https://doi.org/10.1590/0004-282X20190174

Duchenne muscular dystrophy requires treatment also of cardiac, respiratory, cerebral, and orthopedic compromise

A distrofia muscular de Duchenne requer tratamento também do comprometimento cardíaco, respiratório, cerebral e ortopédico

Josef FINSTERER[1]

Dear Editors,

We read, with great interest, the review by Werneck et al. about treatment options so far applied in patients with Duchenne muscular dystrophy (DMD)[1]. The review focuses on drug treatment and concludes that the most effective therapeutic measures are steroids combined with physiotherapy, orthoses, exercise, orthopedic surgery and, eventually, pulmonary or cardiac support. We have the following comments and concerns to add.

A detailed description and discussion of cardiac and respiratory therapy is missing in this review. Since the outcome of DMD patients is mainly determined by the degree of cardiac involvement and involvement of the respiratory muscles, it is crucial to discuss the current status of cardiac and respiratory measures. DMD is frequently associated to progressive dilated cardiomyopathy[2] or ventricular arrhythmias, which require treatment as from the early stages. Therapy of dilated cardiomyopathy in DMD includes angiotensin-converting enzyme inhibitors, beta-blockers, AT-II-blockers, diuretics, levosimendan, atrial ablation, implantation of an implantable cardioverter defibrillator (ICD), mechanical circulatory support with ventricular assist devices, or heart transplantation in case of drug-resistant heart failure. If there is documented atrial fibrillation, severe heart failure, intraventricular thrombus formation, or left ventricular hypertrabeculation with heart failure or atrial fibrillation, also known as noncompaction, oral anticoagulation with vitamin-K antagonists is indicated[3,4]. Treatment of ventricular arrhythmias includes antiarrhythmic drugs, ablation, or ICD implantation.

For weakening respiratory muscles nocturnal or daytime ventilatory support is indicated. If the ventilatory drive is not sufficient to trigger a supportive respirator, mechanical ventilation via a tracheostoma should be considered if it complies with the patient's will and the intentions of the caregivers and the family. In order to prolong the ventilator-free time period of the disease, it is advisable to offer regular respiratory muscle training[5].

Considering DMD patients frequently develop orthopaedic abnormalities[6,7], it is crucial to provide optimal orthopedic support for the outcome to be improved. Orthopedic abnormalities reported in DMD patients include thoracic deformities, scoliosis, bent spine, pelvic obliquity, hip subluxation respectively dislocation, foot deformities, joint contractures, or osteoporosis. Orthopedic involvement may respond to conservative or surgical orthopedic interventions. Orthopedic intervention in form of spinal fixation can be highly beneficial, not only for the correct posture but also for cardiac disease.

Given that some of the DMD patients may develop cognitive impairment or even dementia[8], it can be helpful to provide these patients with anti-dementive drugs and to offer memory trainings.

Overall, a historical review about the treatment of DMD needs to include a detailed discussion about treatment options for cardiac, respiratory, orthopaedic, and central nervous system involvement. To improve the outcome of DMD patients, not only the skeletal muscles but also functions of the heart, respiratory muscles, brain, and skeleton need to be assessed, and eventual dysfunctions need to be adequately treated.

[1]Krankenanstalt Rudolfstiftung, Messerli Institute, Vienna, Austria.
Josef FINSTERER https://orcid.org/0000-0003-2839-7305
Correspondence: Josef Finsterer; E-mail: fifigs1@yahoo.de
Conflict of interest: There is no conflict of interest to declare.
Received on October 10, 2019; Accepted on October 21, 2019.

References

1. Werneck LC, Lorenzoni PJ, Ducci RD, Fustes OH, Kay CSK, Scola RH. Duchenne muscular dystrophy: an historical treatment review. Arq Neuropsiquiatr. 2019 Sep;77(8):579-89. https://doi.org/10.1590/0004-282X20190088
2. Lester G, Femia G, Ayer J, Puranik R. A case report: X-linked dystrophin gene mutation causing severe isolated dilated cardiomyopathy. Eur Heart J Case Rep. 2019 Jun3(2):ytz055. https://doi.org/10.1093/ehjcr/ytz055
3. Hasuike Y, Saito T, Saito T, Matsumura T, Fujimura H, Sakoda S. Cerebral embolism in Duchenne muscular dystrophy after respiratory tract infection - Report of two cases. Rinsho Shinkeigaku. 2018 Oct;58(10):642-5. https://doi.org/10.5692/clinicalneurol.cn-001135
4. Finsterer J, Cripe L. Treatment of dystrophin cardiomyopathies. Nat Rev Cardiol. 2014 Mar;11(3):168-79. https://doi.org/10.1038/nrcardio.2013.213
5. Silva IS, Pedrosa R, Azevedo IG, Forbes AM, Fregonezi GA, Dourado Junior ME, et al. Respiratory muscle training in children and adults with neuromuscular disease. Cochrane Database Syst Rev. 2019;9:CD011711. https://doi.org/10.1002/14651858.CD011711.pub2
6. Apkon SD, Alman B, Birnkrant DJ, Fitch R, Lark R, Mackenzie W, et al. Orthopedic and surgical management of the patient with duchenne muscular dystrophy. Pediatrics. 2018 Oct;142(Suppl 2):S82-S89. https://doi.org/10.1542/peds.2018-0333J
7. Choi YA, Shin HI, Shin HI. Scoliosis in Duchenne muscular dystrophy children is fully reducible in the initial stage, and becomes structural over time. BMC Musculoskelet Disord 2019;20:277. https://doi.org/10.1186/s12891-019-2661-6.
8. Mukherjee S, Roy M, Guha G, Saha SP. Mutation location and cognitive impairment in duchenne muscular dystrophy. J Neurosci Rural Pract. 2018 Jul-Sep;9(3):410-3. https://doi.org/10.4103/jnrp.jnrp_426_17