

Diagnosis of genetic disorders by transcriptome sequencing

Master thesis @ Gagneur lab, I12 - Computational Biology, TU Munich

The lab of Julien Gagneur is looking for a student to perform his/her Master thesis. The goal of our lab is an improved understanding of the genetic basis of gene regulation and its implication in diseases.

Background

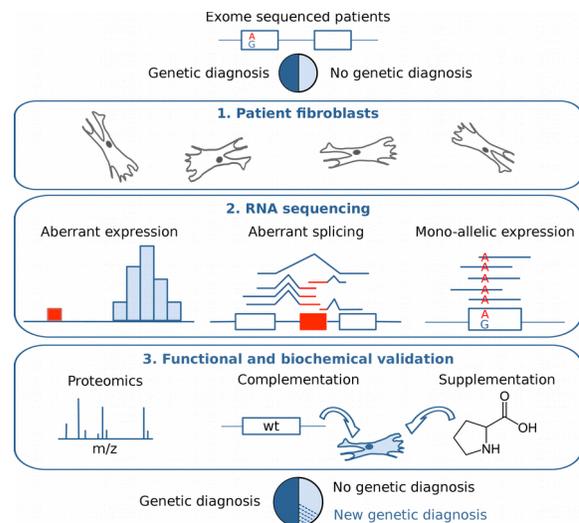
Collectively, rare genetic disorders affect 1 in 17 Europeans. Pinpointing the genetic cause is of major impact for the affected patients and their families: understanding the biological background of the disease, initiating development of therapies, identification of other mutation-carriers among relatives and joining networks of families affected by the same variant. To this end, major progresses have been achieved in the last years with affordable genome sequencing. However, 50 to 75% of rare disease patients do not receive a genetic diagnosis from their genome data alone. We have recently demonstrated the power of combining genome and transcriptome data to boost up genetic diagnosis in a pilot study [[Kremer, Bader et al. 2016 bioRxiv](#)], by revealing defects affecting gene expression of one allele or an entire gene, and aberrant splicing (Figure).

Project Outline

- develop efficient bioinformatic methods for genetic diagnosis
- analyze an unique dataset of combined exome and transcriptome of more than 200 undiagnosed patients
- collaboration with bioinformaticians, biologists and clinicians

Skills to develop

- programming with [R](#) statistical language
- designing interactive patient reports using [Rmarkdown](#), [plotly](#) and [shiny](#)
- adapting [Bioconductor](#) packages and bioinformatic tools
- parallelize your code to leverage the power of compute clusters (i.e. [SLURM](#))
- using [GitLab](#) versioning system to track and document your code



The project is open to bioinformatics, and computer science students. A taste for data science and interest in biological and medical applications is expected. Fluency in English is essential. Our group is a young, international and multidisciplinary group. To apply provide your CV and a brief motivation to:

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