



# Questions and Answers

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## GHEP-OS Spring 2025

### ● WORKSHOP 1

X-chromosomal markers in forensic genetics  
Daniel Kling & Andreas Tillmar

### ● WORKSHOP 2

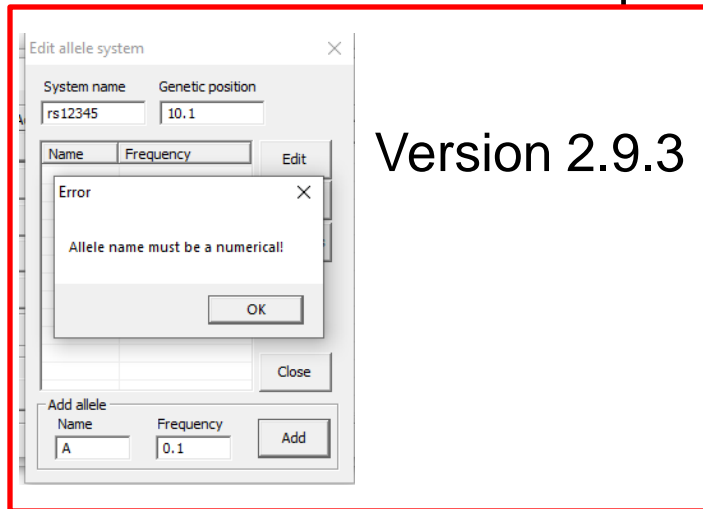
Acreditación en el campo de la Genética Forense y estrategias de validación de ensayos  
Manuel Crespillo Márquez, Rosalía Izquierdo & Estel Enreig Cabanes

### ● WORKSHOP 3

La genética en la Identificación de víctimas a gran escala: comparación de perfiles y evaluación estadística con Familias  
Carlos Vulló & Lourdes Prieto

# Questions and Answers

- Cannot FamLinkX import alleles that are not numeric?



Workaround: Import them from file (normal procedure)



# Questions and Answers

- Should the frequency file downloaded from the FamLink website be used as input in its entirety? If so, where should it be uploaded

## Frequency databases (haplotypes and allele frequencies)

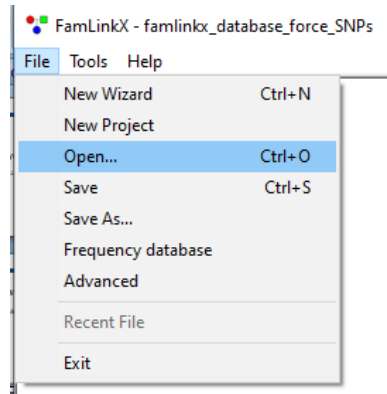
Databases may be found [here](#) or downloaded in the tables below. Size refers to Please note that several populations can appear twice (different sources) and in

### Argus X12

Population	Size	Database	Publication
Sweden	652	<a href="#">SWE</a>	<a href="#">Tillmar A.</a>
Norway	631	<a href="#">NOR</a>	<a href="#">Bergseth et al.</a>
Czech Republic	307	<a href="#">CZE</a>	<a href="#">Zidkova et al.</a>
Germany	1037	<a href="#">GER</a>	<a href="#">Edelmann et al.</a>
Greece	121	<a href="#">GRE</a>	<a href="#">Tomas et al.</a>
Italian	200	<a href="#">ITA</a>	<a href="#">Bini et al.</a>
Sardinia	316	<a href="#">SAR</a>	<a href="#">Robino et al.</a>
Serbia	220	<a href="#">SER</a>	<a href="#">Veselinovic et al.</a>

# Questions and Answers

- Should the frequency file downloaded from the FamLink website be used as input in its entirety? If so, where should it be uploaded



```
[Save file - FamLinkX Version 2.2]
[General parameters]
NewAlleleFrequency = 0.01
Lambda = 652
DatabaseSize = 652 : DatabaseName: Sweden
[[Thresholds]]
0      1e-005
1      1e-005
2      1e-005
3      1e-005
4      1e-005
5      1e-005
6      1e-005
```

The files are FamLinkX projects (.sav format)



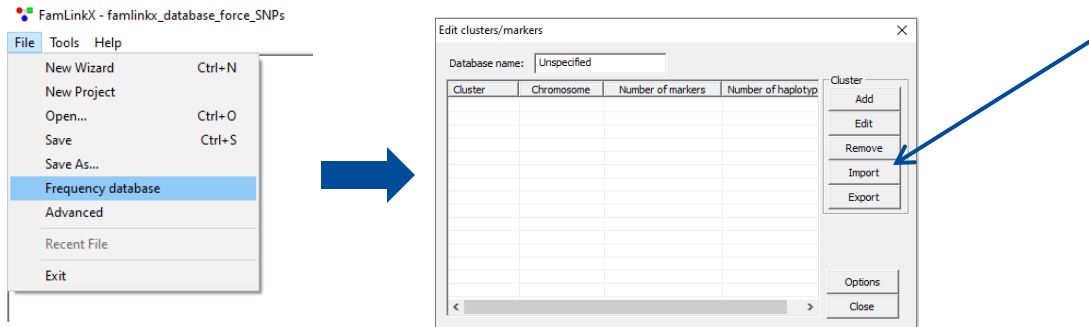
# Questions and Answers

- From the slides in the first session, I noticed there is a section for absolute haplotype frequencies per linkage group and another for allele frequencies. Should we separate the downloaded database into two separate files: one containing only allele frequencies and another with haplotype frequencies?

A single file should be used. Haplotypes are a different format and Allele frequencies are automatically computed from these

# Questions and Answers

- To create a database in the same structure as the one available on the website, is there a specific tutorial or is it the output of a particular software?



See slides from Session 1 on FamLinkX and frequency import

# FamLinkX

➤ Frequency data import format

Haplotypes

Haplotype	DXS10148	DXS10135	DXS8378	Count
Germa1	13.3	28	12.0	1
Germa2	13.3	29	12.0	1
Germa3	14	27	12.0	1
Germa4	16	22.1	10.0	1
Germa5	16.1	27	10.0	1
Germa6	17	27	12.0	1
Germa7	18	18	11.0	1
Germa8	18	27	11.0	1
Germa9	18	27	11.0	1
Germa10	18	27	11.0	1
Germa11	18	27	12.0	1

FamLinkX will automatically detect common STR markers

# FamLinkX

## ➤ Frequency data import format

Haplotype	DXS10148	DXS10135	DXS8378	Count
Germa1	13.3	28	12.0	1
Germa2	13.3	29	12.0	1
Germa3	14	27	12.0	1
Germa4	16	22.1	10.0	1
Germa5	16.1	27	10.0	1
Germa6	17	27	12.0	1
Germa7	18	18	11.0	1
Germa8	18	27	11.0	1
Germa9	18	27	11.0	1
Germa10	18	27	11.0	1
Germa11	18	27	12.0	1

Marker	[cM Kosambi]	Overall mutation rate	Male mutation rate	Female mutation rate
DXS9900	0.31	0.001		
DXS6807	14.76	0.009322404	0.018470994	0.018470994
DXS9895	17.09	0.011684636	0.02309621	0.02309621
DXS9906	17.1	0.001		
DXS10148	19.84	0.004727572	0.007048422	0.002667129
DXS10135	20.03	0.010961627	0.017863682	0.003903
DXS8378	20.21	0.001096763	0.001374888	0.000579003
DXS9902	32.32	0.003241543	0.004436024	0.001921965
DXS6795	44.24	0.001937751	0.003867992	0.003867992
DXS9907	55.32	0.001		
DXS6810	75.12	0.012820264	0.025372465	0.016789775
GATA144D04	78.96	0.001		
DXS10076	85.04	0.001		
DXS10077	86.06	0.001		
DXS10078	85.07	0.001		
DXS10161	89.67	0.001		
DXS10160	89.86	0.001		
DXS10159	90.01	0.01565018	0.030810504	0.030810504

FamLinkX finds genetic position and mutation rates





# Questions and Answers

- Many studies report linkage disequilibrium between loci in different linkage groups. When using a population database where disequilibrium has been identified between loci from different linkage groups, should we use haplotype frequencies based on linkage groups, or is it more appropriate to use haplotype frequencies based on all 12 X-STRs?

We will address this and further question today