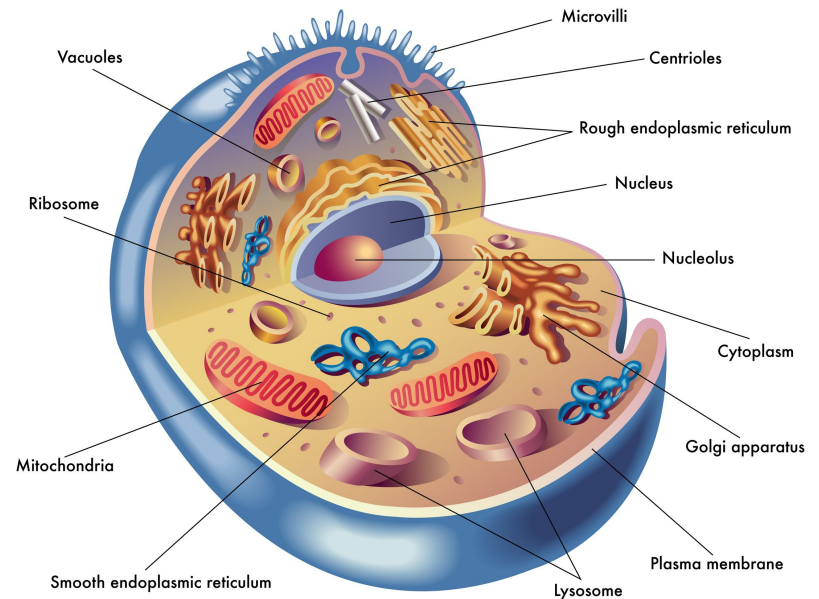
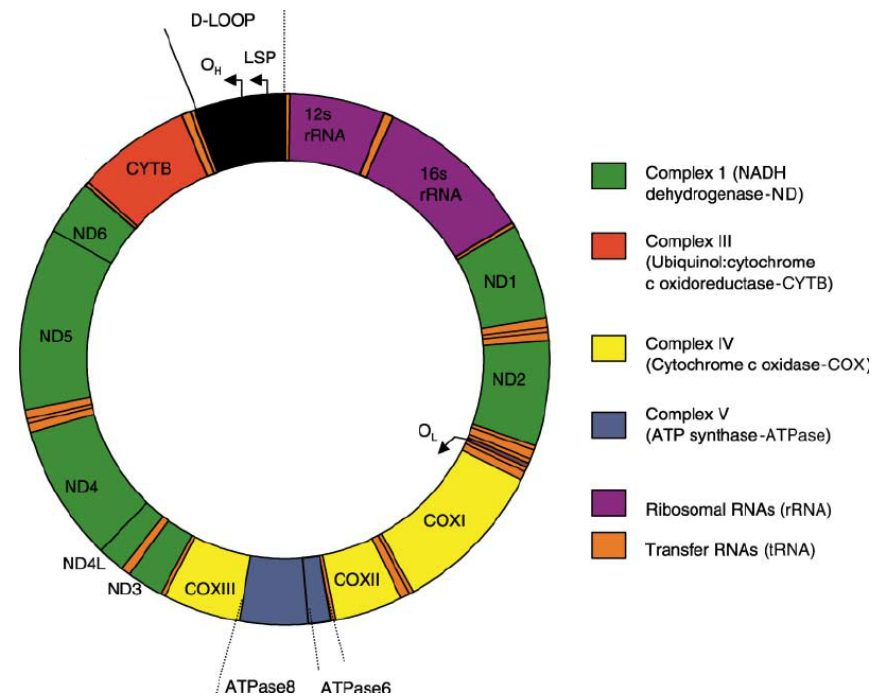
MtDNA - outline

- What is it?
- What do you do with it?
- How do you maximize its value?



mtDNA

- a double-stranded, circular **DNA that is stored in mitochondria outside the nucleus.**
- **mtDNA** is passed down by the mother unchanged, to all her children, both male and female, but while females can pass it on, males can not.
- So it traces the **female line**
- **mtDNA changes relatively slowly – can go back 1000's of years – but still mutations may occur.**
- A **mitochondrial DNA test** can be taken by both men and women.
- mtDNA Haplogroups can be derived

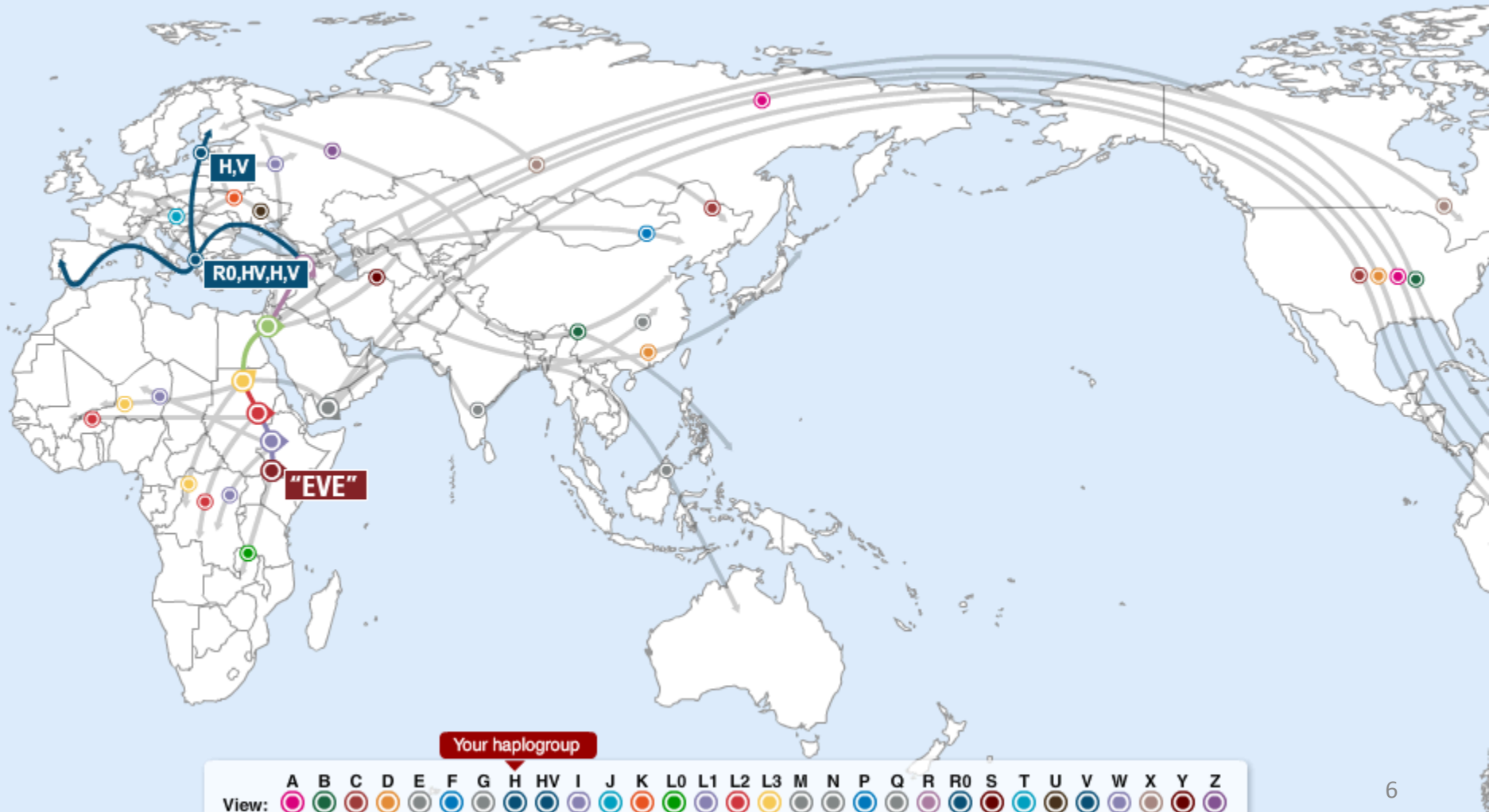


HAPLOGROUP

- Maternally derived haplogroup ‘is a group of maternally related individuals who ‘share several genetic mutations as well as a common (usually ancient) ancestor’
 - Guide to DNA Testing and Genetic Genealogy, Blaine Bettinger, 2016
- H2a1a – K2a2a1 – 2 family members
 - Designated by LETTERS and NUMBERS.
- Y-DNA haplogroups also exist.

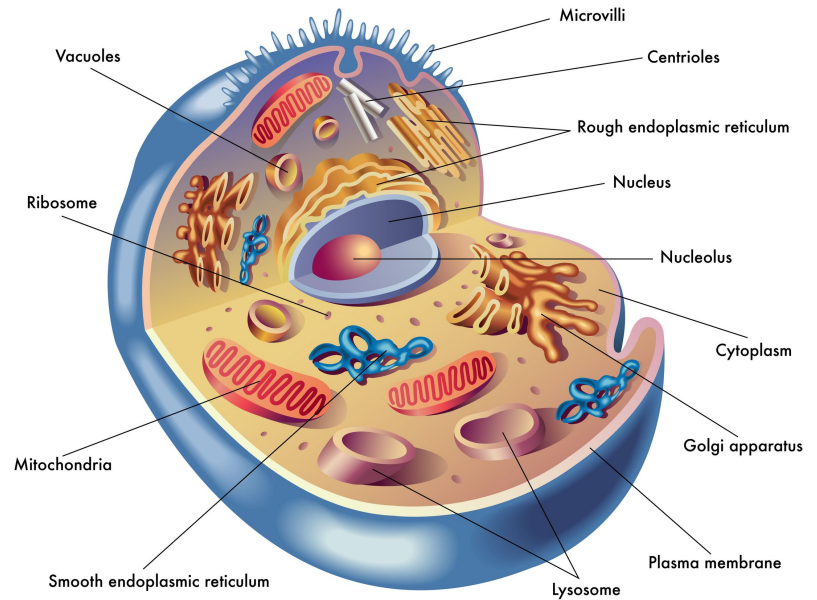
HAPLOGROUP Migration Map

HAPLOGROUP MIGRATION MAP



mtDNA TESTING LIMITATIONS

- **Difficult to pinpoint how closely related 2 individuals with mtDNA matches are since**
- **Individuals with identical mtDNA or identical haplotypes can be related either very recently or as much as several thousand years ago.**
- **You have to use other sources of information to assess significance of a match**



Significance of a match

- Surnames
- Geographic location
- Genetic distance on test – 1,2,3 – mtDNA has
- Check online family trees
- May use Y-DNA on related male relatives.
- Contact other matchees
- Cf to atDNA testing results (use cM's)

mtDNA TESTING

- Family Tree DNA – 2 choices
 - HRV1 & 2 - \$89 (Hypervariable regions) only
 - Full mtDNA sequencing - \$199
- 23andme does SNP testing on selected loci
- Regions HVR1 & 2 – change more quickly so differences between individuals can be discerned.

UPLOAD DNA RESULTS

- **Uploading results to different web sites allows other researchers who share similar DNA markers to connect with you.**
- **Family Tree DNA** - <https://www.familytreedna.com>
 - Ysearch.org – for people who have tested with different companies.
 - <http://www.mitosearch.org> - mtDNA
- **My Heritage** - <https://www.myheritage.com>
 - Analyzes your DNA and compares it to others
 - Sets up a **family tree web site** for you
 - Creates an email for you, with which you can email photos and doc's for automatic uploading.
- Gedmatch - <https://www.gedmatch.com> - free upload

UPLOADING DNA RESULTS

consider including:

- SURNAME information – by place
- Geographic information – family homes
- DNA information – haplotypes, etc.

- Makes it much easier to determine a MATCH
- Privacy could be an issue

HAPLOGROUP PROJECTS – mtDNA & Y DNA

- PROJECTS join researchers with groups from the same haplogroup.
- DNA PROJECTS: Haplogroup, Surname, Geographic
- VALUE OF PROJECTS - Get advice for further testing, compare your results with others, share new information, get help in assessing validity of matches, have possibility of joining subgroups.
- <https://www.familytreedna.com> *project info*
- <https://www.worldfamilies.net/projectfaq#what>
SURNAME PROJECTS – a great resource
- <https://isogg.org/wiki/> -
International Society of Genetic Genealogy

H2a1a (Mitochondrial DNA) PROJECT

- <https://www.geni.com/projects/H2a1a-Mitochondrial-DNA/36000>
- This project is a meeting place for users who share the H2a1a Mitochondrial DNA haplogroup, which means they are related along their maternal lines. Users in this group may want to share their family trees with each other to find overlaps and merge duplicate profiles in order to join or expand the World Family Tree and discover new relatives.

https://www.familytreedna.com/my/ mtdna-matches

- Shirley – K2a2a1 – full sequencing – matches



mtDNA - Matches

FILTER MATCHES

Show Matches For: Regions: Matches Per Page:

Last Name Starts With: (Optional) New Since: [Run Report](#)

HVR1, HVR2, CODING REGIONS - 311 MATCHES

Page: [1](#) [2](#) [3](#) [4](#) [5](#) [6](#) [7](#) [8](#) [9](#) [10](#) ... of 13

Genetic Distance	Name	Earliest Known Ancestor	mtDNA Haplogroup	Match Date
0	Sybil Kleinberg	FMS FF	K2a2a1	5/21/2018
0	Isaac Meilijson Ph.D.	FMS FF Leie Risa Weinblatt	K2a2a1	5/21/2018
0	Evan Lipsitz	FMS FF	K2a2a1	4/18/2018
0	Frits Erik Verhoeven	FMS FF Rebecka Levie Frank b. 24 Nov. 1798	K2a2a1	4/4/2018

mtDNA Uses - SUMMARY

- Ancient origins & migratory patterns
- Finding genetic cousins possible if used along with other DNA studies and traditional genealogical methods.
- Exploring your maternal line
 - Eg, if your atDNA shows a match, you don't know if the relationship is on the maternal or paternal side, mtDNA could clarify it.

CONCLUSIONS

- Full sequencing most useful – slightly more \$\$
- Joining a Haplogroup Project **recommended**
- mtDNA testing useful record of one's female line and ancient origins
- Genetic cousins can be found if corroborated by standard genealogical methods and ancillary genetic testing.

THANK YOU!