Editorial

Copy Number Variants in Neurological Disorder

The past twenty-five years significantly expanded our knowledge of Copy Number Variants (CNVs), genomic imbalances belonging to structural genetic variations, and their role in both human health and disease, particularly in the neurological field. In 1991, Lupski was the first to associate a CNV (a DNA duplication) to a human autosomal dominant neurodegenerative disease, the Charcot-Marie-Tooth Disease Type 1A [1]. In the last years, different technologies succeeded one another, increasing the power of resolution and the regions of application. However, technological and conceptual barriers have hampered the investigation of neurodegenerative diseases from a polygenic point of view, not only for complex neurodegenerative diseases but also for monogenic ones. For example, even if Neurofibromatosis type 1 represents a monogenic autosomal dominant disorder with complete penetrance, it is characterized by a variable expressivity that is hard to address in a genotype-phenotype correlation [2, 3].

In this mini-thematic issue, we aim to describe the progress in the study of these important types of structural variations in medicine, exploring the use of CNVs analysis in neurological disorders, as recently reviewed for Alzheimer's Disease, Parkinson's Disease and Amyotrophic Lateral Sclerosis studies [4-6]. Taken together, the three manuscripts published in the present mini-thematic issue provide the reader an overview of the recent findings regarding inherited neuropathies (Salpietro *et al.*) and adult-onset neuropsychiatric disorders, *i.e.* Schizophrenia and Alzheimer's disease (Lew *et al.*), and emphasize the need of custom technologies, such as a customized exon-centric aCGH, to detect overlapping gene signatures among neurological conditions (La Cognata *et al*.).

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