Construction of coalescent trees on partially fixed pedigrees

Renaud Alie, Sorana Froda & Fabrice Larribe

Université du Québec à Montréal

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Genealogies

- Φ : Phenotypes

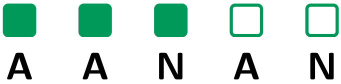
  A : Affected, **N** : Not Affected

- $\mathcal{H}$: Haplotypes

  ■ : Mutant, □ : Non-Mutant

- θ : Unknown parameter

$$\mathcal{L}(\theta) = P\left(\Phi, \mathcal{H} | \theta\right).$$



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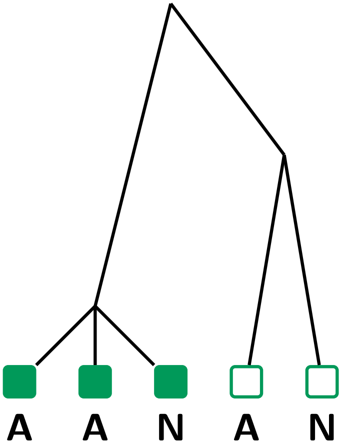
$$\begin{aligned}\mathcal{L}(\theta) &= P\left(\Phi, \mathcal{H} | \theta\right) \\ &= \int_{\mathcal{G}} P\left(\Phi, \mathcal{H} | \mathcal{G}, \theta\right) P\left(\mathcal{G} | \theta\right) \mathrm{d}\,\mathcal{G}.\end{aligned}$$



Diploid, Two-Sex, Wright-Fisher Model

Population size $2N$; N males; N females; $4N$ genes.

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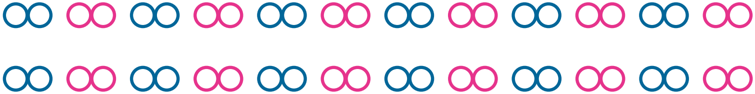

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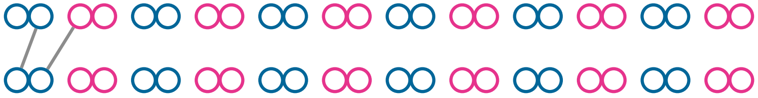

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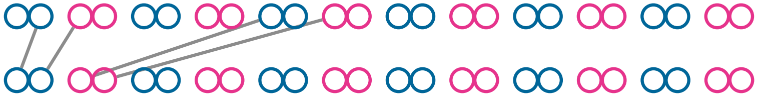

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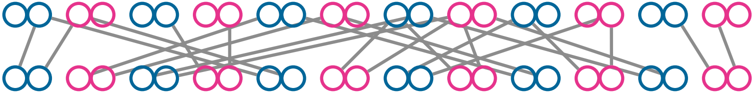

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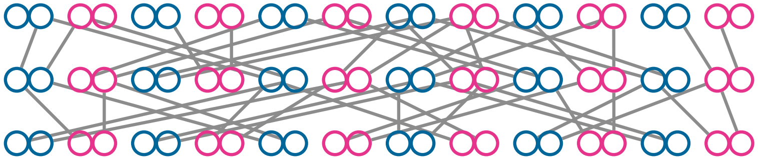

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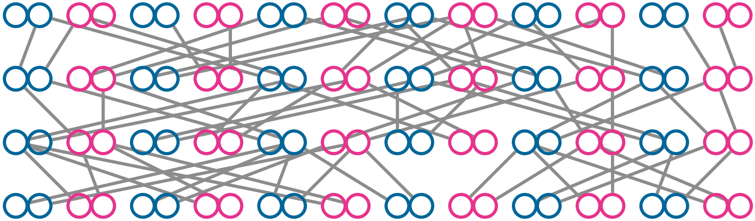

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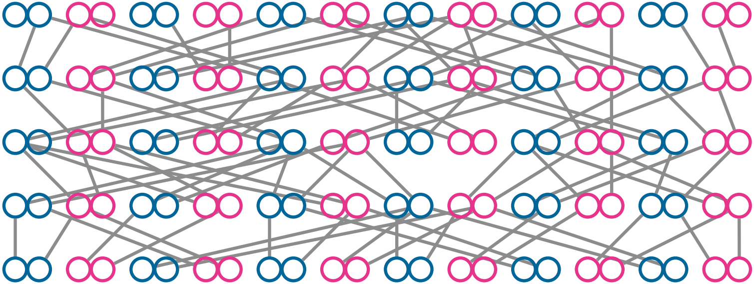

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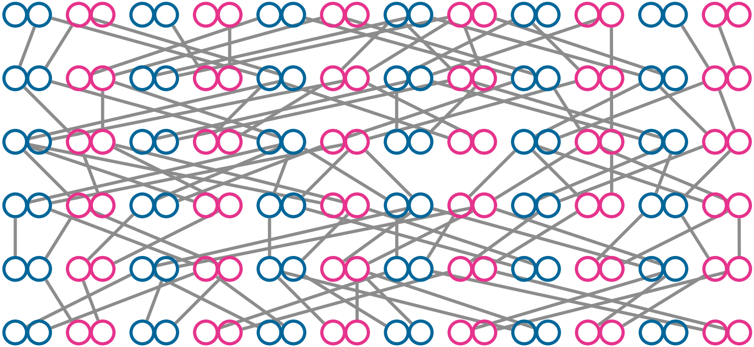

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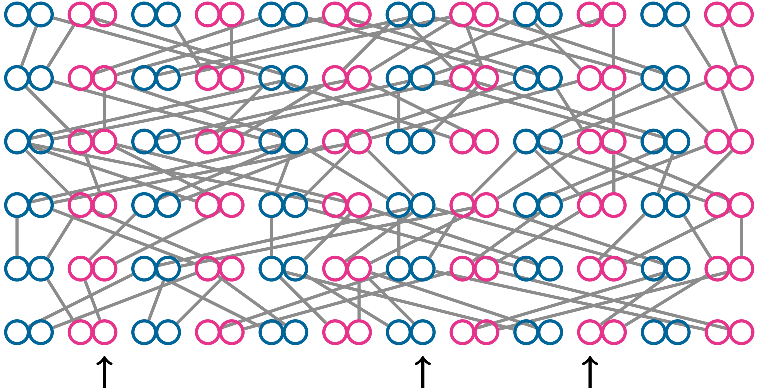

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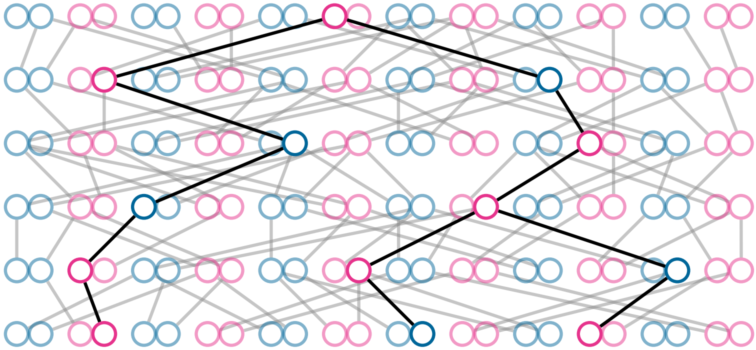

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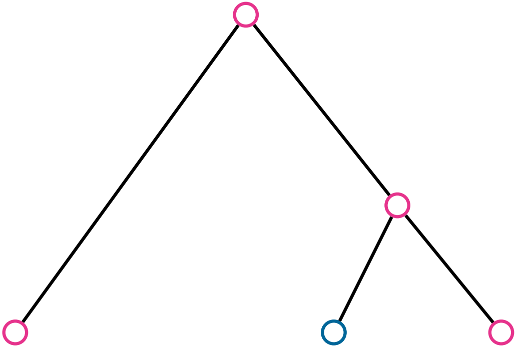

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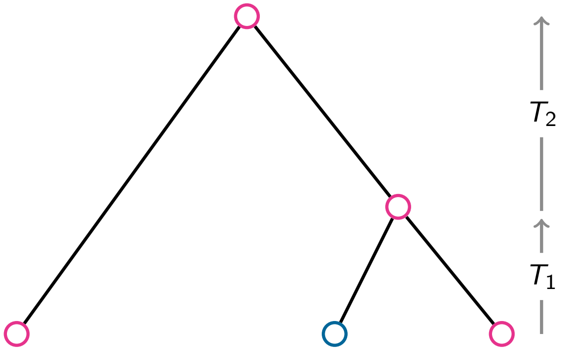

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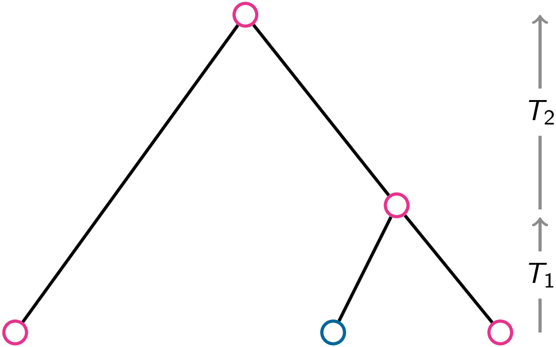
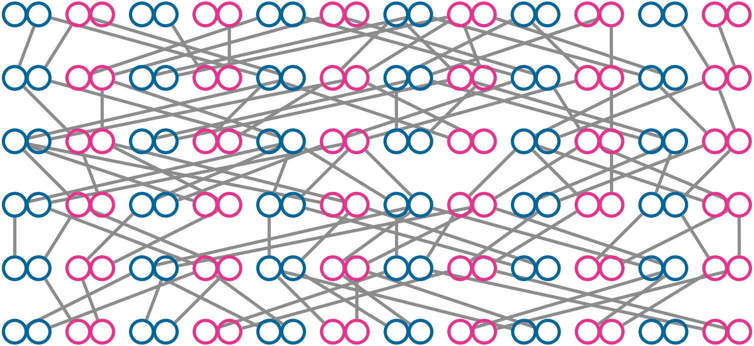
Convergence to the n-coalescent (Möhle [1998])

$$T_1/4N \sim \mathcal{E}xp(3),\ T_2/4N \sim \mathcal{E}xp(1); \quad N \text{ large.}$$



Population Pedigree



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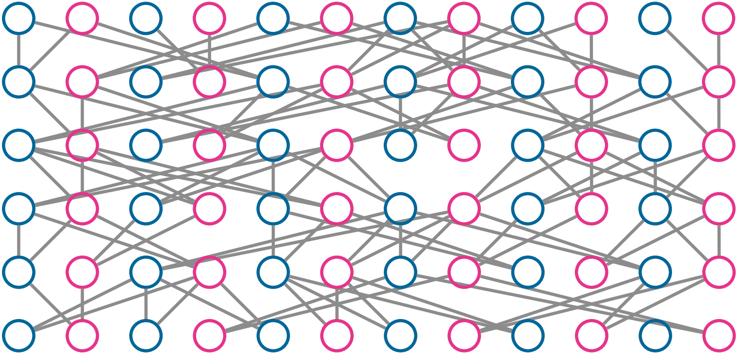

Population Pedigree (Wakeley et al. [2012])

The set of all family relationships among members of the population for every generation.

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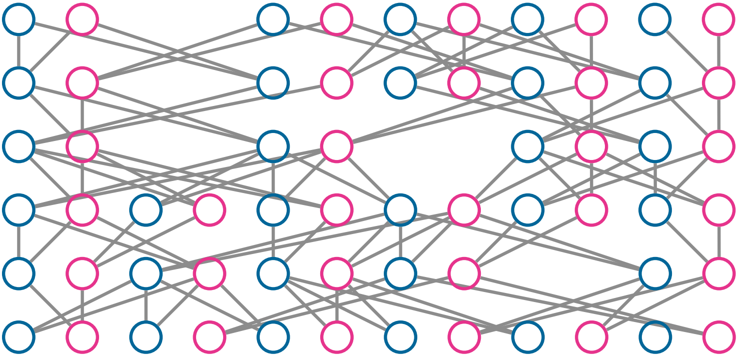

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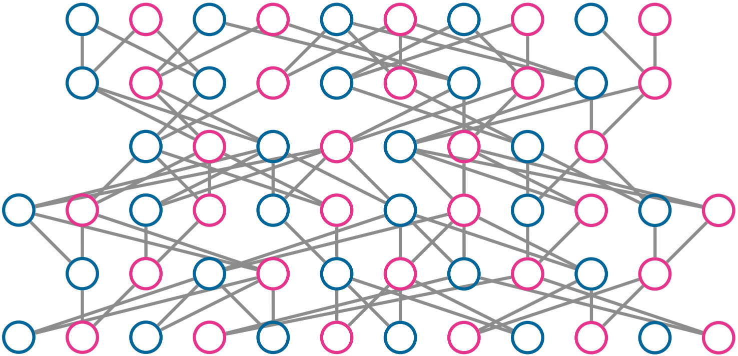

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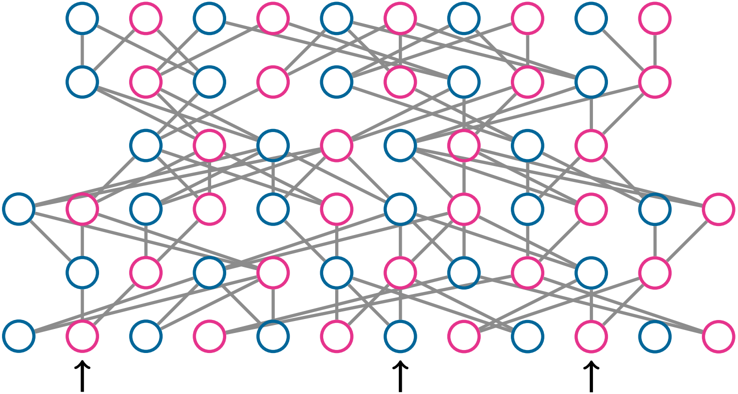

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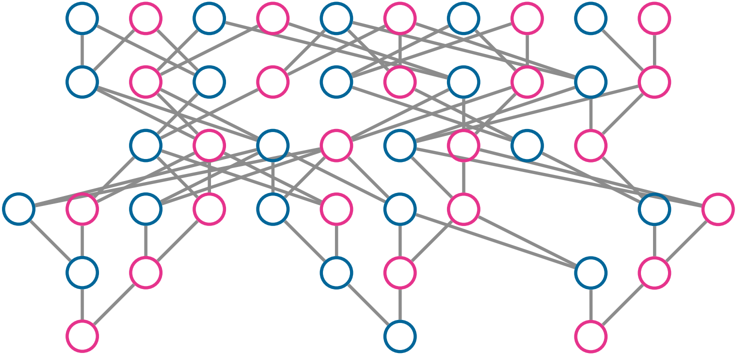

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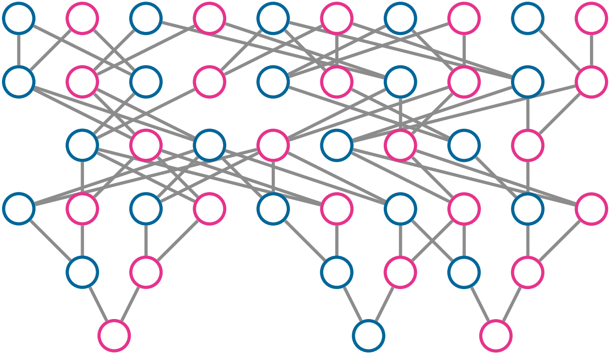

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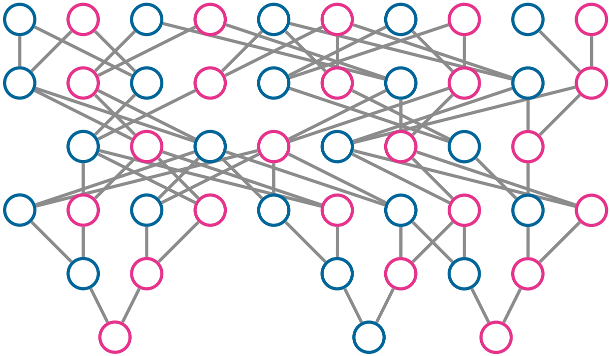

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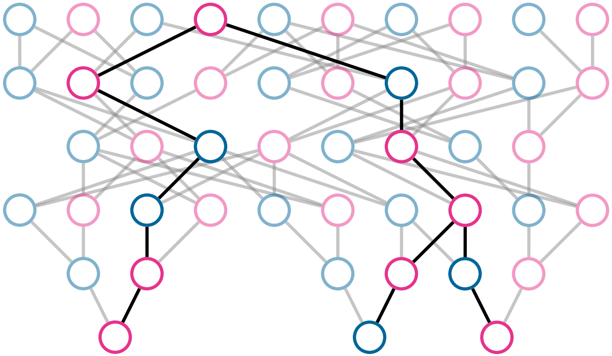

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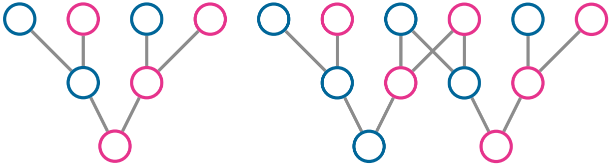

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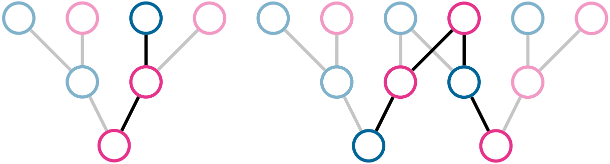

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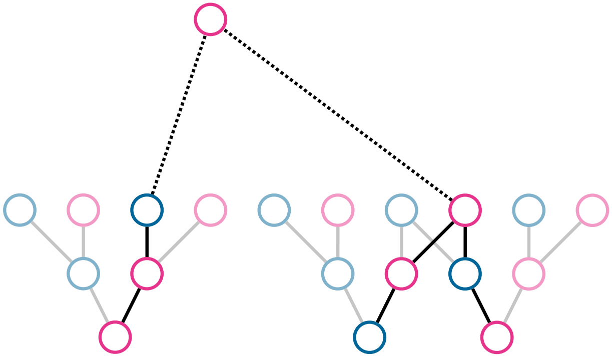

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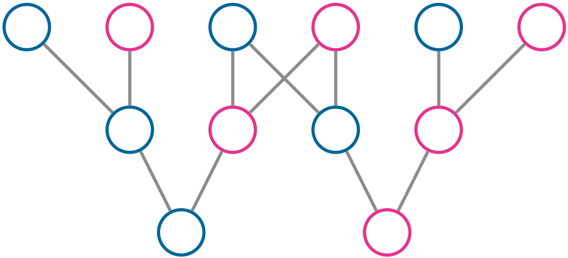

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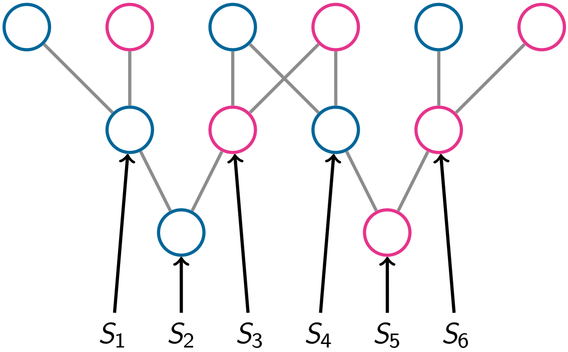

Meiosis Indicator (Thompson [2000])

$$S_i \sim i.i.d.\, \mathcal{B}ernoulli\,(0.5)\,.$$

Mendel's 1st Law

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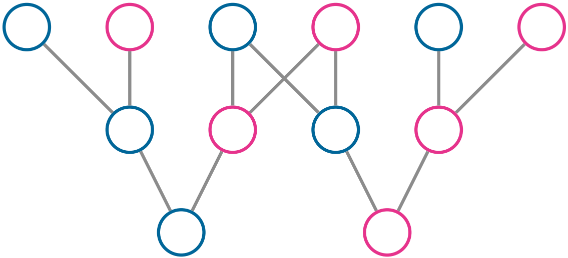

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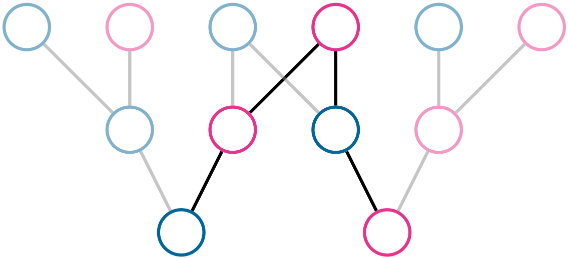

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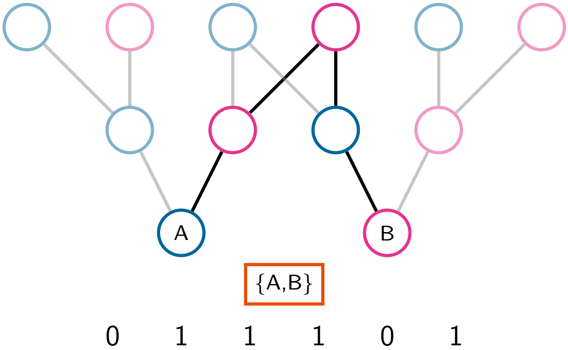

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Partition of genes into subsets that are IBD

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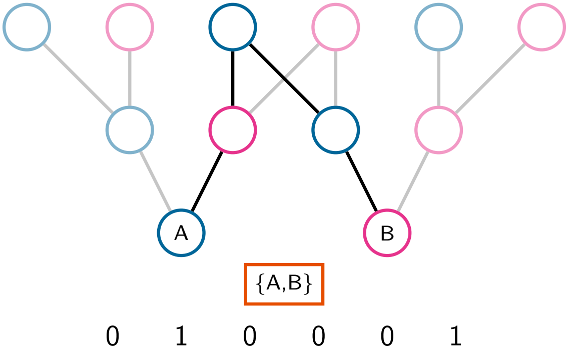

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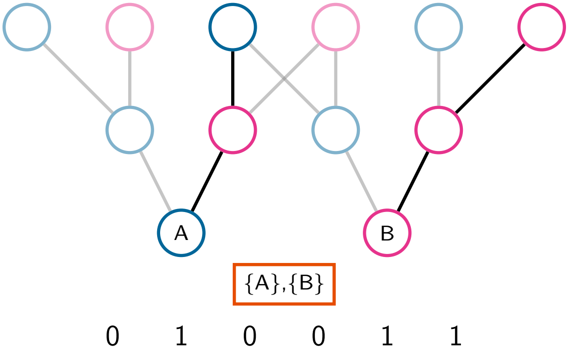

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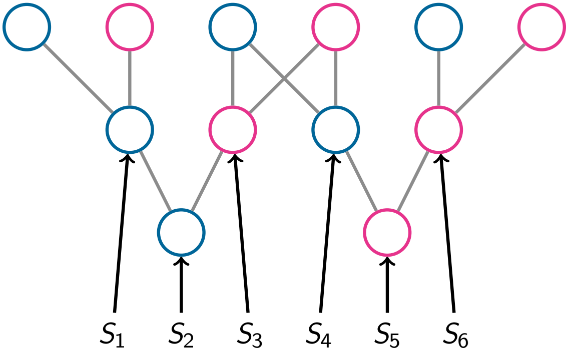

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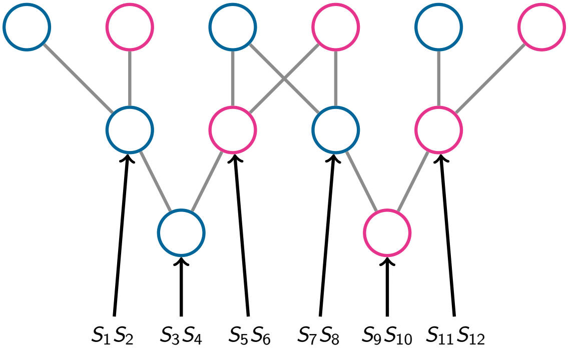

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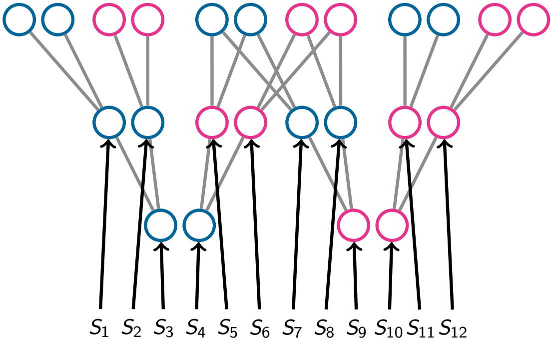

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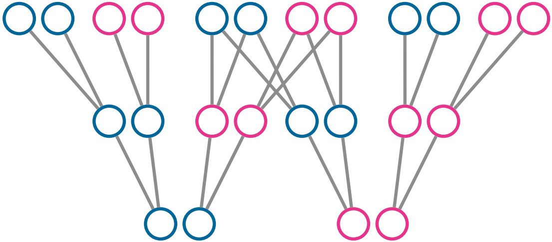

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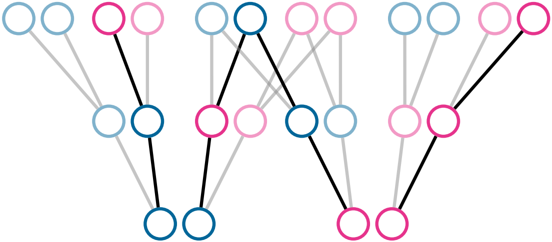

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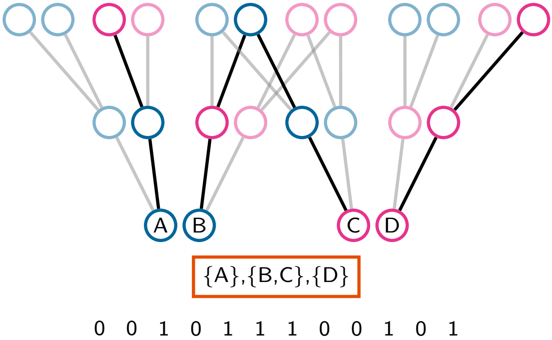

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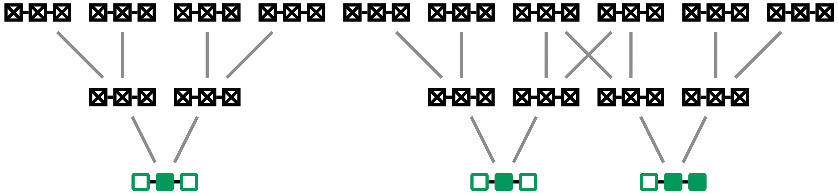

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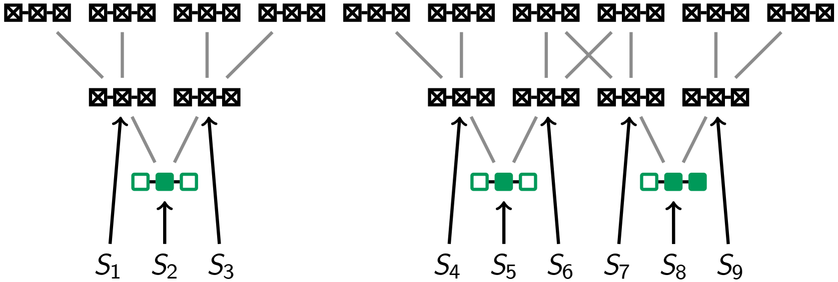

k-locus Meiosis Indicator

$$S_i \in \{0,1\}^k.$$

e.g. No interference (Allen and Darwiche [2008]) :

$$P(S_i = 011) = P(S_i = 100) = 0.5p_{12}(1 - p_{23}).$$

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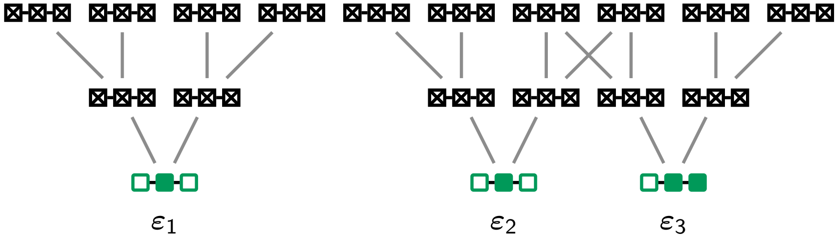

Evidence : ε (Koller and Friedman [2009])

An instantiation of a subset of random variables.

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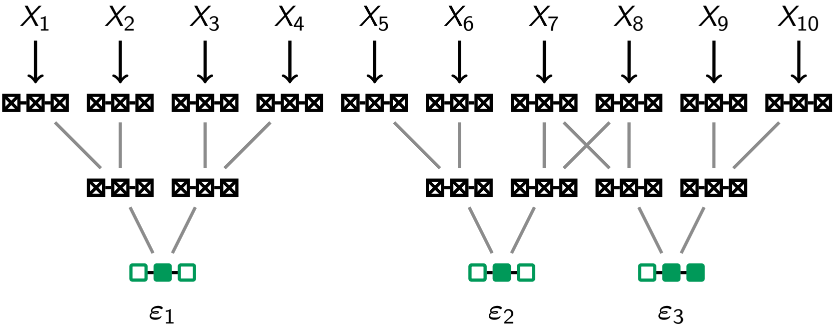

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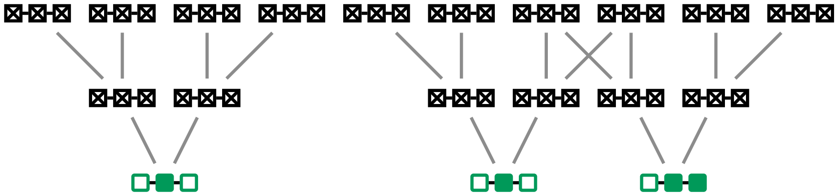

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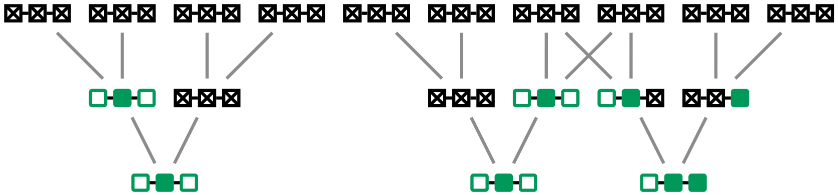

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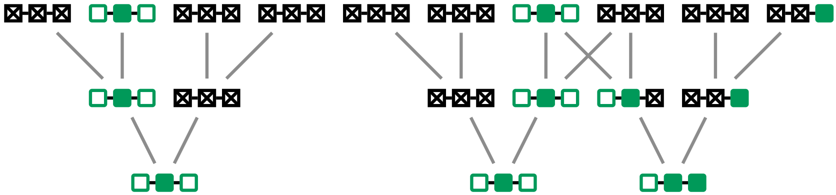

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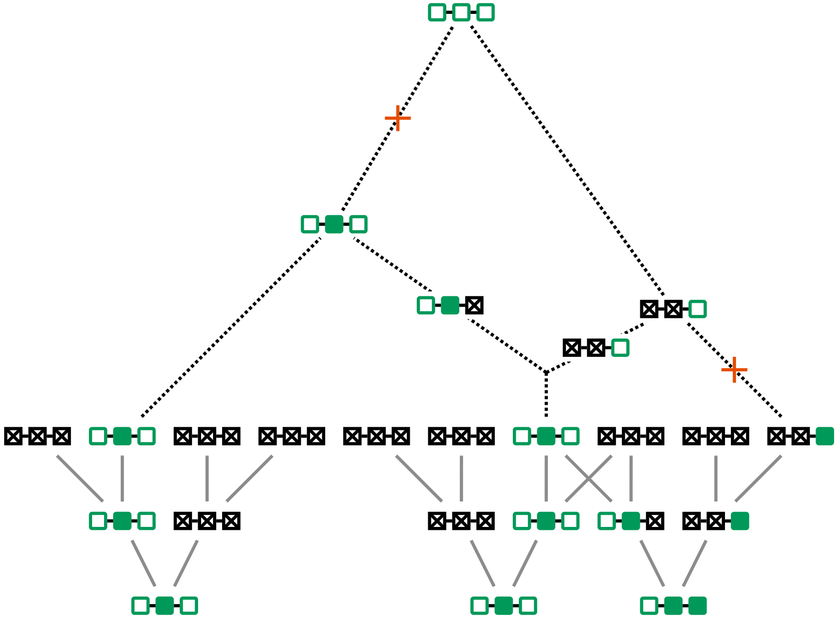

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Founder Prior

- Uniform prior
- Linkage equilibrium
- Linkage disequilibrium

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