PhD student in Scientific Computing focusing on Deep Learning for Genetic Epidemiology

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Uppsala University is a comprehensive research-intensive university with a strong international standing. Our ultimate goal is to conduct education and research of the highest quality and relevance to make a long-term difference in society. Our most important assets are all the individuals whose curiosity and dedication make Uppsala University one of Sweden's most exciting workplaces. Uppsala University has over 54,000 students, more than 7,500 employees and a turnover of around SEK 8 billion.

The Department of Information Technology has a leading position in research and all levels of higher education. Today the department has 280 employees, including 120 academic staff and 110 full-time PhD students. The Department comprises research and education in a spectrum of areas within Computer Science, Information Technology and Scientific Computing. More than 4000 students take one or several courses offered by the Department each year.

The position is hosted by the Division of Scientific Computing within the Department of Information Technology. As one of the world's largest focused research environments in Scientific Computing the research and education has a unique breadth, with large activities in classical scientific computing areas such as mathematical modeling, development and analysis of algorithms, scientific software development and high-performance computing. The division is currently in an expansive phase in new emerging areas such as cloud and fog computing, data science, and artificial intelligence, where it plays key roles in several new strategic initiatives at the University. The division currently hosts 20 PhD students and have awarded more than 80 doctorates. Several PhD alumni from the division are successful practitioners in the field of scientific computing and related areas, in industry as well as in academia around the world.
The Department of Information Technology also heads the Strategic Research Area (SFO) effort eSSENCE. It is a collaborative research programme in e-science between three Swedish universities with a strong tradition of excellent e-science research: Uppsala University, Lund University and Umeå University.

This PhD position is part of the eSSENCE - SciLifeLab graduate school in data-intensive science. The school addresses the challenge of data-intensive science both from the foundational methodological perspective and from the perspective of data-driven science applications. It is an arena where experts in computational science, data science and data engineering (systems and methodology) work closely together with researchers in (data-driven) sciences, industry and society to accelerate data-intensive scientific discovery.

SciLifeLab is a leading institution and national research infrastructure with a mandate to enable cutting-edge life sciences research in Sweden, foster international collaborations, and attract and retain knowledge and talent.

**Project Description**

The deep learning revolution in image analysis has been dependent on developing well-established validated and standardized model architectures, workflows, and pre-trained model coefficients that can be applied to new applications using transfer learning and fine-tuning. All of these are currently lacking for the genetics community.

The deep learning models that do exist for genomics do not readily support training with the large number of genetic variants investigated in most studies, nor the number of individuals, that characterize large-scale study cohorts, such as the UK Biobank. A deep learning approach holds the promise to truly understand the variability and patterns of human genomes, including the ability to directly find connections between genetic variation and disease risk, at scale. Within this project, using the UK Biobank as a starting point, we hope to develop a deep learning toolbox for analyzing large-scale genomic data. Similar to the standard resources for versatile applications that now exist in image analysis, we believe that this could be the start of a new data-driven era for research in genetic epidemiology.

The PhD student will start out with our already published model [A deep learning framework for characterization of genotype data](#), and adapt it to relevant applications and training on the scale of these applications in close collaboration between the
main supervisor, Associate Professor Carl Nettelblad, and the group of the assistant supervisor, Associate Professor Åsa Johansson, at the Department of Immunology, Genetics and Pathology.

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Duties
The duties of a PhD student are primarily directed at their own research education, which lasts four years. The work may also involve, to a limited extent (ca 20%) other departmental duties, such as teaching undergraduate courses and administrative tasks – in which case the position may be extended to a maximum of five years.

Requirements
A PhD position at the Division requires a Master of Science or equivalent in a field that is relevant to the topic of the project, good communication skills and excellent study results, as well as sufficient proficiency in oral and written English. Additional requirements for this position include basic knowledge of, and interest in machine learning, and proficiency in programming (e.g., in Python and possibly modern C++/CUDA).

Additional qualifications
Extra merits with equal merits include knowledge and experience in numerical optimization, deep learning, bioinformatical approaches in genetics, and best practices in software engineering.

Rules governing PhD students are set out in the Higher Education Ordinance chapter 5, §§ 1-7 and in Uppsala University's rules and guidelines.

About the employment
The employment is a temporary position according to the Higher Education Ordinance chapter 5 § 7. Scope of employment 100 %. Starting date 2022-09-01 or as agreed. Placement: Uppsala.

For further information about the position, please contact: Associate Professor Carl Nettelblad, carl.nettelblad@it.uu.se.
Please submit your application by 16 May 2022, UFV-PA 2022/1495.

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Please do not send offers of recruitment or advertising services.

Submit your application through Uppsala University's recruitment system.

Placement: Department of Information Technology
Type of employment: Full time, Temporary position longer than 6 months
Pay: Fixed salary
Number of positions: 1
Working hours: 100 %
Town: Uppsala
County: Uppsala län
Country: Sweden
Union representative: ST/TCO tco@fackorg.uu.se
Seko Universitetsklubben seko@uadm.uu.se
Saco-rådet saco@uadm.uu.se
Number of reference: UFV-PA 2022/1495
Last application date: 2022-05-16

Apply for position