

Brain Changes its Relation to Behavior and Cognitive Features in Neuromuscular Disorders (NMD)

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Editorial

New technologies are advancing such as molecular genetics and brain imaging and their correlation is an active field of investigation both in Neurology and Psychiatry. The correlation between behavior, autism, and cognitive features has been covered in the last three meetings by workshops or Overarching Sessions in ICNMD meetings in the last Editions [1], however, this is an open subject that deserves large attention and needs further investigation, documentation, and research.

Positron Emission Tomography (PET) and Single Photon Emission Computed Tomography (SPECT) have been utilized to find anomalies in the functional connectivity of brain networks and metabolic changes in a number of illnesses, including myotonic dystrophy types 1 and 2 and movement disorders [2]. NMDs are typically linked to pathological brain alterations with cognitive impairment. Particularly, Congenital Muscular Dystrophies (CMDs) have been observed to be associated with skeletal muscle weakness, severe hypotonia, abnormalities in the White Matter (WM), ventricle dilation, and anomalies in cerebral gyration. Adult patients with the dystroglycanopathy 2I subtype (LGMD2I/R9) have substantial ventriculomegaly, subcortical atrophy, and WM periventricular involvement.

A neuro-muscular condition known as myotonic dystrophy type 1 (DM1) is characterized by an unstable CTG triplet expansion in chromosome 19. With beginning in childhood or adulthood, it is the most prevalent form of muscular dystrophy. DM1 is associated with extreme clinical variability, affecting various organs and the CNS. Cerebral involvement in DM1 consists of extensive mental

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impairment, executive dysfunction, and the presence of an avoidant personality. A potential mechanism for cognitive failure is the disconnection of cortical regions caused by alterations in the connectivity of white matter (WM), which appears to be more closely associated with cognitive impairment than grey matter atrophy. Imaging techniques allow for the investigation of white matter changes alterations and brain MR might be used to measure WM alterations [3].

In order to identify abnormalities in the functional connectivity of brain networks and metabolic alterations, Positron Emission Tomography (PET) and Single Photon Emission Computed Tomography (SPECT) were utilized.

A number of NMDs are usually linked to pathological brain alterations with cognitive impairment. In particular, they can be present in Congenital Muscular Dystrophies (CMDs), that present from birth skeletal muscular weakness, severe hypotonia, WM abnormalities, ventricular dilatation, and anomalies in cerebral gyration that are most commonly observed in Fukuyama's disease as well as in Muscle-Eye-Brain disease and Walker-Walburg syndrome. Adult patients with dystroglycanopathy 2I subtype (LGMD2I/R9) have been observed to show mild ventriculomegaly, subcortical atrophy, WM periventricular involvement, and expansion of subarachnoid spaces. Brain imaging characteristics and abnormalities have been observed to be linked to clinical symptoms, with congenital or childhood-onset instances showing more pronounced alterations.

The symptoms of myotonic dystrophy type 2 (DM2) appear to be lightly harsh than those of type 1 (DM1). Cortical atrophy is linked with limited ventricle dilation and WM anomalies in Duchenne and Becker Muscular Dystrophies (DMD, BMD), but mental retardation and autism may also be present.

Delayed Brain myelination characterized the late-onset glycogenosis type II (GSD II) or Pompe infantile variants. An additional characteristic form has been observed in children treated with enzyme replacement therapy that does not cross the blood-brain barrier. Only a small number of cases of oculopharyngeal muscular dystrophy have been reported to impact the central nervous system, which is linked to executive function deficits.

The following NMDs: Dystrophinopathies, dystroglycanopathies, myotonic dystrophies, facioscapulohumeral dystrophy, limb-girdle muscular dystrophies, congenital myotonias, congenital myopathies, and associated words or acronyms are all associated with brain abnormalities.

This editorial focuses on a few specific muscle illnesses while excluding others, such as motor neuron disease, which covers a certain time span (since 2000), when molecular diagnostics were available and brain imaging was popular.

Researchers are aware that the perfect editorial would incorporate every piece of research on brain imaging in NMDs. Researchers present editorially imposed research limits despite the abundance of literature on mitochondrial and neuro-metabolic illnesses that may characterize lesions with

characteristics of stroke, calcification, or pituitary adenoma. To researchers understanding, more than 600 different NMDs have been found as a result of the recent expansion in the research of neuromuscular illnesses.

Due to this, it would not be possible to cover the entire spectrum in a single editorial. Researchers are aware that brain involvement in mitochondrial illnesses is pertinent due to the MELAS syndrome reports.

Researchers intend to offer this new field of investigations for achieving progress through original papers, minireview, and case reports that would deal with this subject in this journal both trying to clarify pathophysiology of pathogenetic mechanism (s) of these multisystemic disorders and encouraging better care of these patients, that can be achieved by a team of dedicated physicians, neuropsychologist and psychiatrists.

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