Since January 2020 Elsevier has created a COVID-19 resource centre with free information in English and Mandarin on the novel coronavirus COVID-19. The COVID-19 resource centre is hosted on Elsevier Connect, the company’s public news and information website.

Elsevier hereby grants permission to make all its COVID-19-related research that is available on the COVID-19 resource centre - including this research content - immediately available in PubMed Central and other publicly funded repositories, such as the WHO COVID database with rights for unrestricted research re-use and analyses in any form or by any means with acknowledgement of the original source. These permissions are granted for free by Elsevier for as long as the COVID-19 resource centre remains active.
upper limbs, after a few months spread to the legs and trunk muscles, sparing the head. Two years later he began experiencing episodes of severe weakness lasting several hours, more common in cold weather, after a hard day’s work or carbohydrate-rich meal. Neurologic examination showed hyperreflexia, paradoxical myotonia and fasciculations. CPK, thyroid hormones, electrolytes levels were normal. Autoimmune disorders were excluded. EMG revealed spontaneous activity at multiple sites but not myotonia. Brain and spine MRI lacked any pathological sign. Multigene NGS analysis demonstrated a missense variant in CACNA1S p.Arg557Cys (c.1669 C>T), not previously reported in the literature, of uncertain pathological significance. Segregation analysis found the same variant in the patient’s mother, who reported a similar clinical picture, pointed to its likely pathogenicity.

Conclusions

This is the first report of a new likely pathogenic variant of the CACNA1S gene, with a clinical phenotype combining paradoxical myotonia and periodic paralysis. Such phenotype was previously associated with variations in the SCNA4 gene, but never in the CACNA1S gene.

doi:10.1016/j.jns.2021.118395

118396
Thymus characteristics in seropositive and seronegative myasthenia gravis

Oyunaa Chimedregzen, Sarangerel Jambal, Munkhbayar Rentsenbat, Byambasuren Dagvajantsan, Mongolian National University of Medical Science, School of Medicine, Neurology, Ulaanbaatar, Mongolia; Reflex Neurological Clinic, Neurology, Ulaanbaatar, Mongolia

Background and aims

Myasthenia gravis (MG) is an autoimmune disorder of neuromuscular junctions. The study of Yi Li et al. has shown that 70–85% of all patients with MG were seropositive and 8–15% seronegative. The high frequency of thymus hyperplasia in seropositive patients. Between 10% to 20% of patients with MG have a thymoma. To our knowledge, any study of MG in Mongolia has not been published yet. To study association between thymus characteristics and serological findings in patients with MG.

Methods

We reviewed a total of 14 adult patients with MG who were diagnosed by serological findings in a five-years period from 2015 to 2020. If Acetylcholine receptor antibody level is greater than 0.40 n/L were assessed seropositive. Thymus characteristics were assessed for chest CT scans. Statistical analysis was performed using SPSS software.

Results

A total 14 patients had Serological tests and Computed tomography. 85.7% (n = 12) patients were seropositive, 14.3% (n = 2) patients were seronegative in a serological test. Thymoma in 3 cases (10% of total MG), all patients were seropositive MG. Thymus hyperplasia in 5 cases, of these 80% of patients were seropositive MG and 20% of patients were seronegative MG. No changes of thymus in 6 cases, of these 83.3% of patients were seropositive MG, 16.7% of patients were seronegative MG.

Conclusions

Similar to other countries, the high frequency of seropositive MG in Mongolia. Thymoma rarely occurred in 10% patients who have MG.

doi:10.1016/j.jns.2021.118396

118397
Acute myopathy at onset of SARS-CoV-2 infection

Anna Maria Simone, Enrico Fileccia, Manuela Costa, Giulia Monti, Massimiliano Devetak, Luca Vaghi, Silvia De Pasqua, Mario Santangelo, AUSL Modena, Ramazzini Hospital, Internal Medicine, Neurology Unit, Modena, Italy

Background and aims

Although various reports indicated the presence of myalgia in 44–70% and “skeletal muscle injury” (increased CK and myalgia) in 23% of hospitalized patients with SARS-CoV-2 infection, the characterization of neuromuscular involvement is still unsatisfactory, and electrophysiologic studies have rarely been performed.

Methods

We describe a case of acute myopathy at onset of SARS-CoV-2 infection.

Results

A 73-year-old woman, with post-infarct ischemic heart disease in her medical history, presented with progressive weakness in the lower limbs and pain, without fever. On admission, neurological examination showed proximal tetraparesis, prevalent in the lower limbs, and reflexes were diminished. CK in the serum was to ~6000 U/L (normal value 10–145 U/L), hepatic enzymes were elevated (GOT 814 U/L, n.v.1–31), urin Hb > 1 (n.v. absent). Polymerase chain reaction (PCR) testing for SARS-CoV-2 was positive. Chest X-ray showed right lower patchy opacities, but oxygen saturation was 94% on room air. Motor nerve conduction studies (NCS) showed mild sensory polyneuropathy; the needle electromyography (EMG) demonstrated myopathic abnormalities with fibrillation potentials in the lower limbs, rapid recruitment on interference pattern and reduced compound muscle action potential amplitude. The therapy with high dose of steroids induced significant clinical improvement. In two weeks, CK levels almost normalized and the patient recovered the ability to walk with assistance.

Conclusions

The severe immune activation known to occur in COVID-19 patients probably plays an important pathophysiological role for onset of rhabdomyolysis, but further studies are needed to elucidate the mechanisms, appropriate treatment, and long-term clinical outcomes of muscular manifestations associated with COVID-19 disease.

References

1. Huang C, Wang Y, Li X, Ren L, Zhao J, Hu Y, Zhang L, Fan G, Xu J, Gu X, Cheng Z, Yu T, Xia J,UNWaR, Liu X, Guo X, Hu J, Shi J, Xia J, Wei Y, Cao Y, Wang J, Gao B (2020). Clinical features of patients infected with 2019 novel coronavirus in Wuhan, China. Lancet 395(10223): 497–506
2. Francescanci M, MD, Barbara Mercier, MD, Guido Primiano, MD, PhD; Salvatore Lucio Cutillo, MD; Giovanni De Pasqua, MD, and Serena Solazzi, MD. Acute myopathic quadriplegia in patients with COVID-19 in the intensive care unit. Neurology 2020;95:492-494.
3. Hu Zhong, MD, PhD; Zainab Chugh, MD; Roberto J Goldstein, MD; Yasazon Aoki, MD; Woodruff W, Vassiloukas, DC; Jonathan R, MD, PhD. COVID-19 associated myositis with severe proximal and bulbar weakness. Muscle Nerve. 2020 Sep;62(3):E37-E40.
4. Lalita Gupta, James B. Liddell, Vikas Agarwal, Hester Choy and Rohit Aggarwal. COVID-19 and myositis – unique challenges for patients. Rheumatology 2020;00:1-4

doi:10.1016/j.jns.2021.118397