

## BioISI - Biosystems & Integrative Sciences Institute

### **Developing machine learning approaches to discover gene-disease associations from biological network data**

Place of work/: Instituto Nacional de Saúde Doutor Ricardo Jorge/Remote

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Establishing gene-phenotype associations for complex, multifactorial diseases is an important problem in computational biology. A paradigmatic complex disease is Autism Spectrum Disorder (ASD). ASD has a very heterogeneous clinical presentation along two main diagnostic dimensions, and dozens of candidate genes have been implicated in this disorder. However, establishing direct correlations between clinical presentation and specific genes has proven very difficult. The main problem may be that, for each affected individual, a specific cluster of altered behaviors is likely related with variants in multiple genes, possibly impacting more than one biological process. Artificial intelligence approaches are needed to deconstruct this complex architecture. This project proposes a combined machine learning and systems biology approach to the identification of gene-phenotype associations. The methods developed in the scope of this project will be applied to the discovery of genes and biological processes associated with Autism Spectrum Disorder (ASD) and, through this, contribute to a better understanding of the genetics of this complex disorder.

This approach consists of building large-scale heterogeneous networks from publicly accessible biological databases and using graph machine learning methods to learn the relationships between the biological entities (genes, diseases, biological processes, etc.) which compose the network. By using link prediction methods will be possible to identify previously unknown relationships between components of interest in the network.

Students selected for this project, after thesis registration, are eligible to apply to the BioISI Junior Programme (supporting 8 students with a 6-month Scholarship(BII), being the selection criterium the academic merit of the candidates.



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Bibliography: (if applicable)