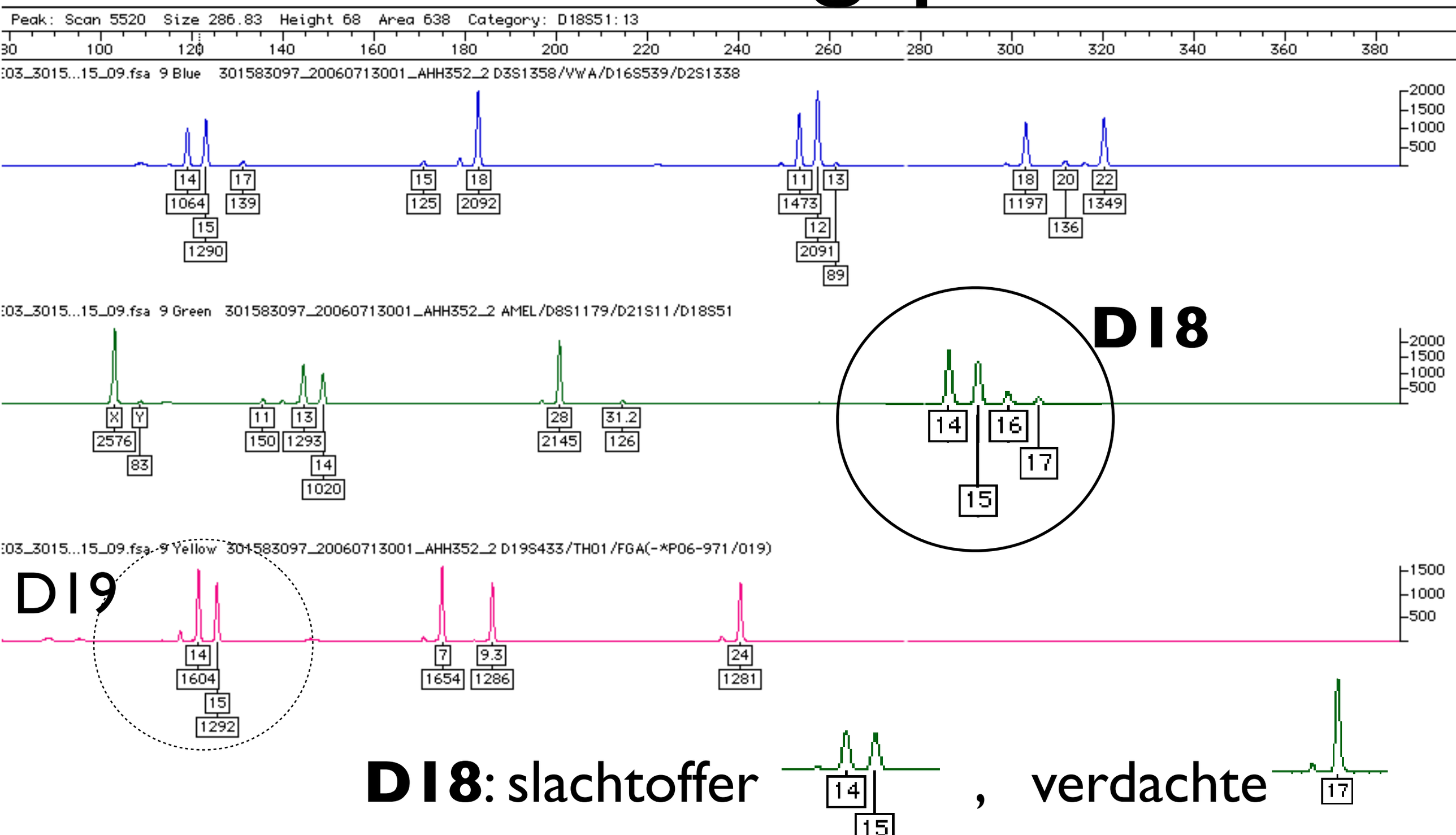
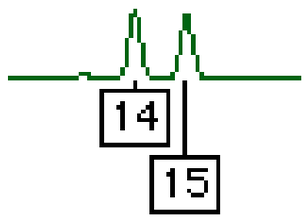
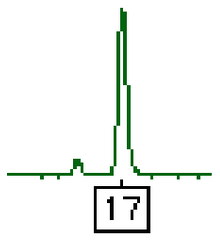
Een problematisch DNA mengspoor



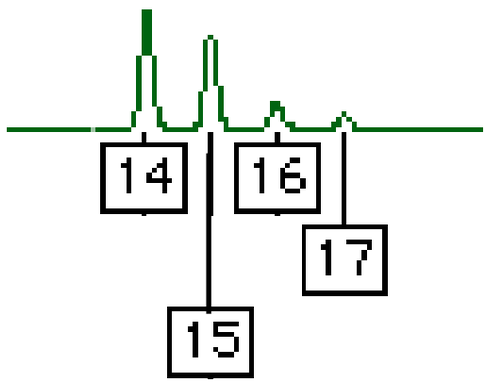
D18: slachtoffer

, verdachte

D18: slachtoffer 

, verdachte 

D18: “mengprofiel” vingernagel slachtoffer



NFI Expert: “Als de spoor van twee personen afkomstig is, kunnen we de verdachte uitsluiten.
Als het van drie personen afkomstig is, niet”.

NFI Expert: “Als de spoor van twee personen afkomstig is, kunnen we de verdachte uitlsuiten.
Als het van drie personen afkomstig is, niet”.

Rest van mengprofiel komt zeer goed overeen met een 90-10 mengsel slachtoffer-verdachte

D18 is meest bekend locus voor *mozaicism* wat daar bij minstens 1 op de 5000 personen voorkomt

Review

Role of Short Tandem Repeat DNA in Forensic Casework in the UK—Past, Present, and Future Perspectives

BioTechniques 32:366-385 (February 2002)

Peter Gill
Forensic Science Service,
Birmingham, UK

ABSTRACT

The analysis of short tandem repeat (STR) DNA sequences is of fundamental importance to forensic science because they have become the recognized standard in constructing national public databases.

DEVELOPMENT OF MULTIPLEXED SYSTEMS

Early multiplexes consisted of few loci that were based on simple short tandem repeats (STRs). The four-locus "quadruplex" was probably the first to be widely used (44); because it consisted of few STRs, the match probability was consequently high—1 in 10 000. In 1996, a six-locus STR system (57,58) combined with the amelogenin sex test (61) was introduced—known as the

ous years, all six loci of the older SGM system were retained in the new Amp*Fl* STR SGM Plus system.

Development and Harmonization of National DNA Databases

The harmonization of STR loci has been achieved by collaboration at the international level. Notably, the European DNA profiling group (EDNAP) carried out a series of successful studies to identify and recommend STR loci for the

Somatic Mutation

If a somatic mutation occurs during embryological development, then two types of cells with different genotypes may coexist, and this leads to a three-banded profile (Figure 4). The peak areas will depend on the relative proportion of the mutant cell and will not be equivalent. This is arguably the most difficult condition to elucidate because it is possible that not all tissues will demonstrate somatic mutation. The incidence of somatic mutation is variable—out of 120000 samples, not one has been observed at the HUMTH01 locus, whereas the incidence is approximately 1 in 5000 at the D18S51 and HUMFIBRA loci. It is possible that some somatic mutations will be indistinguishable from stutters; therefore, these figures are probably underestimates because they are only recorded if unambiguous.

The genetic phenomena described (trisomy, translocation, and primer binding site mutations) can be verified by the analysis of the reference sample, which should also demonstrate the same anomaly unless a tissue-specific somatic mutation has occurred. In the latter case, confirmation may depend on a reference sample that has the same origin as the case stain, although we cannot completely rule out the possibility that the appearance of somatic mutations could vary over time within tissues such as the buccal lining, which consists of rapidly dividing cells.

To summarize, an understanding of the behavior of the DNA profiling system is important to assess potential mixtures. Loci will behave somewhat differently from each other, but it is possible to generalize. Here are some of the key features: (*i*) the smallest peak area of a heterozygote will usually be greater than 60% of the size of its partner (peak area or peak height); (*ii*) within the previous guideline, the high molecular weight peak is often smaller than the low mol-

Groottes van pieken geven ook informatie,
mits we goed rekening houden met

- *Stutter*
- *Dropout*
- *Variatie* grootte na/door amplificatie

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Research articles

Probabilistic modelling for DNA mixture analysis

R.G. Cowell [a,*], S.L. Lauritzen [b], J. Mortera [c]

[a] *Cass Business School, 106 Bunhill Row, London EC1Y 8TZ, UK*
[b] *Department of Statistics, University of Oxford, 1 South Parks Road, Oxford OX1 3TG, UK*
[c] *Dipartimento di Economia, Università Roma Tre, Via Silvio D'Amico, 77, 00145 Roma, Italy*

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Abstract

Taking peak area information into account when analysing STR DNA mixtures is acknowledged to be a difficult task. There have been a number of non-probabilistic approaches proposed in the literature, and some have been incorporated into computer systems, but comparatively little has been published from a probabilistic perspective. Here we briefly review our previous work on using Bayesian networks to analyse two-person mixtures within a probabilistic framework, and present preliminary results obtained for analysing two-person and three-person mixtures that combine peak area information from multiple independent samples.

Keywords: DNA mixtures; Bayesian networks; Multiple traces; Multiple contributors

1. Introduction

In a recent series of papers [1–3] we have presented a probabilistic methodology for analysing peak area information from DNA mixtures based on Bayesian networks. A representative fragment of these networks is shown in Fig. 1

apparatus *after* amplification of the mixture sample. We model the stochastic variations in these areas by Gamma distributions, where the Gamma distribution of the area for allele a depends on the mean μ_a and has expectation proportional to μ_a; similarly for alleles b and c. For further details of the Gamma model and Bayesian networks, and how the probability

Basis model

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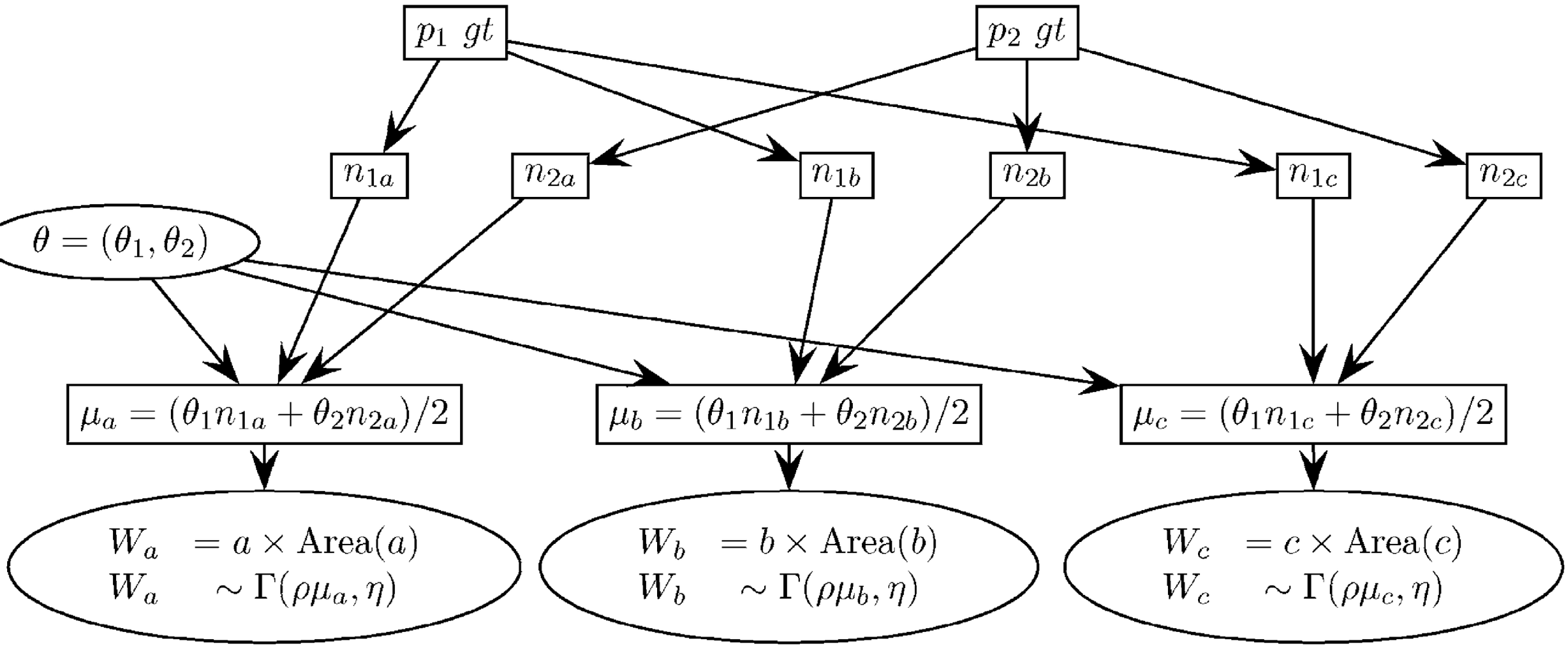
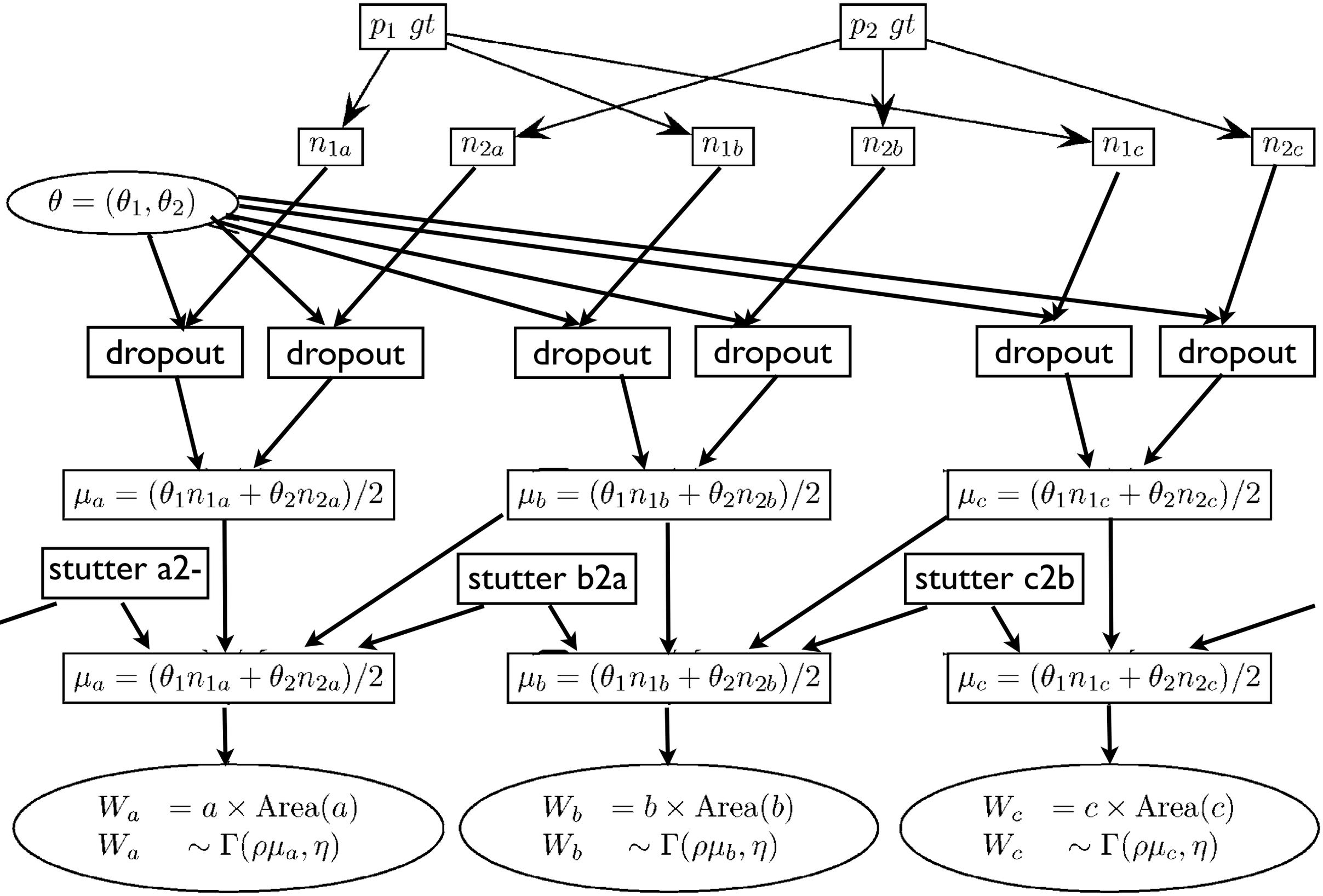
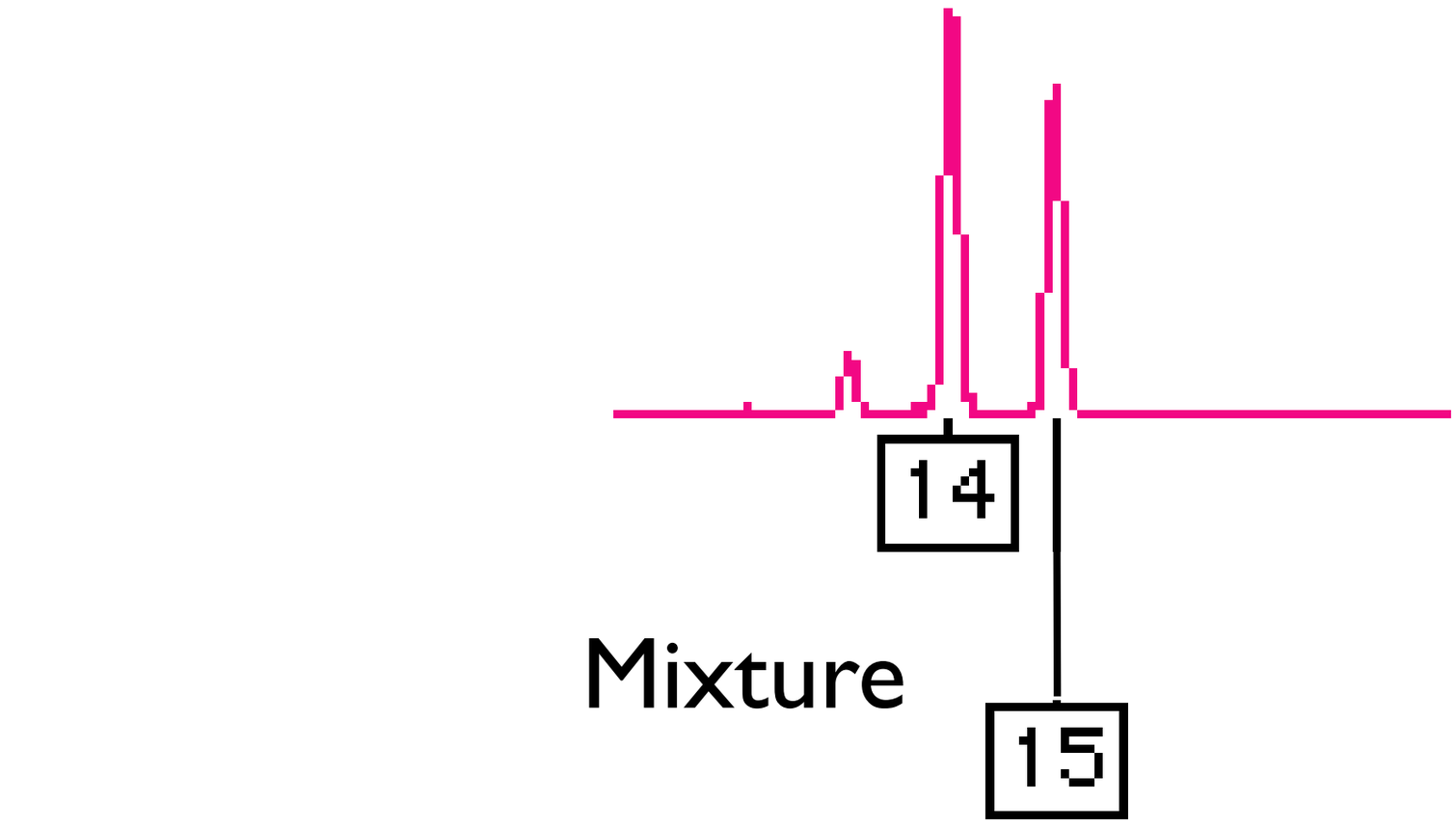


Fig. 1. Bayesian network fragment for modelling peak areas in a mixture.

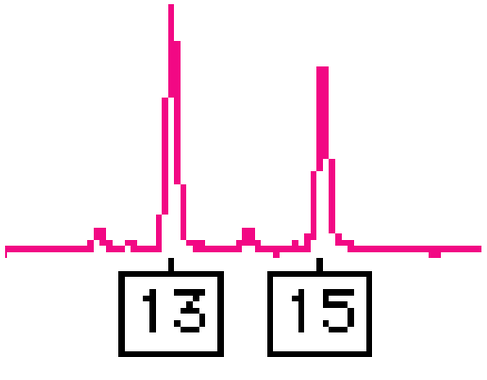
Uitgebreide model



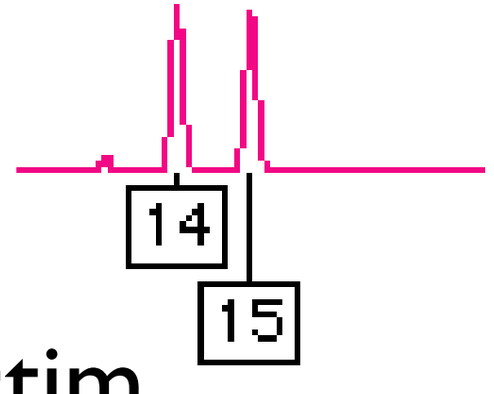
Locus D19 (niet problematisch... (?))



Mixture

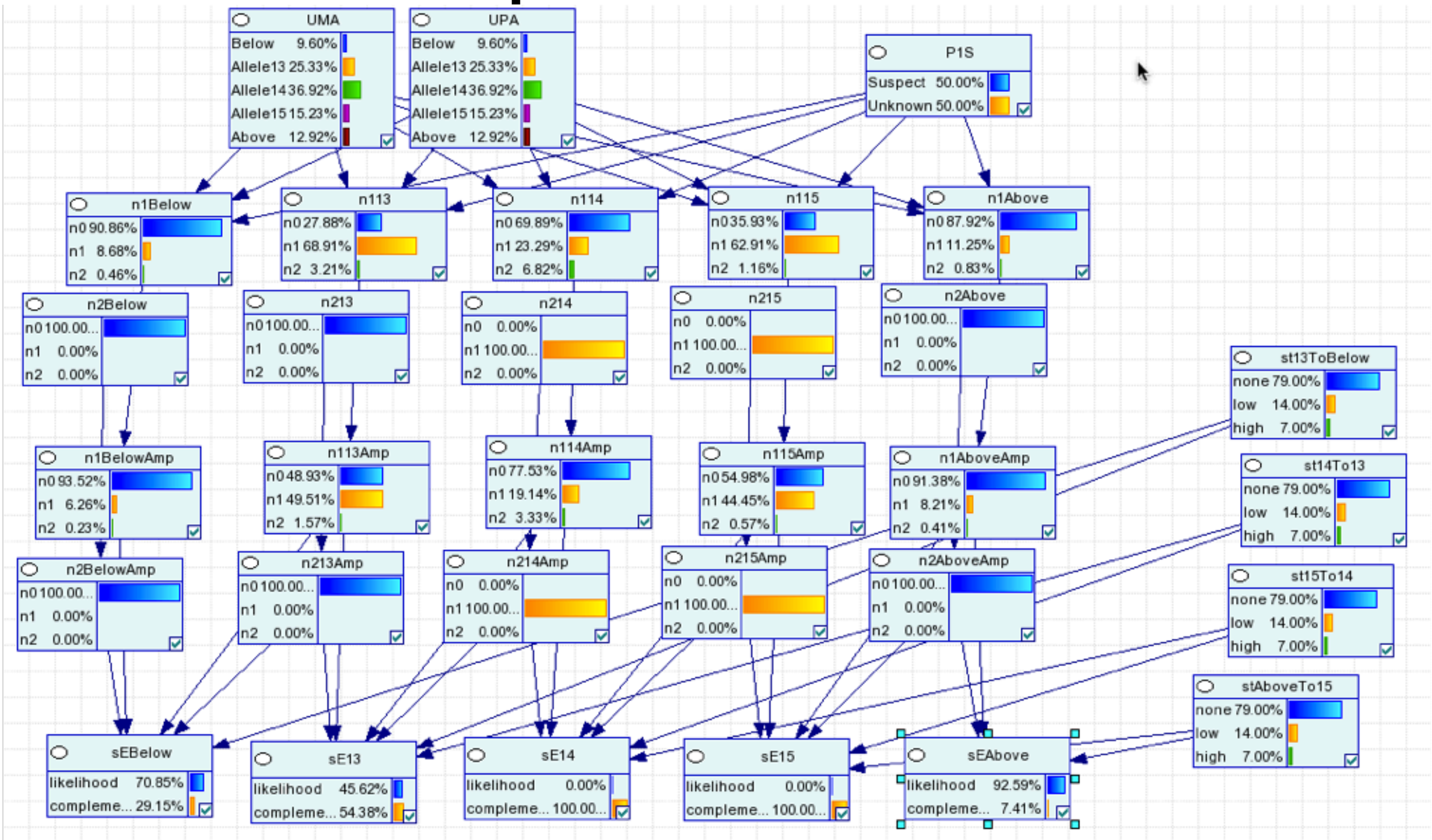


Suspect



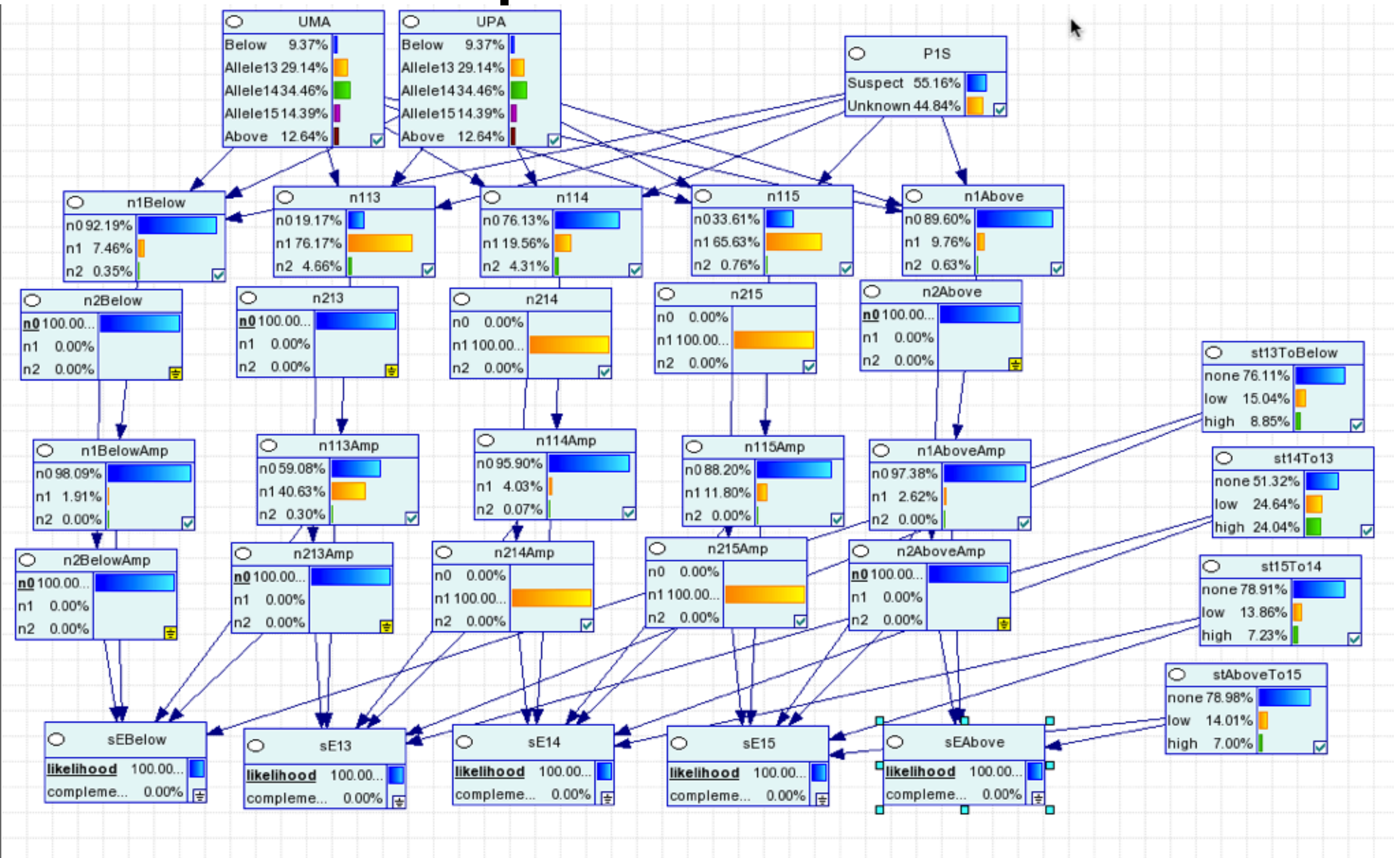
Victim

Locus D19: prior



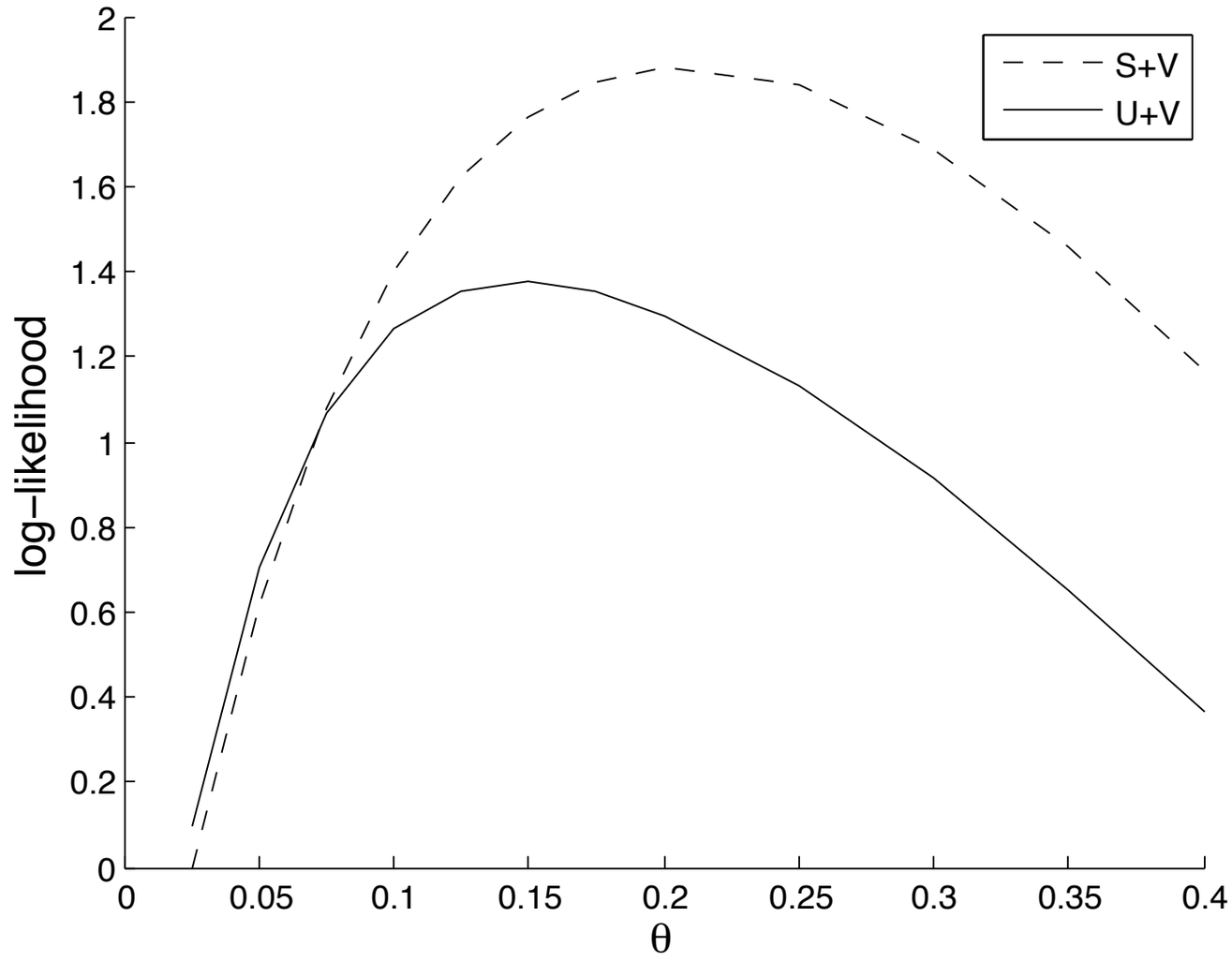
Veronderstel mengverhouding: 90:10

Locus D19: posterior



Veronderstel mengverhouding: 90:10

Wat zegt D19 over de meng-verhouding?



Mengprofiel komt *zeer goed overeen* met een 90-10 mengsel slachtoffer-verdachte, *behalve* bij D18

D18 is meest bekend locus voor *mozaicism* wat bij minstens 1 op de 5000 personen voorkomt

Evidentiele waarde van mengspoor voor slachtoffer +onbekende *versus* slachtoffer+verdachte+mozaicism is sterker dan 1:10 000 (“very strong evidence”)

Evidentiele waarde van mengspoor voor slachtoffer+twee onbekendes *versus* slachtoffer+verdachte+onbekende is sterker dan 1:10 000 <<<<<<

Ervaringen

- Het is mogelijk de afhankelijk van de inferentie op individuele "nuisance parameters" te visualiseren
- *Onderschatting* van statistische variatie leidt tot *overschatting* van evidentiele waarde van resultaat
- Aangezien we niet alle bronnen van variatie meenemen, is "overschatting" van *wel* meegenomen bronnen noodzakelijk, om verantwoord conclusies te trekken