

Familial Aggregation Analysis of Continuous Traits

MSc Thesis

Christian Weichenberger and Francisco Domingues

Eurac Research

The aim of familial aggregation is to identify families where a disease or a binary trait tend to cluster more than expected by chance, which could point to an underlying genetic cause. These individuals are split into "affected" and "non-affected" according to disease status or a binary trait. Examples of disease status and binary traits are "type 2 diabetes" or "high systolic blood pressure".

When investigating continuous traits such as systolic blood pressure, we might want to define as "affected" the individuals with extreme values based on a cutoff criteria. For familial aggregation analysis it would be desirable to also consider individuals in the family with borderline values to account for milder traits in the family.

At Eurac Research we have developed FamAgg^{1,2,3}, an R Bioconductor package which performs familial aggregation analysis on a set of affected individuals given a pedigree. The proposed project aims to extend this methodology to continuous traits. The work involves the planning and design of these improvements, implementation of these improvements in the FamAgg package, and testing and validation of these enhancements.

1. <https://bioconductor.org/packages/release/bioc/html/FamAgg.html>
2. Rainer et al. *Bioinformatics* 2016 32:1583-1585.
<https://doi.org/10.1093/bioinformatics/btw019>
3. Weichenberger et al. *Bioinformatics* 2019 35:69-76.
<https://doi.org/10.1093/bioinformatics/bty541>