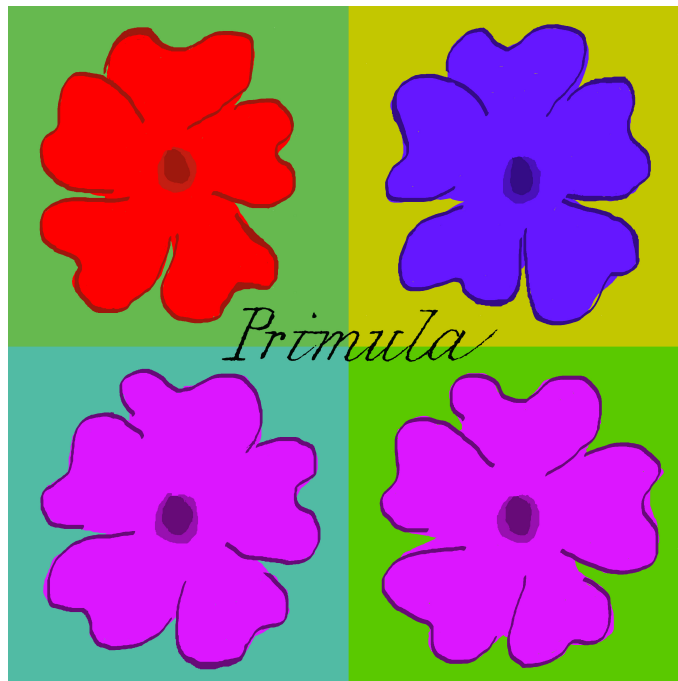# The *Primula* System: user's guide
# Version 3.0
# Example: `Mendel`

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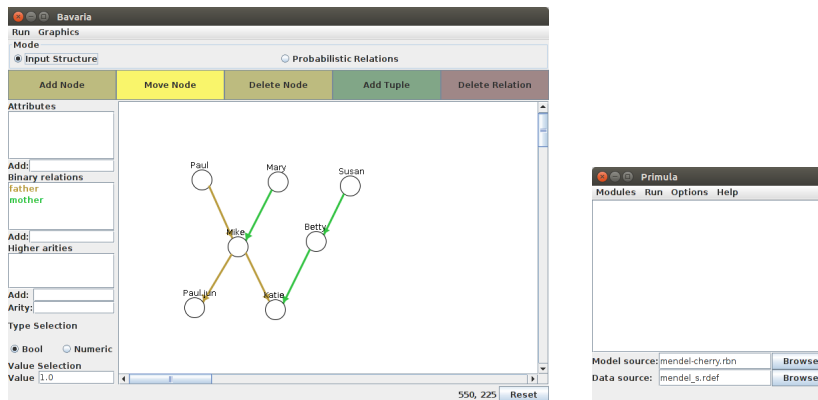*Primula* homepage: www.cs.aau.dk/∼jaeger/Primula

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# Probabilistic Inference: Mendelian Model of Inheritance

Load the model file `Mendel/mendel.rbn` and the data file `Mendel/mendel_s.rdef`.

Select in the *Primula* console Modules:Bavaria to open the graphical data editor. You should now see the two windows:



The *Bavaria* window displays the relational structure contained in `mendel_s.rdef`, which represents a small family tree with 7 individuals.

The model `mendel.rbn` is a model of inheritance for a gene with two alleles $A$ and $a$. A person $P$ can then have one of the genotypes $AA$, $Aa$ or $aa$, which is represented by three corresponding Boolean predicates. The model furthermore uses two auxiliary Boolean predicates *AMother* and *AFather*, which are true for a person if they have inherited allele $A$ from their mother, respectively father. The model represents the random selection of one allele from each parent, together with a small probability of a random mutations. More details can be found in the file `mendel.rbn`.

**Probabilistic inference with Bayesian network in SamIam** (requires that SamIam is installed). The model `mendel.rbn` together with the family tree `mendel_s.rdef` defines a joint probability distribution for the genotype variables of all the persons in the familty tree. To perform probabilistic inferences for this distribution: select Run:Construct Bayesian Network in the Primula console. This will open the SamIam system with the corresponding Bayesian network. We can now compute, e.g., that conditional on the evidence *aa(Mike)=true*, the posterior probabilities for *AA(Katie)*, *Aa(Katie)*, and *aa(Katie)* are, respectively, 0.0018, 0.306, and 0.691. See the SamIam documentation for details.
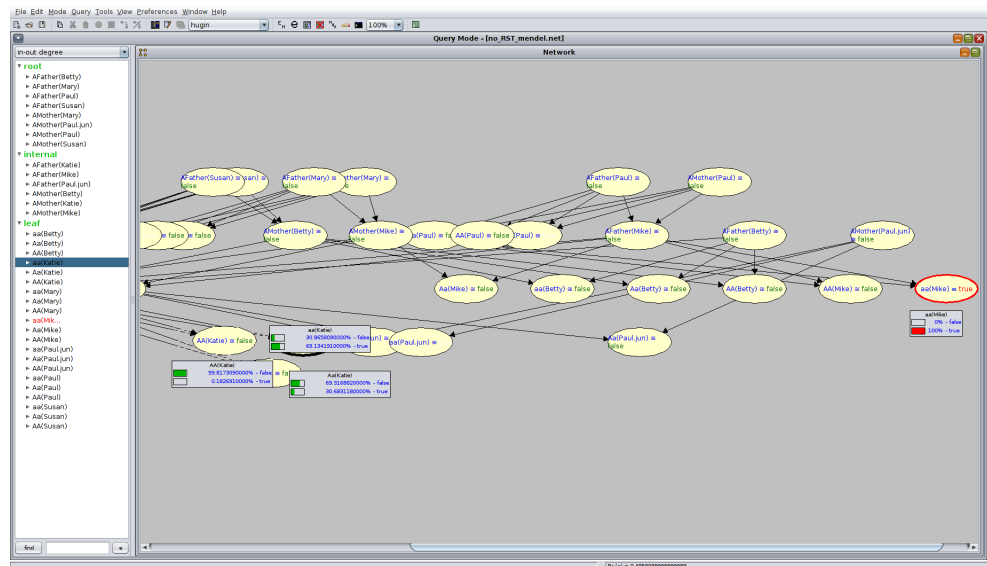
**Probabilistic inference in inference module** Select Modules:Inference Module from the *Primula* console. To enter the evidence *aa(Mike)=true* select the 'Evidence' tab at the bottom, select *aa* in the 'Attributes' list and 'true' in the 'Values' list, and click on *Mike* in the 'Element names' list. To specify the query, select the 'Query' tab at the bottom, and in turn select the *AA*, *Aa*, and *aa* from 'Attributes' and *Katie* from 'Element names'. Select in the bottom panel the MCMC tab and press 'Start'. This will initiate an importance sampling process for estimating the probabilities of the query atoms. The sampling continues until the 'Stop' button is pressed. The 'P' column in the middle panel shows the current approximation of the query probabilities.