

ESWG PAPER CHALLENGE 2021

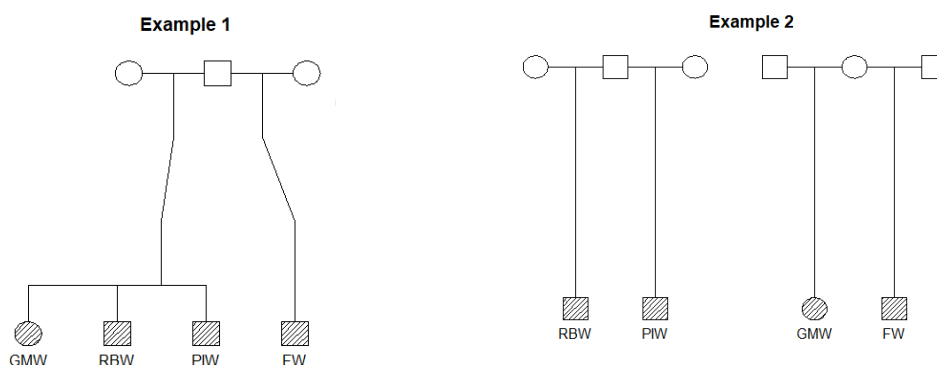
This year's paper challenge consists of a single exercise. In order to obtain the certificate, participants have to submit results. All data is given as files at

https://familias.name/ESWG/ESWG2021_paperchallenge.zip in addition to some details given directly in the cases. Please fill out all answers in the supplied Excel questionnaire.

Case – *Mystery at the Burrow*

In the countryside lay a peculiar old house adorned with chimneys, stretched vertically to accommodate its many inhabitants. According to history there once lived a famous family with more children and grandchildren than one can count. However, recent allegations have been made as to whether or not some of the children were indeed siblings or not. We were able to obtain DNA data from four of the children: GMW, RBW, PIW and FW. You are asked to,

- a) List all possible pedigrees for these four children. That is, given that they can be either full, half siblings or unrelated, provide the number of all potential combinations between the four. For example, the pedigrees below list two such combinations. We refer to the combination where all four individuals are unrelated as the null hypothesis..



- b) Discuss approaches to resolve the case. What approach would your lab take to solve the case? Approaches might include, but not limited to
- 1) Pairwise – Samples are compared in a pairwise fashion and a relationship is considered as confirmed or excluded if the comparison exceeds some threshold.
 - 2) Stepwise – Sample are compared both in a pairwise fashion and in a joint fashion. Starting with pairwise comparisons, where a relationship is considered as confirmed or excluded if it exceeds some threshold. The comparisons are iterated through the inclusion of confirmed matches, until no comparison exceeds the threshold.
 - 3) Joint Bayesian – All relevant pedigrees listed in a) are combined and computations performed for all these.

- c) Using the DNA data provided below, calculate and report the LR comparing the combinations where all individuals are full siblings versus the null hypothesis, see a). What conclusions can be drawn from this LR?

DNA data is given below and in the online files.

Marker	GMW	RBW	PIW	FW
CSF1PO	11,13	12,11	12,11	12,11
D13S317	11,12	11,12	11,11	11,12
D16S539	12,14	12,14	11,14	12,12
D18S51	14,13	14,13	14,13	14,13
D19S433	14,13	13,13	14,13	13,13
D21S11	31,30	28,30	31,31.2	28,30
D2S1338	23,19	25,19	25,23	25,19
D3S1358	17,16	16,16	17,13	17,16
D5S818	11,11	11,11	11,12	11,11
D7S820	11,9	8,12	11,9	8,12
D8S1179	14,13	15,10	15,13	14,13
FGA	20,26	19,22	20,26	19,22
TH01	8,6	8,6	8,6	8,6
TPOX	11,9	11,9	8,11	11,11
D10S1248	15,16	13,14	13,14	15,14
D12S391	17,23	17,23	17,23	19,23
D1S1656	15,14	11,14	15,14	15,14
D22S1045	14,16	16,15	14,16	14,16
D2S441	11,12	11,12	11,11	11,12
SE33	14,27.2	14,27.2	20,27.2	21,27.2
AMEL	X,X	X,Y	X,Y	X,Y

Allele frequencies are given as a file, no population substructure is assumed (i.e. $F_{st}/\theta=0$). We can assume a stepwise mutation model where the mutation rate is 0.001 for all markers, the step rate is 0.1, i.e. it is 0.1 times less likely to observe a two step mutation compared to a single step. Silent alleles and other complicating factors can be disregarded.

- d) Additional information is given stating that GMW, RBW and FW share the same mother. Use this information to reduce the space of possible pedigrees. How many pedigrees remain?
- e) Using the provided DNA data and the complete list of pedigrees listed in **d)**, compute the likelihoods and posterior probabilities for all combinations. Report the top 10 results and your verbal conclusion in the case.