School of Computing, Engineering, and the Built Environment
Edinburgh Napier University

PHD STUDENT PROJECT

Application instructions:
Detailed instructions are available at :
https://www.napier.ac.uk/research-and-innovation/doctoral-college/how-to-apply

Prospective candidates are encouraged to contact the Director of Studies (see details below) to discuss the project and their suitability for it.

Project details

Supervisory Team:

- DIRECTOR OF STUDY: Dr. Luigi La Spada (Email: L.LaSpada@napier.ac.uk)
- 2ND SUPERVISOR: Dr. Maria Elena Candela (The University of Edinburgh)

Subject Group: Cyber Security and System Engineering

Research Areas: Artificial Intelligence, Developmental Biology, Genomics

Project Title: Quantum-Integrated Genomic Models for the Advancement in Early Detection and Understanding of Neurodevelopmental Disorders

Project description:

Developmental disorders (DDs) are defined as a group of conditions manifesting as cognitive, social, and motor dysfunctions The existing literature showcases the potential of Artificial Intelligence (AI) and genomics in advancing our understanding and diagnosis of DDs. Fitzgerald (2015) shows the power of a genotype-driven approach in identifying novel genes associated with developmental disorders, increasing the proportion of diagnosed cases. Tang (2023) applies machine learning to mRNA expression data and identifies key biomarkers for early diagnosis of autism spectrum disorder (ASD). FitzPatrick (2020) highlights the transformative impact of genomic sequencing technologies in making accurate genetic diagnoses in severe developmental disorders. Wright (2018) emphasizes the importance of iterative reanalysis of genomic data, leading to an increased diagnostic yield in developmental disorders. The research gap addressed in this project revolves around the uncharted territory of employing advanced AI techniques to decode the complex genetic landscape associated with DDs. In particular, the objective is to

employ AI to unearth genetic markers, defined as distinct DNA sequences, associated with DDs. Utilizing sophisticated AI paradigms like machine learning (ML) and deep learning (DL), the project is set to analyse voluminous genetic datasets to delineate and comprehend these markers. The initial phase encompasses a thorough examination of prevailing research and genomic data pertinent to DDs. Subsequently, ML and DL will be harnessed to traverse the genetic data, aiming to discover novel markers and decode their functional significance in DDs. Additionally, AI-centric tools will be crafted to authenticate the findings through computational simulations and laboratory examinations. The anticipated outcomes encapsulate the discovery of novel genetic markers linked to DDs, thereby enriching the understanding of these disorders and potentially heralding enhanced diagnostic and therapeutic strategies. This project underscores the efficacy of AI in strengthening the comprehension of the genetic foundations of DDs, steering towards the realms of personalized medicine in neurodevelopmental science.

References:

[1] Fitzgerald, T.W., et al. (2014). Large-scale discovery of novel genetic causes of developmental disorders. Nature, 519, 223 - 228.

[2] FitzPatrick, D.R., & Firth, H.V. (2020). Genomically Aided Diagnosis of Severe Developmental Disorders. Annual review of genomics and human genetics.

[3] Tang, H., Liang, J., Chai, K., Gu, H., Ye, W., Cao, P., Chen, S., & Shen, D. (2023). Artificial intelligence and bioinformatics analyze markers of children's transcriptional genome to predict autism spectrum disorder. Frontiers in Neurology, 14.

[4] Wright, C.F., McRae, J.F., Clayton, S., Gallone, G., Aitken, S., Fitzgerald, T.W., Jones, P., Prigmore, E., Rajan, D., Lord, J., Sifrim, A., Kelsell, R.E., Parker, M., Barrett, J.C., Hurles, M.E., FitzPatrick, D.R., & Firth, H.V. (2017). Making new genetic diagnoses with old data: iterative reanalysis and reporting from genome-wide data in 1133 families with developmental disorders. Genetics in medicine : official journal of the American College of Medical Genetics, 20, 1216 - 1223.

Candidate characteristics

Education:

A first-class honours degree, or a distinction at master level, or equivalent achievements in Biomedical Engineering, Computer Science or Computational Biology, or Bioinformatics

Subject knowledge:

- Artificial Intelligence and Machine Learning
- Statistics and Data Analysis
- Mathematics
- Desirable:
- Genomics and Molecular Biology
- Neuroscience (preferably with a computational orientation)

Essential attributes:

- Research Aptitude
- Computational Proficiency
- Statistical Acumen
- Biological Insight

Desirable attributes:

- Advanced Computational Skills
- Prior Research Experience in Genomics
- Proficiency in Bioinformatics Tools