



ESWG PAPER CHALLENGE 2017

In order to compare statistical calculations among the laboratories, you are encouraged to do this paper challenge. You should submit an LR for each marker and the total LR combining the individual contributions. No algebraic formulas are requested, but may optionally be supplied.

The exercise is divided into two different parts – one concerning autosomal marker data and one concerning X chromosomal marker data. To obtain a certificate participants are obliged to complete *Part 1*, whereas *Part 2* is optional.

The autosomal <u>typing results</u> and frequency data (<u>Plain frequencies</u> and <u>Familias database</u>) are available in the given links. The X-chromosomal <u>typing results</u> and frequency data (<u>Haplotype data</u> and <u>FamLinkX database</u>) are available in the given links. The typing results are also collected into a single file available at <u>http://familias.name/ESWG/ESWG Collected Data 2017.xlsx.</u>

Please use the supplied Excel questionnaire to fill in your answers. As mentioned above, theoretical formulas are not asked for but may be entered into the questionnaire.

Part 1 (Autosomal markers)

Your lab is approached by three females interested to know if they are siblings. You reply by stating that you need some more information restricting the possible scenarios. Given the information that the females are all children, i.e. they have no children of their own (and that we have no inbreeding), how many alternative pedigrees can you construct? For simplicity you may assume that the parents are not themselves related and that the putative siblings all belong to the same generation.

Given further information obtained by the putative siblings, the following hypotheses are listed as plausible alternatives

















Compute the LR comparing the genotype data given each hypothesis using only autosomal data, use H2 in the denominator (Scaling). There is non-DNA evidence favoring H1 and H3, compute the combined posterior probabilities given that H1 and H3 is, a priori, twice as likely as H2. In other words, each hypothesis is twice as likely as H2.

We can assume no subpopulation structure, in other words use θ =0 in the calculations. In addition, mutations can be disregarded. What is your conclusion?







Part 2 (X-chromosomal data) - Optional

Following the results from the autosomal data, your lab decides to run an X-chromosomal analysis to further strengthen your conclusion.

Compute the LR comparing the genotype data given each hypothesis using only X-chromosomal data, use H2 in the denominator (Scaling). There is still non-DNA evidence favoring H1 and H3, compute the posterior probabilities given that H1 and H3 is, a priori, twice as likely as H2.

We can assume no subpopulation structure, in other words use θ =0 in the calculations. In addition, mutations can be disregarded. What is your conclusion?

Combine the data from the autosomal and the X-chromosomal analysis. What is the combined posterior probabilities? Would you suggest any additional testing? What analyses could be useful to shed further light on the case?