

# Neuromuscular disorders and transition from pediatric to adult care in a multidisciplinary perspective: a narrative review of the scientific evidence and current debate

Giuseppe Accogli, Camilla Ferrante, Isabella Fanizza, Maria Carmela Oliva, Ivana Gallo, Marta De Rinaldis, Antonio Trabacca

*Scientific Institute IRCCS “E. Medea”, Unit for Severe disabilities in developmental age and young adults (Developmental Neurology and Neurorehabilitation), Brindisi, Italy*

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## Correspondence

Antonio Trabacca

Head of Unit for severe disabilities in developmental age and young adults (Developmental Neurology and Neurorehabilitation), Scientific Institute IRCCS “E. Medea”, piazza “A. Di Summa”, 72100 Brindisi, Italy. Tel.: +39 083 1349321 (switchboard), +39 0831349643 (direct). Fax: +39 0831349612  
E-mail: antonio.trabacca@lanostrafamiglia.it

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**Objective.** Standards of care and new genetic and molecular therapies have contributed to increasing life expectancy of patients with neuromuscular diseases (NMDs). This review presents the clinical evidence for an adequate transition from pediatric to adult care in patients with NMDs considering both physical and psychosocial aspects and attempts at identifying a general pattern of transition in the literature that can be used for all patients with NMDs. **Method.** A search was performed on PubMed, Embase and Scopus using generic terms that could be referred to the transition construct specifically related to NMDs. A narrative approach was used to summarise the available literature. **Results.** Our review shows that few or no studies explored the transition process from pediatric to adult care in neuromuscular diseases and tried to identify a general pattern of transition applicable to all NMDs. **Conclusions.** A transition process taking into consideration physical, psychological, social needs of patient and caregiver could produce positive outcomes. However, there is still no unanimous agreement in the literature on what it consists of and how to achieve an optimal and effective transition.

**Key words:** transition, neuromuscular disorders, psychological care, social care, healthcare

## Glossary

**Transition:** complex and gradual process, planned movement of young patients (starting at around the ages of 12 and 14) with chronic physical and medical conditions from child-centered to adult-oriented healthcare systems. Physical, psychological and social aspects of young patients and their caregivers are considered. It could have positive or negative outcomes on prognosis. It is an educational process.

**Transfer:** changing of adolescents or young adults with chronic physical or medical conditions from pediatric to adult care (owing to an event or series of events).

**Transitional program:** a series of multidisciplinary measures to prepare adolescents to take charge of their own situation.

## Introduction

Neuromuscular disorders (NMDs) are a genetically and phenotypically heterogeneous group of diseases affecting the neuromuscular system, namely the anterior horn cell, the peripheral nerve, the neuromuscular junction, or the muscle itself. NMDs as a whole are not infrequent, but every single one of them is a rare or orphan disease (prevalence < 1 per 1,500 persons in the U.S. and < 1 per 2,000 persons in Europe). Their causes can be genetic (single gene disorder, polygenic disorder) or nongenetic (infectious, autoimmune, auto-inflammatory)<sup>1,2</sup>. Clinically, the vast majority of NMDs are progressive, impairing motor function and often reducing life expectancy as well as quality of life. Age of disease onset varies. Some diseases are genetically inherited while others manifest in childhood or have an adult onset. For many decades, treatment of NMDs has been exclusively symptomatic but in recent years new genetic and molecular therapies and other focused on pathogenic effects are available or under development, provide hope for mitigating secondary pathophysiological consequences or modifying the underlying genetic defect<sup>3-5</sup>. NMDs need to be treated as systematic diseases due to increased life expectancy resulting from comprehensive standards of care involving multidisciplinary and coordinated care and new therapeutic methods. For this reason, it is possible to witness an expansion of the phenotypic manifestations of the disease with new or previously subtle organ manifestations<sup>6</sup>.

NMDs should be approached from a multiple perspective. For example, spinal muscular atrophies (SMAs) are a group of NMDs characterized by alpha motor neuron degeneration in the spinal cord, resulting in muscle atrophy, weakness and paralysis<sup>7,8</sup>. Given their complexity, a multidimensional approach to the management of SMA is required, and no aspects should be treated independently, such as social and psychological care<sup>8</sup>. Along the same line, Duchenne muscular dystrophy (DMD) – caused by mutations in the dystrophin gene resulting in complete absence or low levels of dystrophin protein<sup>9</sup> – should include a focus on mental and social health. In this multidimensional approach, issues related to transition from pediatric to adult care cannot be overlooked, just because the chances of survival have increased<sup>10</sup>. As reported by Goselink and colleagues<sup>11</sup> transition is the process started by physicians to prepare the child or young adult and their family for transfer from pediatric to adult care. Nonetheless, even though progress in health

care has increased life expectancy, support at home and in the community has not increased<sup>12</sup>.

Transition should be carefully planned. Inadequately planned and gross transition processes could lead to negative outcomes related to mortality, prognosis, psychosocial and educational well-being<sup>13,14</sup>. All transition processes should be tailored specifically<sup>15</sup>.

This paper presents a review on transition from child to adult care to illustrate the importance of an adequate transition process covering all aspects of NMDs. It highlights the clinical evidence and discusses the psychosocial implications underlying positive outcomes in the transition process from pediatric to adult care for patients with neuromuscular diseases. An attempt is made to identify a possible “standardized” protocol that could be used in all patients with neuromuscular conditions.

### *Towards a definition*

Transition is “the purposeful, planned movement of adolescents and young adults with chronic physical and medical conditions from child-centered to adult-oriented health-care systems”<sup>16</sup>. Several definitions of transition can be found in the literature. For example, Moons et colleagues<sup>17</sup> distinguish between transition, transfer or transitional care. Transition is the passage from one phase of a person’s life, physical condition or social role to another. It is also characterized by different stages, milestones, and turning points, and can be defined through process and/or terminal outcomes<sup>18</sup>. It could be considered an educational process that should begin before children reach adolescence<sup>19</sup>. Transfer is the passage from pediatric to adult care of an adolescent or young adult with chronic physical or medical conditions. Transitional care is the set of medical, social, psychological, educational and professional measures to prepare adolescents to take responsibility for their situation.

It is a crucial process that needs to be accurately planned to support families in the process and health care providers to define an appropriate transition plan<sup>20,21</sup>.

The two terms – transition and transfer – cannot be used interchangeably. As reported by Tilton<sup>22</sup>, transition is an extended and complex process that begins in early adolescence and offers a staged and gradual assessment of readiness as well as resources and education. Sawicki et colleagues<sup>23</sup> developed a useful tool for assessing readiness of youth with special healthcare needs.

To avoid worsening health conditions and lack of interest in services, it is necessary to bridge the gap between services for adults and children by identifying the key aspects of transition programs in a multidimensional perspective<sup>24</sup>. The transition process should begin quite early, at around the age of 12 or 14, and should aim to progressively increase a person’s autonomy<sup>25-27</sup>. It does

not end when the adolescent moves to adult-oriented care. In fact, it should be considered as an opportunity to rethink diagnosis and management. In addition, adolescents should undergo psychosocial screening before and after moving to adult-oriented care<sup>28</sup>.

Aho and colleagues<sup>29</sup> highlight how transition from childhood to adult age is an important phase because patients start to perceive the need to understand their life and make their life understood by the others. In addition, patients need to know what coping resources are available and give meaning to life. Therefore, support provided by health care professionals for future management of their disease becomes important.

Transition as a process does not involve solely young patients but also their parents. For example, individual or group psychological interventions could reduce their anxiety and worry about their children<sup>30</sup>. Family involvement is important for a successful transition of youth with disabilities<sup>31,32</sup>.

## Methods

This review specifically focuses on transition for young patients with NMDs. The aim is to identify a “standardized” transition protocol that can be used with all young patients with NMDs. Given the small number of papers available on transition as a unique and non-pathology-specific construct, we opted for a narrative approach vs a systematic review. As reported in the literature<sup>33,34</sup>, there are no recognized guidelines for narrative reviews. A narrative review should however have a structured general framework (rationale, organization of collected information, definition of objectives and goals) and follow a methodology in literature search (database, keywords, inclusion and exclusion criteria, list of references used). It could include a systematic review methodology, but this is not a necessary element. It may or may not include comprehensive search and/or quality assessment, and analysis may be chronological, conceptual, thematic, etc. It could be a good way for identifying omission or gaps<sup>34</sup>.

A search was performed on PubMed, EMBASE, and Scopus using generic terms that could refer to the transition construct for NMDs. Keywords such as Duchenne muscular dystrophy (DMD) or spinal muscular atrophy (SMA), etc. were not included because we were looking for broadly adaptable references to different forms of neuromuscular diseases. Hence, we searched for terms such as “transition”, “neuromuscular diseases”, “neuromuscular disorders”, “transition process”. Non-English language studies and works were excluded. Only papers containing the selected keywords in the title and/or abstract were considered for further analysis. No chronological criteria were applied.

Thirteen articles were selected that specifically reflect the aforementioned criteria. Most of them try to deal with and deepen both the purely physical and the psychosocial (and sometimes ethical) aspects in the transition processes in NMDs. However, only a few try to identify a model applicable to multiple NMDs.

Supplementary Table I reports the main articles containing the selected keywords in the title and/or in the abstract. It shows the aims and main topics of each article. Moreover, through an analysis of the entire text of each article, they were screened according to the topic of this review.

## Results

### *Neuromuscular disorders and transition process*

Interest in transition from pediatric to adult care in NMDs is increasing. Key aspects do not only include medical care but also psychological and social aspects, and increased survival requires rethinking educational, social and psychological services. For a successful transition and care, integration of wider social issues, involvement of young patients and their families in decision-making, support of patient advocacy groups, and proactive care are not to be overlooked<sup>35,36</sup>. To improve patient outcomes, an extensive transition program should focus on the patient’s physical, psychological and social development and not only plan physical transfer from pediatric to adult care<sup>37</sup>. However, particular attention should be paid to the needs and requirements of a wide range of people with different and more or less debilitating conditions in order to avoid negative outcomes. Involving adolescents in planning transition could be a good strategy. For example, a key aspect is independence and it should be determined whether is a goal to be pursued or not<sup>15</sup>. For a young patient with a progressive neuromuscular disease, transition could be an intense period (both from a psychological and physical perspective) in which skills could be lost and dependence on others increases<sup>38</sup>. Furthermore, understanding the personal characteristics as well as the needs and aspirations of young people could help provide the right health care<sup>39</sup>.

The perspectives and needs of patients with NMDs should not be underestimated. Young people with NMDs report a similar view in the different forms of diseases, of the transition process. Care during transition should be consistent and tailored to the individual’s needs<sup>10,40</sup>; access to a wide range of information should be guaranteed. The process should be gradual and timed and should include peer support programs<sup>41</sup>. Therefore, mental and social health is a crucial aspect of the process.

Family members need to be involved in planning. This allows professionals to make choices in line with

their needs and values, involve them in decision-making and provide adequate information in the right ways<sup>38</sup>.

From a physical point of view, it is necessary to consider the progression of many signs and symptoms. For example, progressive muscle weakness as the main manifestation of DMD may become exacerbated in adult age owing to a lack of adequate and regular physical activity<sup>42</sup>. Therefore, a multidisciplinary rehabilitation assessment with interventions across all disease stages is recommended<sup>9,42,43</sup>. Respiratory problems are another example. All or almost all therapies for respiratory management (e.g. monitoring of respiratory muscle function and the timely use of lung volume recruitment, assisted coughing, nocturnally assisted ventilation, and subsequent daytime ventilation) should be used even before transition from pediatric to adult care providers<sup>44</sup>.

#### *Health and physical care*

For patients with NMDs, transition from pediatric to adult care is a critical phase. Key findings suggest that this process remains problematic with a gap between pediatric and adult care services, including significant differences in clinical practice and culture with a tendency for the adult physician to focus more on specific medical aspects as opposed to a global, interdisciplinary view. The transition process should ensure a plan for ensuring continuity of care with pediatricians<sup>10</sup>.

The pediatric team should initiate the process gradually by educating patient and caregiver, identifying the practitioner in charge, updating key management plans, addressing key problem areas, and assessing readiness to transfer at each visit<sup>45</sup>. An up-to-date and accessible medical record may be useful and help the team to collaborate actively and jointly<sup>46</sup>.

Monitoring patients with chronic conditions allows to observe recurring themes present and relevant in most NMDs, attributed to the specific muscle or nerve disease and its effect on other areas of the body<sup>47</sup>.

Brown and colleagues<sup>26</sup> suggest that young patients and their caregivers should be involved in transition planning (at least once a year) through scheduled visits starting from age 13. Indeed, as mentioned above, the recommended age for an effective transition is between 12 and 14 years old<sup>25,27</sup>. These visits should address: the young person's medical condition; current medications and any side effects; genetic counseling and reproductive implications of the condition; issues related to sexuality and puberty; psychological well-being; any additional tests or assessments to be performed prior to transfer; a current assessment of the young person's understanding of his/her diagnosis and prognosis (if with severe cognitive impairment, plans are established for legal guardianship). Physicians should provide transparent information and

guidance to patients on the management of the terminal phase of NMDs, too. But often this type of communication occurs at a late stage<sup>48</sup>.

During adolescence, therefore, transition from childhood to adulthood, symptoms and signs may progress. For this reason, rehabilitation or support should be provided. The main areas of intervention may concern the muscular and skeletal system, the respiratory system, cardiological aspects and consequences, gastrointestinal and nutritional problems, the endocrine system, kidneys problems, orthopedic problems<sup>42,43,49-55</sup>. Patients with NMDs should attend follow-up visits on a regular basis, otherwise their risk of developing complications typical of NMDs or of their progression increases.

Emergency care planning is another key component of transition. Appropriate and timely interdisciplinary emergency management is critical for survival in these patients. Emergency cards (ECs) are provided for this purpose to patients and caregivers as a way to ensure timely and appropriate relief for these patients with unique and complex medical needs<sup>56</sup>.

Obviously, then, transition from pediatric to adult care in patients with NMDs is complex and requires effective organization and planning.

#### *Patients and caregivers: psychological care and implications*

For a successful and useful transition from pediatric to adult care, psychological aspects in neuromuscular diseases should not be underestimated. For example, patients with SMA often experience anxiety, depression, and social isolation in response to increased symptom-related stress. However, mental health remains one of the unmet needs<sup>7,57</sup>.

A high prevalence of depression and anxiety was found among school-age patients (age 8-18 years) with SMA and other muscular disorders. Prevalence seems to differ across school types, academic delay, household SES, clinical characteristics of disease, caregivers' mental health and expectations. No gender-related differences emerged<sup>58,59</sup>. This confirms once more the importance of making the transition process consistent with the young patient's needs and their family's.

With regard to dystrophies, it has been suggested that dysfunctions in dysbindin due to alterations in dystrophin could increase the risk of depression<sup>60,61</sup>. Some studies point to an increased incidence of anxiety and obsessive-compulsive disorder among children and adolescents with DMD<sup>62-64</sup>. Other emotional, affective and behavioural disorders, such as aggression, may also be present<sup>65,66</sup>. Physical symptoms may increase stress, especially in DMD vs Becker muscular dystrophy (BMD), the latter causing a less severe physical impairment<sup>67</sup>.

Affective disorders should not be regarded as secondary conditions because they are not caused by mental retardation or physical disability. Furthermore, environmental and/or social factors, such as bullying, can affect mental health in NMDs <sup>68,69</sup>.

Families often do not know what to do during the transition process, and young patients sometimes avoid telling their families how they feel about their disease or try to hide them to protect their parents <sup>12,70</sup>. Parents, on their part, feel useless or hopeless, which interferes with daily life. This may increase the risk of a major depressive episode <sup>71</sup>. Erby and colleagues <sup>72</sup> highlight that parents are afraid to explain the terminal nature of the disease to their children, and these fears could impact communication within families about care planning issues.

Because of the limited nature of screening, regular mental health assessments ensure continuity of care and provide longitudinal measures of the psychological state of the patient and families <sup>73</sup>. As previously reported, caregivers may experience stress as well as emotional and physical burden. One study reported more than fifteen components of caregivers burden, including stress, pain, anxiety, depression, sleep quality, and sexual function <sup>74</sup>.

In addition, periodic neuropsychological assessments are advisable. There may be a correlation between molecular defects in genes mainly responsible for muscle diseases and cognitive impairment or between neurological findings and intellectual disability <sup>75</sup>. Therefore, active collaboration between professionals, patients and caregivers is necessary to ensure optimal transition in this critical period.

### *Social care and implications*

The importance of social skills and social support during transition processes is an understudied topic in the literature. A combination of psychoeducational, psychosocial, relationship-centered, family-based education, support, communication, problem solving, and skills development interventions is recommended <sup>76</sup>. It appears that poor social and interpersonal behavioural skills are more significant than an increase in depressive and anxious behavior in boys with DMD <sup>77</sup>. In the same study, many boys with DMD display relevant social problems, such as poor peer relationships and immaturity. Furthermore, patients with NMDs could experience an important change in their social relationships, characterized by feelings of loneliness and isolation <sup>78</sup>.

Especially adolescents express the need for meaningful relationships with professionals, family and friends looking for greater support in daily life <sup>79</sup>.

Children with NMDs may show a particular behavioural phenotype with difficulties in social interaction and communication and probable issues in theory of mind <sup>80</sup>.

DMD and BMD boys appear to have a higher level of social and communication difficulties compared with children with other muscular disorders or with the general population <sup>81</sup>.

Poor social support could lead to problematic behaviours. As reported by Fee and Hinton <sup>82</sup>, problematic behaviours decrease with increased social involvement, which acts as a protective factor and also leads to a reduction in internalizing problems. Social closeness with peers with similar life experiences can enhance the physical and psychological well-being of young people with NMDs (or, in general, with disabilities) <sup>83</sup>.

Finally, turning to the needs of young people, information on more intimate aspects should be collected, including desire for intimacy, sexuality and family planning. In point of fact, the need to explore one's identity arises in adolescence, but it becomes manifest above in early adulthood, when individuals start to think how to structure their life – both private and professional <sup>84</sup>. There are not many studies that focus on employment needs of young people with NMDs during transition into adulthood. As reported by Lindsay et al. <sup>85</sup>, transition plans from an occupational point of view are insufficient. Young people with NMDs experience severe disadvantages during transition to adulthood, especially in pursuing meaningful occupations, such as school, volunteering, employment, social and recreational activities. Thus, unemployment, social isolation and depression are widespread among this population. An important intervention in this area could be geared to increase self-esteem of young patients with NMDs <sup>40</sup>.

## **Discussion**

This review highlights how transition from pediatric to adult care in NMDs is a process not just encompassing physical aspects but also psychological and social ones. It thus required a multidisciplinary approach (Fig. 1). The literature on transitional care is still fragmented and vague, although the increasing importance of psychosocial factors <sup>10,86,87</sup>.

However, it is indisputable that correct physical programming is essential for a transition with positive outcomes. Periodic and continuous checks regarding the medical condition, pharmacological and genetics and a preparation of the patient regarding his own diagnosis and prognosis are very important aspects. Collaboration between professionals is an essential condition for a successful transition. A gap in transition planning emerges for most adolescents, both with and without mental, behavioural and/or developmental disorders <sup>88</sup>.

The findings of this review, even if not systematic, can be grouped into three macro-areas of interest: health

and physical needs, young patients' and caregivers' psychological needs, and social aspects.

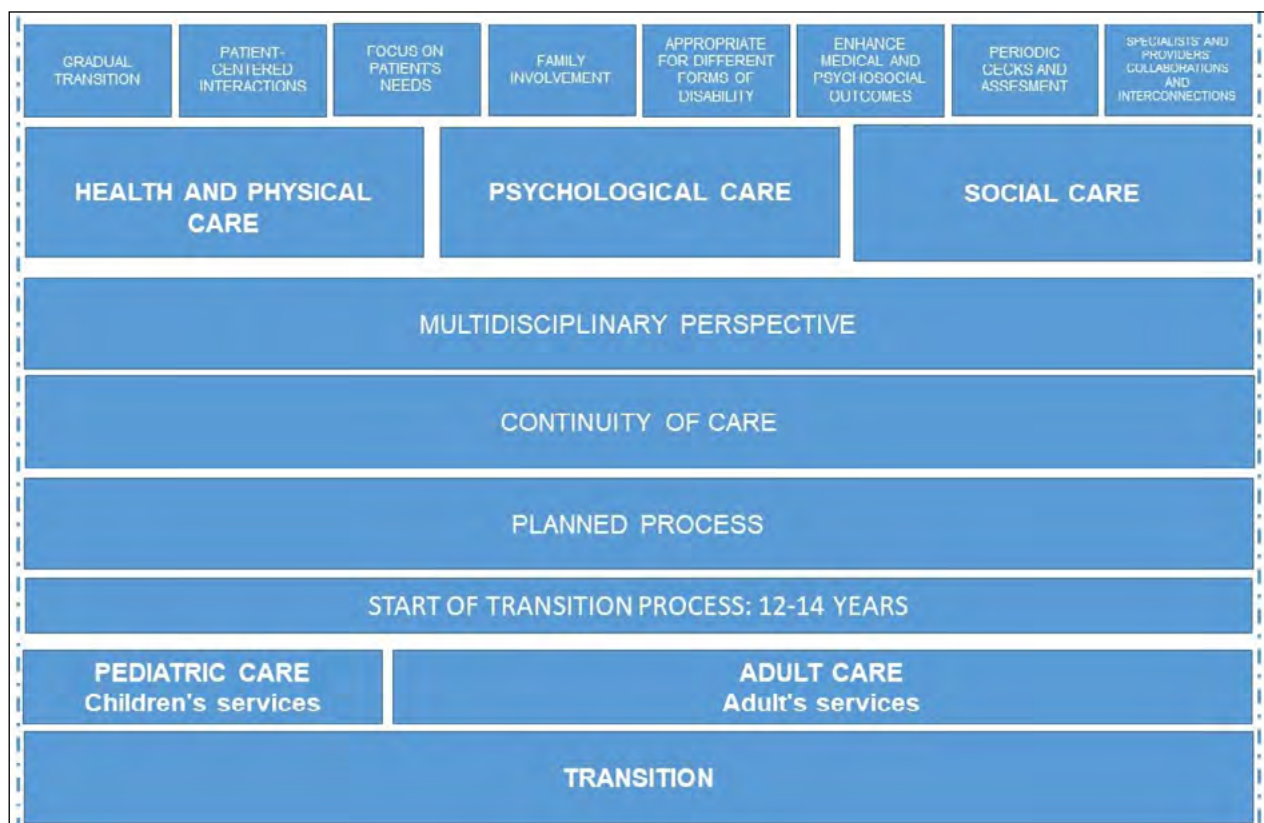
Regarding psychological care, young patients are more likely to show internalizing and externalizing problems (such as anxiety, depression, aggression) during this phase, as seen in some studies<sup>59</sup>. Moreover, anxiety seems to mediate the relationship of negative coping and transition readiness in youths<sup>89</sup>. Furthermore, social involvement – in general – and social closeness with peers – in particular – are related to the patients' well-being<sup>82,83</sup>.

Another aspect that should not be overlooked is caregiver distress<sup>71</sup>. Parental or caregiver stress increases in the transition period mainly due to problems with social interaction<sup>90</sup>.

Clearly, children and adolescents have different needs and demands (medical, psychological and social) which need be reconciled. For an optimal transition, physical care, emotions, social involvement, education, sexuality, family implications, job possibilities, and advocacy need to be addressed. Improving transition from adolescent to adult health care may be considered a real goal to achieve good care outcomes for individuals with chronic conditions<sup>91</sup>. This shows once again how an optimal transition process involves the collaboration of several professionals<sup>45</sup> and active participation of patients and caregivers.

## Conclusions

This work has some limitations. Indeed, this is a non-systematic review of the literature. For this reason, it lacks an explicit intent to maximize scope or analyse data collected, and therefore there could be some bias<sup>34</sup>, but our main purpose was to stress that transition should not only be planned taking into consideration physical aspects but also psychological and social aspects, namely implementing a multidisciplinary model of intervention<sup>92</sup>. We found that few or no studies explored the transition process from pediatric to adult care in neuromuscular diseases and attempted to identify a general pattern of transition applicable to all NMDs. Studies in the literature provide fragmented findings due to the different manifestations of NMDs and do not delve into topics such as sexual activity, family planning or the social impact of neuromuscular diseases<sup>10</sup> or do the psychosocial aspects<sup>8,93</sup>. Most studies do not report longitudinal or outcome data on successful transition in patients with NMDs. It might be useful to start drafting unique general guidelines for the transition process from pediatric to adult care that can take into account physical, psychological and social aspects and that can be applied to all patients with NMDs. Rather than drawing up general guidelines, the greatest



**Figure 1.** Successful factors for transition from pediatric to adult care in a multidisciplinary perspective.

difficulties could arise on a practical level. For example, it may be difficult to interconnect and coordinate a large number of individuals or to afford the costs, given that patients would have to be followed up by multiple specialists or specialists may not be adequately trained in the field of transition <sup>6</sup>.

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#### Conflict of interest statement

The authors declare no conflict of interest.

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#### Author's contributions

GA, AT: study conceptualization and design. CF, MCO, IG, IF and MDR: literature search and data extraction. GA and AT: original draft of the manuscript. AT and GA: critical revision of the manuscript. Finally, all the authors approved the final version of the manuscript.

#### Ethical consideration

The research was conducted ethically, with all study procedures being performed in accordance with the requirements of the World Medical Association's Declaration of Helsinki.

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**Supplementary Table I.** Articles containing the selected key words in the title and/or in the abstract. Main purpose and consistency with the topic of this review.

Authors	Year	Title	Journal	Aim	Main topic and coherence with this review
Fleischer et al.	2022 <sup>6</sup>	Essen transition model for neuromuscular diseases	Neurological Research and Practice	Structuring a transition process with an interdisciplinary collaboration to improve patients' quality of life	Presentation of a structured transition process for some neuromuscular pathologies established at Essen University Medical Center. Psychosocial aspects are not addressed in detail
Cheng et al.	2020 <sup>38</sup>	Transition of patients with neuromuscular disease and chronic ventilator-dependent respiratory failure from paediatric to adult pulmonary care	Paediatric Respiratory Reviews	Identifying difficulties and needs in transition for patients with neuromuscular diseases and chronic respiratory failure.	In-depth analysis of pshysical and psychological aspects, but mainly focus on breathing difficulties
Willis	2020 <sup>25</sup>	Transition from pediatric to adult care for young adults with chronic respiratory disease	Respiratory Care	Increasing clinician awareness of health care transitions of young adults with chronic respiratory disease	Detailed definition of transition and description of a successful transition. Focuses are on chronic respiratory problems that could be also caused by NMDs



**Supplementary Table I.** Continues.

<b>Authors</b>	<b>Year</b>	<b>Title</b>	<b>Journal</b>	<b>Aim</b>	<b>Main topic and coherence with this review</b>
Menon et al.	2022 <sup>45</sup>	Clinical profile and multidisciplinary needs of patients with neuromuscular disorders transitioning from paediatric to adult care	Neuromuscular disorders	Description of the spectrum of neuromuscular diseases evaluated through a pediatric to adult neuromuscular transition program and various issues requiring specific services. The study suggests the need of multidisciplinary clinical care	Focus on the physical and psychosocial aspects (the latter are not very detailed). Particular attention on issues of different services. Importance of a multidisciplinary clinical perspective
Paguinto et al.	2020 <sup>95</sup>	Multidisciplinary perspectives and practices of wheelchair prescription for children with neuromuscular conditions	Disability Rehabilitation. Assistive Technology	Understanding healthcare professionals' clinical perspectives and practices when recommending wheelchair equipment for the first time for pediatric neuromuscular disorders	Transition as the passage from deambulation to wheelchair in children with progressive NMDs
Burns et al.	2014 <sup>96</sup>	The cerebral palsy transition clinic: administrative chore, clinical responsibility, or opportunity for audit and clinical research?	Journal of Children's Orthopaedics	Clear communication needs with children with cerebral palsy in transition to adult services	Definition of transition and focus mainly on physical aspects and little on the psychological ones. Focus only on cerebral palsy
Ambrosini et al.	2019 <sup>94</sup>	"Be an ambassador for change that you would like to see": a call to action to all stakeholders for co-creation in healthcare and medical research to improve quality of life of people with a neuromuscular disease	Orphanet Journal of Rare Diseases	Investigating the position of neuromuscular patients in a European foundation of patient organisations with respect to health care and medical conditions to identify and address gaps and bottlenecks	Focus on "shared decision making" (SDM) and on patient involvement. Particular attention on patient involvement and on psychosocial aspects
Wasilewska et al.	2020 <sup>42</sup>	Transition from childhood to adulthood in patients with Duchenne Muscular Dystrophy	Medicina	Overview of healthcare needs related to the transition from pediatric care to adult care in patients with DMD	Focus on physical and psychosocial aspects of transition, but only on DMD patients



**Supplementary Table I.** Continues.

<b>Authors</b>	<b>Year</b>	<b>Title</b>	<b>Journal</b>	<b>Aim</b>	<b>Main topic and coherence with this review</b>
Sonneveld et al.	2013 <sup>97</sup>	Gaps in transitional care: what are the perceptions of adolescents, parents and providers?	Child: Care, Health and Development	Exploring perspectives in adolescents with chronic conditions, their parents and providers on transitional care. Exploring the extent to which such perspectives are disease-specific	Other pathologies in addition to neuromuscular ones are taken as example (juvenile rheumatoid arthritis, diabetes Type I). Main focus on participants' perspective and their experiences with transitional care
Baldanzi et al.	2016 <sup>98</sup>	Hard ways towards adulthood: the transition phase in young people with myotonic dystrophy	Acta Myologica	Identifying areas of unmet needs and targeted health objectives that ensure support to myotonic dystrophy type 1 (DM1) population	Main focus on psychosocial aspects during transition process. However, it focuses on DM1
Paguinto et al.	2020 <sup>99</sup>	"It's not just the wheelchair, it's everything else": Australian parents' perspectives of wheelchair prescription for children with neuromuscular disorders	Disability and Rehabilitation	Investigating parents' perception or their experiences of their child's transition to wheelchair equipment	Transition seen as the passage to wheelchair for children with neuromuscular disorders. Focus on parents' perspectives and experiences
Tripodoro & De Vito	2015 <sup>48</sup>	What does end stage in neuromuscular diseases mean? Key approach-based transitions	Current Opinion in Supportive and Palliative Care	Providing a definition of end stage in neuromuscular diseases, highlighting the implications for patients, family and healthcare team	Transition seen as the move to supportive and palliative care. Focus on physical and psychosocial issues
Lu et al.	2019 <sup>100</sup>	Transition to adult care in sleep medicine	Paediatric Respiratory Reviews	Considering the common barriers to transition and reflect on the specific barriers relating to patients managed by the sleep medicine team	Main focus on barriers in transition process, but particular attention on sleep medicine