PhD - Machine learning to integrate genome, transcriptome and patient outcome in neurological disease

UMC Utrecht

Project outline. Work to decode the genetics of amyotrophic lateral sclerosis (ALS), a devastating neurological disease that strikes 1 in 300 people. You will contribute to analyses of one of the largest genetic datasets in the world and combine the state of the art in machine learning, computational genetics and multi-omic profiling of human tissue.

Techniques

- Statistical programming & data analytics (R, python)
- Deep learning (Keras)
- High performance computing
- Human genetics (disease gene discovery, genome interpretation)
- Integrative transcriptomics (bulk/ single cell RNAseq)
- Wetlab: RNAseq (bulk/single cell), target validation

Additional background. A complete understanding of genetic risk factors could revolutionize ALS treatment options and enable effective genetic counseling for patients and their families. To achieve this, the ALS community initiated project MinE. This global consortium (coordinated by the Veldink group in UMC Utrecht) has generated whole genome sequencing for >10,000 patients and healthy controls. The challenge for this full-time 4 year PhD position will be applying novel methods to determine which of the hundreds of millions of DNA variants identified by project MinE are relevant to ALS. The project can be 70% computational + 30% wetlab work OR 100% computational depending on the applicant.

Candidate. We're looking for an enthusiastic candidate eager to engage in challenging high impact research. You will have an interest in big data, problem solving and analytical thinking. We welcome applicants from **any analytical discipline** including but not limited to bioinformatics, genetics, neuroscience, data science, mathematics, engineering etc. Teamwork and basic English are essential. **You are not required to speak Dutch** (all meetings are in English). Preference will be given to applicants with prior experience in genomics, machine learning, biostatistics and/or programming. Wetlab experience relevant to DNA/RNA sequencing library preps (bulk/single cell/ ChIP) is desirable but not essential.

Work Environment. Utrecht University (UMC Utrecht) is consistently ranked among the top 15 universities in Europe (1st in Netherlands) and is an internationally recognized centre of excellence for ALS research and healthcare. You will be embedded within the Brain Centre and an extensive research programme that spans bioinformatics, genetics, translational neuroscience, stem cell models, environmental risk factors, neuroimaging, electrophysiology, patient care and clinical trials. The project supervisors have an extensive background in relevant disciplines (see publications by K. Kenna/ J.Veldink), and have led multiple high impact ALS gene discoveries (*Nature Genetics x3*, *Neuron* x2). For more general information about genomics/ computational biology in Utrecht see ubc.uu.nl.

Questions/ Applications (CV & cover letter)

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Project Start Date: 2020