ESWG PAPER CHALLENGE 2020

This year's paper challenge is divided into four different parts. In order to obtain the certificate for participation, at least two has to be completed. All data is given as files at http://familias.name/ESWG/ESWG2020_paperchallenge.zip in addition to some details given directly in the cases. Please fill out all answers in the supplied Excel questionnaire.

Case 1 – Complex kinship case – An inheritance claim

The first case deals with an inheritance dispute, where a cousin (paternal) to a sole heir of rich woman, whom is recently deceased, claims that she is also the long lost (maternal) half sister. The heir strongly disputes that her paternal cousin (their fathers are full brothers) could also be her maternal half sister. Data for 32 autosomal STR markers is given in the table below.

Marker	Heir	Cousin
D3S1358	16,16	16,17
TH01	6,7	6,6
D21S11	28,29	28,30.2
D18S51	17,17	16,17
PENTA_E	7,17	7,10
D5S818	12,13	10,13
D13S317	12,12	11,12
D7S820	8,11	9,12
D16S539	9,12	8,11
CSF1PO	11,13	10,13
PENTA_D	9,14	10,14
VWA	16,17	16,17
D8S1179	13,14	12,12
TPOX	9,10	9,11
FGA	23,23	23,24
D19S433	13,14	12,14
D2S1338	20,24	20,24
D10S1248	12,14	14,16
D1S1656	11,17.3	11,15.3
D22S1045	16,16	15,17
D2S441	14,14	14,15
D12S391	17,17	17,20
SE33	25.2,27.2	25.2,27.2
D7S1517	19,19	19,21
D3S1744	15,18	16,18
D2S1360	22,23	22,23
D6S474	16,16	13,17
D4S2366	10,11	9,11
D8S1132	17,18	17,19
D5S2500	12,15	10,15
D21S2055	26,34	19.1,34

D10S2325	11,13	8,11

a) Plot the pedigrees and discuss what type of markers could be used to solve the case (Not reported in the questionnaire).
b) Compute the LR for the given autosomal markers comparing,

   H1: The heir and her paternal cousin are maternal half sister.

   H2: The heir and her paternal cousin are unrelated on their maternal side.

For b) we can ignore complicating factors such as mutations, silent alleles and linkage.

Data is also available for X chromosomal markers, the data is given in the table below.

Marker	Heir	Cousin
DXS10148	18, 25.1	25.1, 25.1
DXS10135	20, 23	23, 27
DXS8378	10, 11	10, 11
DXS7132	13, 13	13, 13
DXS10079	19, 22	19, 22
DXS10074	16, 16	16, 16
DXS10103	16, 18	16, 17
HPRTB	13, 14	13, 14
DXS10101	25.2, 28.2	25.2, 31
DXS10146	27, 28	27, 28
DXS10134	34, 37	37, 38
DXS7423	13, 14	13, 15

c) Compute the LR for the X-chromosomal data using the same hypotheses as in a). Mutations and silent alleles can be ignored while linkage and linkage disequilibrium should be accounted for if relevant.
d) Combine the LRs from b) and c) into a total LR. What is your verbal verdict in the case?

Case 2: Mixture with relatives

The second case involves a blood sample from an abortion, where the results display a mixture of two persons, the mother and her child. The alleged father is sampled as well as mother. In addition, we have access to a single source sample from the unborn child. Data is given in the table below.

Marker	Mother	Father	Child	Mixture
D3S1358	14,16	15,15	14,15	14,15,16
TH01	9,9.3	9,9.3	9,9.3	9,9.3
D21S11	29,34.2	29,30	29,30	29,30,34.2
D18S51	14,18	14,15	14,18	14,18
PENTA_E	10,12	5,13	12,13	10,12,13
D5S818	9,12	12,13	9,12	9,12
D13S317	11,12	10,12	10,11	10,11,12
D7S820	9,10	8,12	8,10	8,9,10
D16S539	11,13	11,13	11,13	11,13
CSF1PO	10,12	12,12	10,12	10,12
PENTA_D	10,12	10,13	10,10	10,12
VWA	17,17	17,20	17,17	17,17
D8S1179	10,16	12,12	12,16	10,12,16
TPOX	8,11	8,9	8,11	8,11
FGA	25,25	20,21	21,25	21,25
D19S433	13,15	14,15	13,15	13,15

The hypotheses we will consider is given by,

H1: The alleged father is the biological father of the child (either the single source or the mixture).

H2: The alleged father is unrelated to the child.

Maternity for the mother can be assumed in both hypotheses. There will be some slight variations when the mixture is considered, see details in the exercises below.

In all computations we will disregard any further complicating factors such as mutations, population substructure etc. Frequency data as well as the genotypes (single source only) are also given as files at http://familias.name/ESWG/ESWG2020_paperchallenge.zip

a) Compute the LR comparing using the single source sample for the child.
b) Now consider the mixture (abortion sample), compute the LR in the same manner. Use the sample for the mother as a known contributor in the mixture (both hypotheses).
c) Consider the mixture again, compute the LR as in b), but disregard the known profile of the mother (both hypotheses).

Case 3: Expanded marker panels – On the effect of linkage

A and B have the same mother, but it is disputed whether they also share the same father. In this case, genotypes for both STRs and SNPs are available, of which many are located on the same chromosome.

DNA profiles, allele frequencies and the genetic locations of the included markers are available for the likelihood ratio calculation (see zip file, link given in the beginning of this document).

Set up hypotheses and calculate the combined LR as well as the combined posterior probability (assuming a flat prior). Provide a verbal statement.

Case 4: Multiple hypotheses – A quest for the "best" answer

Child1, Child2 and Child3 have the same mother. For legal reasons the paternal relationship between the children needs to be establish. The question is whether all of the children share the same father, or if two of the children share the same father, or if all children have different fathers. DNA data is given below.

Marker	Child1	Child2	Child3
CSF1PO	12, 12	12, 12	9, 11
D13S317	8, 12	8, 12	8, 12
D16S539	11, 13	11, 13	11, 12
D18S51	22, 22	14, 18	18, 18
D19S433	12, 14	14, 15.2	14, 15.2
D21S11	29, 32.2	29, 32.2	29, 33.2
D2S1338	17, 18	18, 21	17, 18
D3S1358	16, 16	15, 16	15, 16
D5S818	11, 12	12, 13	12, 12
D7S820	8, 8	8, 8	8, 12
D8S1179	14, 14	14, 14	13, 14
FGA	22, 22	22, 22	20, 22
TH01	8, 9	8, 9	7, 9
TPOX	8, 12	8, 11	11, 12
D10S1248	15, 17	14, 16	14, 16
D12S391	17, 21	17, 21	17, 17
D1S1656	16, 17	16.3, 17	16, 17
D22S1045	11, 15	11, 15	11, 16
D2S441	10, 14	10, 14	10, 14
SE33	30.2, 33	30.2, 32	26.2, 30.2

Set up all relevant hypotheses given the above specifications and calculate the combined posterior probability assuming a flat prior. Allele frequencies are given as a file, no population substructure is assumed (i.e. theta=0). NB! mutation model, mutation rates, silent allele frequencies needs to be assigned.