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Optimizing multiple sclerosis diagnosis using gene expression and genomic association data

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The diagnosis of multiple sclerosis (MS) is based on the neurological symptomatology in combination with the presence of central nervous system lesions disseminated in time and space. However, the clinical, imaging and/or laboratory findings of patients with MS may mimic a wide array of other vascular, inflammatory and demyelinating diseases, hereby defined as non-MS. This overlap may pose a significant diagnostic challenge especially in the process of diagnosis at the early disease stage. We utilized findings of large-scale Genome Wide Association Studies (GWAS) to develop a blood gene expression based classification tool to assist in the diagnosis during the first demyelinating event suggestive of MS. We merged knowledge of 110 MS susceptibility genes gained from MS GWAS studies together with our experimental results of differential blood gene expression profiling between 80 MS patients and 31 non-MS patients. Multiple classification algorithms were applied to this cohort to construct a diagnostic classifier that correctly distinguished between MS and non-MS patients. The overall accuracy of the constructed 42-gene classifier was tested on an independent patient population consisting of diagnostically challenging cases including non-MS patients with positive MRI findings and achieved a correct classification rate of $76.0 \pm 3.5\%$. The presented diagnostic classification tool complements the existing diagnostic McDonald criteria by assisting in the accurate exclusion of other neurological diseases at presentation of the first demyelinating event suggestive of MS.

Biography

Gurevich Michael is the Head of the Neuro-genomics Laboratory at Sheba Multiple Sclerosis Center, Israel. His research primarily focuses on The study of translational medicine and functional genomics in neurological diseases and multiple sclerosis. He has gained extensive knowledge and experience working with DNA microarray technology, discovering multiple sclerosis related molecular disease pathogenesis and finding biomarkers that may assist in disease diagnosis, monitoring and prognosis of clinical outcome.

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