

# Additional exercises

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All data files are available at [http://familias.name/additional\\_exercises.zip](http://familias.name/additional_exercises.zip) . All names etc. are fictional and do not correspond to actual persons unless otherwise stated.

## Exercise 1 – Plotting and simulations

Mary is interested to find out if she shares the same father as her maternal cousin. She tells us that her full sister and her mother are willing to give their samples for testing. She tells us that her paternal uncle is reluctant and would rather not participate and asks if she should convince him.

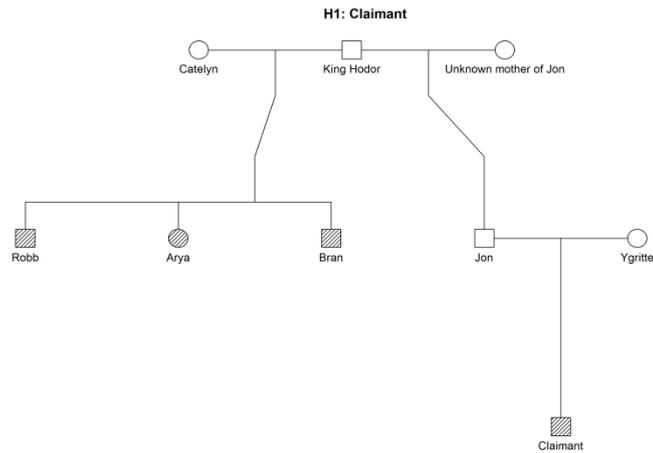
- a) Construct and plot the complete pedigree with all mentioned individuals as well as the alternative hypothesis using the information above.
- b) Find out what individuals that are necessary to collect samples from in order to reach a conclusion (say  $LR > 1000$ ) with a 90% certainty. We can assume Mary and her cousin are typed. Use the default parameters defined in the input frequency database (e.g. mutation rates and theta correction set to zero).

## Exercise 2 – A claimant to the throne

This case involves an ancient tale from a realm known as Westeros. The history has it that a claimant to the throne presents himself. He proposes that he is the grandchild of the deceased king. At the time, the allegations could not be ascertained and the claimant was regarded as a false pretender.

Using modern DNA techniques we are able to obtain samples and results from three known descendants of the King as well as from the Claimant.

- a) Import frequency data from a Westeros population sample and DNA data for the involved individuals.
- b) Conduct a kinship analysis to uncover whether the Claimant is related to the three descendants of the king the way he claims. The pedigree corresponding to the claimant's allegations is depicted below. In the alternative hypothesis, the Claimant is unrelated to the descendants who are themselves still full siblings. Use the default parameters defined in the input frequency database.



**Figur 1. Pedigree corresponding to the main hypothesis in Exercise 2.**

- c) Following the results, your lab decides to perform a simulation study to evaluate whether including further relatives could improve the results. We may assume we have already analyzed all available genetic markers. Evaluate the following scenarios,
- I. Exhumation of the mother of the descendants (Catelyn).
  - II. Exhumation of the mother of the Claimant (Ygritte).
  - III. Exhumation of the King (Hodor).
  - IV. Exhumation of all the above mentioned individuals.

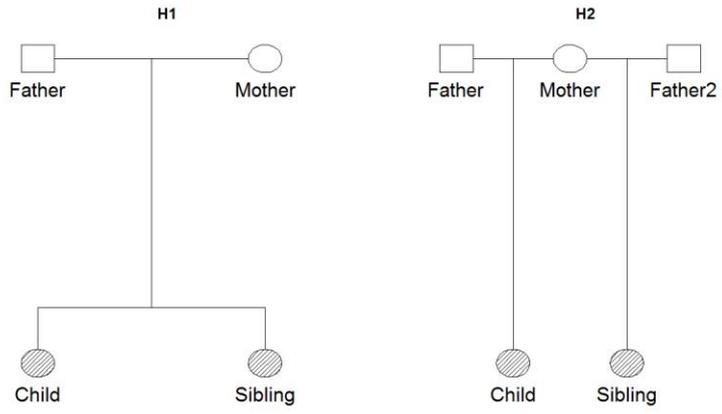
Report the median of your simulations as well as the probability that the LR will exceed 1000 for each given scenario (assuming the Claimant is related to the descendants as proposed). Perform at least 1000 simulations.

- d) What is your conclusion regarding the four possible scenarios? For instance, can we reach a more certain conclusion by only exhuming one extra individual (I-III) or do we need to exhume all three of them (IV).

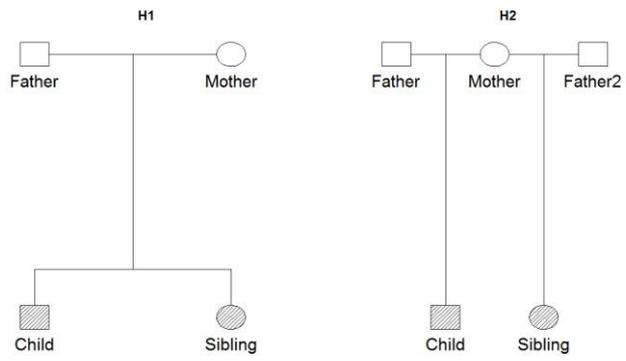
### **Exercise 3 – The utility of X-chromosomal markers**

Use your knowledge about inheritance patterns to decide if X-chromosomal markers are relevant in the following scenarios.

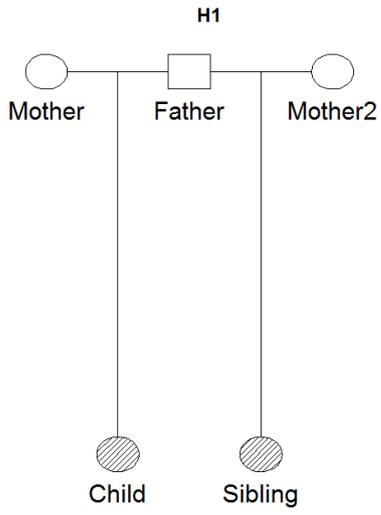
- a) Case 1



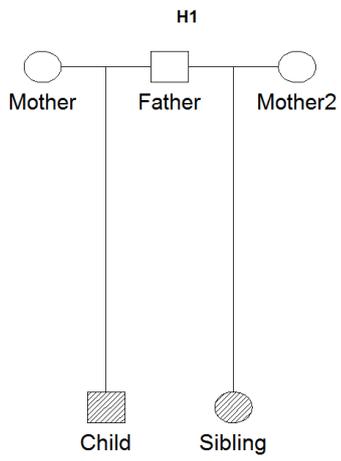
b) Case 2



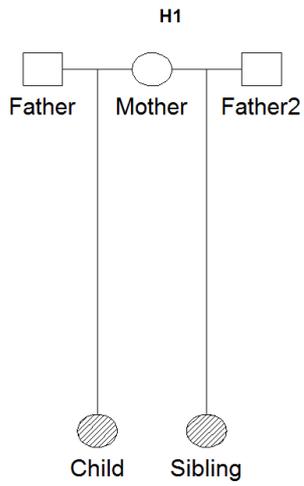
c) Case 3



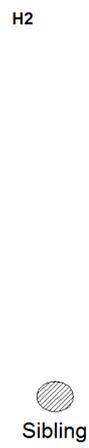
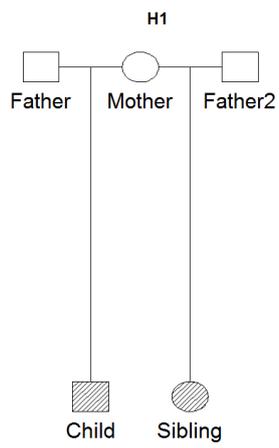
d) Case 4



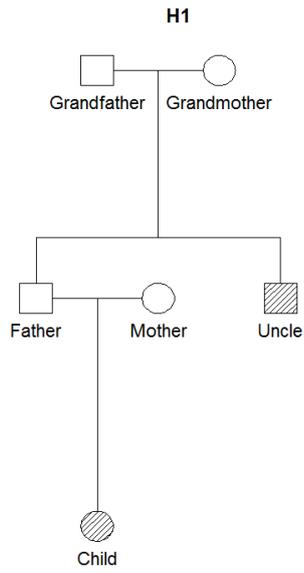
e) Case 5



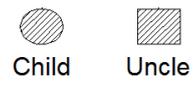
f) Case 6



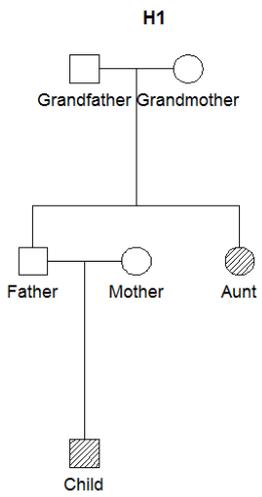
g) Case 7



**H2**



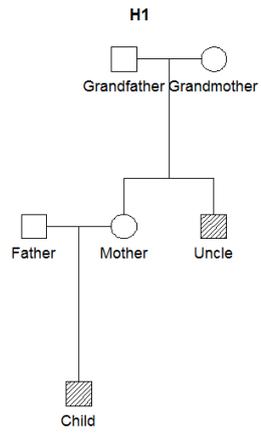
h) Case 8



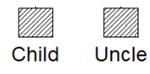
**H2**



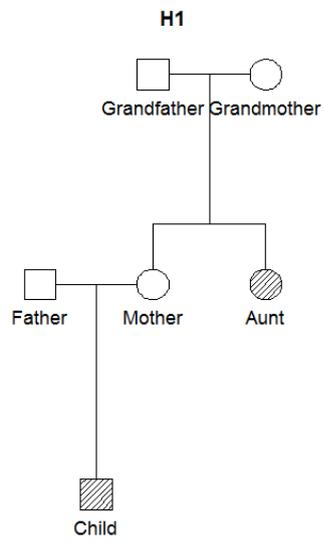
i) Case 9



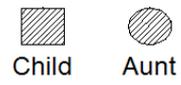
**H2**



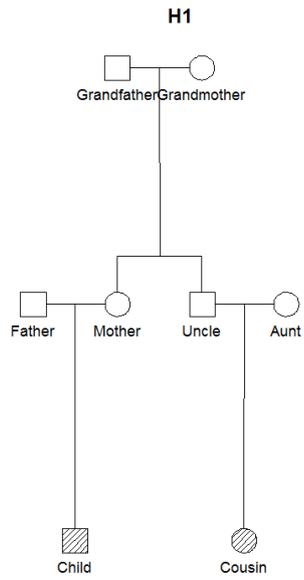
j) Case 10



**H2**



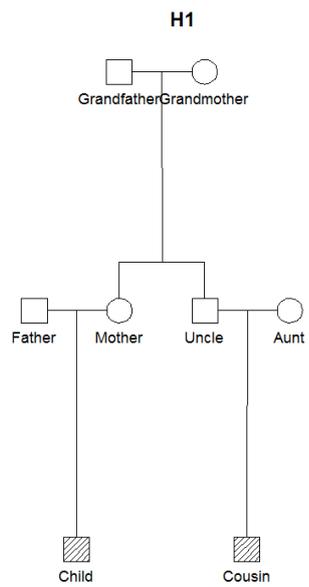
k) Case 11



**H2**



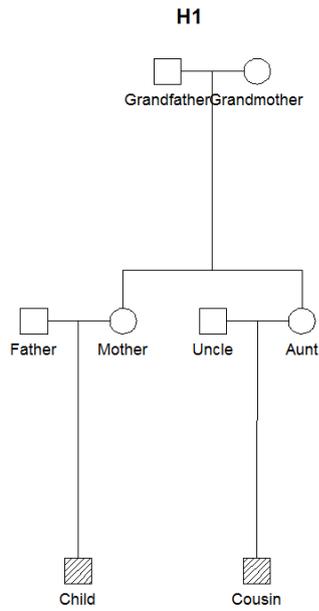
l) Case 12



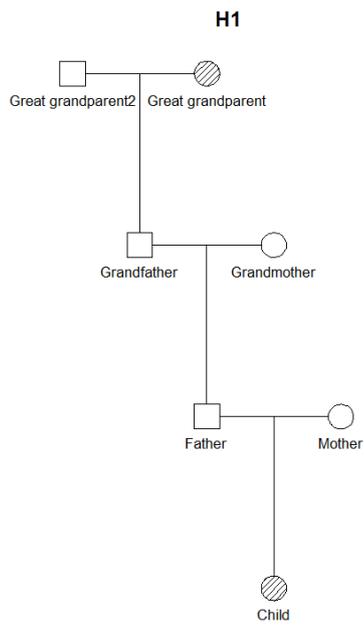
**H2**



m) Case 13



n) Case 14



### Exercise 4 – Duckburg

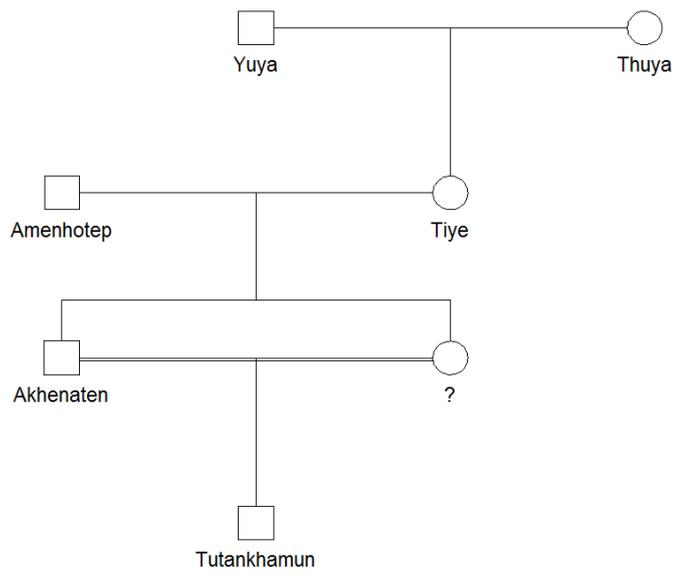
A woman named Daisy, residing in the city Duckburg, asks for your expertise in a case where she needs to determine the relationship between three girls, April, May and June. She has been able to obtain X-chromosomal profiles for the girls and herself.

- a) Import frequency data from a Duckburg population sample and DNA data for the three girls. Mutations are unheard of in Duckburg and all rates are zero to reflect this fact.  
\* Estimate the lambdas for the clusters defined in the frequency database using the built-in function in FamLinkX and the R software. You may skip this part and use lambdas 315, 208, 111 and 208 for the four clusters respectively.
- b) Our first task is to decide what hypotheses to test. We have information that they all share the same mother, but whether or not they share the same father is uncertain. In practice we would now need to test all the different combinations where two of the girls are full siblings and the third girl maternal half sibling to those two. However, Daisy further tells us that she is also certain that April and May shares the same father, Gladstone, while she is uncertain if he is also father of June or if another man, is the father (both are unavailable for testing). Compute the LR for the two relevant pedigrees (This part does not involve DNA data from Daisy).
- c) Assume the three girls are full siblings and construct the pedigree to test whether or not Daisy is their maternal aunt.

### **Exercise 5 – Sorting out the pharaohs**

Genetic data from some of the ancient Egyptian pharaohs are available through <https://jamanetwork.com/journals/jama/fullarticle/185393>. We will here try to sort out the relationships between them.

- a) Import a modern Egyptian frequency database and the DNA data. Review the data.
- b) Perform a blind search including only the typed individuals. Include parent/child, full siblings and half siblings. Use default search parameters. Review the results.
- c) Try to reconstruct the pharaonic pedigree by assigning parent/child relationships to all matches where the kappa values favors this relationship.
- d) Compare the results in c) with the suggested pedigree (Figure 2 in the above link and illustrated below). Does your reconstruction correspond to the illustration? Did you find any additional relationship connections?



Figur 2. Suggested pedigree for the Egyptian pharaohs described in Exercise 5.

- e) Plot a dendrogram using the Export matrix (distance) function in Familias and the R software.