

ISFG-GHEP Online School 2024
October 7, 14, 21, 28

Kinship and pedigree analysis: Methods and applications

Teachers

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Oct 7: Theory of relatedness

- 16:00–17:00 [Introduction to pedigrees, QuickPed and R](#) (MDV)
- 17:00–17:45 [Exercises I.](#) ([Solutions](#))
- 17:45–18:00 *Break*
- 18:00–19:00 [Measures of relatedness](#) (MDV)
- 19:00–19:45 [Exercises II.](#) ([Solutions](#))
- 19:45–20:00 *Wrap-up*

Oct 14: Kinship testing

- 16:00–17:00 [Introduction to forensic kinship testing](#) (TE)
- 17:00–17:45 [Exercises III.](#) ([Solutions](#))
- 17:45–18:00 *Break*
- 18:00–19:00 [Kinship testing with Families](#) (TE)
- 19:00–19:45 [Exercises IV.](#) File needed: [kinship-riddle.fam.](#) ([Solutions](#))
- 19:45–20:00 *Wrap-up*

Oct 21: Relatedness inference

- 16:00–17:00 [Realised relatedness: Why are some siblings more alike than others?](#) (MDV)
- 17:00–17:45 [Exercises V.](#) ([Solutions](#))
- 17:45–18:00 *Break*
- 18:00–19:00 [Pedigree reconstruction](#) (MDV)
- 19:00–19:45 [Exercises VI.](#) ([Solutions](#))
- 19:45–20:00 *Wrap-up*

Oct 28: Disaster victim identification

- 16:00–17:00 [DNA-based disaster victim identification](#) (TE)
- 17:00–17:45 *Exercises VII*
- 17:45–18:00 *Break*
- 18:00–19:00 [Practical DVI with Diviana](#) (MDV)
- 19:00–19:45 *Exercises VIII*
- 19:45–20:00 *Wrap-up*

Home page

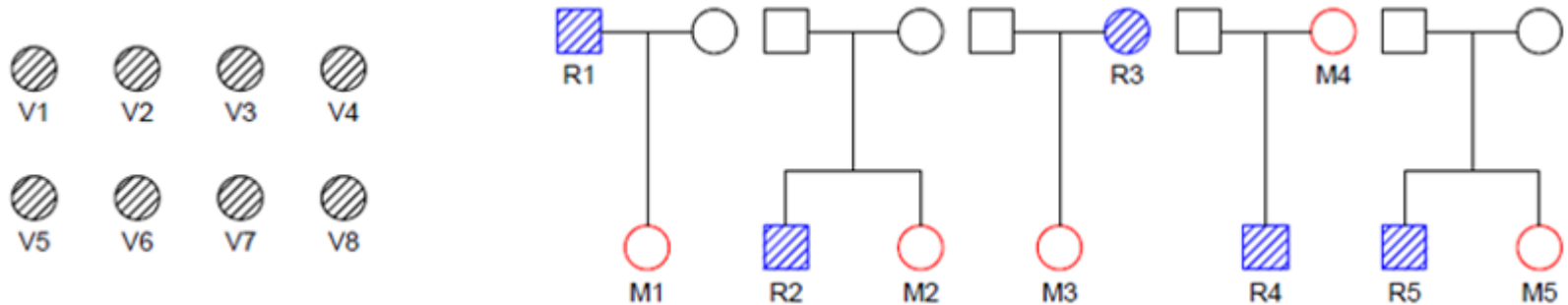
https://magnusdv.github.io/pedsuite/articles/web_only/course-ghep2024.html



+ **dvir**

DNA based disaster victim identification

Familias DVI module



Thore Egeland

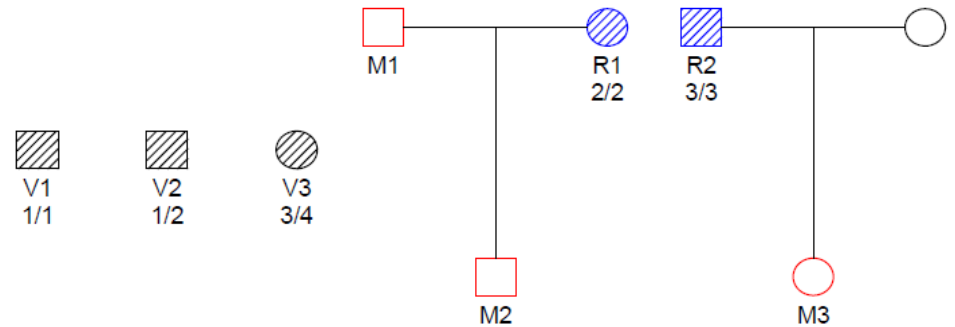
ISFG-GHEP Online School 2024

Kinship and pedigree analysis: Methods and applications

Disaster Victim Identification (DVI)

- DVI

- Match list of unidentified persons against a list of missing persons
- Special case:
 - Missing Person Identification



- Cases

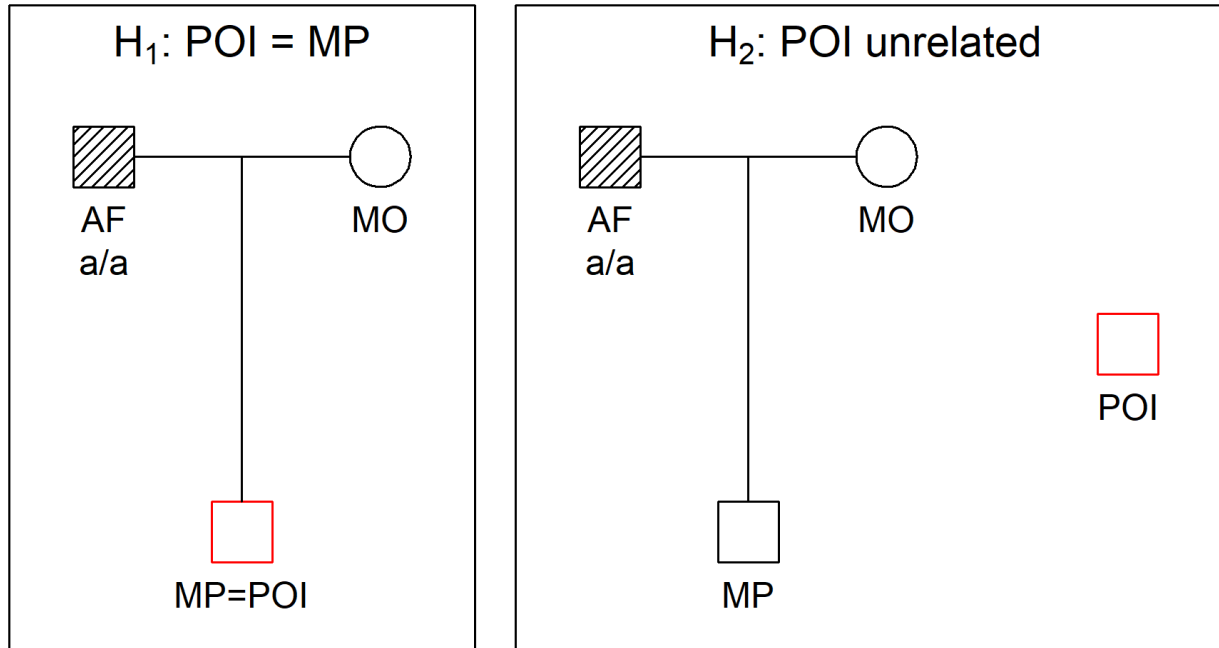
- Missing grandchildren of Argentina
- World Trade Center attack
- Spitsbergen civil aircraft disaster
- Balkan conflicts
- Drowned immigrants
- Thailand tsunami

- Methods and software

- Exclusion and inclusion power
- Treat *each* victim (PM approach) or *each* family (AM approach) at a time
- Joint approach. GLR
 - library(dvir)

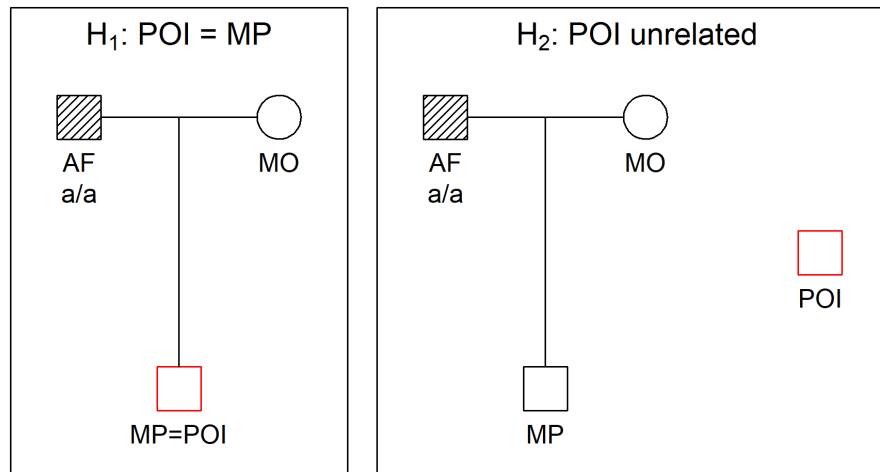


First: Missing Person Identification



- Special case of DVI:
 - ✓ Only one *missing person* (MP) in each reference family
 - ✓ Possibly many reference families and many *persons of interest* (POIs)
 - ✓ Each case treated independently

Exclusion power



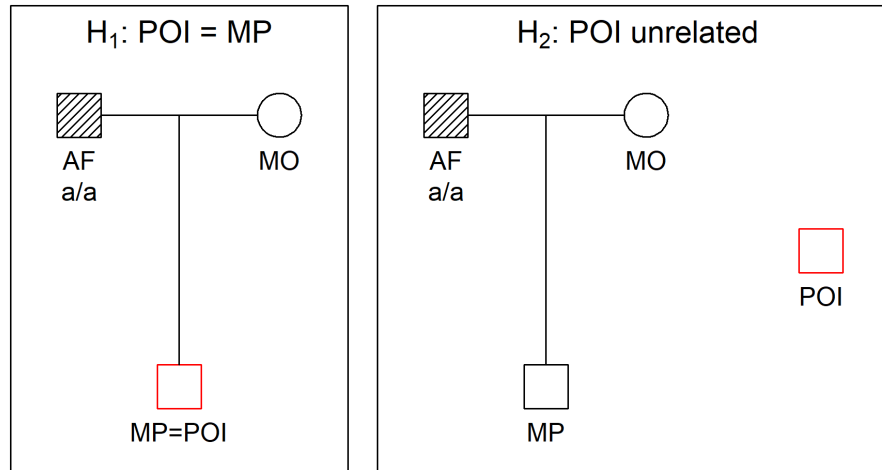
- Sufficient data to *exclude* an unrelated POI?
- **Exclusion power**

$$EP = P(\text{data incompat. with ped} \mid POI \text{ unrelated})$$

- Can be computed exactly
(Egeland, Pinto, Vigeland, 2014).

In forrel:
> exclusionPower()
> missingPersonEP()

Inclusion power

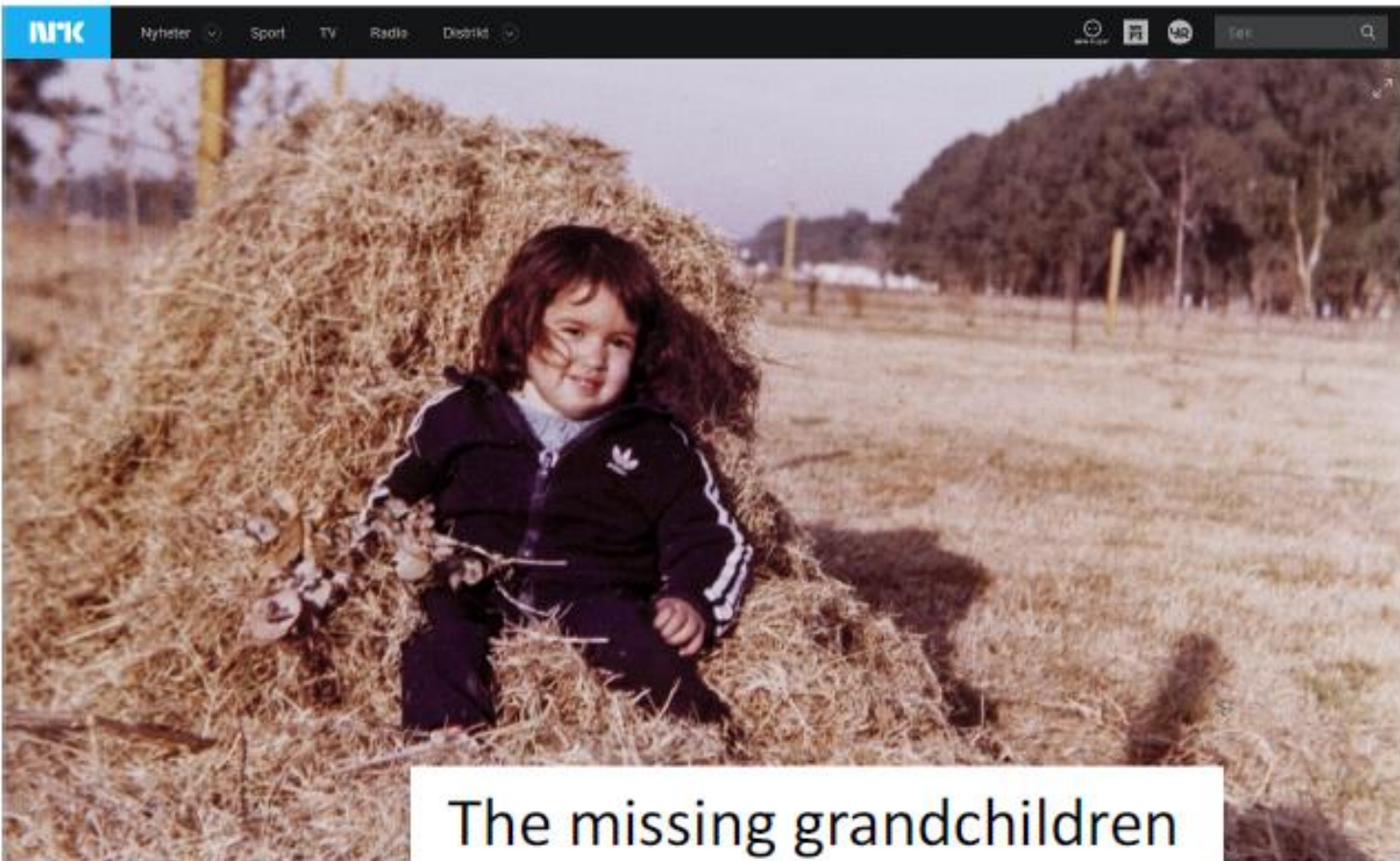


- Sufficient data to give a positive match if POI = MP?
- **Inclusion power** (or *exceedance probability*):

$$IP_{10000} = P(LR > 10000 \mid POI = MP)$$

- Can be computed by simulation!
 - Unconditional → average for all pedigrees of this type
 - Conditional → probability for this particular case

In forrel:
> missingPersonIP()



The missing grandchildren of Argentina

Argentina 1976 – 1983: Dirty War

- Military dictatorship
- *War* against guerrillas

- Opponents killed or disappeared

- *About 500 children abducted*
 - kidnapped with their parents or born in captivity
 - parents killed
 - raised by police or military families

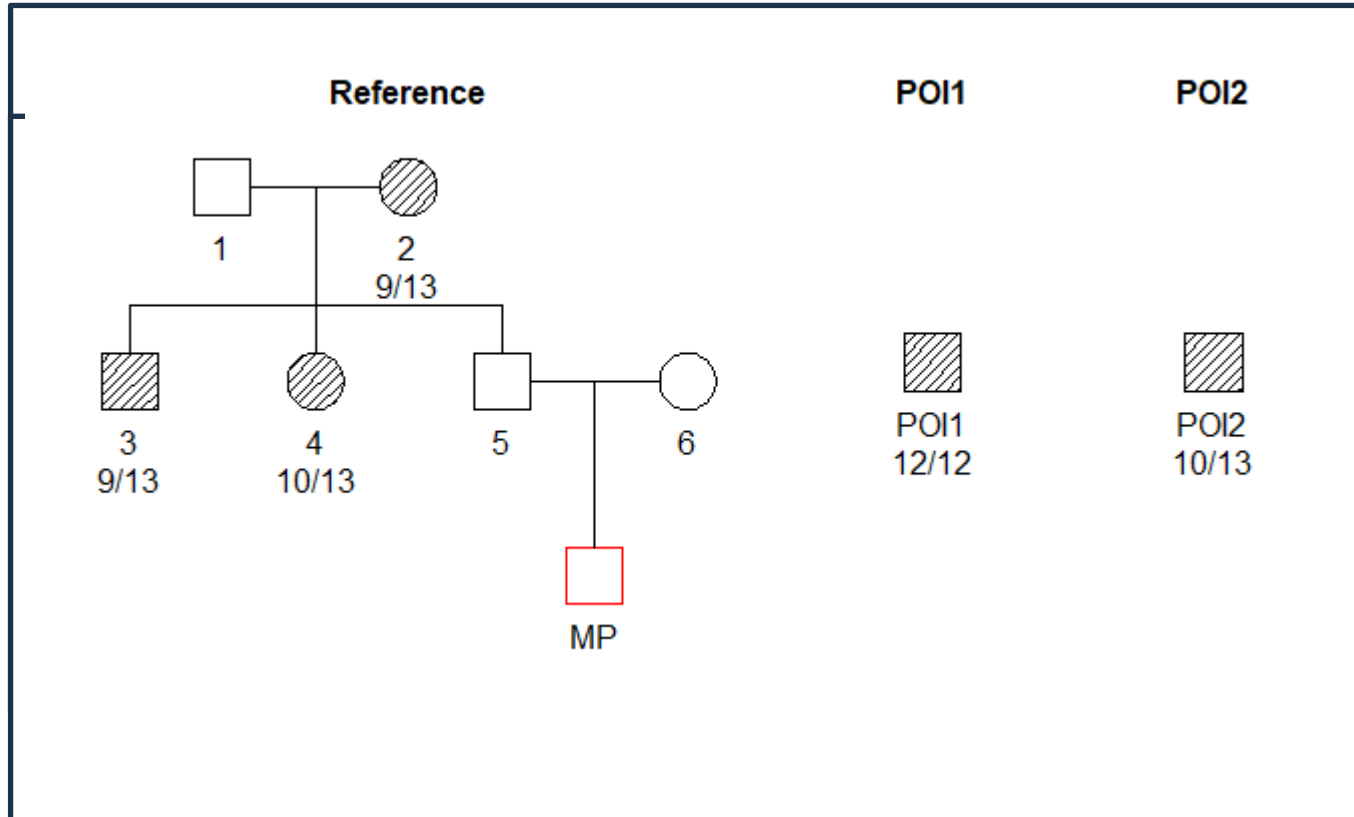


History

- 1977: *Grandmothers of Plaza de Mayo*
 - weekly marches ever since
 - <https://www.abuelas.org.ar>
- 1983: Mary-Claire King, US geneticist
 - Collaboration with the Grandmothers
 - HLA typing + blood groups
 - First grandchild recovered using genetics
- 1987: National Genetic Data Bank (BNDG)
- 2007: Argentine Forensic Anthropology Team
- 2023: In total 132 reunifications by BNDG



Exclusion and inclusion



- POI1 = 12/12 is **excluded** if mutations are ignored since the alleles of his father are among 9/10/13
- POI2 is
 - ✓ **not excluded**
 - ✓ **included** if $LR > 10000$

Power analysis

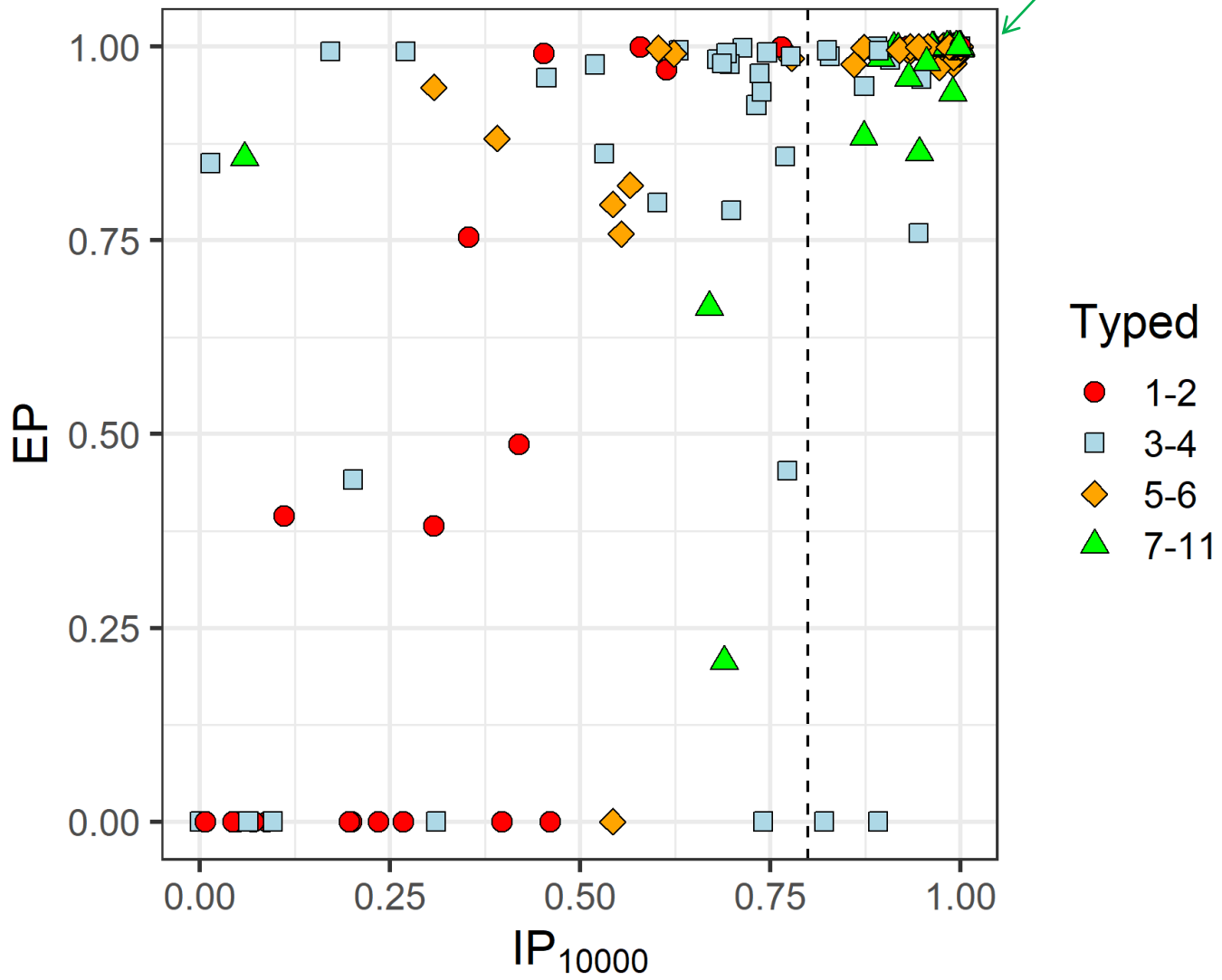
- Power evaluation of ~200 families in the BNDG database
 - most of them unsolved

Typed	Families	Parent(s) typed	2 nd degree only
1	11	5	5
2	20	5	15
3	29	7	20
4	41	6	33
5	29	7	14
6	26	5	14
7	14	0	8
8	14	0	9
9	5	0	3
10+	7	0	5
Total	196	35	126

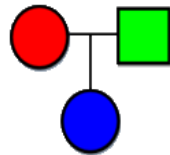
Missing	Total	mtDNA	Y
Male	27	27	25
Female	17	17	-
Unknown	152	142	108
Total	196	186	133

- For each family: Compute IP_{10000} and EP

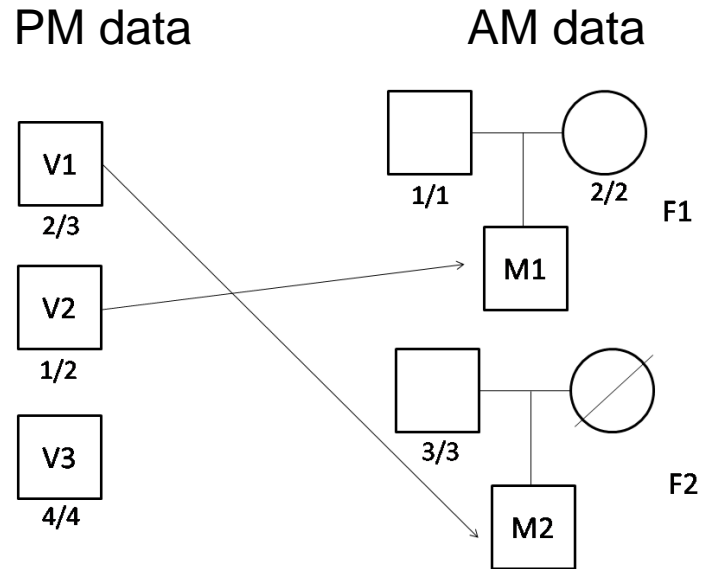
Results



DVI in Familias



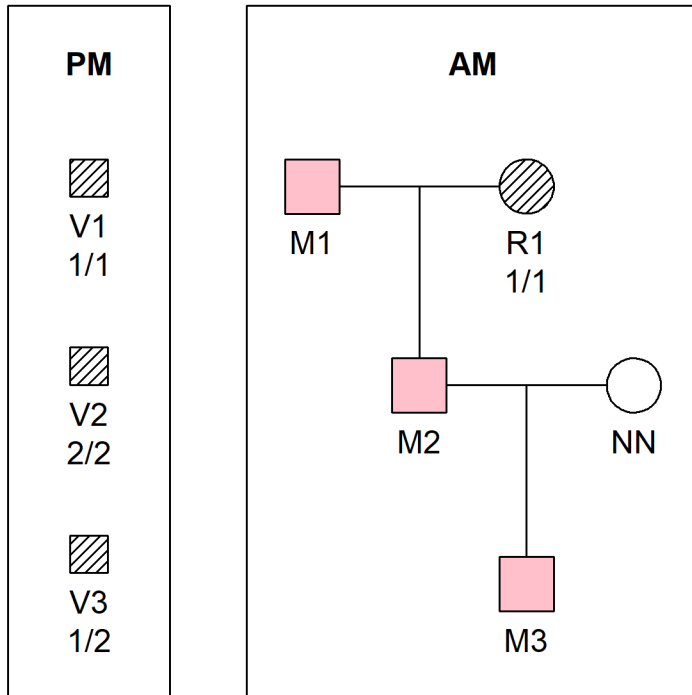
DVI approaches



Approaches

- ✓ **PM driven**: Each PM (victim) sample treated separately
- ✓ **AM driven**: Each AM (missing person) treated separately
- ✓ **Joint**: Simultaneous, consistent solution (dvir, DIVIANA)

Challenge: Joint solution sometimes needed

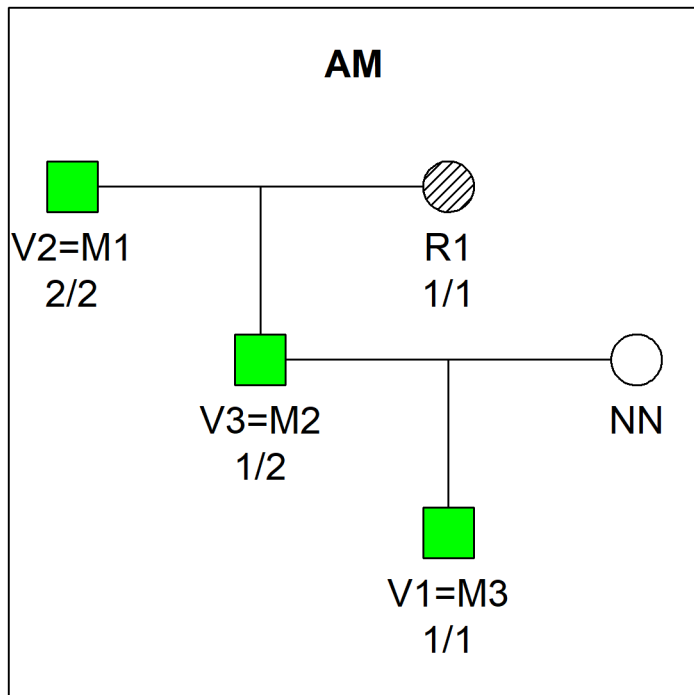


	M1	M2	M3
V1	1	20	10.5
V2	1	0	0.5
V3	1	10	5.5

Pairwise LRs indicate $V1 = M2$, but

- $V1 = M2$ is impossible if $V2$ is among the missing

Joint solution



Joint likelihoods, sorted

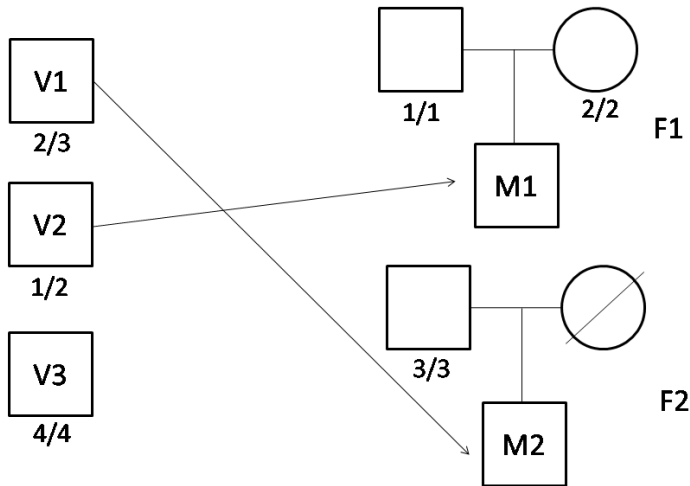
	V1	V2	V3	loglik	LR
1	M3	M1	M2	-15.67181	2000.0
2	M2	*	M1	-17.97439	200.0
3	M2	*	M3	-17.97439	200.0
4	*	M1	M2	-17.97439	200.0
5	M3	*	M2	-18.66754	100.0

6	*	M3	M2	-18.66754	100.0

19	*	*	*	-23.27271	1.0

34	*	M3	*	-Inf	0.0

DVI in Familias, AM or PM approach



- 1. General DNA data:**
A marker with alleles 1, 2, 3, 4 (all with freq = 0.25)
- 2. Unidentified samples (PM):**
Three samples V1, V2 and V3 and their genotypes.
- 3. Reference families (AM):**
Two reference families F1, F2 with missing individuals (sons here) M1 and M2.
Marker data for available relatives (mother and father for F1, only father for F2.)
- 4. Search.** Perform and interpret the results.

Familias - Tutorial

Daniel Kling, Lourdes Prieto
Thore Egeland

http://familias.name/tutorial/familias_tutorial_spanish.pdf
http://familias.name/tutorial/familias_tutorial_english.pdf

Daniel Kling, Lourdes Prieto and Thore Egeland

Contents

1. [Basics. A paternity case in four steps](#)
2. [Complications](#)
3. [R Familias, paramlink, and plotting](#)
4. [Simulation](#)
5. [Blind search](#)
6. [DVI \(Disaster Victim Identification\)](#)
7. [Familial searching](#)
8. [Input files \(formats\)](#)
9. [Output files and reports](#)
10. [Advanced](#)
11. [Miscellaneous](#)

Basic steps

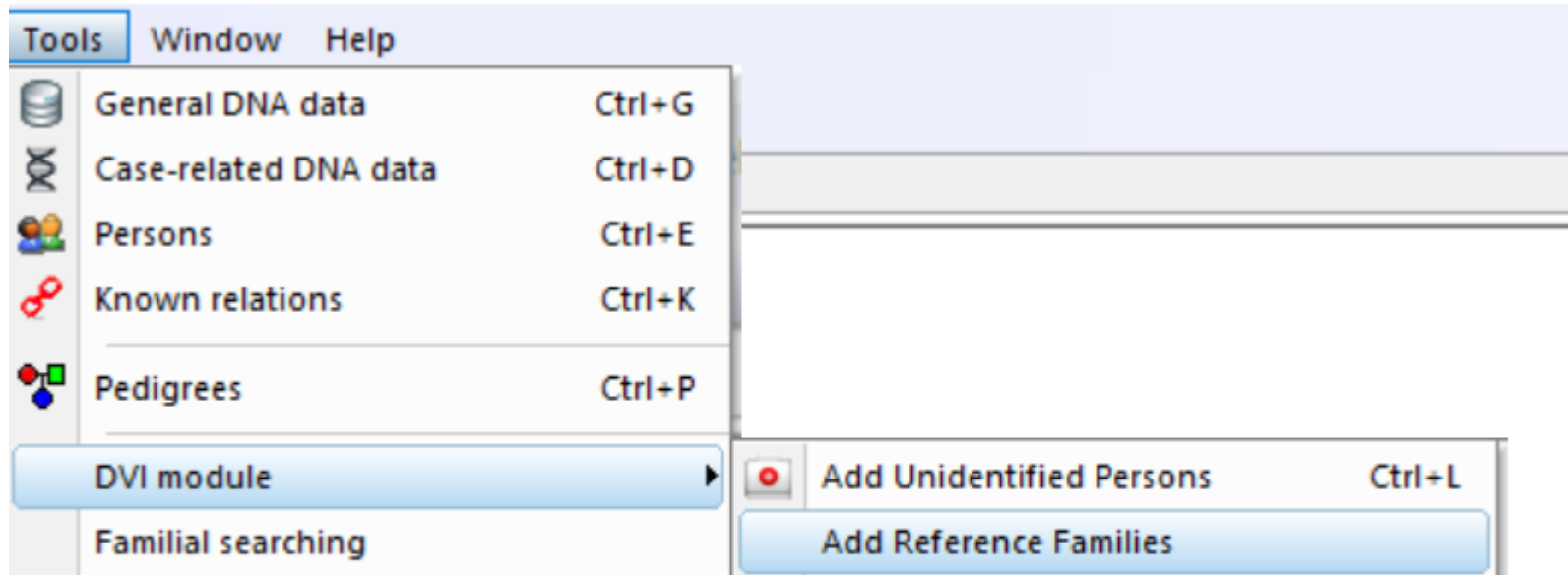
1. **General DNA data.** Input of database, i.e., allele frequencies, etc.
 - ✓ Described in Lecture 4: *Kinship testing with Familias*
2. **Unidentified samples (PM)**
3. **Reference families (AM)**
4. **Search.** Perform and interpret the results.

Step2: Unidentified samples (PM)

The screenshot displays the 'Familias' software interface. The main window is titled 'Familias - Exercise.3.1'. The 'Tools' menu is open, showing options such as 'General DNA data', 'Case-related DNA data', 'Persons', 'Known relations', 'Pedigrees', 'DVI module', 'Familial searching', 'Blind search', and 'Merged profiles'. The 'DVI module' option is selected, and a sub-menu is open, showing 'Add Unidentified Persons', 'Add Reference Families', and 'Search'. The 'Add Unidentified Persons' option is selected, and a dialog box titled 'DVI module - Add unidentified person(s) (PM)' is displayed. The dialog box contains a '#Missing persons' field with the value '3' and a checked 'Use #PM' checkbox. Below the dialog box is a table with the following data:

ID	Gender	DNA data
V1	Male	M1: 2, 3;
V2	Male	M1: 1, 2;
V3	Male	M1: 4, 4;

Step 3: Reference families (AM)



DVI module - Edit reference family



Name:

Persons

Name	Gender	DNA data
Father1	Male	M1: 1, 1;
Mother1	Female	M1: 2, 2;

Pedigrees (Relationships)

Missing person gender:

Missing persons: 1

Name	#Relations
Reference pedigree	2
F1	2

Step 4: Search. Perform and interpret the results

The screenshot shows a software interface with a 'Tools' menu open. The menu items are:

- General DNA data (Ctrl+G)
- Case-related DNA data (Ctrl+D)
- Persons (Ctrl+E)
- Known relations (Ctrl+K)
- Pedigrees (Ctrl+P)
- DVI module** (highlighted)
- Familial searching
- DVI module - Results

Sub-menu items for 'DVI module':

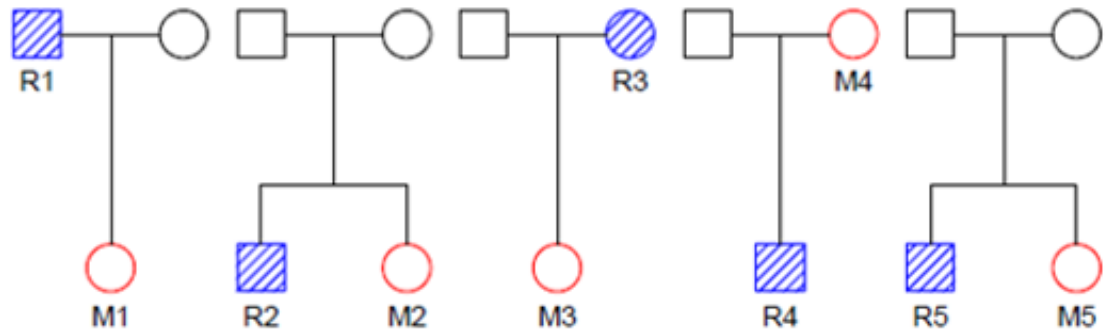
- Add Unidentified Persons (Ctrl+L)
- Add Reference Families

Summary:

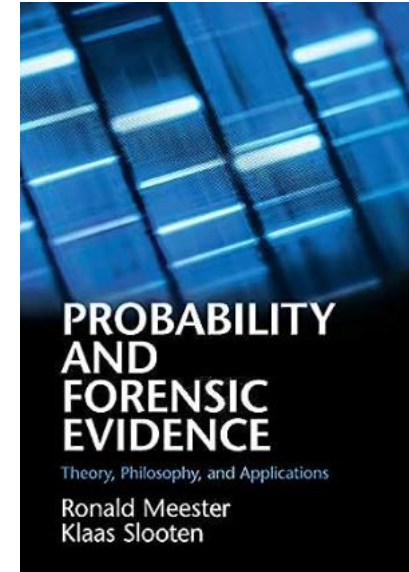
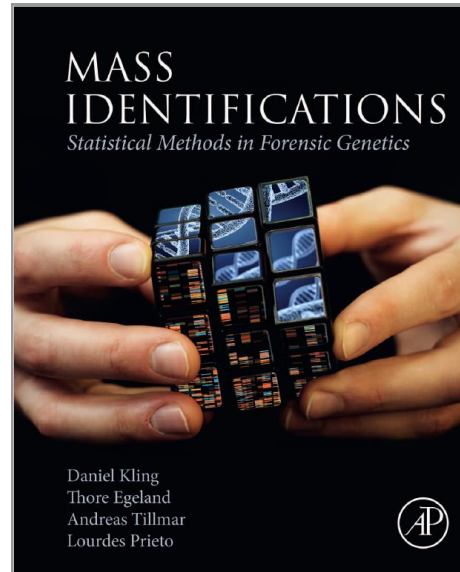
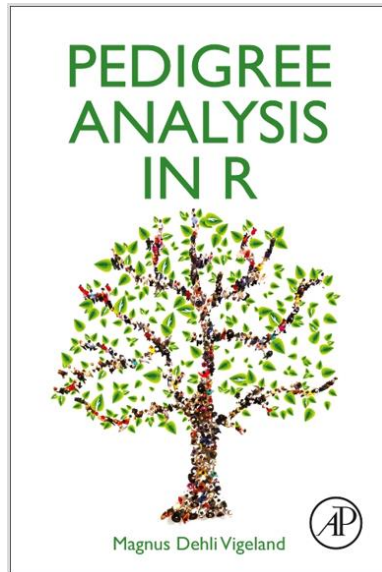
Project name is: Untitled
Number of matches: 6
Number of PM samples: 3
Number of AM families: 2

Family id	Unidentifi...	Prior	Posterior	LR
M1	V1	0.25	0	0
M1	V2	0.25	0.888889	8
M1	V3	0.25	0	0
M2	V1	0.25	0.666667	2
M2	V2	0.25	0	0
M2	V3	0.25	0	0

Exercise: plane crash



Biased selection of references



Article | [Open Access](#) | Published: 01 July 2021

Joint DNA-based disaster victim identification

Magnus D. Vigeland [✉](#) & Thore Egeland

Scientific Reports **11**, Article number: 13661 (2021) | [Cite this article](#)