Sveriges lantbruksuniversitet
Swedish University of Agricultural Sciences

Department of Animal Breeding and Genetics

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Simulation-based inference of complicated monogenic inheritance patterns

Goal

The project aims to prototype methods for simulation-based inference for monogenic traits with complicated inheritance patterns, such as variable penetrance, sex-specific effects, sex-linkage or transmission distortion.

Background

The inheritance of monogenic disorders is not simple, even if only one causative allele is involved. Diseases often have variable penetrance, dominance may be incomplete, effects may be sex-specific and autosomal as well as sex-linked. Inferring the mode of inheritance if often more a matter of researcher judgement from incomplete data than modelling. However, with increasing data sizes and efficient computational modelling, there are new opportunities.

Project description

This project will build computational models of monogenic disorders with complicated modes of inheritance—e.g., variable penetrance, incomplete dominance, sex-specific effects—to be able to simulate data for given inheritance parameters. Then, these will be combined with random sampling of parameters and summary statistics for simulation-based inference. Methods will be tested on simulated pedigrees with known ground truth.

Useful previous knowledge: Some background in population, quantitative or evolutionary genetics. R and C++ programming. Interest in veterinary or medical genetics.

Start time: Flexible

Contact

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