



Rare Neurologic Diseases and Neurological Sciences: a report for the celebration of the 2020 Rare Diseases Day

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Introduction

In the recent years, a great interest has been given to rare diseases, that are over 6000 different pathological conditions, affecting more than 300 million people around the world, more than 70% with genetic pathogenesis, and more than 50% with neurologic or muscle involvement. For years, research projects have been developed, discovering many genes related to the pathogenesis of many disorders and also new therapies.

In the same time, in order to promote the better knowledge of these conditions, many actions have been developed, able to improve the information between doctors, families, and also institutions.

One of these is the Rare Diseases Day, that is celebrating every year, on the last day of February (this year on the February 29th).

Rare Disease Day is a great example of how progress continues to be made in this area. Beginning in 2008, which events only in 18 countries, Rare Disease Day has taken place every year with events being held in over 100 countries in 2019.

Neurological Sciences, as done on the past year, participates to the Rare Diseases Day activities promoting the knowledge and the attention on rare neurologic disorders, reporting the main articles published on the journal in 2019 related to Rare Neurologic Diseases.

We hope that this action may help our readers to be more familiar with rare neurologic diseases, facilitating their early diagnosis and promoting new researches on therapies.

In 2019, our journal published 61 articles related to rare neurologic diseases out of a total of more than 450 articles, the majority (34) referring the clinical aspects of many different diseases, 10 amyotrophic lateral sclerosis, 12 new genetic

mutations, 8 pathogenetic aspects, 5 recommendations, guidelines, and reviews, and 2 therapies.

Clinical aspects of Rare Neurologic Diseases

The heterogeneity of brain FDG-PET metabolic patterns in patients with C9orf72 mutation was described by Castelnovo et al. [1]. A neurobehavioral phenotype of familial Creutzfeldt–Jakob disease lead to Giovagnoli et al. [2] to discuss an impairment of theory of mind; a fronto-temporal type of dementia was reported associated to Langerhans cell histiocytosis by Garg et al. [3]. Body composition analysis in patients with myotonic dystrophy types 1 and 2 was described by Peric et al. [4]; a case report of cerebellar ataxia associated with anti-glutamic acid decarboxylase antibodies was reported by Villa et al. [5]. Late-onset and fast progressive neuropathy and cardiomyopathy was associated to Val32Ala transthyretin gene mutation (Cazzato et al.) [6]. A rare cause of Horner's syndrome, cervicothoracic spinal root cysts, was described by Karti et al. [7]. Clinical, radiological features, and surgical strategies for 23 NF1 patients with intraorbital meningoencephalocele were reported by Niu et al. [8], and Santoro et al. [9] in a child with neurofibromatosis type 2 and unilateral moyamoya discussed on further evidence of cerebral vasculopathy in NF2 and SCN11A variant as possible pain generator in sensory axonal neuropathy (Ginanneschi et al.) [10]. Desensitization of two young patients with infantile-onset Pompe disease and severe reactions to alglucosidase alfa was described by Gragnaniello et al. [11]; an overview on degenerative and acquired sporadic adult onset ataxia was reported by Lieto et al. [12] and by Teive et al. [13]. Lieto et al. [14] also reported a longitudinal study of a cohort of MSA-C patients in South Italy discussing their survival and clinical features. A rare case of pure sensitive Parsonage–Turner syndrome was described by Pessa et al. [15]. Nesti et al. [16] reported a complex multisystem phenotype associated with the mitochondrial DNA m.5522G > A

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mutation. Escorcio-Bezerra et al. [17] focused the attention on tonic pupils as an unusual autonomic involvement in chronic inflammatory demyelinating polyneuropathy (CIDP), while Kobayashi et al. [18] suggested that ischemic stroke was associated to sarcoidosis for the presence of arterial wall enhancement. Qin [19] showed association of HIF1A and Parkinson's disease in a Han Chinese population by molecular inversion probe analysis, and Yin et al. [20] by a meta-analysis reported the relationship with BDNF Val66Met polymorphism and cognitive impairment in Parkinson's disease. Basal ganglia calcifications (Fahr's syndrome) and related condition were discussed by Donzuso et al. [21]. Malaquias et al. [22] described GNAO1 mutation presenting as dyskinetic cerebral palsy. Liu et al. [23] reported a case of recurrent vomiting extending the spectrum of neuronal intranuclear inclusion disease; carrier frequency of spinal muscular atrophy in Thailand was described by Dejsuphong et al. [24], and Savcun Demirci [25] described the effect of taping on scapular kinematics of patients with facioscapulohumeral muscular dystrophy. An atypical clinical manifestations of Miller Fisher syndrome was reported by Jung [26].

Amyotrophic lateral sclerosis

Amyotrophic lateral sclerosis (ALS) is the most frequent rare neurologic diseases. This is reflected also in the article published in 2019 in our journal and reported in Table 1.

Table 1 Articles on ALS published in Neurological Journal in 2019

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| Dardiotis et al. [27], ERCC6L2 rs591486 polymorphism and risk for amyotrophic lateral sclerosis in Greek population. |
| Luo et al. [28], Efficacy and safety of edaravone in treatment of amyotrophic lateral sclerosis—a systematic review and meta-analysis |
| Sivaramakrishnan et al. [29], Recumbent stepping aerobic exercise in amyotrophic lateral sclerosis: a pilot study |
| Amin Lari et al. [30], A review of electrophysiological studies of lower motor neuron involvement in amyotrophic lateral sclerosis |
| Hancevic et al. [31], Attenuation of ALS progression during pregnancy—lessons to be learned or just a coincidence? |
| Verde et al. [32], PON1 is a disease modifier gene in amyotrophic lateral sclerosis: association of the Q192R polymorphism with bulbar onset and reduced survival |
| Sprovieri et al. [33], A novel S379A TARDBP mutation associated to late-onset sporadic ALS |
| Yang et al. [34], UNC13A variant rs12608932 is associated with increased risk of amyotrophic lateral sclerosis and reduced patient survival: a meta-analysis |
| Park et al. [35], Under-recognized primary spontaneous pneumothorax in ALS: a multicenter retrospective study |
| Corrado et al. [36], Analysis of the GCG repeat length in NIPA1 gene in C9orf72-mediated ALS in a large Italian ALS cohort |
| Santus et al. [37], Orthostatic tremor and behavioral frontotemporal dementia: a case report with 7 years of follow-up. |

Table 2 Articles with new genetic mutations of Rare Neurologic Diseases published in 2019 in Neurological Sciences

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| Koh et al. [38], Decreasing 123I-ioflupane SPECT accumulation and 123 I-MIBG myocardial scintigraphy uptake in a patient with a novel homozygous mutation in the <i>ZFYVE26</i> gene |
| Abdulkareem et al. [39], A novel homozygous nonsense mutation in <i>CCDC88A</i> gene cause PEHO-like syndrome in consanguineous Saudi family |
| Zhang et al. [40], A novel missense mutation in the <i>ABCD1</i> gene of a Chinese boy diagnosed with X-linked adrenoleukodystrophy: case report |
| Genc et al. [41], Methylation of cation–chloride cotransporters <i>NKCC1</i> and <i>KCC2</i> in patients with juvenile myoclonic epilepsy |
| Kaur et al. [42], L-type calcium channel blockers and a symptom complex mimicking de Melo-Souza's syndrome |
| Oliva [43], A novel <i>SLC20A2</i> gene mutation causing primary familial brain calcification in an Ukrainian patient |
| Du [44], A novel mutation in the <i>CSF1R</i> gene causes hereditary diffuse leukoencephalopathy with axonal spheroids |
| Liu et al. [45], G327E mutation in <i>SCN9A</i> gene causes idiopathic focal epilepsy with Rolandic spikes: a case report of twin sisters |
| Catania et al. [46], DNMT1-complex disorder caused by a novel mutation associated with an overlapping phenotype of autosomal-dominant cerebellar ataxia, deafness, and narcolepsy (ADCA-DN) and hereditary sensory neuropathy with dementia and hearing loss (HSN1E) |
| Ferrante et al. [47], Identification of a novel <i>NOTCH3</i> mutation in an Italian family affected by a mild form of CADASIL |

New genetic mutations

Since the majority of Rare Neurologic Diseases has a genetic cause, the research in the discovering of new mutation on the basis of the different phenotypic presentations has a big interest. Article describing new genetic mutations published in Neurologic Sciences are reported in Table 2.

Pathogenetic mechanisms

Rare Neurologic Diseases are also useful for studying the different pathogenetic mechanisms underlying the symptoms. These investigations have also been published on Neurologic Sciences in 2019 and reported in Table 3.

Table 3 Articles on pathogenetic mechanisms of Rare Neurologic Diseases published in Neurologic Sciences in 2019

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| Wu [48], Inflammatory cytokines expression in Wilson's disease |
| Smolinski et al. [49], Brain volume is related to neurological impairment and to copper overload in Wilson's disease |
| Rucco et al. [50], Mutations in the <i>SPAST</i> gene causing hereditary spastic paraplegia are related to global topological alterations in brain functional networks |
| Ginanneschi et al. [10], <i>SCN11A</i> variant as possible pain generator in sensory axonal neuropathy |
| Wang et al. [51], Abnormal nuclear aggregation and myotube degeneration in myotonic dystrophy type 1 |
| Alpaydin-Baslo et al. [52], Is Hirayama a Gq1b disease? |
| Bohi et al. [53], Knuckle pigmentation, peripheral neuropathy, madness and abnormal movement: is it B12 deficiency? |
| Pianese et al. [54], Maternal germline mosaicism in Fabry disease |

Table 4 Miscellanea articles on Rare Neurologic Diseases published in Neurologic Sciences in 2019**Therapies**

Sansone et al. [55], Intrathecal nusinersen treatment for SMA in a dedicated neuromuscular clinic: an example of multidisciplinary and integrated care

Levy [56], Ten years of iron chelation in a patient with superficial siderosis

Guidelines and reviews

Maggi et al. [57], Italian recommendations for diagnosis and management of congenital myasthenic syndromes

Basantsova et al. [58], Small-fiber neuropathy definition, diagnosis, and treatment

Picillo et al. [59], Validation of the Italian version of the PSP Quality of Life questionnaire

Vita et al. [60], Genetic neuromuscular disorders: living the era of a therapeutic revolution. Part 1: peripheral neuropathies

Vita et al. [61], Genetic neuromuscular disorders: living the era of a therapeutic revolution. Part 2: diseases of motor neuron and skeletal muscle

Other findings related to Rare Neurologic Diseases are reported in Table 4.

Conclusions

All our data confirm the strong interest of the neurologic community in Rare Neurologic Diseases, representing an important area of scientific interest and innovation. With this editorial, we want to contribute to focus the attention on the important role of Rare Neurologic Diseases in the Neuroscience research and improve the knowledge of this important but still neglected area of Neurology.

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Compliance with Ethical Standards

Conflict of interest None.

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