

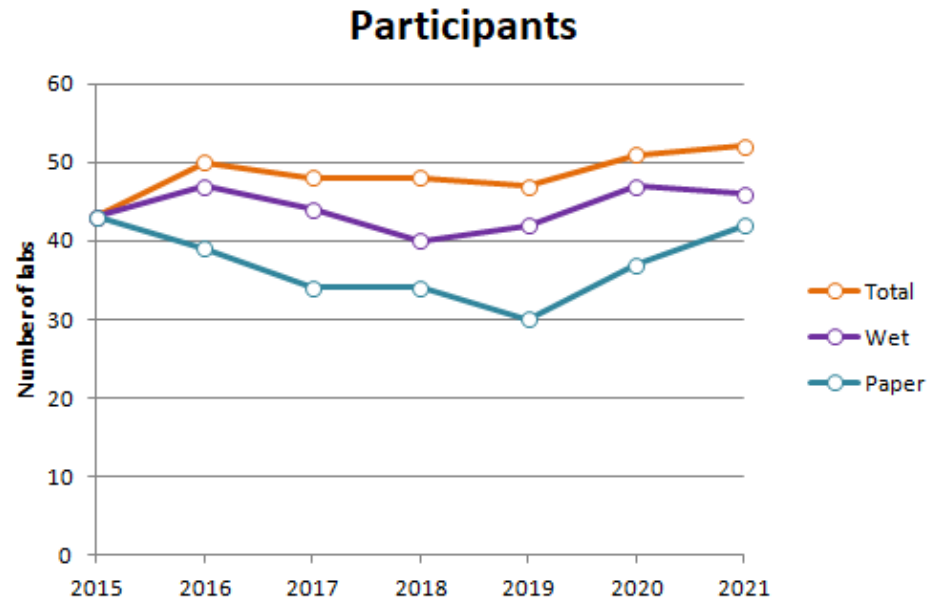
- Proficiency test 2022

Daniel Kling – Head Organizer

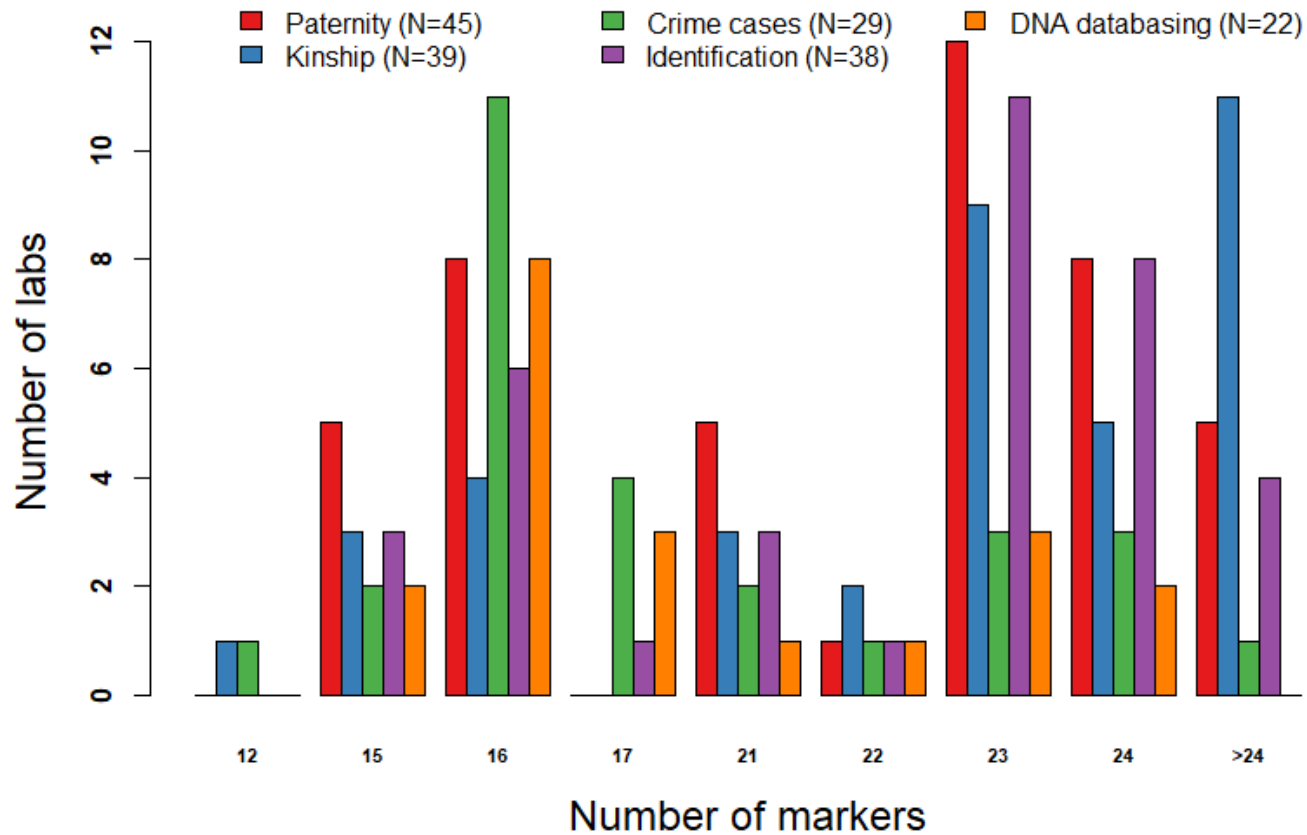
daniel.kling@rmv.se

Summary

- Summarizing statistics for 2021
- 51 labs participated
- 37 completed paper challenge
- 47 completed wet exercise

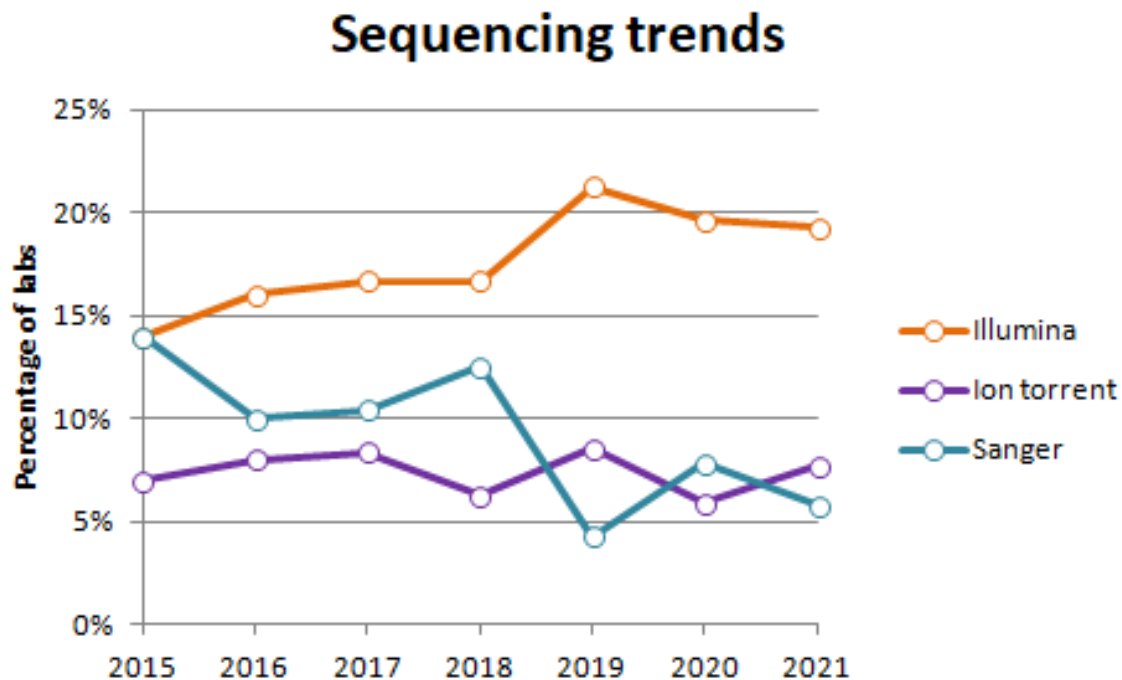


Questionnaire – Markers used



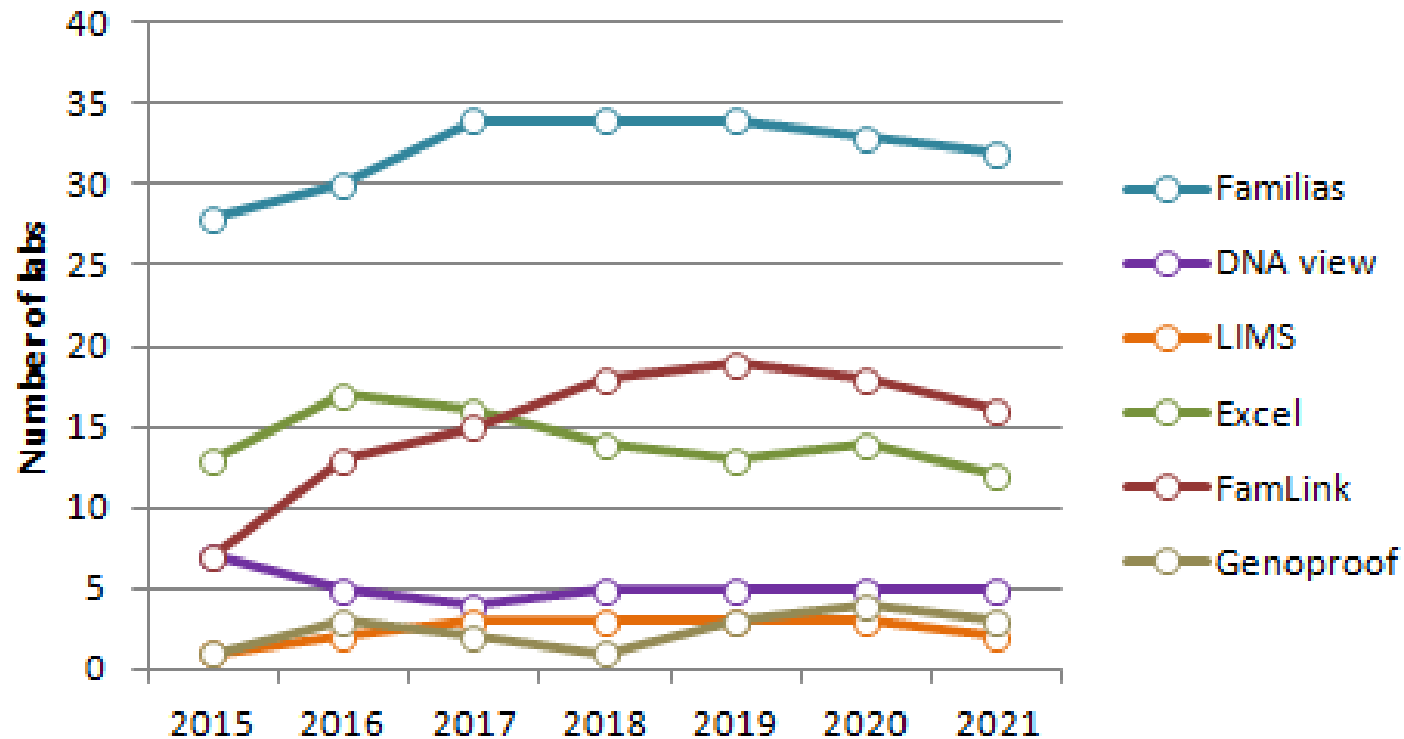
Questionnaire – Sequencing trends

- 17 labs (32%) own sequencing instrument.



Questionnaire – Software trends

Use of software



Questionnaire – Linked markers

Not accounted for: 10

Not used: 14





Exclude one: 16

Accounts for: 9

Similar numbers as 2020

WET EXERCISE

Wet exercise - Background



ESWG WET EXERCISE 2022

This year's wet exercise includes a child (sample labeled Child) seeking his/her biological father. Conduct a paternity test for the putative father (sample labeled Alleged father).

Use a frequency database appropriate for a European population. Report the likelihood ratios (LR) for the individual genetic markers included in the tests as well as the combined LR. State which frequency database you have used for the calculations. Similar to previous years, all results should be reported in the electronic spreadsheet questionnaire.

Samples and procedure

The samples (two in total) consist of blood on FTA cards (diluted spots). We recommend direct amplification with buffers available from vendors (alternatively direct amplification with modern multiplexes). Other extraction procedures have not been tested.

Please perform the DNA tests according to your procedures for kinship analysis and report the data and conclusions in the questionnaire attached to the information email. If different kits are included in the analysis and any discrepancies between overlapping markers occur, please state the difference(s) in the comment field.

Due date

The due date is August 31th, results submitted after this deadline may be dismissed.

Wet exercise - Summary

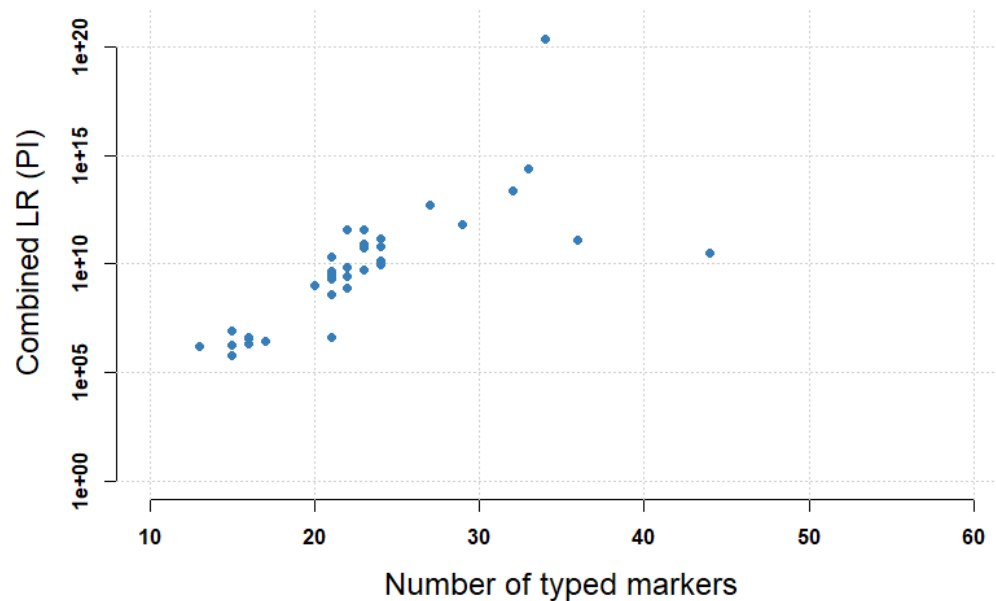
- Overall very concordant results (despite the use of potentially different databases)
- 46 labs participated (43 submitted results)
- Consult the Excel summary for details
- For the wet exercise some labs' results have been highlighted (red or orange) which indicates a result that deviates. Certificates will still be issued.

Combined LR (reported)	8,19E+06	5,60E+12	2,20E+10	4,93E+06						6,24E+14	1,62E+10						9,08E+09	1,5E+11			3,18E+11	3,46E+11
Combined LR (product)	3,58E+06	5,19E+12	9,53E+09	1,77E+06	5,33E+10	1,88E+09		8,10E+06	4,54E+09	2,26E+14	6,83E+09	0,00E+00		2,14E+10	0,00E+00	2,56E+09	4,04E+09	6,55E+10	0,00E+00	3,52E+11	1,35E+11	2,37E+20
Number of markers -->	16	27	24	15	23	21		15	21	33	22	0		21	0	22	21	24	0	22	24	34
D3S1358	2,43	2,29	2,44	2,32	2,78	2,88	2,33	2,30	2,30	2,76	2,38		2,38	2,50		2,38	2,25	2,31			2,34	2,48
D19S433	3,18	3,59	2,71	2,55	3,13	2,88	2,33		3,80	3,88	3,54		3,54	3,08		2,55	3,08	3,73		3,08	3,28	2,47
D2S1338	2,35	2,47	3,26	2,99	2,27	1,77	2,94		2,50	4,86	2,38		2,37	2,25		2,35	2,24	2,66		2,25	3,65	2,20
D22S1045	0,72	0,64	0,72	0,72		0,80	0,76		0,70	0,86	0,65		0,65	0,81		0,82	0,75	0,67		0,81	0,74	0,72
D16S539	9,70	9,02	9,97	7,74	5,83	8,86	9,85	7,80	11,10	9,46	10,30		10,30	11,31		8,96	9,82	12,02		11,30	6,88	9,48
D18S51	2,03	2,00	1,62	1,60	2,08	1,81	2,00	2,20	1,80	1,88	2,03		2,03	2,03		2,19	1,80	1,81		2,03	2,32	2,46
D1S1656	4,73	4,89	4,26	3,80		6,60	6,38		3,40	4,73	5,01		5,01	6,93		3,43	4,60	4,65		6,94	6,27	7,00
D10S1248	8,45	8,22	9,09	6,88		7,02	7,23		11,10	8,68	9,03		9,03	8,92		6,68	6,24	12,24		8,93	7,54	120,25

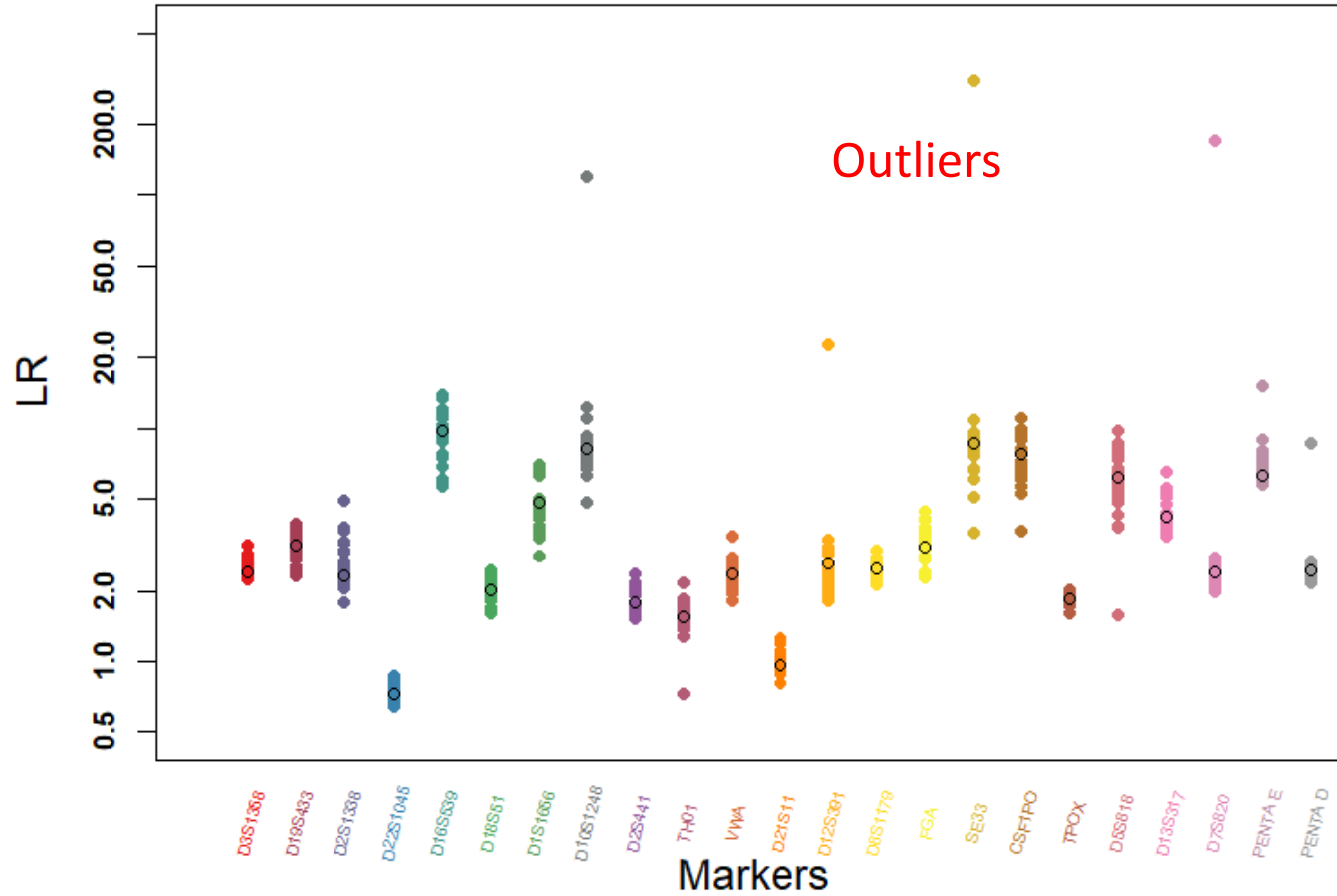


Wet exercise

- A single alleged father
- No labs reported SNP markers this year
- One lab with outliers in several markers (see next slide)



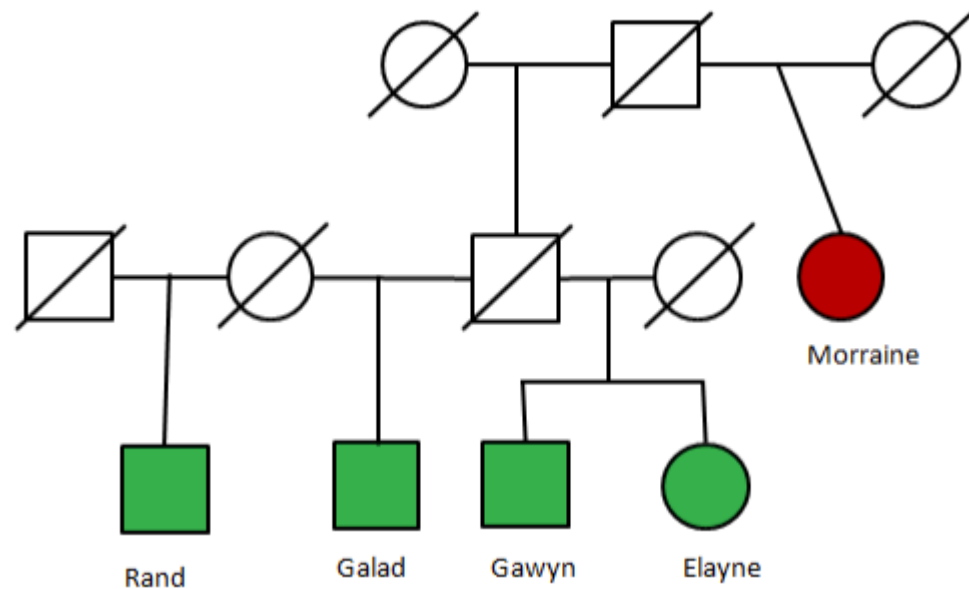
Wet exercise – per marker LR variation






PAPER CHALLENGE

Paper challenge - Background

- The origin of shawl
- Four relatives
- Extended pedigree



Paper challenge – Setup

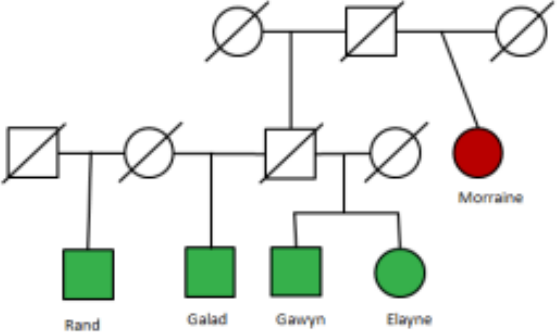


ESWG PAPER CHALLENGE 2022

This year's paper challenge consists of a single exercise. In order to obtain the certificate, participants have to submit results. All data is given as files at https://familias.name/ESWG/ESWG2022_paperchallenge.zip in addition to some details given directly in the cases. Please fill out all answers in the supplied Excel questionnaire.

Case – The Wheel of Time turns...

An excavation outside an ancient massive tower with an appearance of burnished steel, uncovers a mystical blue shawl. The shawl radiates power and bears the inscription 'Alys' which is a known pseudonym of the legendary Morraine Damodred. Using modern DNA techniques we are able to obtain STR profiles from personal items of known relatives of Morraine as well as from the shawl itself. The family tree is depicted below indicating the connection between Morraine and the relatives (samples with DNA data in green).

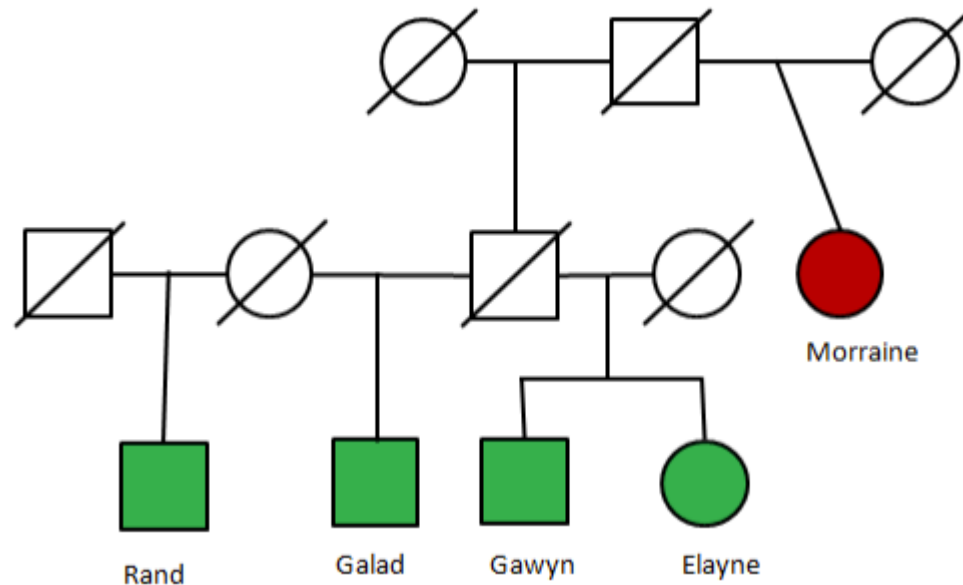


Rand Galad Gawyn Elayne

Paper challenge – Part a)

- State the IBD coefficients between all pairs of individuals in the pedigree, use [IBD0, IBD1, IBD2]

Two individuals can have 0, 1 or 2 alleles IBD for any given marker

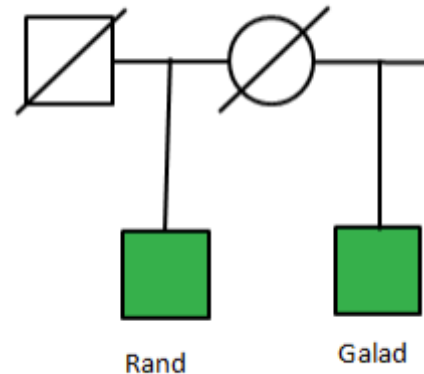


Note that Rand is only related to Galad

Paper challenge – Part a)

- State the IBD coefficients between all pairs of individuals in the pedigree , use [IBD0, IBD1, IBD2]

Note: Rand and Galad can share more alleles IBS which does not mean they are IBD.

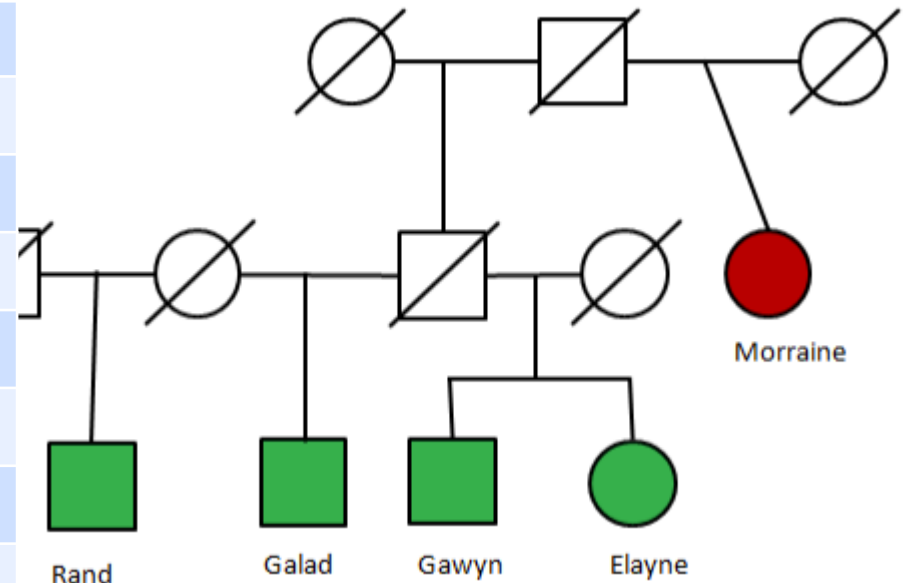


Rand and Galad are half siblings. They are expected to share 0 alleles IBD with 50% probability and 1 allele IBD with 50% probability

Paper challenge – Part a)

- State the theoretical IBD coefficients between all pairs of individuals in the pedigree, use [IBD0, IBD1, IBD2]

Individual 1	Individual 2	IBD coefficients
Rand	Galad	[0.5, 0.5, 0]
Rand	Gawyn	[1, 0, 0]
Rand	Elayne	[1, 0, 0]
Rand	Morraine	[1, 0, 0]
Galad	Gawyn	[0.5, 0.5, 0]
Galad	Elayne	[0.5, 0.5, 0]
Galad	Morraine	[0.75, 0.25, 0]
Gawyn	Elayne	[0.25, 0.5, 0.25]
Gawyn	Morraine	[0.75, 0.25, 0]
Elayne	Morraine	[0.75, 0.25, 0]



Note that Rand is only related to Galad

Paper challenge – Part b)

- Use a software or manual calculations to compute LR comparing full siblings and half siblings with unrelated. Two LRs per comparison

Demonstration in Familias

Paper challenge – Part b)

- Use a software or manual calculations to compute LR comparing full siblings and half siblings with unrelated. Two LRs per comparison

Individual 1	Individual 2	LR full siblings	LR half siblings
Rand	Galad	15314	7989.05
Rand	Gawyn	0.0006	0.13
Rand	Elayne	0.07	3.53
Rand	Shawl	0.0000	0.02
Galad	Gawyn	19.13	763.04
Galad	Elayne	72.30	5406.33
Galad	Shawl	0.29	75.48
Gawyn	Elayne	186190000000	3568455.90
Gawyn	Shawl	7.26	405.81
Elayne	Shawl	0.06	76.31

Paper challenge – Part b)

➤ Using Familias and the blind search function we also find,

Individual 1	Individual 2	IBS0	IBS1	IBS2	Kinship coefficient	Theoretical kinship
Rand	Galad	0.15	0.65	0.2	0.195	0.125
Rand	Gawyn	0.45	0.4	0.15	0.05	0
Rand	Elayne	0.3	0.55	0.15	0.0875	0
Rand	Shawl	0.4	0.6	0	0	0
Galad	Gawyn	0.25	0.65	0.1	0.1325	0.125
Galad	Elayne	0.2	0.75	0.05	0.155	0.125
Galad	Shawl	0.3	0.65	0.05	0.105	0.0625
Gawyn	Elayne	0	0.5	0.5	0.2875	0.25
Gawyn	Shawl	0.25	0.65	0.1	0.1175	0.0625
Elayne	Shawl	0.3	0.7	0	0.0925	0.0625

Paper challenge – Part c)

- Use a software or manual calculations to compute a joint LR where all the available relatives are used to determine the potential origin of the shawl.

Demonstration in Familias

Paper challenge – Part c)

- Use a software or manual calculations to compute a joint LR where all the available relatives are used to determine the potential origin of the shawl.

LR = 1065

Paper challenge – Summary

- Extended pedigree
- Video will be available through <https://familias.name/ESWG/>

Proficiency test – Future

- Not determined who will organize next – ESWG board and members will decide at the meeting in London 2023

- Proficiency test 2022

Daniel Kling – Head Organizer

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