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Case Report

The importance of palliative care in Duchenne muscular dystrophy: a reflection

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ABSTRACT

Duchenne muscular dystrophy is a progressive disease involving early morbidity and mortality of children. Hence, it becomes essential to address the evolving issues comprehensively according to palliative care principles. Apart from physical issues, psychological, ethical and spiritual concerns affect the quality of life throughout the symptomatic period including the end of life. This case depicts the incomplete management of progressive physical issues as well as psychological, ethical and spiritual concerns of a ten-year-old child and his family. Lack of proper communication and inadequate knowledge about end-of-life care adversely impacts the quality of life of such patients. Understanding of Palliative Care principles helped to gain insight about the lacuna in clinical care of such patients. Knowledge and incorporation of Palliative Care principles is extremely important for any clinician dealing with progressive diseases like Duchenne Muscular Dystrophy and many more. Application of these principles has the potential to enhance the quality of life of patients and their caregivers significantly throughout the course of the disease.

Keywords: Muscular dystrophy, Palliative care, Quality of life, Needs

INTRODUCTION

Duchenne muscular dystrophy (DMD) is a genetic disorder affecting the musculoskeletal, cardiopulmonary, gastrointestinal, endocrine and nervous system.[1-3] It leads to early morbidity and mortality of the affected male child, thus significantly disrupting the quality of life (QOL) of the patient as well as the whole family. Identifying their needs with a comprehensive care is required at each level of clinical management.

CASE REPORT

A 10-year-old male child visited our OPD two years back. The Physician observed him walking without support with a waddling gait, toe walking and lumbar lordosis. This gave a picture of Muscular Dystrophy. He was referred from pediatrics, to physical medicine & rehabilitation (PMR). The history as stated by the parent's revealed difficulty in walking with frequent falls, and difficulty in standing from a sitting or supine position from the last 4-5 years. The symptoms were gradually progressive. There were no complaints of weakness in the upper limbs, bending of back, difficulty in speaking, swallowing or breathing. There was no history suggestive of any family member having similar complaints or having diagnosed of any muscular disease earlier. The examination revealed hypertrophy or pseudo hypertrophy of bilateral calf with a positive Gower's sign, and gait as mentioned above. There were no contractures, scoliosis or any sensory deficit. The power in bilateral hip girdle muscles and bilateral knee extensors was in the range of grade 3-4, with the power in

the trunk muscles being fair. Rest of the muscles had normal power. The cardiovascular and respiratory system examinations were found to be normal. The child was diagnosed as DMD based on genetic testing, revealing a deletion in the dystrophin gene, with raised serum creatine phosphokinase and lactate dehydrogenase levels. The other tests including routine hemogram, liver, kidney and thyroid function tests were depicted as normal. He had already received immunization with the Influenza and Pneumococcal vaccine, and was started on oral steroids and calcium supplements since the last few months, from pediatric neurology.

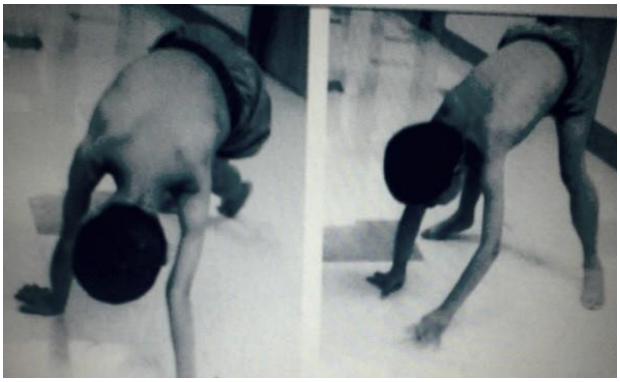

Figure 1: Patient of Duchenne muscular dystrophy with proximal muscle wasting and positive Gowers sign.

As the diagnosis was evident, the Physician chalked out a rehabilitation management plan. It was inclusive of sub maximal non-fatigable strengthening exercises, range of motion & active assisted exercises for bilateral upper and lower limbs, and trunk muscles. The Physician also advised aerobic exercise, deep breathing exercises, energy conservation and joint protective techniques, positioning and postural care, and prevention of falls. He was not advised for any walking orthotics as there were no contractures and the toe walking was due to weakness in the proximal group of muscles. Providing with any walking orthotics would have been detrimental to his muscles and have caused further damage.

The parents were well aware of the prognosis, so the Physician didn't explain much further and insisted the child to continue the exercises along with his studies, and asked them to revisit after 6 months. Following this, he was sent to the occupational and physical therapist for learning the exercises. The patient came for follow up after 8 months, and it was astonished to see him on the wheelchair, being obese. He had developed knee and ankle contractures. The Physician hunched the obesity to be probably due to steroid intake and inactivity. The parents stated that he was not doing exercises well and eating too much lately. It was stressed upon them briefly on the importance of exercises, and sent them to a social worker for procuring a pediatric wheelchair. They were asked to follow up in pediatric Neurology for the dose of steroids to be continued.

DISCUSSION

When it was reflected back with the new knowledge of Palliative Care, we found various gaps in our management for this patient. There was a lacuna in the management plan in relation to the physical, psychosocial, spiritual, communication and ethical aspects as follows:

Physical aspect

What did the physician do? The physician looked after maintaining the strength of the muscles and care for the joints, positioning and support for daily activities. This was for preventing the development of any deformity while helping the child to maintain the functional level for as long as possible.[1] What did the physician not do? The physician did not look after his bone health. As he was prone for osteoporosis owing to steroid intake and inactivity, the physician could have assessed his fragility with certain investigations like serum calcium, phosphate, alkaline phosphatase, kidney function test, parathyroid hormone level and dexa scan. The Physician could have explained them the probability and cause for decline in bone health, and how to prevent fractures and falls.[2]

As the patient was growing obese, the physician could have advised for a diet plan according to the requirement based on the energy expenditure.[1] It could have been stressed more to both parents and child, regarding the importance for exercise, activities and positioning. The Physician could have checked how they have been performing the exercises. They could have been informed regarding dealing with adrenal issues that could occur due to abrupt stopping of steroid therapy and during stressful situations.[1] The Physician could have advised about the occurrence of other physical deterioration in the later years of his life like swallowing and breathing difficulty, increased respiratory secretions and cardiac failure. Due to swallowing difficulty, weight loss could be experienced after the initial weight gain due to steroid intake and inactivity. The physician could have advised them to consult if they observe any such symptoms, and the ways for maintaining nutrition and hydration at that time.[2,3] The Physician could have referred them to Pulmonary Medicine for assessment of respiratory function and interventions for the same.[2] The patient was already undergoing a follow up from Cardiology. The Physician could have stressed on the prevention of infections which may lead to adrenal insufficiency and deterioration in muscular strength and functioning of the whole body.[1]

Psychosocial aspect

What did the physician do? The physician listened to the parents' worries and acknowledged their concerns regarding their child's treatment. It was mentioned about the muscular dystrophy support groups. What did the Physician not do? The physician did not address the concerns of the parents in detail, or to the point of their satisfaction. Their emotional and social health was not

assessed objectively. It was not communicated regarding the respite measures. The Physician did not stress the importance of involvement in social activities or maintenance of social relationships for both the parents and the child. The child was not questioned about his emotional concerns and difficulties in social involvement, and continuing education. The Physician did not make recommendations for associating with other such patients and their families to exchange their experiences. The Physician did not assess the siblings' emotional needs and concerns.[3-5]

Spiritual aspect

What did the physician do? The physician listened to the spiritual issues and pain of the parents with their concerns regarding the reduced longevity of their child. What did the physician not do? The physician did not assess the spiritual issues of the parents and child objectively.[4,6] The Physician could not address the parents' questions regarding their child's age and life, related to a non-treatable disease and why did it happen to their child only. The Physician did not consider spirituality to be an important aspect to be addressed for such patients. The Physician didn't give them enough time to express all their spiritual concerns. The Physician did not address the spiritual issues of the siblings. The Physician could have guided them according to their spiritual or religious beliefs.

Ethics

What did the physician do? The Physician explained regarding the prognosis and purpose of rehabilitation. What did the physician not do? The Physician didn't convey about the available treatment, ongoing research, complications that may arise from treatment, and how to prevent it.[7,8] The Physician didn't convey about the end of life issues, and how to ensure care and support during that phase.

Communication

What did the physician do? The Physician communicated about the rehabilitation plan to the parents, and necessary precautions regarding musculoskeletal care to be done. The Physician communicated regarding continuing education and all the activities with precautions which the child would do, like other children. The Physician communicated about the prognosis and to review early if any new symptom occurs or aggravates. What did the physician not do? The Physician did not give enough time to let the parents come up with all their concerns related to the disease, their emotions, fears, spiritual questions, their hopes and expectations, and their own global QOL. The Physician did not ask the child if he had any questions related to his health condition, and any other concerns related to his social life, education, future prospects, spirituality and emotional health.[6] The Physician did not communicate with the siblings of the child for their concerns, and about relationship with their parents and the patient. The Physician did not communicate about the architectural and other accommodative changes at home and school that may have required to be done once the patient loses ambulation. [3] The Physician did not explain to both the parents and the child about the changes related to health, sexuality, activities of daily living, education and vocation, to be made during the transition from childhood to adolescence and further to adulthood. The Physician did not discuss about the future prospects for the child in relation to his disability.[3] The Physician did not communicate regarding appropriate referral to Palliative Medicine from the initial stage of him being diagnosed with the disease. The Physician did not discuss regarding their concerns and fears related to death. The Physician did not explain them in detail about the relevant support groups & registries, and how to access them.[9] The Physician did not discuss the management as a group together with other members of the PMR team inclusive of the physical and occupational therapist, psychologist, social worker and rehabilitation nurse, along with the patient and his parents. The Physician could have conducted group sessions inclusive of other such patients along with the child and his parents.

CONCLUSION

To conclude, the important points required to be inculcated for a total care are as follows: helping the patients and their families to understand completely about the disease, its management plan, prognosis and future prospects. Giving enough time to the patients and families to let them express their concerns, doubts and fears. Objective assessment of the QOL, psychosocial and spiritual aspect along with the physical domain. Involving the patients and their families as active members in guiding the management plan and end of life care. Ensuring the spiritual aspect and concerns about death to be addressed as is possible, which is often ignored. Allowing other members of the management team to give equal contributions regarding the management and care of the patients and their families. Group therapies for such patients and their families. Collaborating with other specialties involved in the care of patient like pediatric neurology, palliative medicine, pulmonary medicine, cardiology, along with PMR, to come under one roof for the smooth transitioning of patient and their families at each level. Organizing joint clinics and conferences. Spiritual workshops for both the patients and medical professionals. Communicating well.

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REFERENCES

1. Birnkrant DJ, Bushby K, Bann CM, Apkon SD, Blackwell A, Brumbaugh D, et al. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management. Lancet Neurol. 2018;17(3):251-67.
2. Birnkrant DJ, Bushby K, Bann CM, Alman BA, Apkon SD, Blackwell A, et al. Diagnosis and management of Duchenne muscular dystrophy, part 2: respiratory, cardiac, bone health, and orthopaedic management. Lancet Neurol. 2018;17(4):347-61.
3. Birnkrant DJ, Bushby K, Bann CM, Apkon SD, Blackwell A, Colvin MK, et al. Diagnosis and management of Duchenne muscular dystrophy, part 3: primary care, emergency management, psychosocial care, and transitions of care across the lifespan. Lancet Neurol. 2018;17(5):445-55.
4. Bell M, Biesecker BB, Bodurtha J, Peay HL. Uncertainty, hope, and coping efficacy among mothers of children with Duchenne/Becker muscular dystrophy. Clin Genet. 2019;95(6):677-83.
5. Magliano L, Patalano M, Sagliocchi A, Scutifero M, Zaccaro A, D'Angelo MG, et al. I have got something positive out of this situation: psychological benefits of caregiving in relatives of young people with muscular dystrophy. J Neurol. 2014 ;261(1):188-95.
6. Pehler SR, Craft-Rosenberg M. Longing: the lived experience of spirituality in adolescents with Duchenne muscular dystrophy. J Pediatr Nurs. 2009; 24(6):481-94.
7. Nelson SF, Crosbie RH, Miceli MC, Spencer MJ. Emerging genetic therapies to treat Duchenne muscular dystrophy. Curr Opin Neurol. 2009;22(5): 532-8.
8. Lim KR, Maruyama R, Yokota T. Eteplirsen in the treatment of Duchenne muscular dystrophy. Drug Des Devel Ther. 2017;11:533-45.
9. Wang RT, Nelson SF. What can Duchenne Connect teach us about treating Duchenne muscular dystrophy? Curr Opin Neurol. 2015;28(5):535-41.

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