

# Discussion “Relatedness inference with forensic applications”

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# Genealogy: iterative pedigree construction, new searches, possible?

- 5th cousins are quite likely not to share any IBD
- But people have very many 5th cousins: likely to share DNA with some of them
- Can an iterative search be envisaged using the genealogical marker sets, e.g. when one family member is found, place it in a pedigree in its most likely spot (perhaps try several) and search again?
- In general: pedigree search as with autosomal markers

(Forensics: LR, genotype distributions, works for any pedigree; genealogy: IBD based classification, extendable?)

# IP and EP

- Inclusion and exclusion power are useful statistics for predictive purposes
- But is exclusion the goal? If (all) mutations are allowed for, which is probably best (to deal with actual mutations, but also with technical profile artefacts, clerical errors,..) then  $EP=0$ , but many LR's will be  $\ll 1$ , and the corresponding hypotheses will not be interesting to follow up on.
- Perhaps one could envisage: if  $LR < t$ , drop the case; if  $LR > T$ , declare match; if  $t < LR < T$ , further investigate. Thresholds: according to costs? Or FPR/FNR?
- Need to decide what to do if LR remains between  $t$  and  $T$  ? Report if  $LR > t'$  for some  $t < t' < T$ ?

(Familial searching in NL:  $t=1000$ ,  $T=10^9$ ,  $t'=10^5$ ).




# Reporting: which LR?

- Imagine missing parent-child pair in pedigree
  - Victim 1 & Victim 2: LR of  $10^{15}$  for (V1=M1,V2=M2) versus M1&M2 unrelated to V1&V2, and V1, V2 unrelated. Should we report that number?
  - LR for (V1 parent/child of V2) versus (V1 unrelated to V2) will also be large, say  $10^7$ .
  - Then LR for (V1=M1,V2=M2) versus (V1 is parent/child of V2) is  $10^8$ ? Perhaps better report LR= $10^8$ ?
  - The parent-child pair may even have a large LR in the pedigree primarily due to being parent-child, not particularly for being this specific pair ( $10^9$  instead of  $10^{15}$ , say).
- In general: does one consider in the algorithm the unplaced victims as unrelated? Also if there likely exist family relationships between them?

# Reporting: posterior prob's

- Also seems dangerous, since in order to obtain posterior probabilities, there shouldn't be any other not-investigated possibilities. This is not always the case. Only odds available then. Similar remarks as for previous slide.
- It is standard practice in paternity labs, and it is sometimes required by law that posterior probabilities are reported
- Seems logical from the point of view that the decision maker is not primarily interested in what the evidence is, but in what the truth is.
- How do we get the point across that we can not always know the truth with absolute certainty. Sometimes we may get so certain that this point is purely philosophical; but not always.

# Ethical issues; whose responsibility?

- Technology is developed based on what we find acceptable/desirable
  -  It turns out to have an application we hadn't thought of; we do it anyway: function creep
- Then,
- The new application changes our idea of what we find acceptable/desirable: ethics creep
- E.g. DNA databases for direct matching.  we can find relatives in it. Familial searches!
- New matches are found and crimes solved. But often there is no match in a familial search.
- Genealogical databases invented for the public to find their relatives and see how much Norwegian/Dutch/whatever DNA they have. *Much* more reach than forensics, but data are more sensitive too.
-  we can also use these databases for a familial search! Similarly: children of sperm donors who wished to remain anonymous, try and find them via genetic genealogy.
- Question: Perhaps we should acknowledge this mechanism and develop tools that can *only* do the originally intended application? Or are we ok with this evolution which after all is a spontaneous one?

Are there many examples? Who should discuss this, think ahead? Scientists, philosophers, parliament, public discussion?