

# mitoBench: An interactive visual workbench for population genetics on mitochondrial DNA



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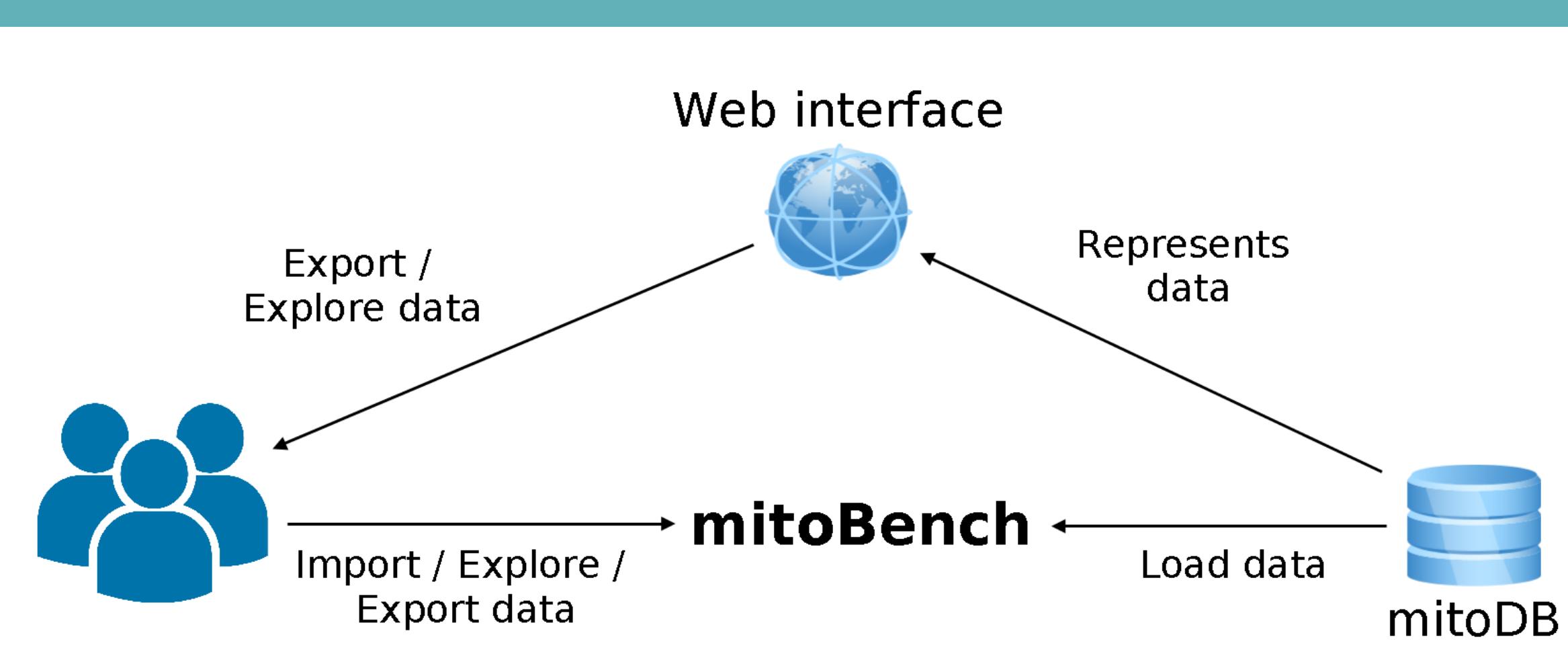


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## Motivation & Goals

- Especially in the research field of ancient human DNA, mitochondrial DNA (mtDNA) is often the only proxy available to study extinct populations and their relationship with modern populations
- Tools for the analysis typically rely on different file formats → requiring manual interaction with the data for downstream analysis
- mitoBench: workbench to ease file conversions, interactively analyzing human mitochondrial genomes and visualize results
- mitoDB: database for mitochondrial reference data to provide a central reference database that can be easily accessed via the workbench

## Workflow



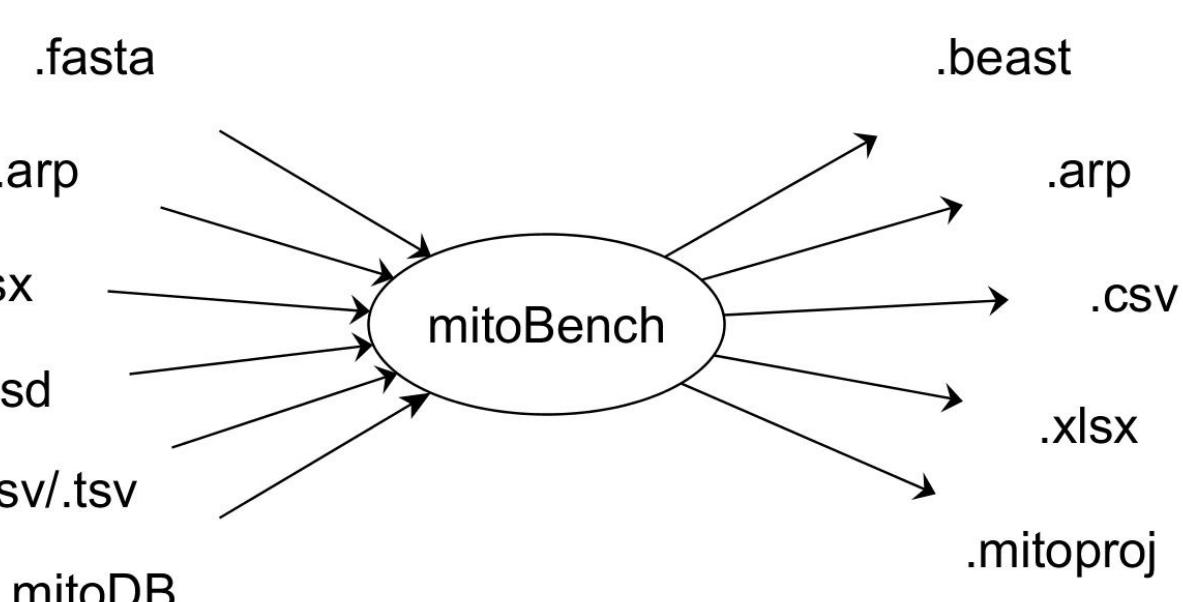
## mitoDB

- Web-Frontend with Vaadin Java Framework
- Backend with PostgreSQL, providing sequence information and meta-data
- Curated data upload with meta-data
- Retrieval possible through WebUI and mitoBench



## mitoBench

### File conversions



⇒ connect the workbench with existing analysis methods such as BEAST, Arlequin, PhyloTree and others

### Data representation as Table

ID	MTSequence	Haplogroup	Time Period	continent	country	country_region	culture_type
JK2975	GATCA...	R	Roman Period	Africa	Egypt	Beni Suef	Ancient
JK2976	GATCA...	U	Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient
JK2972	GATCA...	T1a5	Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient
JK2916	NNCA...	R0	Pre-Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient
JK2972	GATCA...	HT1a2a	Roman Period	Africa	Egypt	Beni Suef	Ancient
JK2918	GATCA...	I2a2e	Roman Period	Africa	Egypt	Beni Suef	Ancient
JK2984	GATCA...	T1a5	Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient
JK2337	GATCA...	I2a2b	Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient
JK2963	GATCA...	M1a1	Pre-Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient
JK2885	GATCA...	R2Y	Pre-Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient
JK2887	GATCA...	I2a1a1	Pre-Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient
JK2903	GATCA...	R0a1a	Roman Period	Africa	Egypt	Beni Suef	Ancient
JK2332	GATCA...	H1	Roman Period	Africa	Egypt	Beni Suef	Ancient
JK2328	GATCA...	H1a1	Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient
JK2330	GATCA...	M1a1	Roman Period	Africa	Egypt	Beni Suef	Ancient
JK2962	GATCA...	H1a1c1	Pre-Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient
JK2820M	GATCA...	R0a2f	Pre-Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient
JK2996	GATCA...	H1V2	Pre-Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient
JK2883	GATCA...	T1a	Pre-Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient
JK2879	GATCA...	U3b	Roman Period	Africa	Egypt	Beni Suef	Ancient

### Data filtering / Statistics

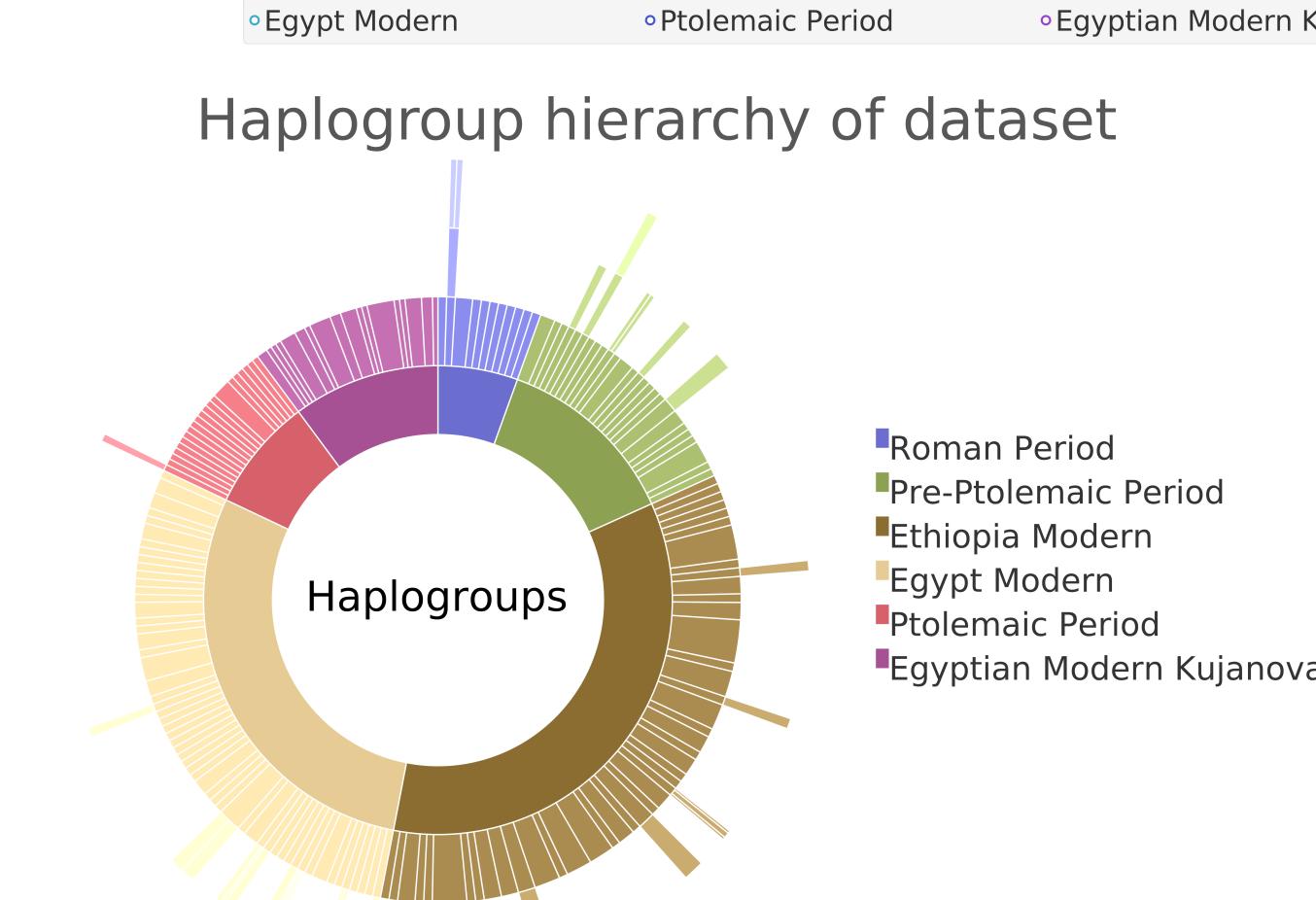
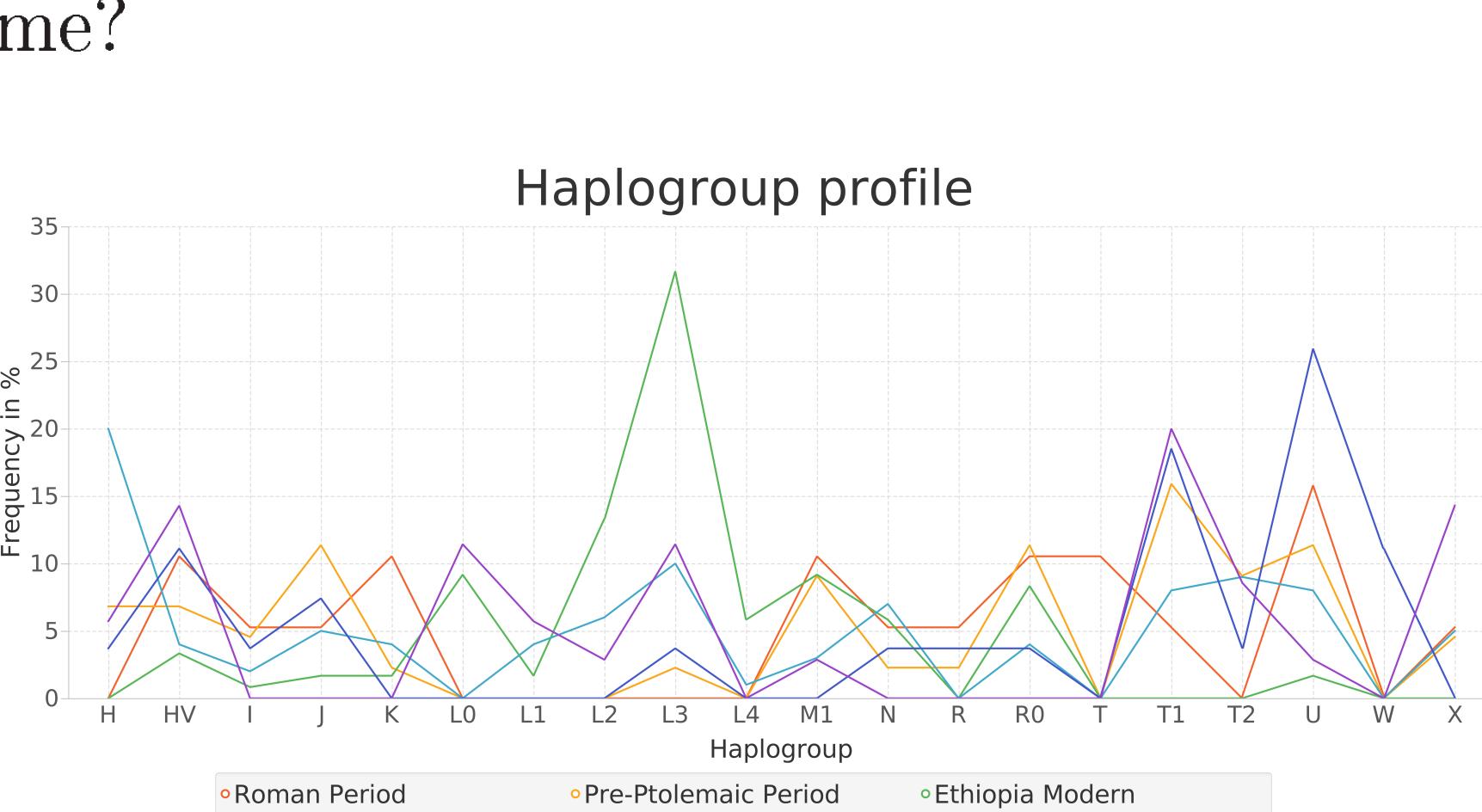
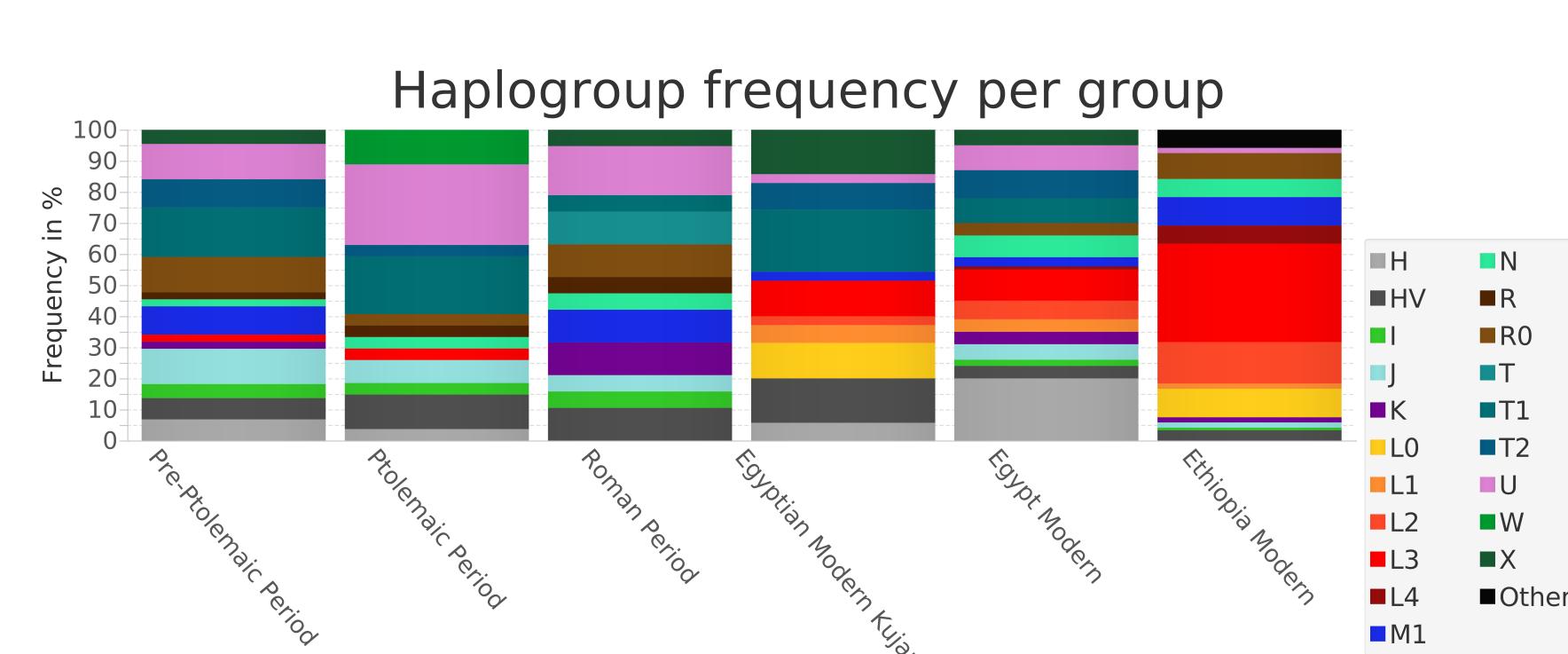
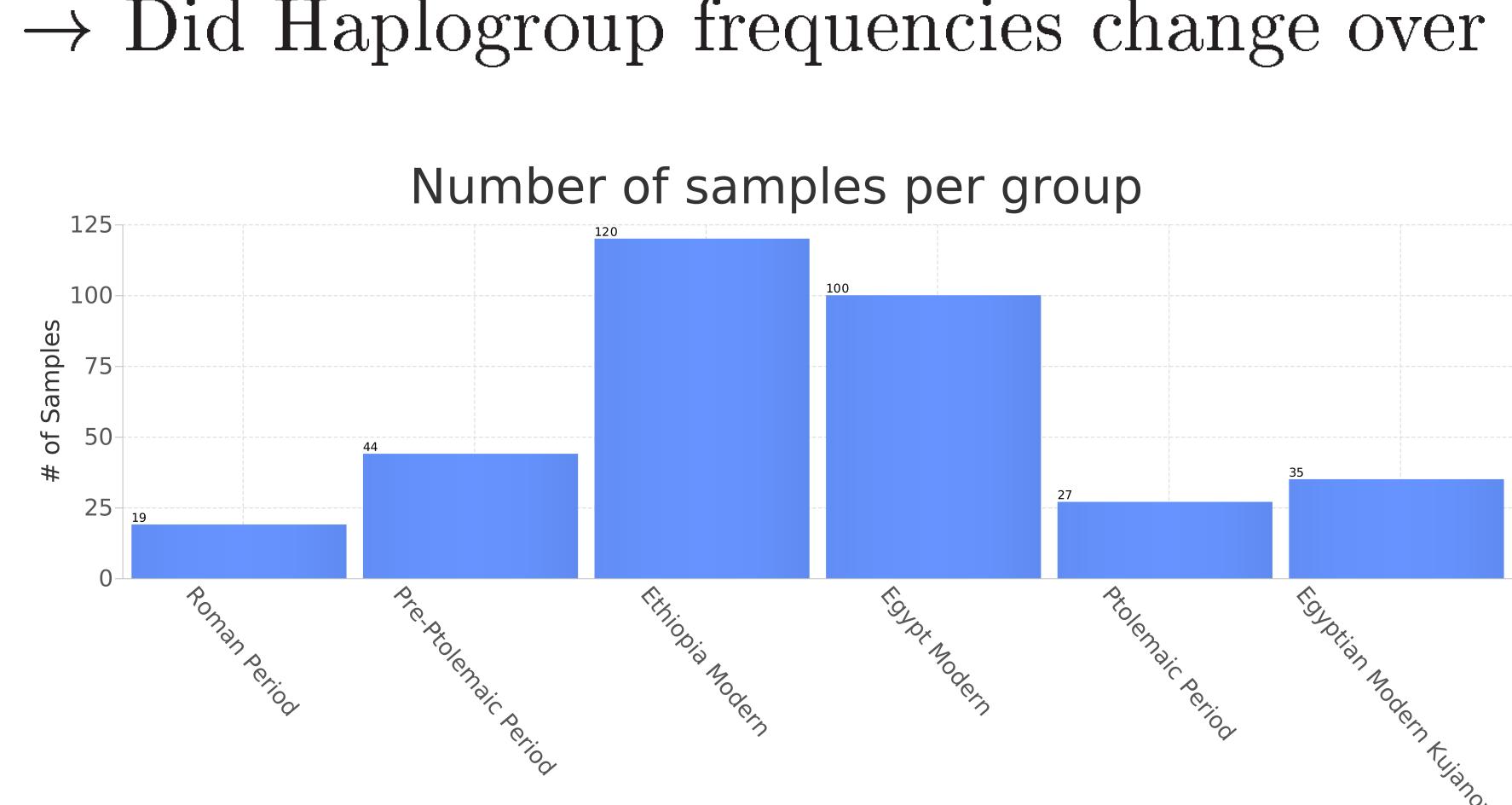
- Haplogroup filtering / frequencies
- Mutation filtering / frequencies

Haplogroups based on [PhyloTree<sub>mt</sub>](#)

### Data visualizations

Mitochondrial analysis of 90 ancient Egyptian mummies (Schuenemann & Peltzer et al. 2017, accepted)

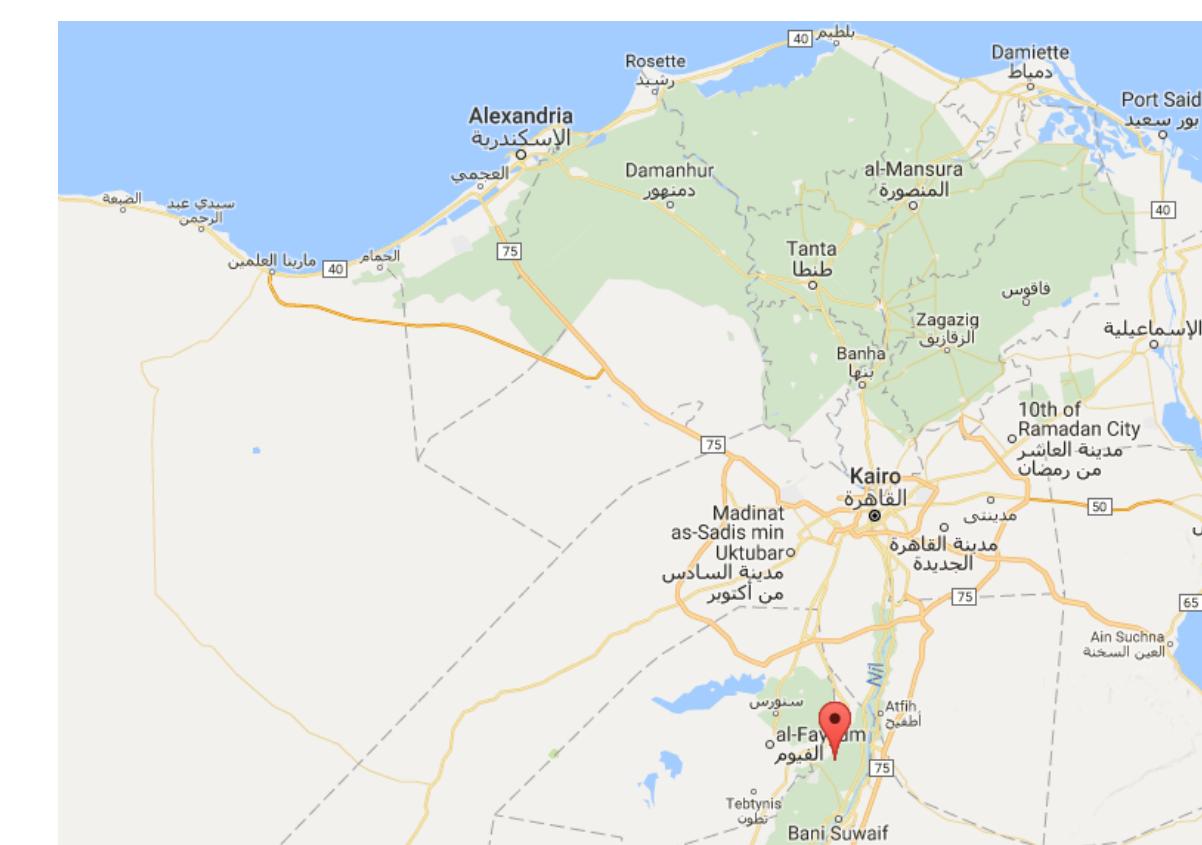
→ Did Haplogroup frequencies change over time?



## Outlook

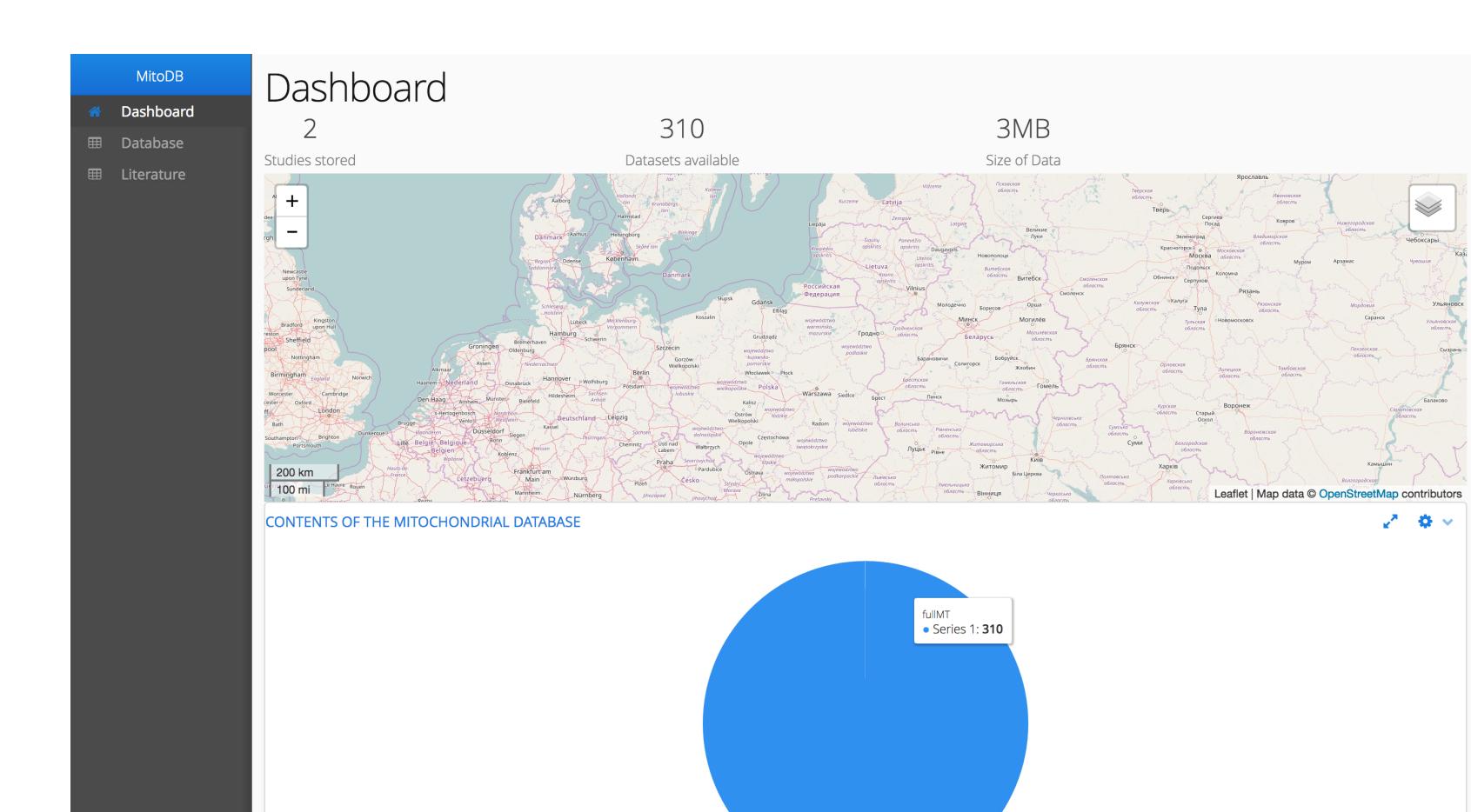
### mitoBench

- Provide methods for downstream analysis (e.g.  $F_{ST}$  calculations, Founder Analysis)
- Offer more visualizations (e.g. geographical maps)



### mitoDB & web interface

- Export/import functionality
- web-based dashboard to explore database



## References

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