

Kinship analysis with DBLR™



The Kinship module introduced in DBLR[™] v1.1

is an extremely powerful tool that allows you to assign likelihood ratios for a wide range of scenarios. By dragging and dropping, you can set up scenarios that may include incestuous pedigrees. You can allocate reference profiles or mixtures to selected individuals within the pedigree, then calculate the likelihood of the profiles given the scenario. If you have more than one scenario a likelihood ratio (*LR*) will be assigned.

THE MODULE CAN BE USED:

- for disaster victim identification and paternity testing.
- to assign *LR*s when the relationships of individuals within mixtures differs between propositions.
- to infer the genotypes of missing individuals using a pedigree.

Here we demonstrate several applications of the Kinship module within $\mathsf{DBLR}^{\mathsf{m}}.$

UNIDENTIFIED REMAINS

In this example, an unidentified body has been found. A partial single source DNA profile has been obtained from the remains. It is suspected that the remains have originated from a female, who has been missing for many years. There is no direct reference profile available for the missing woman.



FIGURE 1: Screen capture from pedigree builder in DBLRTM showing the putative relationship of the deceased under alternate scenarios

You have a putative reference profile from the biological mother (also deceased) of the missing woman, but it is a mixed DNA profile (collected from a comb). There is no reference profile from the biological father of the missing woman but there is a single source reference profile from his biological father (the paternal grandfather).

This relationship is shown graphically as a pedigree in the pedigree builder within the user interface of DBLR^M (*Figure 1*) where the individuals with reference profiles are filled (squares for males and circles for females). An alternate explanation is that the deceased is not related to members within the pedigree. This is represented by the pedigree on the right. By assessing both scenarios an *LR* can be calculated.

PATERNITY: WHO'S YOUR DADDY?

In our second example, a simple disputed paternity case is described. Reference DNA profiles are available from a child (C), his biological mother (M), and the putative biological father (PF).

The proposed scenarios are displayed graphically in Figure 2, again using the pedigree builder within DBLR[™].

As for all scenarios within the Kinship module of DBLR™ the inputs can be:

- text files (for example from FaSTR[™] DNA) for single source profiles,
- · .csv summary files, or
- STRmix[™] deconvolutions for mixed or single source profiles (including partial profiles).

Cases investigated using the Kinship module could also include incestuous relationships.



FIGURE 2: Pedigree for a disputed paternity case

DISPUTED RELATEDNESS BETWEEN PROPOSITIONS

When calculating LRs for mixed DNA profiles, the different contributors to the mixture are traditionally assumed to be unrelated to each other. Using the Kinship module within DBLR^M we can assign likelihoods for mixtures where the donors are assumed to be related. Any relationship can be tested.

In this example, we have a mixed DNA profile from two contributors. In addition, we have a reference profile from one person of interest (POI). There is no





FIGURE 3: Pedigrees demonstrating alternate propositions where H2 considers that the DNA originated from two full siblings, who are unrelated to the POI

evidence that the two donors are related and under the prosecution proposition it is assumed that they are not. The defence have proposed that the two donors are full siblings, who are unrelated to the POI. We use the Kinship module within DBLRTM to assign an *LR* considering the following propositions:

H1: The DNA originated from the POI and one other individual, unrelated to the POI

H2: The DNA originated from two full siblings, who are unrelated to the $\ensuremath{\text{POI}}$

CONTACT

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This is shown in Figure 3. Under H1, unrelated individuals A and B are donors to the mixture and the POI is individual B. Under H2, full brothers C and D are donors to the mixture and the POI is not a contributor and not related to the brothers.

The Kinship module is very flexible and likelihoods for various other propositions may be assigned, including assuming they are related given both scenarios, either given the same relationship or a different one (for example, full siblings versus half siblings).

INFERRING GENOTYPES OF MISSING PEOPLE IN A PEDIGREE

In cases where the person of interest is missing, their genotype may be inferred from the genotypes of known family members. This is a type of familial searching, however the Kinship module within DBLR[™] is a much more powerful method where you are not limited to pairwise relationships with known individuals (parents, children, or siblings) but can combine the data from known references in a pedigree.



FIGURE 4: Pedigree demonstrating that the genotype of ? will be inferred using the mother (M) and sibling profiles (C2 and C3)

Consider a child abduction case. You don't have a suitable reference profile for the child to load to a missing person's database. You do have the reference profiles of the child's biological mother and two full siblings. The pedigree is shown in Figure 4, where the individuals with reference profiles are filled.

The genotype of the missing child (? within the pedigree) can be inferred using the genotypes of the biological mother and two biological brothers. The weighted genotypes can be exported and used to search or load to a database (e.g. remains or reference samples from missing persons).

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