iSFG
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DIVIANA: A free, user-friendly app for complex DVI cases

Magnus Dehli Vigeland Thore Egeland

Thore Egeland

2nd INTERNATIONAL SYMPOSIUM ON DNA SUPPORTED MISSING PERSONS AND DISASTER VICTIM IDENTIFICATION
Tbilisi, Oct 10, 2024

X/José Ballesta

Plan

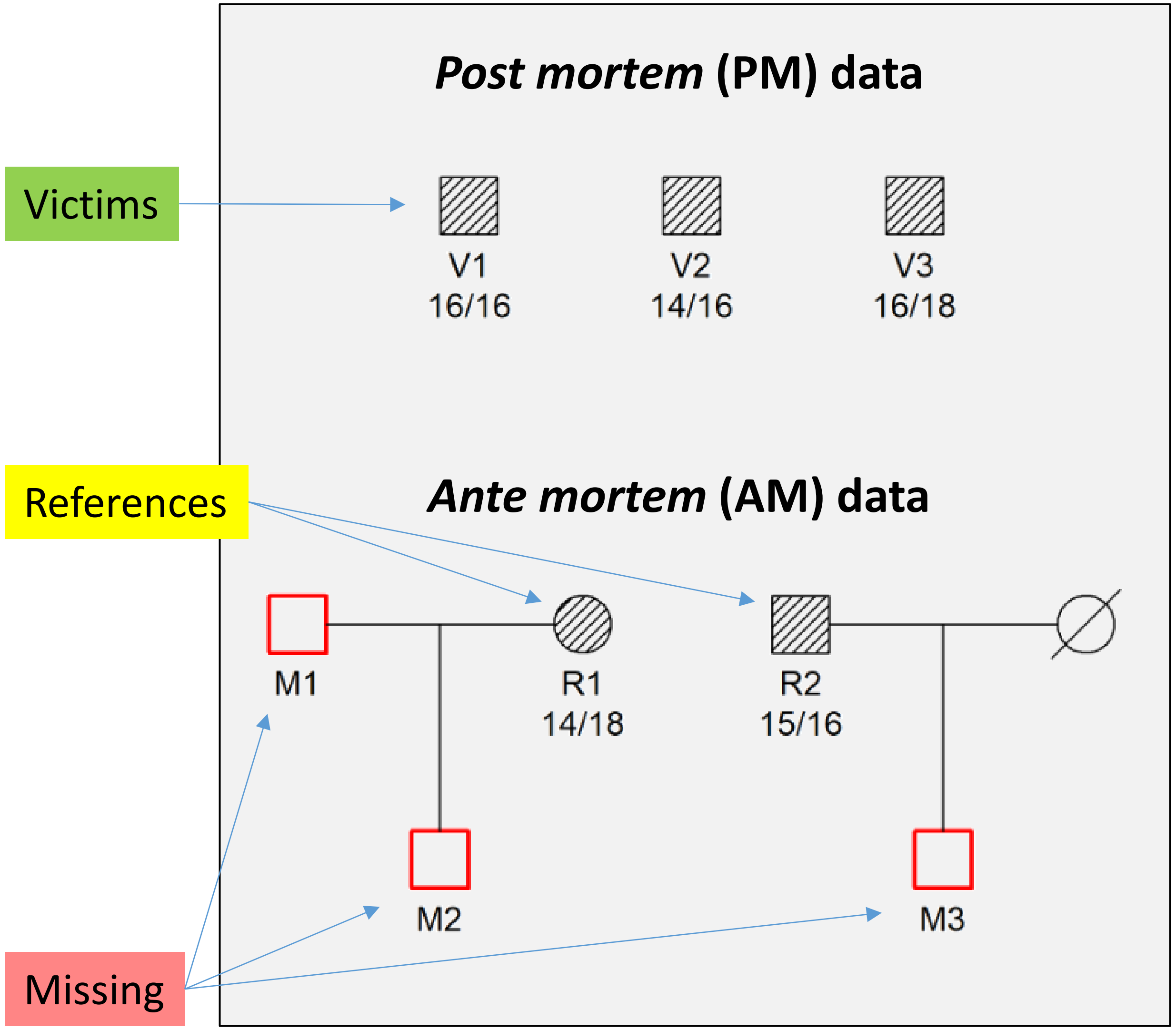
- Disaster victim identification
  - Terminology and standard methods

- Challenging cases
  - Families with multiple missing

- New statistical tools
  - Generalised likelihood ratio (GLR)
  - Relatedness triangle

- New software: **DIVIANA**

Disaster victim identification (DVI)



***Post mortem* (PM) data**

***Ante mortem* (AM) data**

Computational approaches

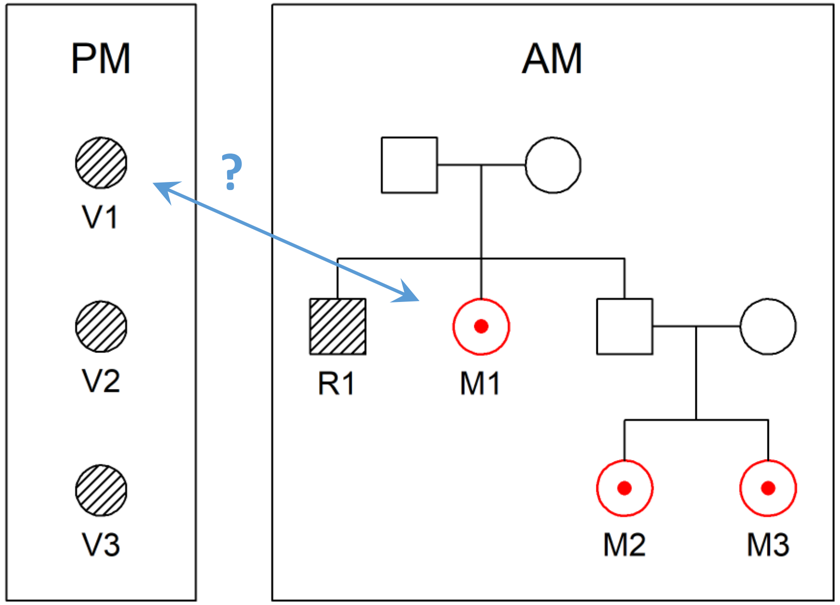
- Pairwise LR

$$\begin{array}{c|ccc} & M1 & M2 & M3 \\ \hline V1 & 1 & 0 & 0.39 \\ V2 & 1 & 16142 & 0 \\ V3 & 1 & 0 & 6266 \end{array}$$

- Joint / global

Vigeland & Egeland (Scientific Reports, 2021):
Joint DNA-based disaster victim identification

Joint analysis of DVI data



Victims and R1 typed with 23 STRs.

Pairwise LR

$$\begin{array}{c|ccc} & M1 & M2 & M3 \\ V1 & 121.3 & 398.6 & 398.6 \\ V2 & 0.6 & 36.6 & 36.6 \\ V3 & 0.00 & 1.3 & 1.3 \end{array}$$

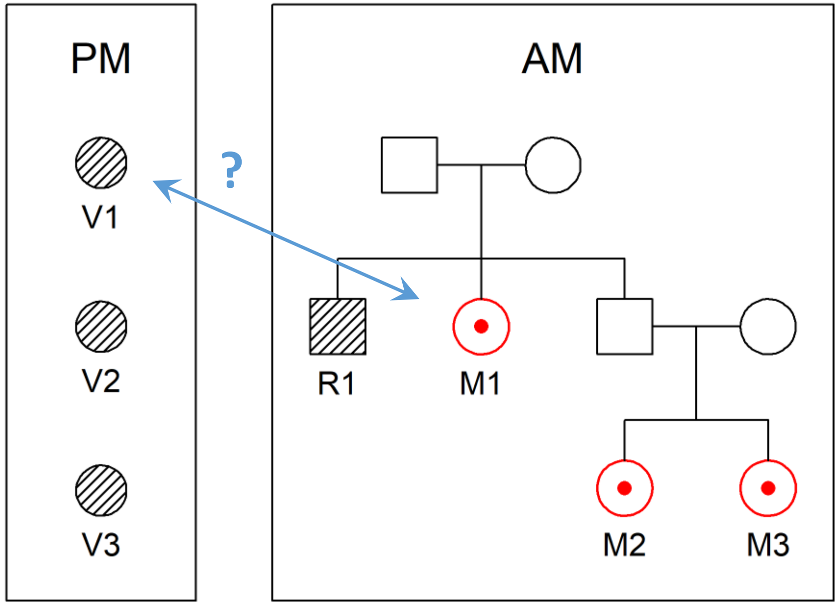
Joint likelihoods

	V1	V2	V3	loglik
1	M1	M2	M3	-257.7
2	M1	M3	M2	-257.7
3	*	M2	M3	-268.9
4	*	M3	M2	-268.9
5	M2	M1	M3	-272.6
6	M3	M1	M2	-272.6
7	M1	*	M2	-276.5
8	M1	*	M3	-276.5
9	M2	M3	M1	-276.6
⋮				
32	*	*	*	-286.0
33	*	M1	*	-286.5
34	*	*	M1	-292.2

A tool for interpreting joint results:

Generalised likelihood ratio (GLR)

$$\mathrm{GLR} = \frac{\max_{i\in H_1} L_i}{\max_{j\in H_2} L_j}$$



Victims and R1 typed with 23 STRs.

H_1: V1 = M1
H_2: V1 ≠ M1

$$\mathrm{GLR} = \frac{L_1}{L_3} = \frac{e^{-257.7\ldots}}{e^{-268.9\ldots}} = 70\,582$$

Strong evidence

Joint likelihoods

	V1	V2	V3	loglik	
1	M1	M2	M3	-257.7	✓
2	M1	M3	M2	-257.7	✓
3	*	M2	M3	-268.9	✗
4	*	M3	M2	-268.9	✗
5	M2	M1	M3	-272.6	✗
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7	M1	*	M2	-276.5	✓
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⋮					
32	*	*	*	-286.0	✗
33	*	M1	*	-286.5	✗
34	*	*	M1	-292.2	✗

Interpretation of $GLR = x$

$$\mathrm{GLR} = \frac{\max_{i \in H_1} L_i}{\max_{j \in H_2} L_j}$$

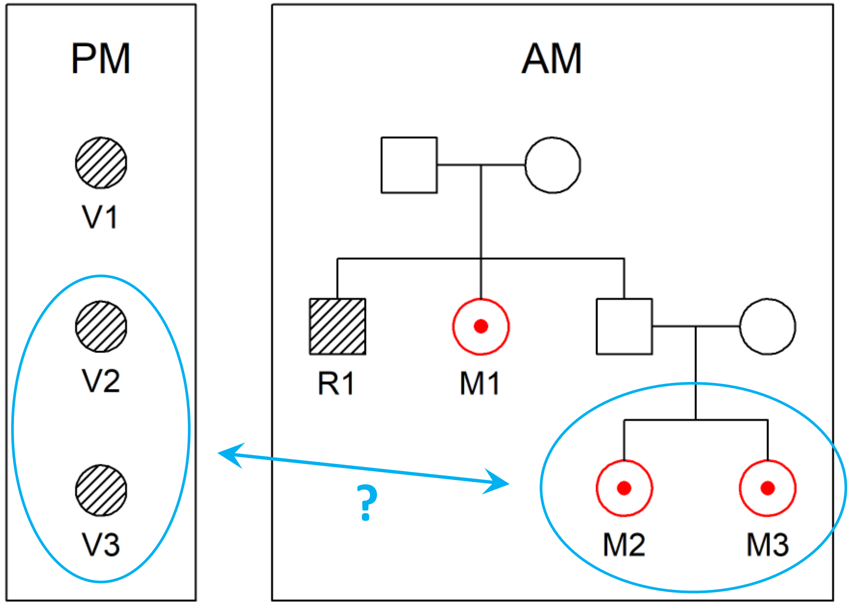
The best explanation of the data given H1

is $\boldsymbol{x}$ *times more likely*

than the best explanation given H2

Symmetric matches

$$\mathrm{GLR} = \frac{\max_{i \in H_1} L_i}{\max_{j \in H_2} L_j}$$



Cannot separate these! →

Joint likelihoods

	V1	V2	V3	loglik
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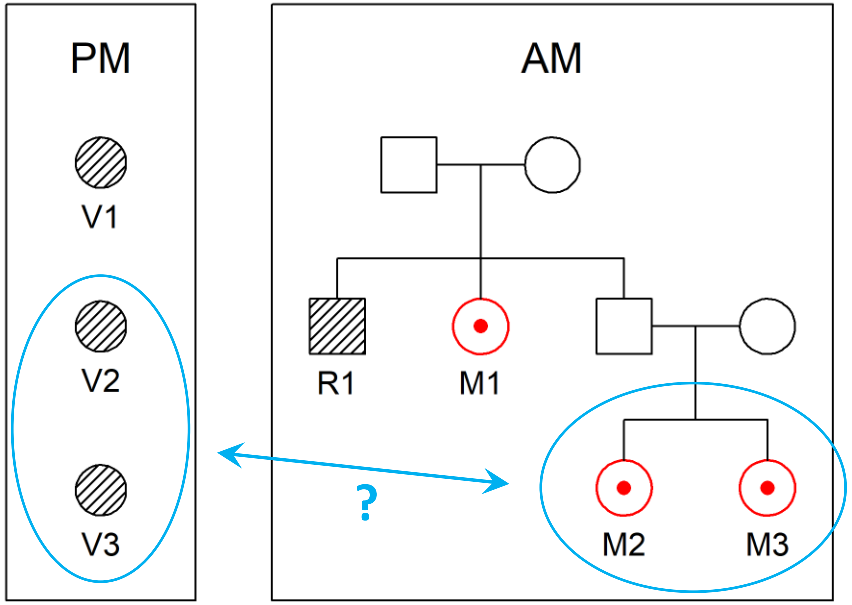
Symmetric matches

$$\text{GLR} = \frac{\max_{i \in H_1} L_i}{\max_{j \in H_2} L_j}$$



H_1: {V2,V3} = {M2,M3}

H_2: {V2,V3} ≠ {M2,M3}

Joint likelihoods

	V1	V2	V3	loglik	
1	M1	M2	M3	-257.7	✓
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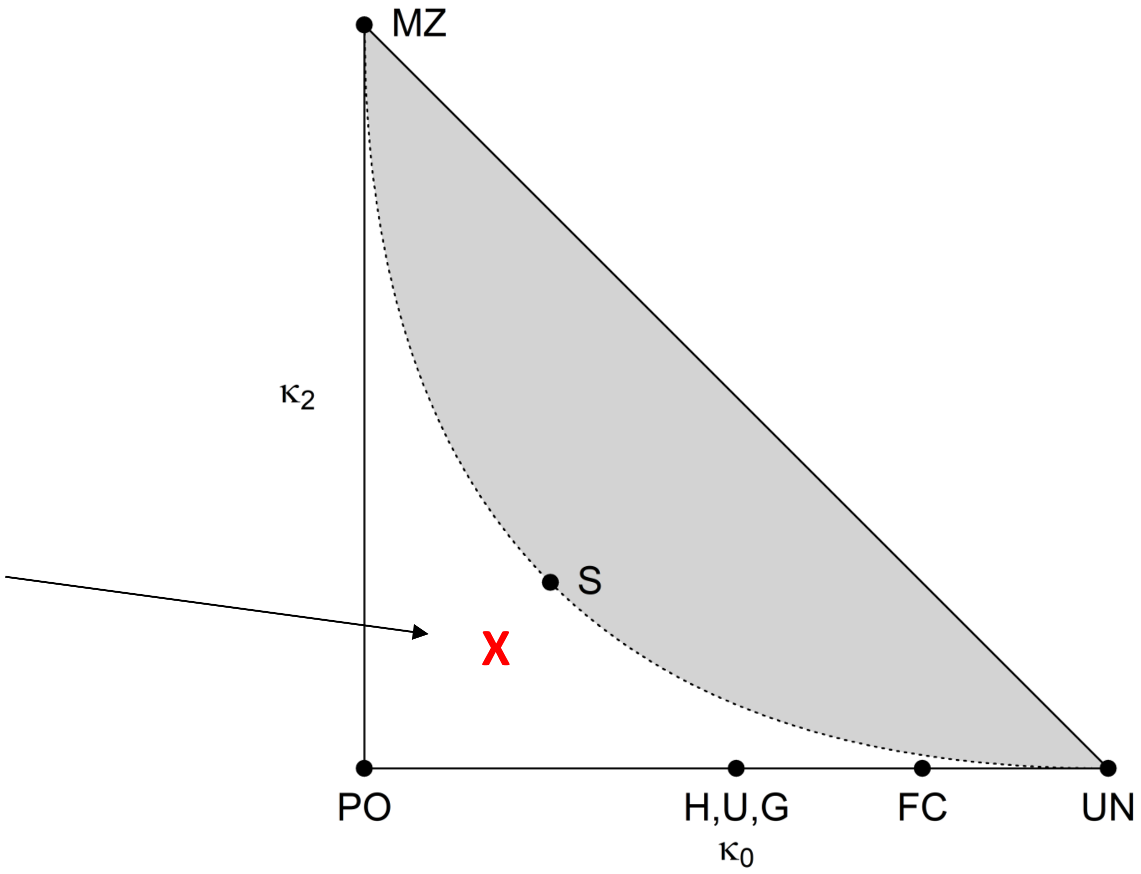
$$\text{GLR} = 2.9 \times 10^6$$

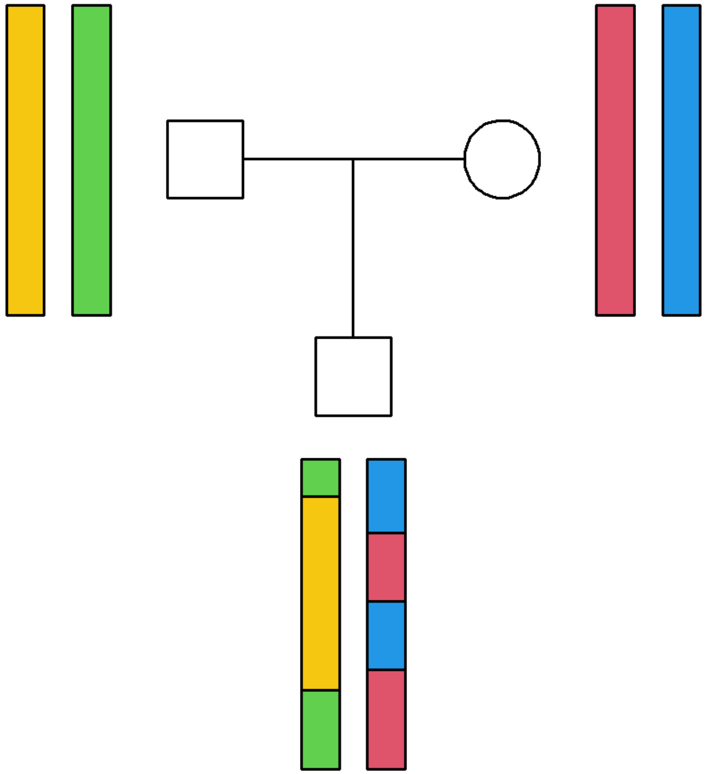
Strong evidence!

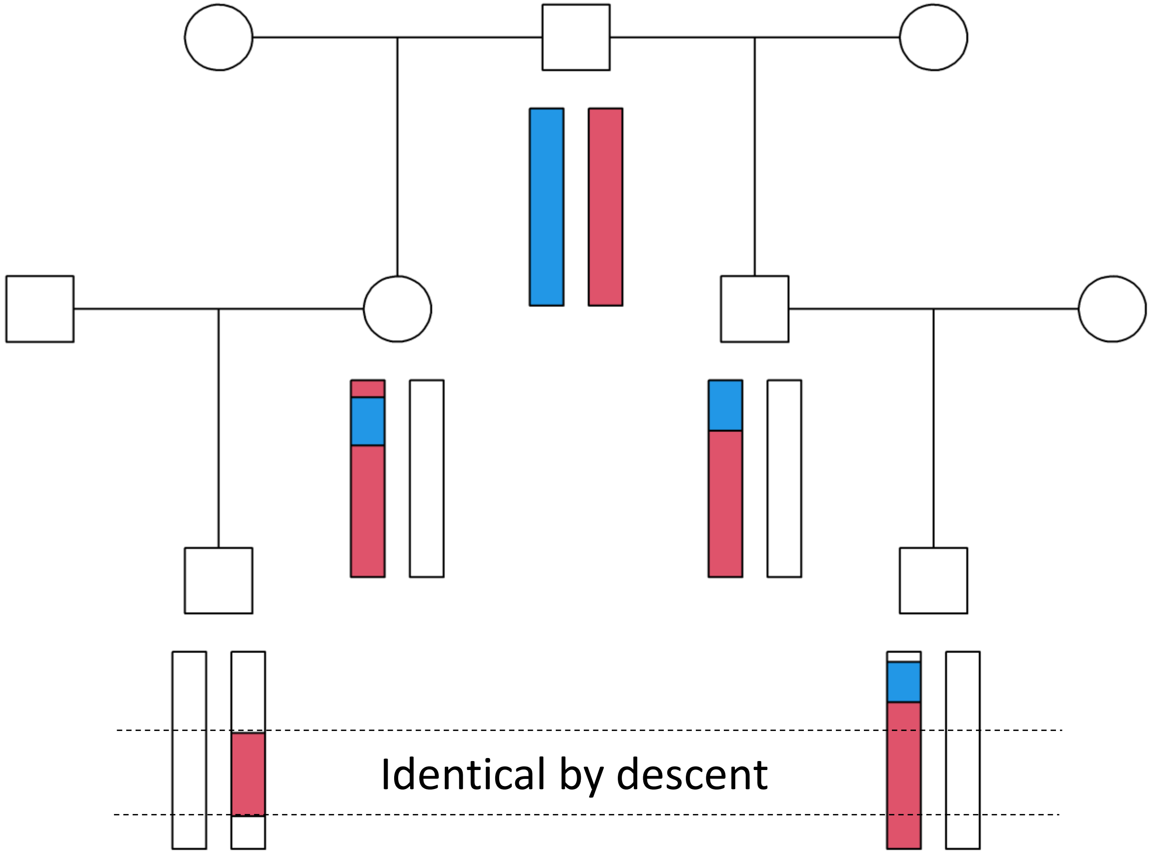
Conclusion: V2 and V3 are M2 and M3 (but we don't know who-is-who)

The relatedness triangle

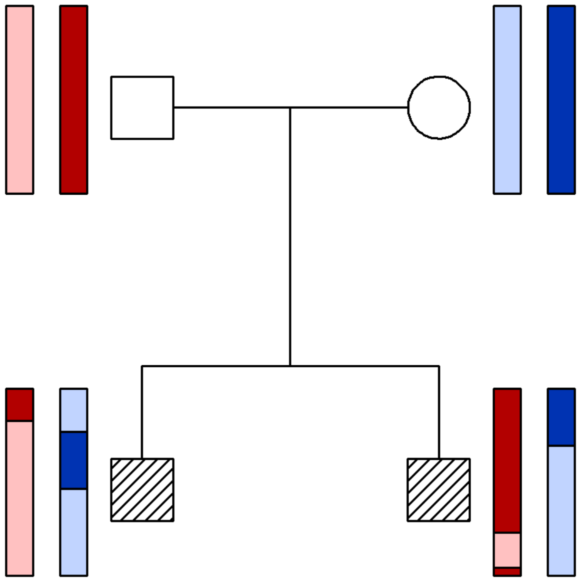


Identity by descent





The "kappa" coefficients

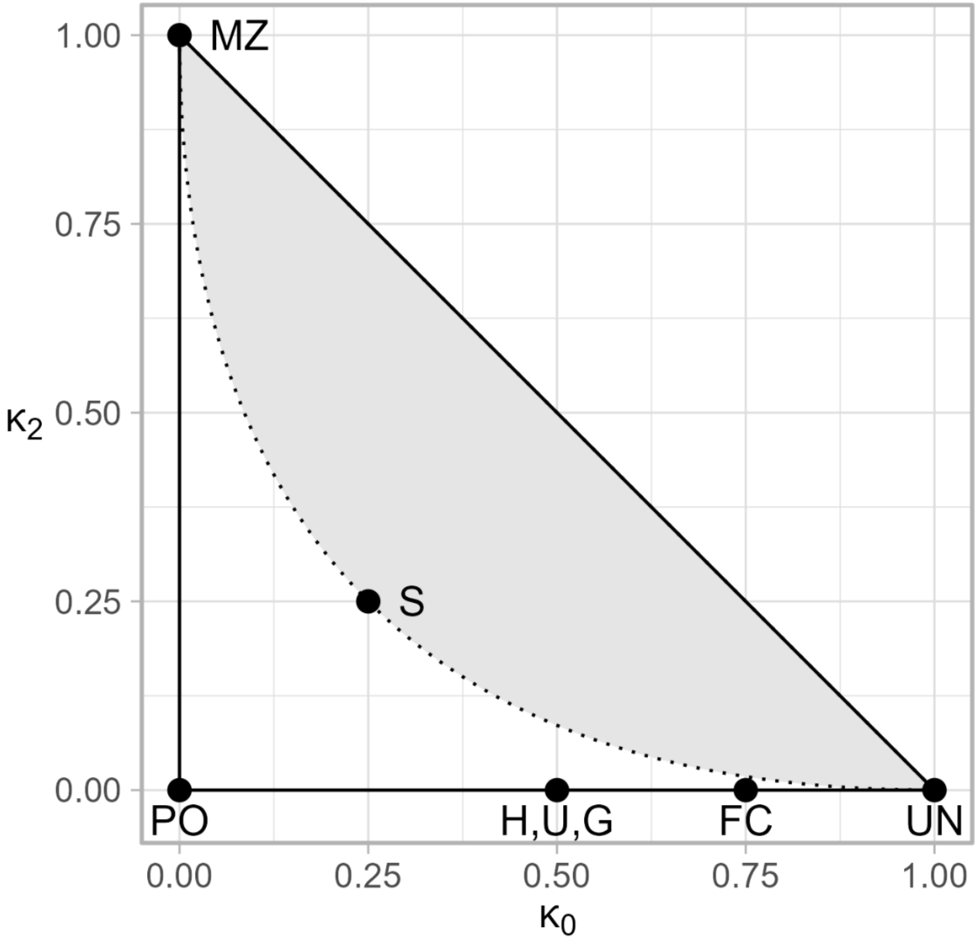


K_0 = proportion of genome with IBD=0

K_1 = proportion of genome with IBD=1

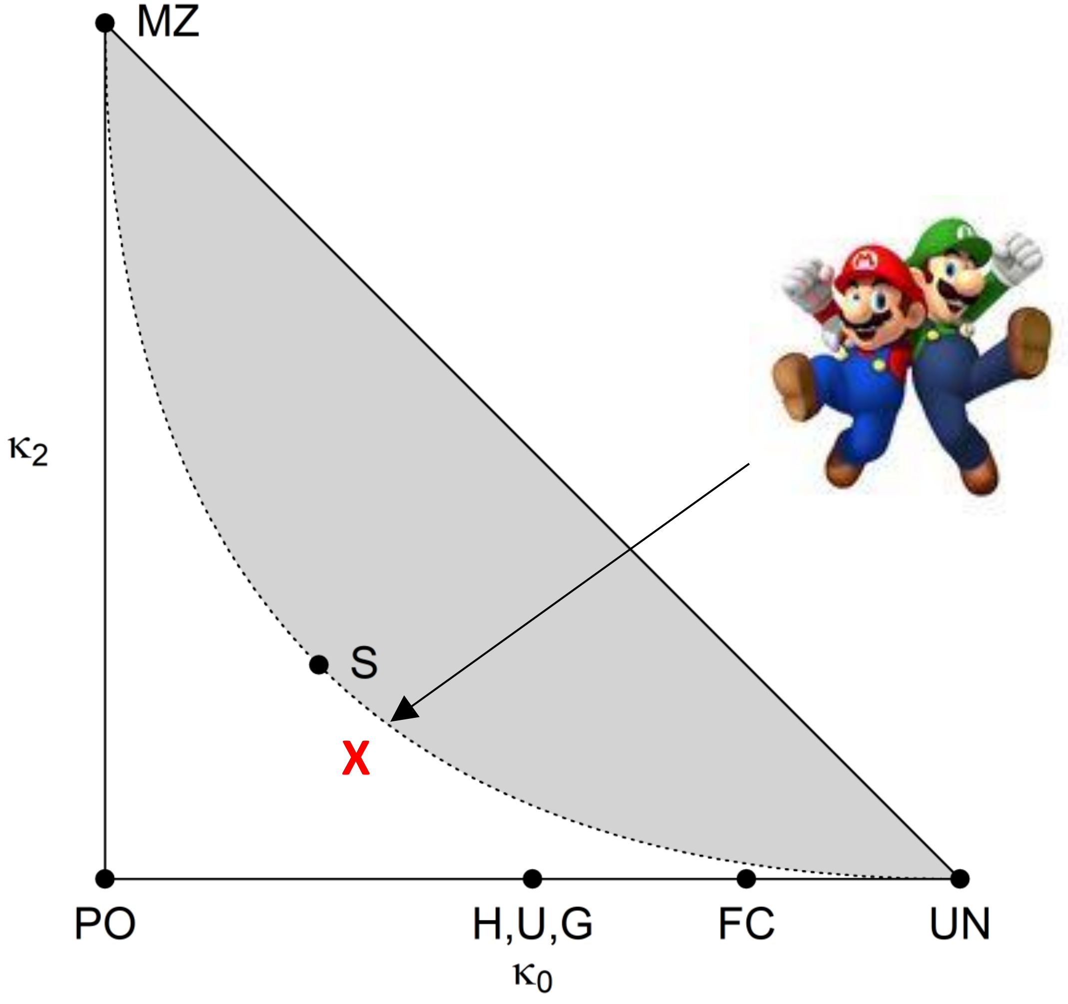
K_2 = proportion of genome with IBD=2

$$K_0 + K_1 + K_2 = 1$$

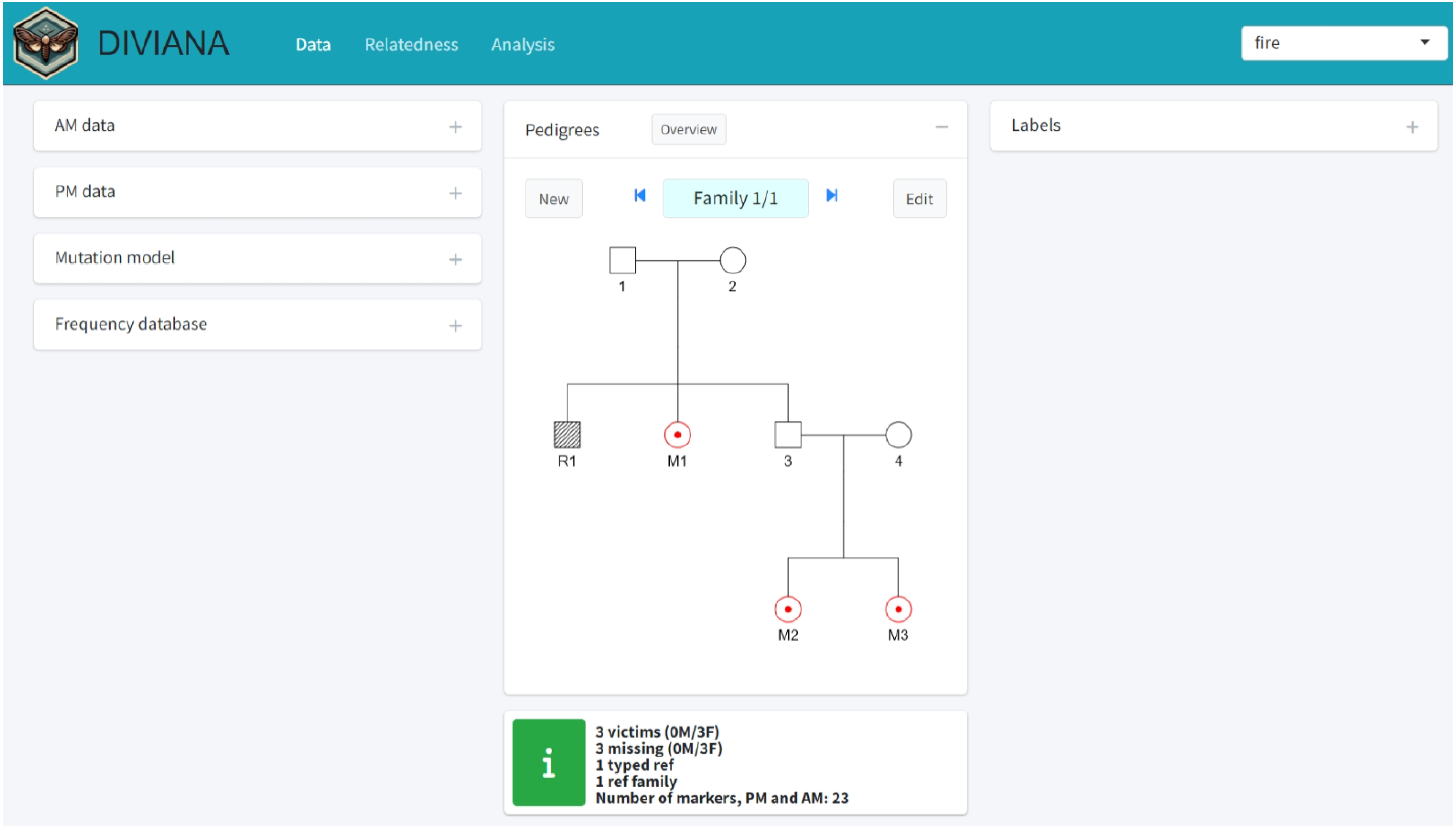


Maximum likelihood estimation

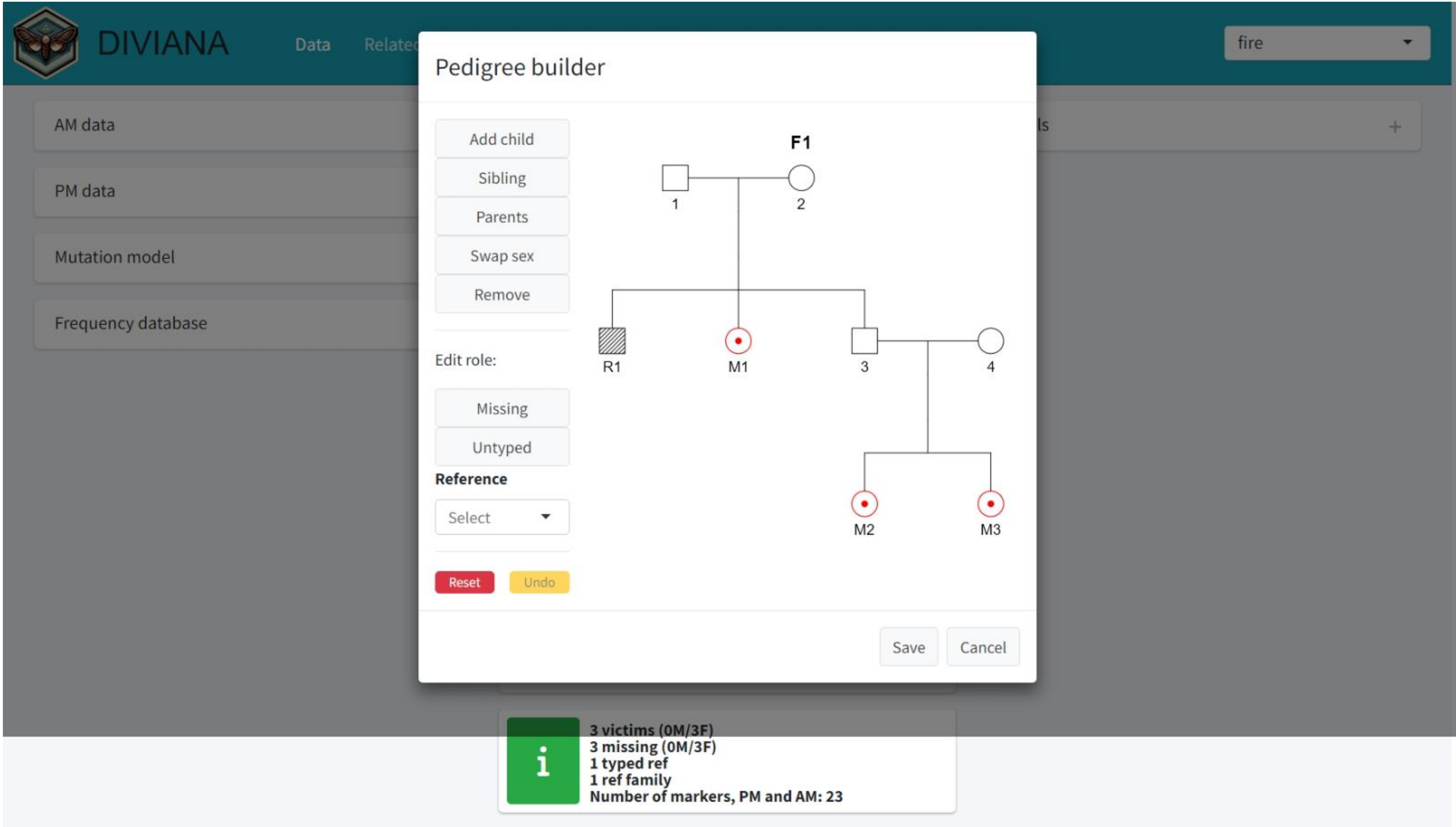
- Thompson (1975)
- Input: Genotypes from two individuals
- Output: Most likely point in the triangle



DIVIANA



3 victims (0M/3F)
3 missing (0M/3F)
1 typed ref
1 ref family
Number of markers, PM and AM: 23



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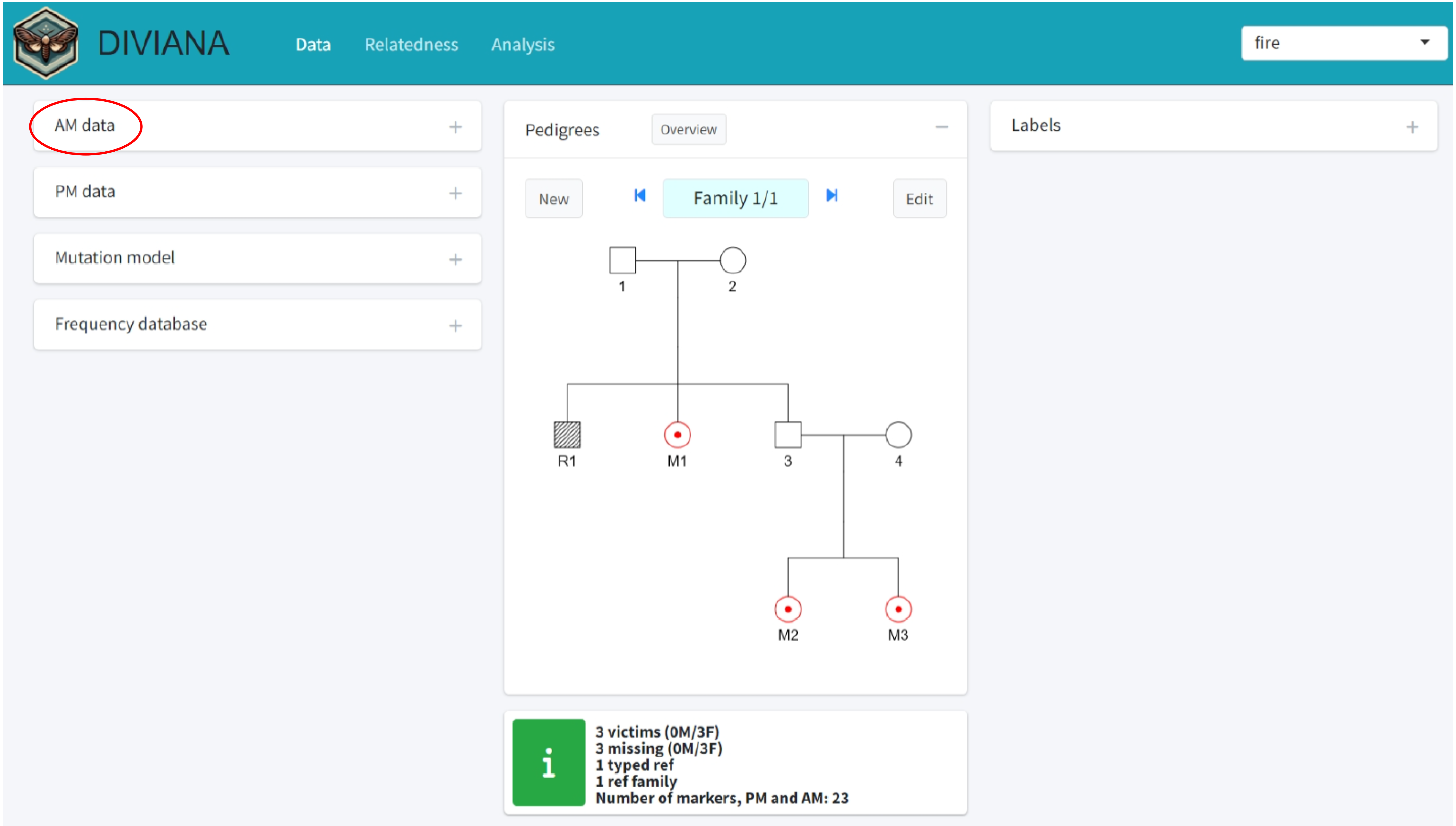

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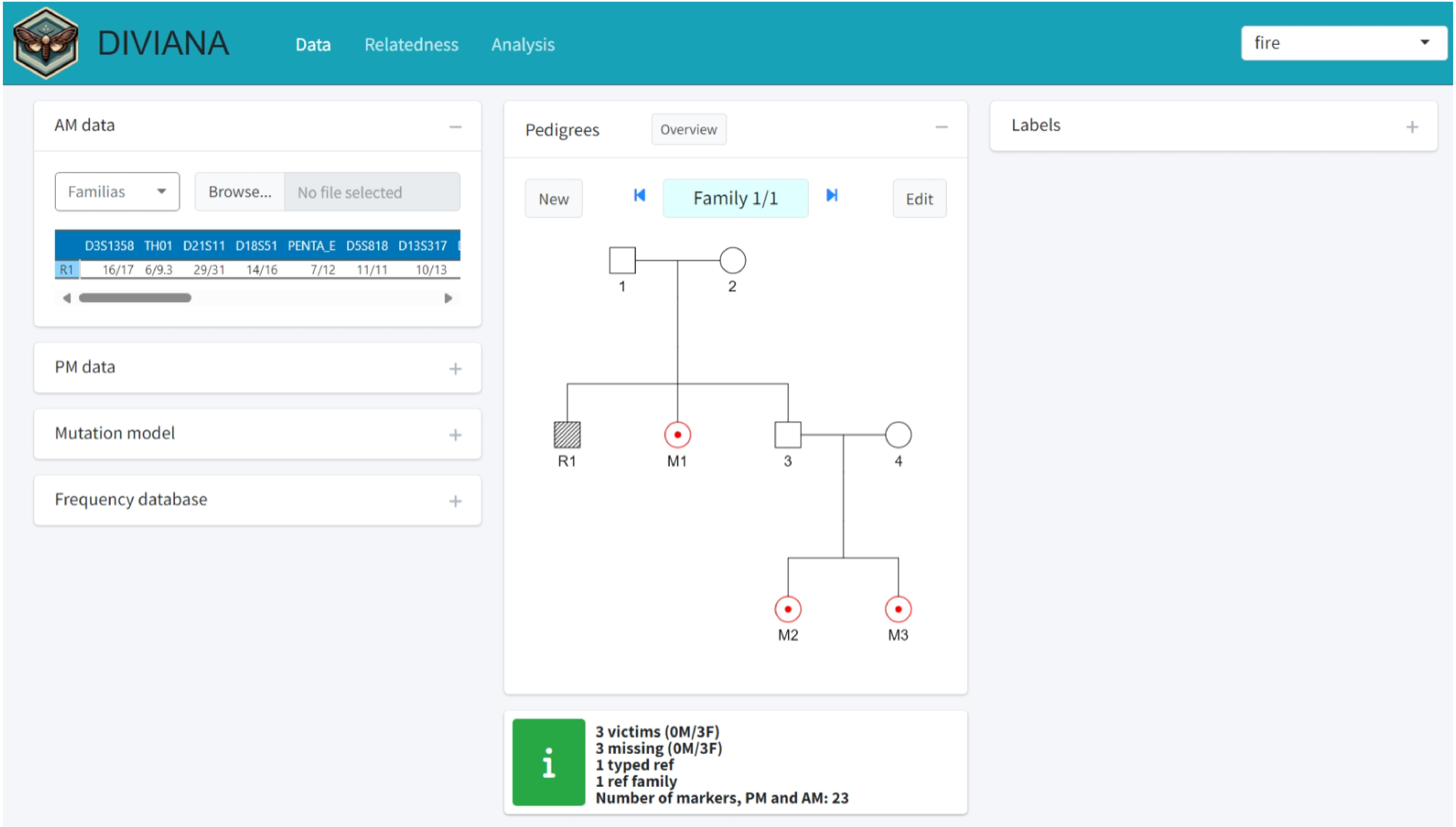

	D3S1358	TH01	D21S11	D18S51	PENTA_E	D5S818	D13S317
R1	16/17	6/9.3	29/31	14/16	7/12	11/11	10/13

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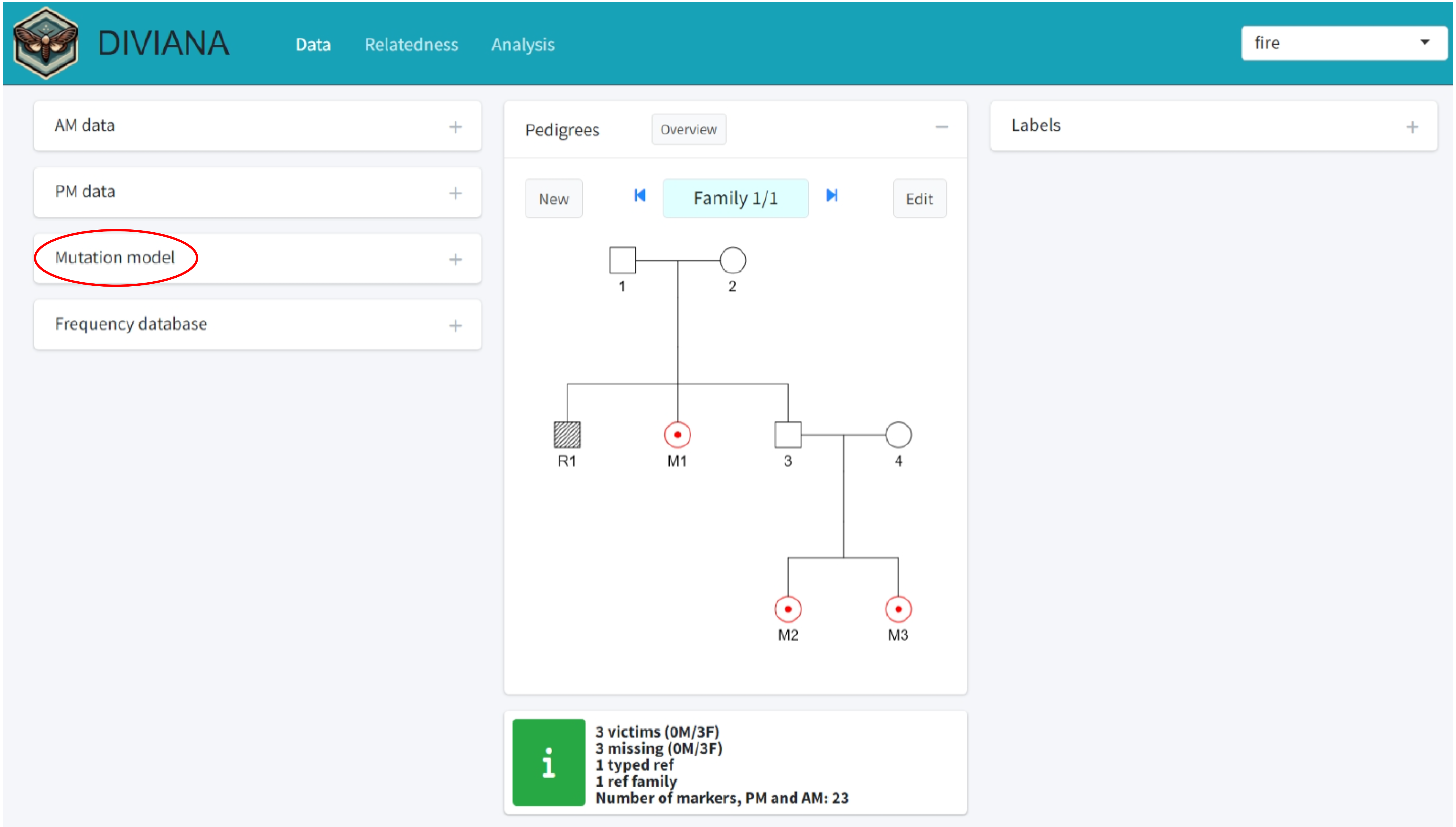

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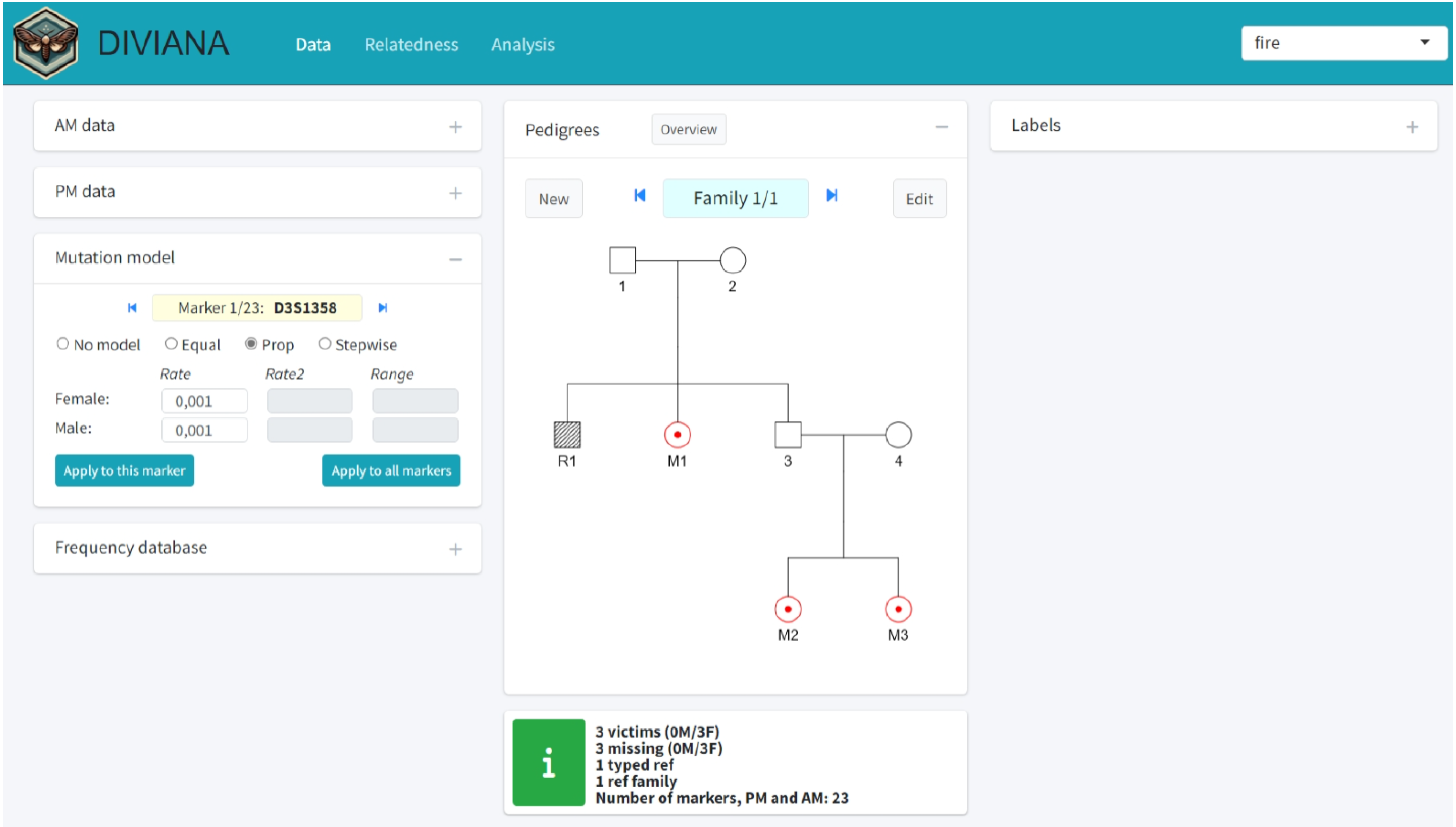

Family 1/1

Marker 1/23: **D3S1358**

○ No model ○ Equal ● Prop ○ Stepwise

	Rate	*Rate2*	*Range*
Female:	0,001		
Male:	0,001		

3 victims (0M/3F)
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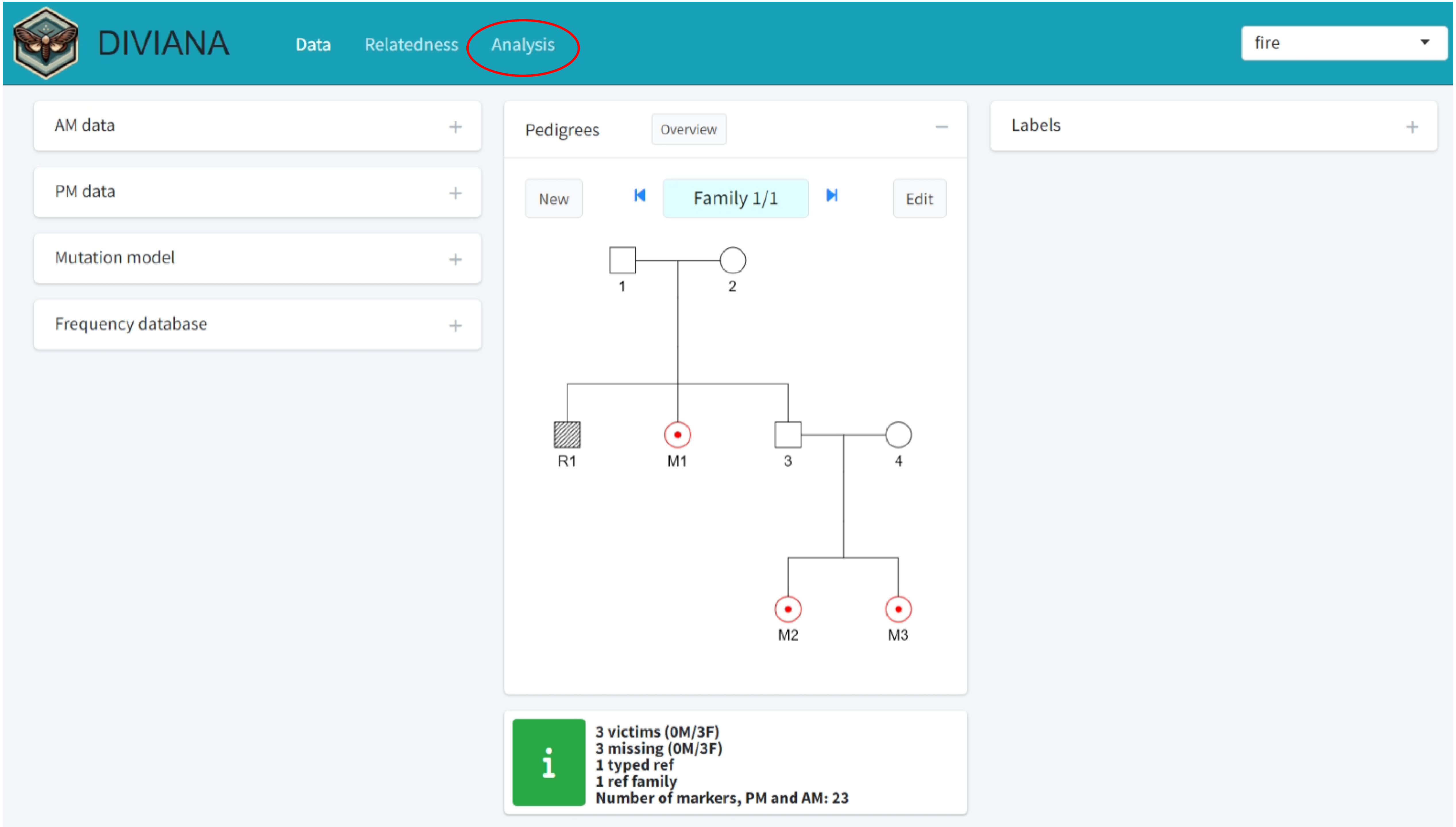
Number of markers, PM and AM: 23



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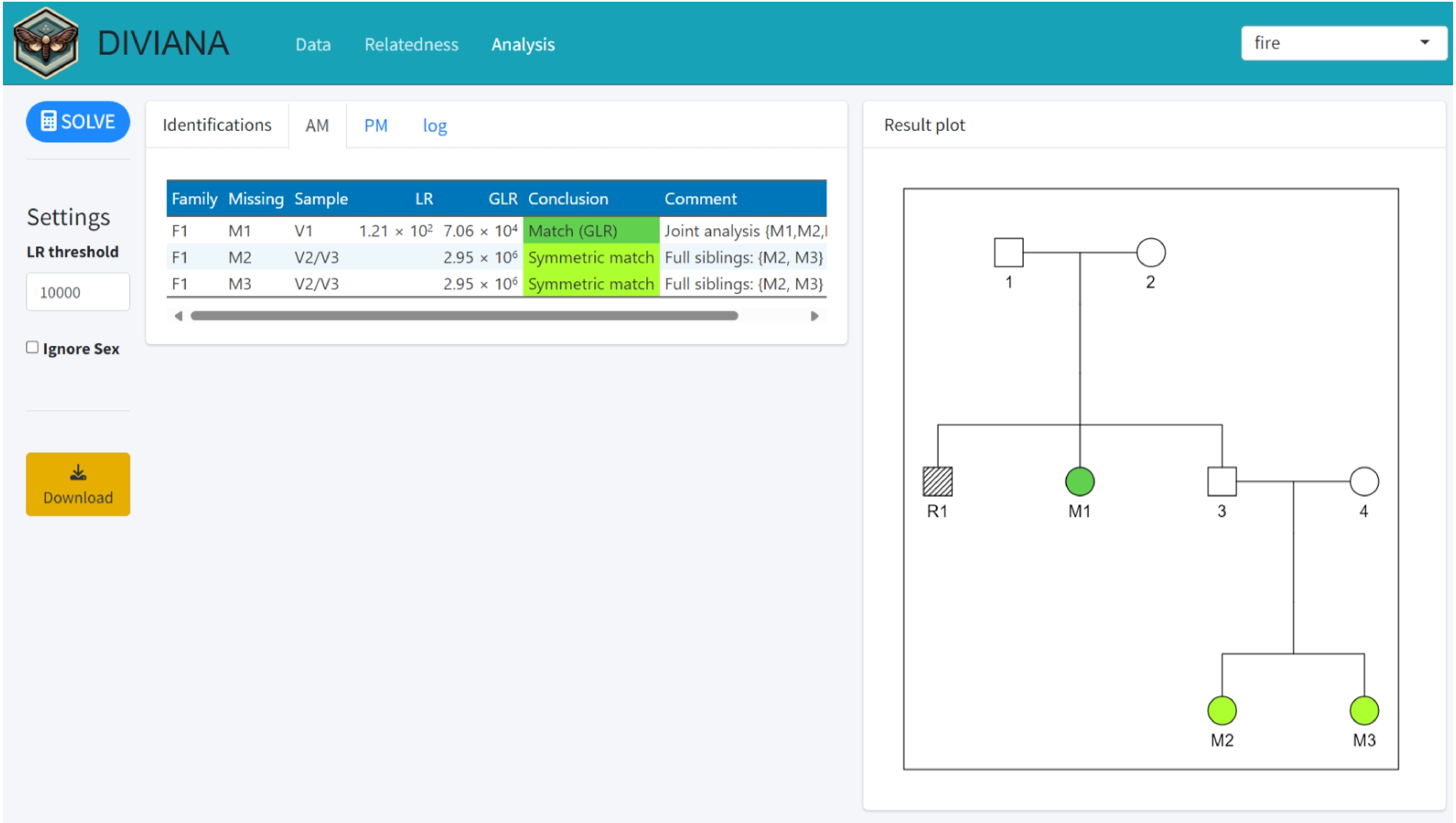


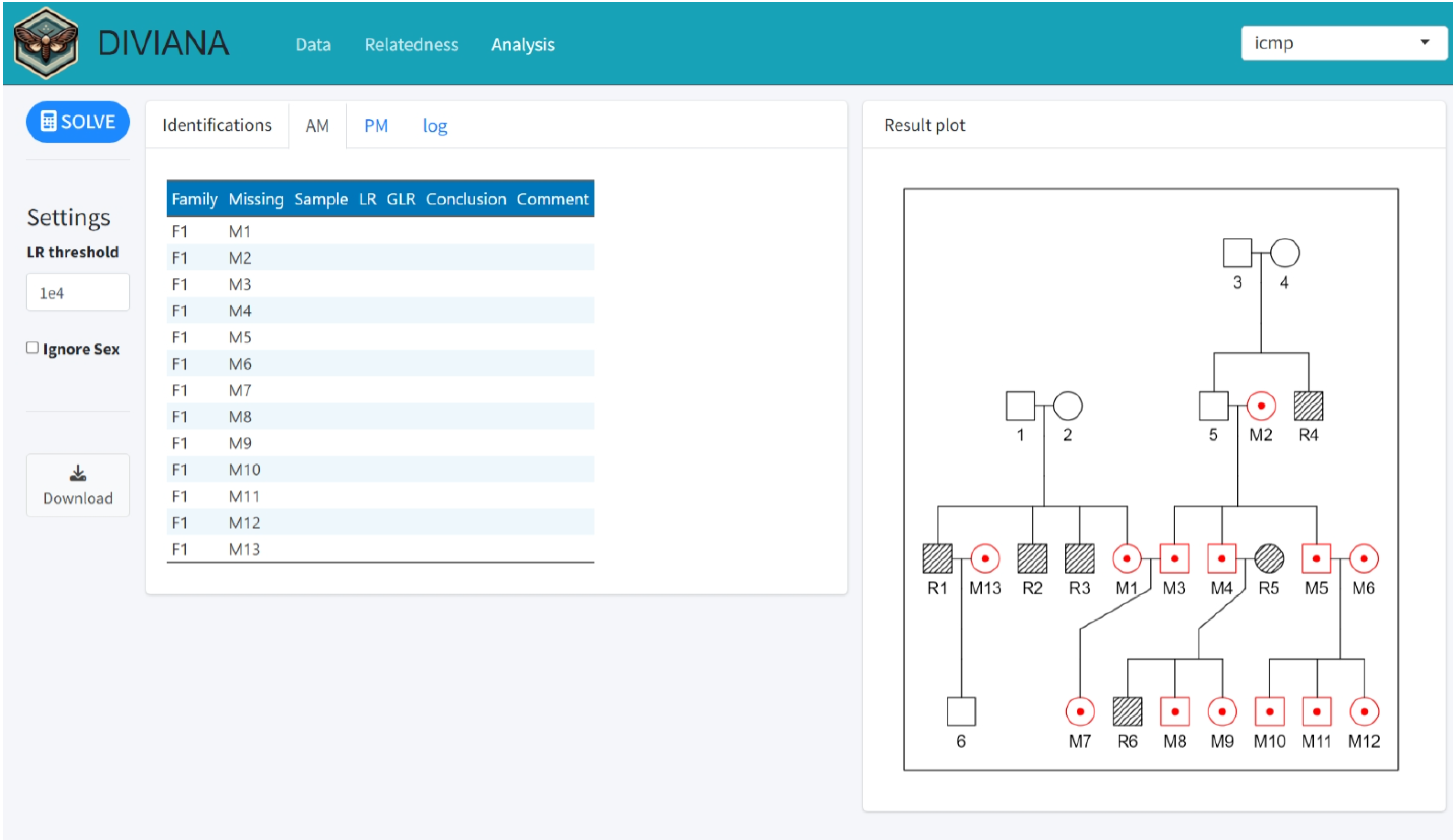
Settings

LR threshold

10000

☐ **Ignore Sex**

Family	Missing	Sample	LR	GLR	Conclusion	Comment
F1	M1	V1	1.21×10^{2}	7.06×10^{4}	Match (GLR)	Joint analysis {M1,M2,
F1	M2	V2/V3		2.95×10^{6}	Symmetric match	Full siblings: {M2, M3}
F1	M3	V2/V3		2.95×10^{6}	Symmetric match	Full siblings: {M2, M3}



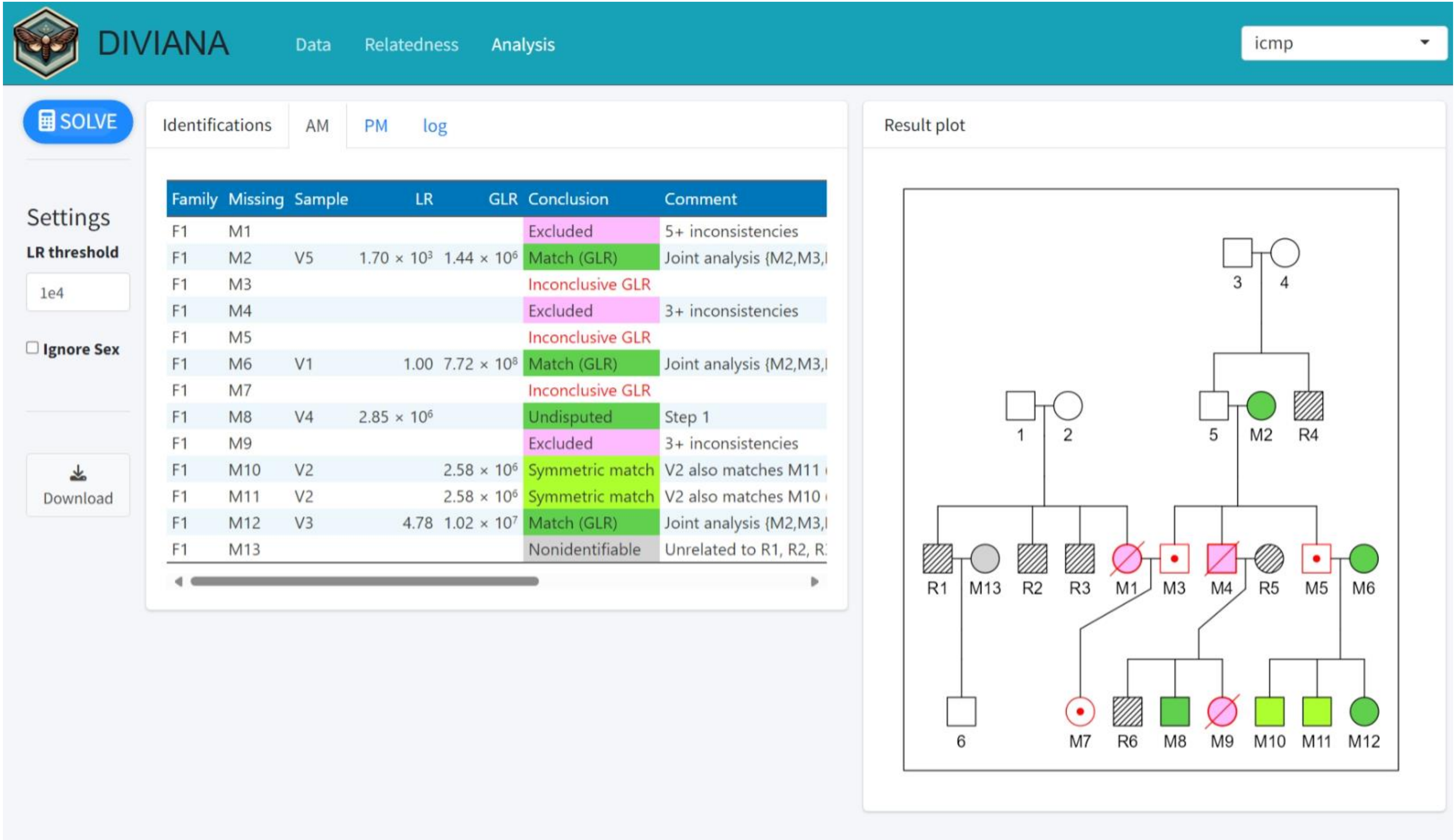
Settings

LR threshold

1e4

☐ **Ignore Sex**

Family	Missing	Sample	LR	GLR	Conclusion	Comment
F1	M1					
F1	M2					
F1	M3					
F1	M4					
F1	M5					
F1	M6					
F1	M7					
F1	M8					
F1	M9					
F1	M10					
F1	M11					
F1	M12					
F1	M13					



Settings

LR threshold

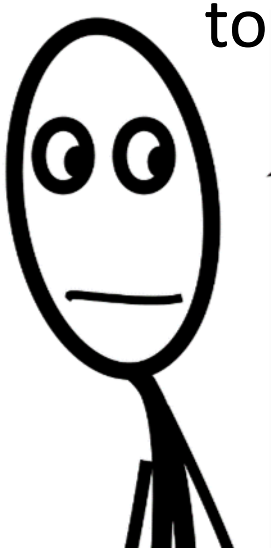
1e4

☐ **Ignore Sex**

Family	Missing	Sample	LR	GLR	Conclusion	Comment
F1	M1				Excluded	5+ inconsistencies
F1	M2	V5	1.70×10^3	1.44×10^6	Match (GLR)	Joint analysis {M2,M3,
F1	M3				Inconclusive GLR	
F1	M4				Excluded	3+ inconsistencies
F1	M5				Inconclusive GLR	
F1	M6	V1	1.00	7.72×10^8	Match (GLR)	Joint analysis {M2,M3,
F1	M7				Inconclusive GLR	
F1	M8	V4	2.85×10^6		Undisputed	Step 1
F1	M9				Excluded	3+ inconsistencies
F1	M10	V2		2.58×10^6	Symmetric match	V2 also matches M11
F1	M11	V2		2.58×10^6	Symmetric match	V2 also matches M10
F1	M12	V3	4.78	1.02×10^7	Match (GLR)	Joint analysis {M2,M3,
F1	M13				Nonidentifiable	Unrelated to R1, R2, R

Live demo!

Gosh, I'd love
to learn more!



Beta version online

https://magnusdv.shinyapps.io/diviana

GitHub

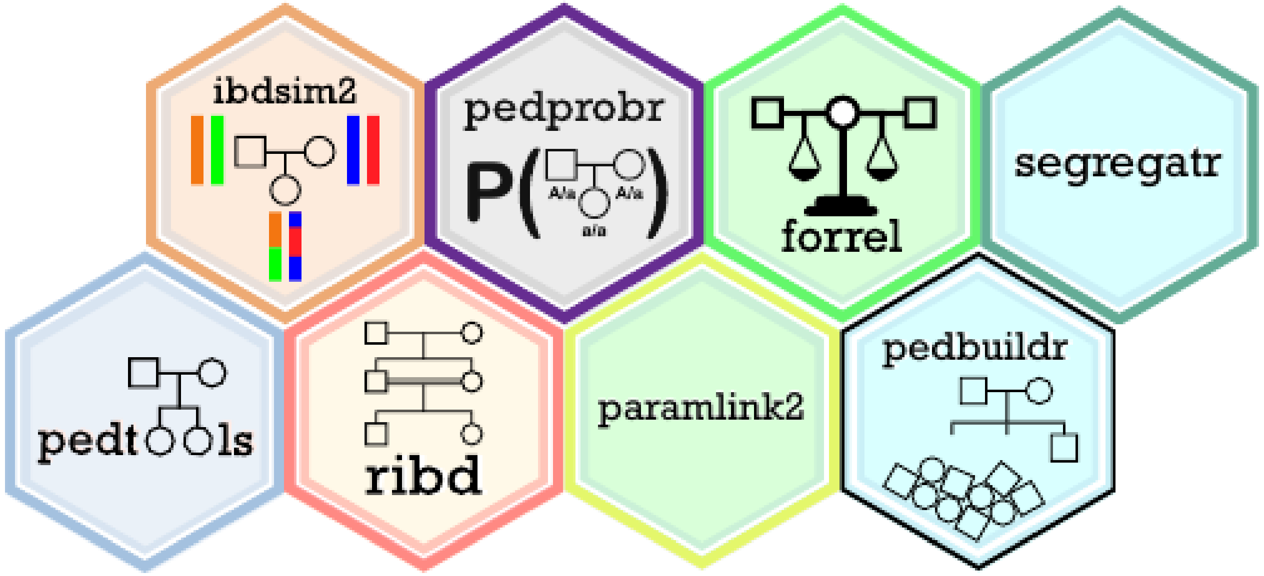
https://github.com/magnusdv/diviana

GLR paper

Kinship cases with partially specified hypotheses

Egeland & Vigeland (submitted, in review)

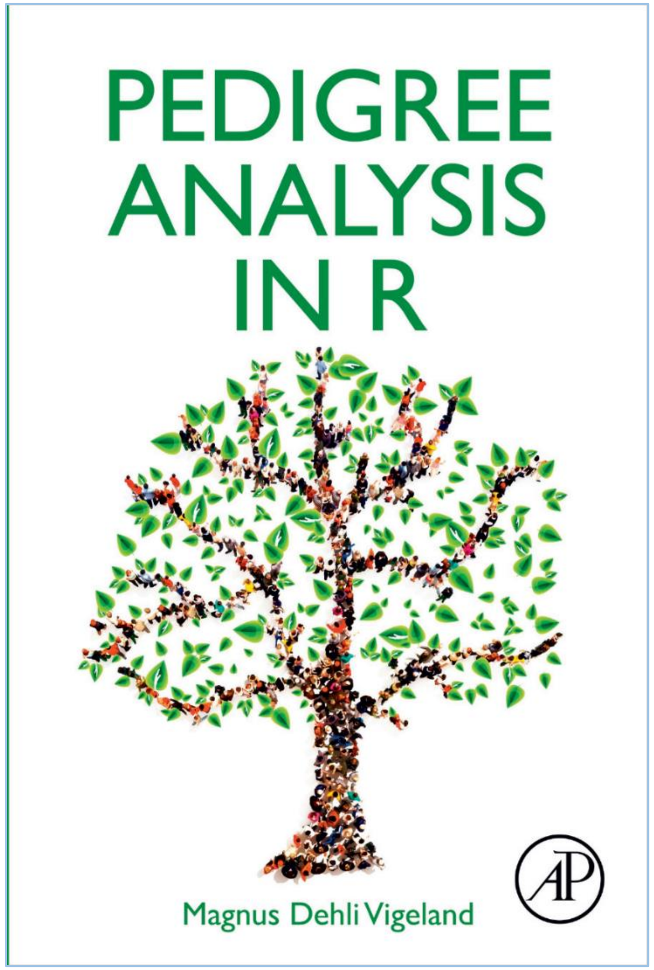
The **pedsuite**:
A collection of packages for pedigree analysis in R



Home page: https://magnusdv.github.io/pedsuite

Source code: https://github.com/magnusdv

Developers: Magnus D. Vigeland, Thore Egeland



PEDIGREE ANALYSIS IN R

Magnus Dehli Vigeland

Diviana eudoreella