

ESWG PAPER CHALLENGE 2023

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/ESWG2023_paperchallenge.zip in addition to some details given directly in the cases. Please fill out all answers in the supplied Excel questionnaire.

Case – A story from far away

Ancient scrolls discovered in dessert caves depicts a story about genealogy. According to the drawing two children (Luke and Leia) are in search for their unknown paternal grandfather who according to the legend was a great force who could change the destiny itself. An individual named Thef Orce is suggested as the unknown grandfather of the two children but from what can be eluded, no methods to establish the genealogy existed at the time of drawing. Amazingly, on the walls of the caves, archeologists are able to decipher DNA profiles of T. Orce as well as Luke and Leia. Using modern biostatistical approach we are tasked to establish the genealogy.

- a) The pedigree depicting the main hypothesis is illustrated below. Compute the pairwise LR_s between individuals in the pedigree (include full and half siblings). Can any conclusions be drawn based on these results?

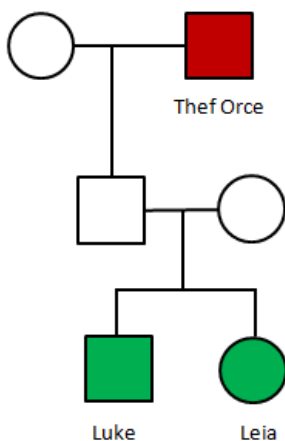


Figure 1. Pedigree illustrating the main hypothesis (H1). Data is available for the individuals with either green or red fill.

- b) Compute the LR comparing the pedigree in the illustration above with the alternative hypothesis where Luke and Leia are unrelated to Thef Orce [in the pedigree].

DNA data is given below and in the online files for a set of 20 autosomal STRs.

Marker	<i>T. Orce</i>	<i>Leia</i>	<i>Luke</i>
CSF1PO	10, 10	10, 12	11, 12
D13S317	11, 12	12, 13	11, 11
D16S539	11, 11	13, 12	11, 11
D18S51	18, 11	14, 12	11, 12
D19S433	14, 16.2	14, 12	16.2, 14
D21S11	28, 29	31.2, 28	28, 28
D2S1338	19, 22	20, 17	19, 17
D3S1358	16, 18	18, 18	18, 18
D5S818	11, 13	11, 13	11, 13
D7S820	12, 10	7, 11	7, 11
D8S1179	13, 13	14, 13	13, 12
FGA	22, 25	20, 25	20, 23
TH01	9.3, 9	9, 6	6, 6
TPOX	8, 8	8, 8	8, 8
D10S1248	15, 14	13, 13	14, 13
D12S391	17, 19.3	19.3, 18	19, 20
D1S1656	15, 17.3	15, 14	17, 14
D22S1045	15, 15	15, 16	15, 16
D2S441	11, 10	10, 10	10, 10
SE33	27.2, 19	27.2, 14	27.2, 12
AMEL	X,Y	X,X	X,Y

Allele frequencies are given as a file, no population substructure is assumed (i.e. $\theta=0$). We can disregard mutations (i.e. mutation rate=0). Silent alleles and other complicating factors can also be disregarded.

A more distant story (Optional)

In the ruins of an old temple, a lost archive is found. From the shelves we are able to obtain the Y-chromosomal profile of a paternal grandfather of T. Orce. See pedigree below.

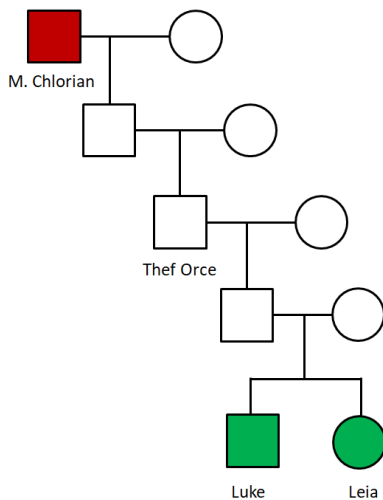


Figure 2. Pedigree illustrating the main hypothesis (H1). Data is available for the individuals with either green or red fill.

- c) [Optional] The Y-profile of Luke is extracted from the walls of the cave. In total, data for 22 Y-STRs are available. Estimate the weight of evidence that Luke and his great great grandfather M. Chlorian are paternally related as depicted in the pedigree above. In the calculations you can assume that the haplotype of Luke has never been observed while the haplotype of M. Chlorian has been observed 10 times. The size of the Y-database is 100,000. For simplicity we can assume that the mutation rate is equal to 0.001 for all included markers and that there is an equal chance for a loss or gain of a tandem repeat in the mutation model. Further assume that there is a 90% chance for a mutation to be single step, 9% two step and so forth.

Marker	<i>M. Chlorian</i>	<i>Luke</i>
DYS19	14	14
DYS389I	13	14
DYS389II	29	30
DYS390	24	24
DYS391	10	10
DYS392	11	11
DYS393	12	12
DYS385	14,17	14,17
DYS437	15	15
DYS438	10	10
DYS439	12	12
DYS448	21	21
DYS456	15	15
DYS458	16	16
DYS635	21	21
YGATAH4	10	10
DYS481	22	22
DYS533	11	11
DYS549	12	12
DYS570	17	17
DYS576	15	15
DYS643	11	11



For part c) no data files are available online. Data is only provided in the table above.

- d) [Optional] Ultimately, we are able to decipher the genetic code for an expanded SNP panel from both the caves and the temple with data for Luke and M. Chlorian. We are stunned to realize there is a 100% overlap with the recently published FORCE panel with data for 3,931 autosomal kinship markers. Compute the pairwise LR for the alleged relationship between the two individuals (depicted in the pedigree above). All necessary information is given in the files linked to at the top of this document. For brevity, genetic data is omitted from this document.