ORIGINAL ARTICLE

Cardiovascular Compromise in Children with Neuromuscular Disease with Respiratory Disturbance: An Audit for Better Follow-Up

João Rato[1], Mónica Rebelo[2], Rui Anjos[1], Teresa Moreno[3], Teresa Bandeira[4,5], Ana Saianda[4], Rosário Ferreira[4]

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Abstract

Introduction: Neuromuscular diseases can be followed by cardiomyopathy and/or arrhythmias that have prognostic consequences. Although respiratory failure is the most frequent cause of death in this group of patients, heart disease is independent of ventilatory compromise and should be investigated. We aimed to investigate the cardiovascular follow-up and diagnosis of a population of patients with neuromuscular disease with respiratory compromise.

Methods: Digital files of patients with neuromuscular diseases with respiratory compromise, in active follow-up at a pediatric pulmonology unit of a Portuguese tertiary hospital, were reviewed. Data on demographic, neuromuscular diseases and cardiovascular follow-up characteristics were analyzed.

Results: All 49 patients with neuromuscular diseases were analyzed. The median age was 14.3 (1.1-25.1) years and 28 (57%) were males. All of the patients had some type of respiratory compromise. Cardiac evaluation was performed in 35 (71%) patients, with at least six to 12 months of interval, and the following diagnoses were ascertained: dilated cardiomyopathy in three patients with Duchenne muscular dystrophy; right ventricular hypertrophy, mild ascending aortic dilatation and left ventricular diastolic dysfunction in three patients with congenital muscular dystrophy; frequent ventricular ectopic beats and left ventricular diastolic dysfunction in two patients with myotonic dystrophy. Only three patients with neuromuscular diseases with predictable cardiovascular involvement had not been evaluated at our center.

Discussion: Most of the patients with neuromuscular diseases with a predictable cardiovascular involvement had been evaluated in a cardiology consultation. The cardiovascular changes present in the group of patients evaluated agree with those described in the literature and the patients are followed-up on according to the recommendations. The diagnosis and monitoring of these cardiovascular changes are mandatory in order to control their progression and impact.

Keywords: Adolescent; Cardiovascular Diseases/etiology; Child; Muscular Dystrophies/congenital; Follow-Up Studies; Heart Diseases/etiology; Neuromuscular Diseases/complications; Portugal; Respiration Disorders/etiology

Introduction

Neuromuscular diseases are a heterogeneous group of diseases affecting skeletal muscle, with distinct phenotypes, but overlapping characteristics.[1,2] The survival of these patients, although often conditioned by respiratory failure, has increased due to the evolution of home respiratory care, especially ventilator support.[3,4] Consequentially, in the last few years, cardiovascular disease has assumed a greater importance in the prognosis of these diseases, highlighting the relevance of its research.[2] Cardiovascular involvement is independent of the involvement of other organs and may be the predominant characteristic, and it can be manifested by ventricular dysfunction and progressive heart failure, arrhythmias, and risk of sudden death, or both.[1] It is particularly characteristic of muscular and myotonic dystrophies and is well described in the guidelines for a multidisciplinary approach of these diseases.[2,5-7]

The aim of this study was to evaluate the referral to the cardiology clinic and describe any cardiovascular diseases in a population of pediatric patients with neuromuscular disease and some type of respiratory compromise. To our knowledge, this is the first study to evaluate the adequacy of the cardiology referral of

1. Pediatric Cardiology Department, Hospital de Santa Cruz, Centro Hospitalar Lisboa Ocidental, Carnaxide, Portugal
2. Pediatric Cardiology Department, Department of Pediatrics, Hospital de Santa Maria, Centro Hospitalar Universitário de Lisboa Norte, Lisboa, Portugal
3. Pediatric Neurology Unit, Department of Pediatrics, Hospital de Santa Maria, Centro Hospitalar Universitário de Lisboa Norte, Lisboa, Portugal
4. Pediatric Respiratory Unit, Department of Pediatrics, Hospital de Santa Maria, Centro Hospitalar Universitário de Lisboa Norte, Lisboa, Portugal
5. Faculdade de Medicina, Universidade de Lisboa Centro Académico de Medicina de Lisboa, Lisboa, Portugal
Corresponding Author
João Rato
https://orcid.org/0000-0002-1063-7174
joaorato4@gmail.com
Serviço de Cardiologia Pediátrica, Hospital de Santa Cruz, Avenida Prof. Reinaldo dos Santos, 2790-134 Carnaxide, Portugal
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pediatric neuromuscular patients, in accordance with the international follow-up recommendations.

Methods

In December 2015, we conducted a cross-sectional study by reviewing the electronic medical records of patients with neuromuscular diseases with active follow-up at a pediatric pulmonology unit of a tertiary hospital in Portugal. Data on demographic characteristics, such as age and sex, baseline neuromuscular disease diagnosis, respiratory disease (respiratory compromise was classified as ventilatory failure, nocturnal hypoventilation, bulbar dysfunction or ineffective cough, according to sleep studies, blood gases, and/or lung function tests), and cardiology evaluation, cardiovascular diagnosis, treatment, and monitoring, were collected and analyzed. All patients with neuromuscular diseases were included and characterized in the present study, even if a pediatric cardiology evaluation had not been performed. The need for cardiovascular evaluation and subsequent referral to the pediatric cardiology clinic as well as the adequacy of this referral according to the international recommendations was also evaluated. All patients referred to cardiology evaluation had at least one electrocardiogram and echocardiogram.

Standard statistical analysis was performed using IBM SPSS Statistics for Windows, Version 23.0.

Results

Forty-nine patients with neuromuscular diagnoses were included. The median age was 14.3 (1.1-25.1) years and 28 (57%) were male. Table 1 shows the neuromuscular diseases of the sample. We identified 26 patients with muscular dystrophy, 10 with spinal muscular atrophy, seven with congenital myopathies, four with myotonic dystrophy, one with congenital myasthenia, and one with congenital hypomyelination neuropathy.

Respiratory disease

All of the patients had some degree of respiratory compromise. Respiratory compromise included global respiratory failure in 12 patients (24%), nocturnal hypoventilation in 38 (78%), cough and bronchial cleaning disorder in 42 (86%), and bulbar dysfunction in 14 (29%). A sleep study was performed in all patients and 34 (69%) presented an obstructive sleep disorder. Spirometry was performed and presented conclusive results in 33 patients (16 patients either did not perform it or had inconclusive results): 15 (31%) patients had a very severe restrictive pattern, 10 (20%) patients had a severe restrictive pattern, five (10%) patients had a moderate restrictive pattern, and three (6%) patients presented no abnormalities. Thirty (61%) patients were on chronic home ventilation, of which 27 patients were in non-invasive home ventilation and three were in invasive ventilation by tracheostomy.

Table 1. Demographic characteristics of the patients evaluated by type of neuromuscular disease (n = 49)

			Respiratory disease			
Neuromuscular disease	**n (%)**	**Actual age (years) median (min-max)**	**Global respiratory insufficiency n (%)**	**Nocturnal hypoventilation n (%)**	**Cough disorder n (%)**	**Bulbar dysfunction n (%)**
Muscular dystrophies						
Duchenne muscular dystrophy	11 (23)	16 (13-22)	2 (4)	8 (16)	11 (23)	5 (10)
Congenital muscular dystrophy	8 (16)	11 (5-15)	2 (4)	6 (12)	7 (14)	1 (2)
Limb girdle muscular dystrophies	3 (6)	14 (14-17)	0	1 (2)	1 (2)	0
Laminopathies	3 (6)	5 (5-15)	2 (4)	2 (4)	3 (6)	0
Becker muscular dystrophy	1 (2)	14	0	1 (2)	1 (2)	0
Spinal muscular atrophy	10 (21)	10 (1-19)	2 (4)	10 (20)	10 (20)	2 (4)
Congenital myopathies						
Central core	3 (6)	2 (1-25)	2 (4)	3 (6)	2 (4)	2 (4)
Nemaline	2 (4)	18 (17-19)	2 (4)	2 (4)	2 (4)	2 (4)
Myotubular	1 (2)	10	0	0	1 (2)	0
Not Specified	1 (2)	13	0	1 (2)	1 (2)	1 (2)
Myotonic dystrophy	4 (8)	12 (6-21)	0	3 (6)	1 (2)	0
Congenital myasthenia	1 (2)	16	0	1 (2)	1 (2)	0
Congenital hypomyelination neuropathy	1 (2)	2	0	1 (2)	1 (2)	1 (2)

Max - maximum; min - minimum.

Cardiovascular evaluation

Thirty-five (71%) patients underwent a cardiovascular evaluation, which included electrocardiogram (ECG) and transthoracic echocardiogram (echo) (M mode, two-dimensional and Doppler) in all cases. Changes were found in eight of the 35 patients evaluated (23%), all with muscular or myotonic dystrophies (Table 2).

Both patients with dilated cardiomyopathy and those with left ventricular diastolic dysfunction were treated with different combinations of angiotensin converting enzyme inhibitors (ACE inhibitors), beta-blockers, and diuretics. The patient with ventricular extrasystoles was treated with a beta-blocker.

All patients were being followed-up in the pediatric cardiology clinic every six to 12 months, except for two cases, with dilated cardiomyopathy, in which more frequent evaluations were performed due to the clinical situation.

In our cohort of neuromuscular diseases patients, 11 did not undergo cardiovascular evaluation since, according to the literature, their type of neuromuscular disease presented either no association or only an extremely rare association with heart disease. This group of neuromuscular diseases includes congenital myopathies, spinal muscular atrophy, congenital myasthenia, and congenital hypomyelination neuropathy. In addition, three patients with neuromuscular diseases with a predictable cardiovascular involvement (all with muscular dystrophies) were not evaluated in cardiology consultations at our center; two were also followed in other hospitals, since they lived far away from our center, and we could not have access to the clinical registries on cardiovascular evaluation, and one had irregular compliance and poor attendance to the follow-up visits scheduled.

Table 2. Cardiac diagnosis of patients with cardiovascular disorders (n = 8)

Neuromuscular disease	Cardiovascular disorders (n)
Muscular dystrophies	
Duchenne muscular dystrophy	Dilated cardiomyopathy (3)
	Left ventricular diastolic dysfunction (1)
Congenital muscular dystrophy	Right ventricular hypertrophy (1)
	Mild dilation of the ascending aorta (1)
Myotonic dystrophy	Left ventricular diastolic dysfunction (1)
	Frequent ventricular extrasystoles (1)

Discussion

This study confirms that cardiac involvement in neuromuscular diseases is frequent, especially in muscular dystrophies and myotonic dystrophies. The incidence of Duchenne muscular dystrophy, the most frequent neuromuscular disease and one of the most severe, with early clinical manifestations, is 1:3,500 male newborns and the prevalence from 6:100,000 men.[2,8] Among other factors, household nocturnal ventilation, in particular, increased the average life expectancy from 19 to 25 years.[3] This increase in longevity was reflected in the greater role of cardiomyopathy as the cause of death.[6] In fact, congestive heart failure or sudden death is the cause of death in 10% to 20% of patients with Duchenne muscular dystrophy.[6] Heart disease in these patients is mainly characterized by dilated cardiomyopathy, but may also present as ventricular hypertrophy or arrhythmias,[1] as described in our study. Changes in the electrocardiogram and echocardiogram occur early in the disease course and can be identified at about 10 years of age.[1]

Cardiovascular follow-up and treatment in Duchenne muscular dystrophy are oriented by several international recommendations.[2,5-7] These are clearly affirming that a proactive strategy of early diagnosis and treatment is essential to maximize the duration and quality of life of these patients.[5] The proposed follow-up is summarized in Table 3. Regarding treatment, traditional first-line drugs are ACE inhibitors.[5] Some guidelines recommend pharmacological intervention with these first-line drugs from the age of 10, regardless of the degree of cardiovascular disease, with the addition of beta-blockers after the initiation of ACE inhibitors and the existence of ventricular dysfunction.[9]

Becker muscular dystrophy has a milder phenotype and a better prognosis.[1] It has an incidence of 1:18,450 and a prevalence of 2.4:100,000.[8,10] The average life expectancy is about 45 years.[1] Cardiovascular involvement is common and increases with age,[1] and heart disease is estimated as the cause of death in about 50%.[7] Becker muscular dystrophy has a high rate of cardiac transplantation, of 25% within five months after cardiomyopathy diagnosis.[11]

In our population, three out of 11 patients with Duchenne muscular dystrophy have dilated cardiomyopathy, the most common cardiac change in these patients, and are medicated with different combinations of drugs recommended in this pathology, namely ACE inhibitors and beta-blockers[5] as well as diuretics as needed. The age of the patients with cardiovascular disease ranges from 17 to 22 years and the age of the remainder patients is from 13 to 22. The patient with Becker muscular dystrophy, aged 14, had no diagnosis of heart disease at the time. A patient with Duchenne muscular dystrophy was not evaluated due to the family nonadherence to the recommended medical follow-up. The periodicity of evaluation of these patients with Duchenne muscular dystrophy is from six months to

one year, which is in accordance with the international guidelines (Table 3).

Laminopathies are a group of diseases caused by mutation in the *lamin A/C* gene, with variable phenotype and prevalence still unknown.[1] In these diseases, arrhythmias represent the main cardiac manifestation, with a high risk of sudden death.[12] The average life expectancy is 45 years.[12] Our study included three patients with laminopathies, two of whom were evaluated by cardiology with no heart disease. They are followed-up in the pediatric cardiology clinic with intervals of one year, as recommended (Table 3). The other patient resides in another country and has an erratic follow-up in our center.

Limb girdle muscular dystrophies are a group of several disorders, inherited as autosomal dominant or recessive traits,[1] with symptoms of the autosomal dominant forms being relatively mild compared with the recessive ones.[13] The overall prevalence ranges from 1:23,000 to 1:150,000.[8,13] Cardiac involvement depends on genetic mutation, and may be common in some forms being characterized by both cardiomyopathy and arrhythmias.[1] It is recommended that all patients are evaluated in a specialized cardiology clinic at regular intervals (Table 3).[7] In the present study, three patients had limb girdle muscular dystrophies, all were assessed by cardiology and none had heart disease.

Congenital muscular dystrophy is also a heterogeneous group characterized by muscle weakness present in the neonatal period or up to six months after birth.[1] Almost all forms are autosomal recessive, and the prevalence, morbidity, and mortality depend on the type of congenital muscular dystrophy.[1] Heart disease can be present in several forms of the disease, and all patients must be evaluated by pediatric cardiology.[1] This study included eight patients with congenital muscular dystrophy. Only one was not evaluated in the cardiology clinic for having erratic follow-up in our center and living in an autonomous region. Three of the patients evaluated had cardiovascular anomalies: one patient with left ventricular diastolic dysfunction and was treated with a beta-blocker, another with right ventricular hypertrophy and the last one with mild dilatation of the ascending aorta. The first anomaly may be part of the spectrum of ventricular dysfunction characteristic of neuromuscular diseases and the first

Table 3. Recommended cardiovascular evaluation in patients with neuromuscular diseases

Neuromuscular diseases	Recommended cardiovascular evaluation
Dystrophinopathies	
Duchenne muscular dystrophy	≤ 10 years: ECG + echo biannual (or annual[5]) > 10 years: ECG + echo annual Annual Holter if cardiovascular dysfunction Treat cardiovascular dysfunction
Becker muscular dystrophy	> 10 years: ECG + echo biannual Annual ECG + echo + Holter if cardiovascular anomalies Treat cardiovascular dysfunction, consider heart transplant
Emery-Dreifuss muscular dystrophy	
X-linked recessive pattern	Annual ECG + Holter, echo each five years Pacemaker or implantable cardioverter defibrillator may be indicated
Autosomal dominant pattern	Annual ECG + Holter, echo each two years Annual ECG + echo + Holter if cardiovascular anomalies Pacemaker or implantable cardioverter defibrillator may be indicated
Limb girdle muscular dystrophies	ECG + echo biannual or annual Holter may be indicated Treat cardiovascular dysfunction Pacemaker or implantable cardioverter defibrillator may be indicated
Facioscapulohumeral muscular dystrophy	ECG + echo at diagnosis Clinical dependent reassessment
Congenital muscular dystrophy	ECG + echo at diagnosis Clinical and specific mutation dependent reassessment
Myotonic dystrophy	
Type 1	Annual ECG, biannual Holter, echo each five years Annual ECG + echo + Holter if cardiovascular anomalies Pacemaker or implantable cardioverter defibrillator may be indicated
Type 2	Annual ECG Annual ECG + echo + Holter if cardiovascular anomalies

ECG - electrocardiogram; echo - transthoracic echocardiogram.
Adapted from Hermans MC, et al. Hereditary muscular dystrophies and the heart. Neuromuscul Disord 2010;20:479-92,[1] and American Academy of Pediatrics Section on Cardiology and Cardiac Surgery. Cardiovascular health supervision for individuals affected by Duchenne or Becker muscular dystrophy. Pediatrics 2005;116:1569-73.[6]

manifestation of the development of cardiomyopathy. Both right ventricular hypertrophy and mild ascending aortic dilatation may only be an incidental finding in this context, but they must be followed-up by the specialty.

Myotonic dystrophy is an autosomal dominant disease with two types of presentation,[1] myotonic dystrophy type 1 being the most serious phenotype. Myotonic dystrophy type 1 is the most common muscular dystrophy in adults, with a prevalence ranging from 2.1-14.3:100,000.[14] The median life expectancy is about 60 years for patients with early manifestations in adulthood and 35 years for those with congenital disease.[15] Arrhythmias are the most frequent cardiac manifestation.[16] Mild to moderate ventricular dysfunction is rarer.[17] The risk of sudden death is high.[1] Myotonic dystrophy type 2 typically occurs in adulthood, with a milder phenotype.[1] Arrhythmias are present in about 20% of affected patients.[18]

In our population, four patients, aged 2-21 years, have myotonic dystrophy. All of them were evaluated by pediatric cardiology. One had frequent ventricular extrasystoles and was medicated with a beta-blocker and another was diagnosed with left ventricular diastolic dysfunction and has been given an ACE-inhibitors. As already mentioned, this ventricular dysfunction may be the first manifestation of the development of cardiomyopathy that, although rarer in the context of this pathology, may occur.[17]

The remaining neuromuscular diseases included in this study, congenital myopathies and spinal muscular atrophy, are only rarely associated with heart disease.[19,20] Finally, both congenital myasthenia and congenital hypomyelination neuropathy are extremely rare diseases with no known association with cardiovascular changes. Some of these patients had cardiac evaluation because they presented isolated signs or symptoms that needed clarification or pre-surgical evaluations, and no cardiac changes were found. It is recommended that each patient should be assessed individually and referred for cardiology only when there is suspicion of heart disease.

This study is limited by its retrospective design, which conditioned access to some potentially relevant information. However, the data now presented confirm the previous findings about the relevance of elective referral to a specialized cardiology clinic of patients with neuromuscular diseases and summarizes the relevance of intervention by type of disease.

This is the first study performed to evaluate the proper cardiology referral of pediatric patients with neuromuscular diseases. Future areas of research should evaluate the long-term outcomes of such referral, and if it is maintained throughout time both in this population and in new patients.

Cardiac involvement is frequent in some neuromuscular diseases, and patients should be systematically referred, evaluated, and monitored in pediatric cardiology clinics. The diagnosis of cardiovascular anomalies is fundamental in order to control its progression. Referral of patients with neuromuscular diseases, in which cardiac involvement is rare, should only be performed if cardiac disease is suspected. Finally, the complexity and rarity of neuromuscular diseases imply that experienced multidisciplinary teams should be involved in the follow-up of these patients in order to avoid missing the diagnosis of other organs and systems involvement.

WHAT THIS STUDY ADDS

- Highlights the importance of the systematical referral of patients with neuromuscular diseases, in which cardiac involvement is frequent, to pediatric cardiology evaluation.
- Diagnosis and monitoring of the cardiovascular anomalies of these patients is fundamental to optimize the follow-up of disease progression.
- Provides a model for an audit of proper cardiology referral of pediatric patients with neuromuscular diseases and a protocol for their recommended cardiovascular evaluation.

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

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Protection of human and animal subjects

The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki).

Provenance and peer review

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Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

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Comprometimento Cardiovascular em Crianças com Doença Neuromuscular com Alteração Respiratória: Uma Auditoria para Melhor Acompanhamento

Resumo:

Introdução: As doenças neuromusculares podem acompanhar-se de miocardiopatia e/ou arritmias com consequências prognósticas. Embora a insuficiência respiratória seja a causa mais frequente de morte neste grupo de doentes, a doença cardíaca é independente do comprometimento ventilatório e deve ser investigada. Neste estudo, procurámos analisar o seguimento e diagnóstico cardiovascular de uma população de doentes com doenças neuromusculares com comprometimento respiratório.

Métodos: Foram revistos os processos eletrónicos de doentes com doenças neuromusculares com comprometimento respiratório, em seguimento ativo numa unidade de pneumologia pediátrica de um hospital terciário. Foram analisados dados acerca das características demográficas, doenças neuromusculares e seguimento e diagnóstico cardiovascular.

Resultados: Todos os 49 doentes com doenças neuromusculares foram analisados. Mediana da idade 4,3 (1,1-25,1) anos, 28 (57%) do sexo masculino. Todos os doentes tinham algum tipo de comprometimento respiratório. A avaliação cardíaca foi realizada em 35 (71%) doentes, com pelo menos seis a 12 meses de intervalo, e os seguintes diagnósticos foram verificados: miocardiopatia dilatada em três doentes com distrofia muscular de Duchenne; hipertrofia ventricular direita, dilatação ligeira da aorta ascendente e disfunção diastólica do ventrículo esquerdo em três doentes com distrofia muscular congénita; extrassistolia ventricular frequente e disfunção diastólica do ventrículo esquerdo em dois doentes com distrofia miotónica. Apenas três doentes com doenças neuromusculares com previsível envolvimento cardiovascular não foram avaliados no nosso centro.

Discussão: A maior parte dos doentes com doenças neuromusculares com previsível envolvimento cardiovascular foi avaliada em consulta de cardiologia. As alterações cardiovasculares presentes no grupo de doentes avaliados estão de acordo com as descritas na literatura e os doentes são acompanhados de acordo com as recomendações. O diagnóstico e monitorização destas alterações cardiovasculares são obrigatórios a fim de controlar a sua progressão e impacto.

Palavras-Chave: Adolescente; Cardiopatias/etiologia; Criança; Distrofias Musculares; Distúrbios Respiratórios/etiologia; Doenças Neuromusculares/complicações; Portugal; Seguimentos