Towards endophenotyping multiple sclerosis

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Multiple sclerosis (MS) is a disease of the central nervous system, with inflammatory and neurodegenerative pathological features. The clinical course is variable, varying from mild neurological deficit to severe disability. It is unfortunately currently not possible to give an individual patient a reliable prognosis of the extent of neurological deficit later in the disease course. The pathophysiological mechanisms that cause this variability in clinical expression of MS are probably heterogeneous. Genetic factors have long been established as contributing risk factors for developing MS, and it is also plausible that genetic variation between individual patients is related to a variable disease course. In this thesis we aimed to provide further insight into the clinical variety of MS using a stratification approach based on imaging and pathological phenotypes. Furthermore, in order to identify possible genetic mechanisms that might influence disease expression, we extended our research into genetic association studies using imaging phenotypes. With this translational approach this thesis suggests that endophenotyping provides a new direction for understanding the heterogeneity of MS.

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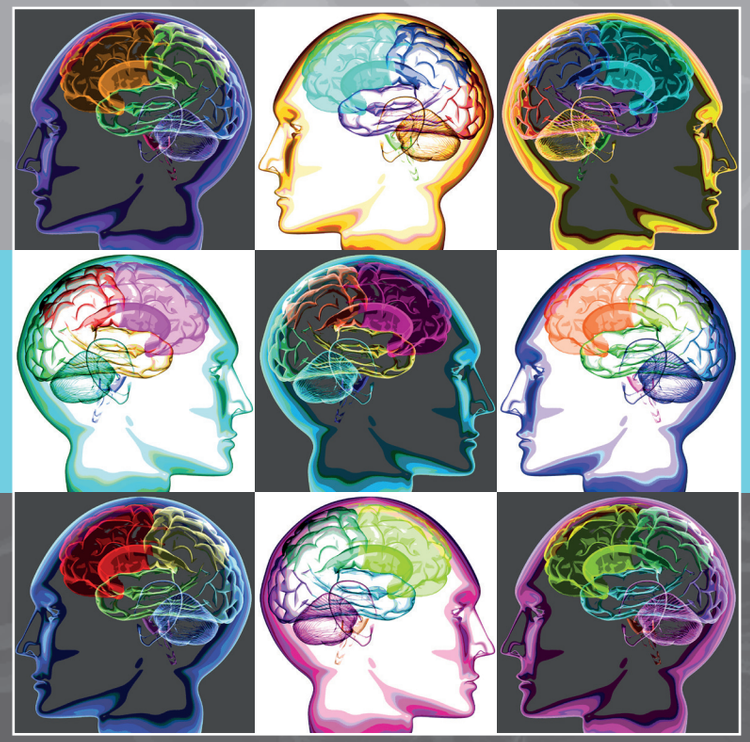

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