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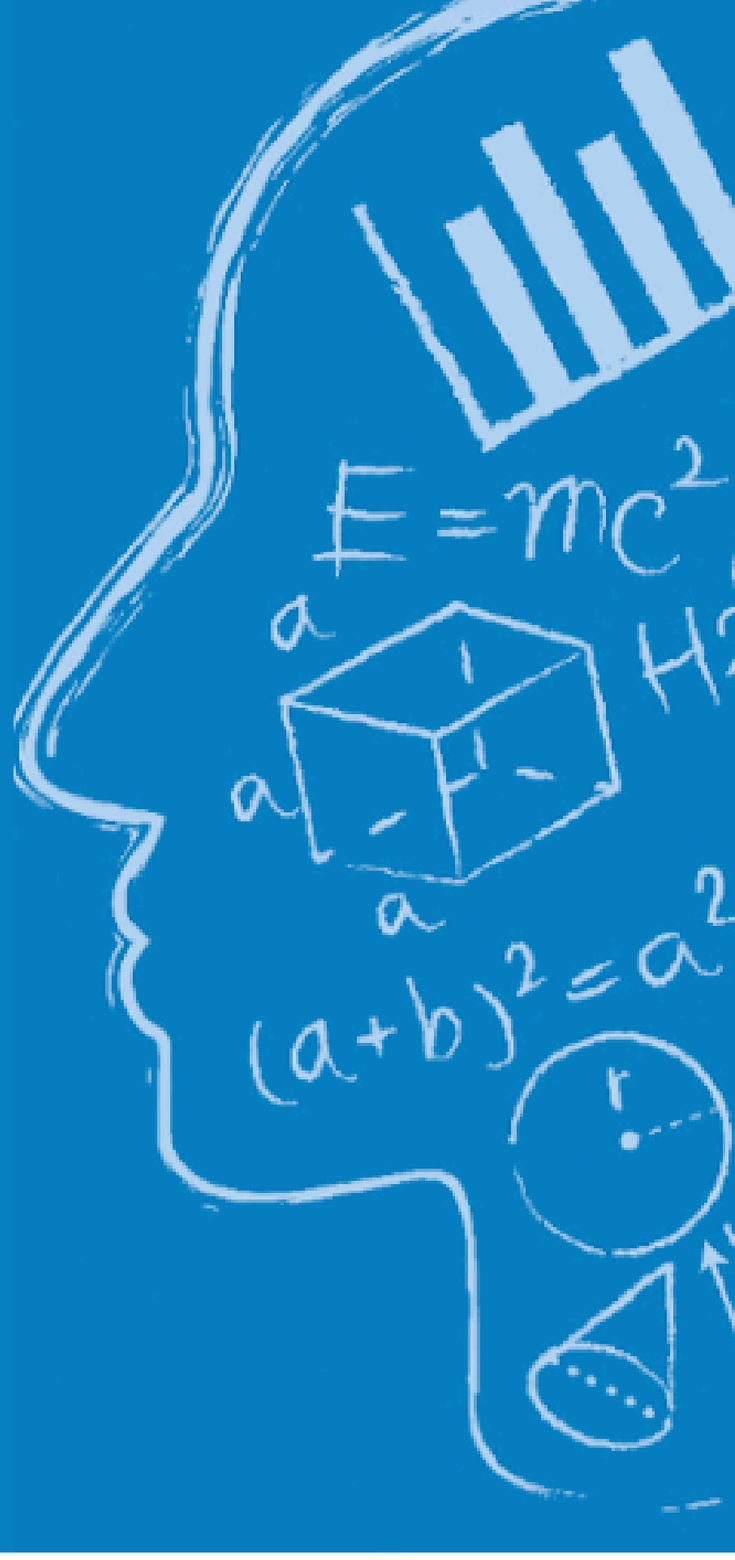

IMPACT OF MISDIAGNOSIS IN CASE-CONTROL ASSOCIATION STUDIES: THE CASE OF MYALGIC ENCEPHALOMYELITIS/CHRONIC FATIGUE SYNDROME

ABSTRACT:

Misdiagnosis can occur when different case definitions are used by clinicians (relative misdiagnosis) or when failing the genuine diagnosis of another disease (misdiagnosis in a strict sense). In complex diseases, such as myalgic encephalomyelitis/chronic fatigue syndrome (ME/CFS), this problem translates to a recurrent difficulty in reproducing research findings. To explore these effects, we simulated data from case-control studies under the assumption of misdiagnosis in a strict sense. We estimated the power to detect a genuine association between a potential causal factor and ME/CFS and demonstrated how current research studies may have suboptimal power. To address the implications of these findings, suggestions for how power can be improved are given and explained within the context of the disease.

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João is a Biomedicine and Computational Biology researcher currently working between the Institute of Molecular Medicine, Faculty of Medicine, University of Lisbon, and the Faculty of Mathematics and Information Science, Warsaw University. He is also a member of the Centre of Statistics and its Applications from the University of Lisbon. His previous studies were a BSc in Biology and a MSc in Biostatistics. He is interested in bioinformatics applications, autoimmunity, and diseases with complex diagnosis such as myalgic encephalomyelitis/chronic fatigue syndrome (ME/CFS), a multifactorial illness with no biomarker and a heterogeneous affected population. For his recent research, he has been working with DNA methylation and gene expression data from ME/CFS patients to infer on population-related risks and using serology and immunological cellular compartments' data to study alternative cohort stratification and immunological profiling methods, dealing with the effects of misclassification trying to identify signatures for specific groups of patients.