Research Article

Parental Healthcare-Seeking Behaviors of Filipinos with Duchenne Muscular Dystrophy

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Background: Duchenne muscular dystrophy (DMD) is the most common X-linked dystrophinopathy characterized by a chronic progressive neuromuscular degeneration. Early diagnosis and interventions may delay or prevent its complications. A factor that affects prompt diagnosis is the healthcare-seeking behavior of the parents. With the ultimate goal of improving care and outcomes of patients with DMD, this study aimed to assess caregivers' healthcare-seeking behavior in a developing country.

Methods and Results: Using a prospective cross-sectional study, factors affecting the parental healthcare-seeking behavior of patients with DMD were investigated. A total of 55 parents participated. There is delay between the mean age (4 + 2.3 years) when first symptom was noted and the first medical consult (6.6 + 2.3 years). Furthermore, there is a delay of 4.5 + 1.1 years from the symptom onset to DMD confirmation as majority opted observation and traditional medicine prior to physician consult. Upon medical consultation, multiple variables also affected arrival to a diagnosis. Facilitating factors include the first physician handling the patient doing a diagnostic evaluation or alleviating caregiver concern, a positive family history, and physical therapist or pre-school teacher recognizing the problem. Impeding factors include cultural beliefs, financial constraints, alternative diagnosis or non-action by the physician, parental non-compliance to medical advice, and family logistics.

Conclusion: The identified factors affecting prompt arrival to diagnosis should be the target of local healthcare systems as areas of development in order to translate to timely treatment and intervention and thus a better quality of life for patients with DMD.

Keywords: Duchenne muscular dystrophy, health-seeking behavior

INTRODUCTION

Duchenne muscular dystrophy (DMD) is the most commonly encountered X-linked recessive neuromuscular condition with an estimated prevalence of 1 in 3500 to 5000 male births. Together with Becker muscular dystrophy (BMD), it is in the spectrum of a group of diseases called dystrophinopathies. [1]

Dystrophinopathies are a result of genetic mutations, primarily exon deletions leading to out- or in-frame mutations resulting to absent or a partially functional dystrophin protein, respectively.[2] Due to this mutation, patients with DMD develop a progressive muscle weakening leading to increased disability. Despite the technological advances, the mean age at diagnosis (4.7 years old) has remain unchanged.[3]

Care for patients with DMD will require a multidisciplinary approach involving several specialists. The condition undergoes through progressive five clinical stages – 1) pre-symptomatic, 2) early ambulatory, 3) late ambulatory, 4) early non-ambulatory and 5) late non-ambulatory.[4] The annual total medical cost of this disorder per patient may range from €4 420 (Php 24 300) in the Stage 1 to €68 968 (approximately Php 3.8 M) in the Stage 5 of the disease.[5] In a developing country like the Philippines, aside from the financial burden to achieve recommended

care, further challenges may include the availability of the genetic tests, accessibility to novel pharmacologic therapies and multidisciplinary care.

Early diagnosis provides the best opportunity for maximum benefit from the current standard of care and access to novel therapies.[4,6,7] However, certain challenges are present even before arrival to a specialty clinic including the parental health-seeking behavior.

Health-seeking behavior is defined as all the activities undertaken by the individuals who perceive to have a health problem for the purpose of finding an appropriate remedy. A model was used to describe and characterize the health-seeking behavior of parents. There are four stages of this help seeking behavior - 1) problem recognition, 2) decision to seek help, 3) service selection and 4) service utilization. This model suggests that all families pass through a unidirectional movement along each stages which is controlled by a complex interplay of factors such as 1) predisposing characteristics, 2) social networks, 3) cultural factors, 4) enabling resources and 5) barriers to care.[9]

Given such model, the authors opted to characterize the parental health-seeking behaviors of the caregivers of DMD patients in order to identify the challenges and recommend targeted strategies in facilitating early diagnosis, prompt intervention and optimal outcomes among Filipino DMD patients.

METHODS

This study was approved by the Institutional Review Ethics Board. Utilizing a prospective cross-sectional design, our study investigated the factors affecting the healthcare-seeking behavior of parents or caregivers of pediatric patients aged less than 19 years old diagnosed with Duchenne muscular dystrophy seen at the Multidisciplinary Neuromuscular Clinic of the University of the Philippines – Philippine General Hospital (UP-PGH). With informed consent, the caregivers were requested to participate in an interview.

The study identified the facilitating and impeding factors that affected their healthcare-seeking behaviors. Descriptive statistics were used to summarize the demographic and clinical characteristics of the patients. Frequency and proportion were used for categorical variables.

RESULTS

Demographic Profile of the Patients

The study recruited 55 respondents who were parents of children with genetically-confirmed DMD. Majority (52.73%) belongs to the 6 to 10-year old group while 40% are already adolescents. Almost half (47%) are in their 4th stage of the illness at the time of study.

Majority of the participants are Roman Catholics and 95% percent are enrolled in a government educational institution. Two patients attend an individualized education program (**Table 1**).

Demographic Profile of the Parents or Caregivers

All caregivers interviewed in the study are parents of the patients (n=55). Half belong to the young adult age group while the rest are of middle age. About one-half of the family resides in the urban area within 50-100 km

distance from the multidisciplinary clinic at the UP-PGH. One-fifth of the cohort resides at a distance of more than 250 km.

Table 1. Demographic profile of patients

Characteristics		n	%
Age *(n = 55)*	0 to less than 3 years old	0	0
	3 to less than 6 years old	4	7.27%
	6 to less than 10 years old	29	52.73%
	10 to less than 19 years old	22	40%
Ethnicity *(n = 55)*	Bisaya	5	9.09%
	Bicolano	6	10.91%
	Ilocano	3	5.45%
	Ibanag	1	1.82%
	Tagalog	40	72.73%
Religion *(n = 55)*	Roman Catholic	41	74.55%
	Christian	9	16.36%
	Iglesia ni Cristo	2	3.64%
	Jehovah's Witness	1	1.82%
	Muslim	1	1.82%
	Protestant	1	1.82%
Current form of education *(n = 55)*	School	0	0
	Homeschooling	41	74.55%
	Individualized Education Program	2	3.64%
	None (out of school)	12	21.82%
Type of school *(n = 43)*	Private	2	4.65%
	Public	41	95.35%
Current stage of illness *(n = 55)*	Stage 1	2	3.64%
	Stage 2	14	25.45%
	Stage 3	13	23.64%
	Stage 4	26	47.27%
	Stage 5	0	0

Forty-three percent of parents are college graduates while one-third finished high school. Six out of 10 participants come from low and lower middle income families with an annual income of less than Php 400 000. Nuclear family set-up is observed in half of the participants. More than half of the patients with DMD have two to three caregivers at the same time. Secondary caregivers included grandparents, siblings, aunts, uncles and hired staff. The mean duration of care the primary caregivers in this cohort has spent until the diagnosis was made is 7 years. Those with secondary caregivers have a mean duration of 3 years until confirmation of DMD. (**Table 2**)

Problem Recognition Profile of the Caregivers

The mean age of onset of the symptom of the patients included in this study is 4 (±2.3) years old. Symptom onset is between age 3 to 6 years old in nearly half of boys.

On the other hand, the mean age on first medical consult is at 6.6 (±2.31) years old. This is a 2.6 years delay from the mean age of the onset of symptoms. Ninety-five percent of the patients are in the first two stages of the disease on first medical consult. The remaining were first brought to medical attention only upon reaching the Stage 3 of the illness.

Table 2. Characteristics and demographic profiles of caregivers

Characteristics		n	%
Number of caregivers the patient had from birth to time of diagnosis	1	23	41.82%
	2 to 3	32	58.18%
	4 to 5	0	58.18%
	More than 5	0	0
Mean Duration (years) per caregiver (For patients with >1 caregiver)	Primary Caregiver	7.44	
	Secondary Caregiver	2.94	
Number of caregivers	1	23	41.82%
	2 to 3	32	58.18%
	4 to 5	0	0
Age	Young adult (20 less than 40 years old)	28	50.91%
	Middle age (40 to less than 60 years old)	27	49.09%
	Old age (60 years old and above)	0	0
Type of Residential Area	Urban	30	54.55%
	Rural	25	45.45%
Distance away from hospital	<20 km	5	9.09%
	20 – 50 km	5	9.09%
	50 – 100 km	25	45.45%
	100 – 200 km	8	14.55%
	>250 km	12	21.82%
Educational Attainment	Elementary undergraduate	0	0
	Elementary graduate	0	0
	Highschool undergraduate	2	3.64%
	Highschool graduate	19	34.55%
	College undergraduate	8	14.55%
	College graduate	24	43.64%
	Vocational course	2	3.64%
Annual Total Family Income	<Php 250 000 (20 000/month)	14	25.45%
	(Low income)		
	Php 250 000 – Php 400 000 (20 000 – 33 000)	21	38.18%
	(Lower middle income)		
	Php 400 000 – Php 800 000 (33K to 66K)	7	12.73%
	(Middle income)		
	Php 800 000 – Php 2 000 000 (66K – 166K)	13	23.64%
	(Upper Middle income)		
Family type	Nuclear	29	52.73%
	Solo Parenting	5	9.09%
	Extended	21	38.18%

The most common first symptoms noted by their parents are the abnormal gait and the Gower's sign with some noting concomittant non-motor manifestations. Such non-motor symptoms include behavioral issues, cognitive delay, learning and attentional issues and speech delay. Most of the parents the persons recognizing the problems are the parents in 90%

The mean time interval between first notice of these symptoms until confirmation of DMD is 4.5 ($\pm$1.1) years. One-third of the patients had their confirmatory test done at an interval of one to less than three years from the onset of signs and symptoms.

Table 3. Problem recognition of caregivers

Factors		n	%
Age of patient at the onset of symptom	0 to less than 3 years old	15	27.27%
	3 to less than 6 years old	27	49.09%
	6 to less than 10 years old	13	23.64%
Age of patient on first medical consult	0 to less than 3 years old	4	7.27%
	3 to less than 6 years old	9	16.36%
	6 to less than 10 years old	40	72.73%
	>10 years old	2	3.64%
Stage of Illness on 1st medical consult:	Stage 1	30	54.55%
	Stage 2	22	40%
	Stage 3	3	5.45%
	Stage 4 and 5	0	0
First symptom noted	**MOTOR**		
	Abnormal gait	46	83.64%
	Calf pseudohypertrophy	10	18.18%
	Decreased endurance	4	7.27%
	Difficulty climbing stairs	7	12.73%
	Frequent falling or clumsiness	21	38.18%
	Gower's sign on rising from floor	32	58.18%
	Gross motor delay	4	7.27%
	Hypotonia	4	7.27%
	Inability to keep up with peers	1	1.82%
	Loss of motor skills	1	1.82%
	Muscle pain or cramping	2	3.64%
	Toe walking	25	45.45%
	Difficulty running or climbing	4	7.27%
	NON-MOTOR		
	Behavioral issues	17	30.91%
	Cognitive delay	7	12.73%
	Poor weight gain	12	21.82%
	Learning and attentional issues	7	12.73%
	Speech delay or articulation difficulties	2	3.64%
Person who first noticed the problem	Parents / Caregivers	49	89.09%
	Relatives	6	10.91%
	Friends	1	14.29%
	Non-physician healthcare professional	1	5.56%
Time interval between note of symptoms and confirmation of Duchenne muscular dystrophy	Less than 1 year	0	0
	One to less than 6 months	1	1.82%
	Six to less than 12 months	9	16.36%
	One year to less than three years	17	30.91%
	Three to less than six years	16	29.09%
	More than 6 years old	12	21.82%

Factors Affecting Decision to Seek Help

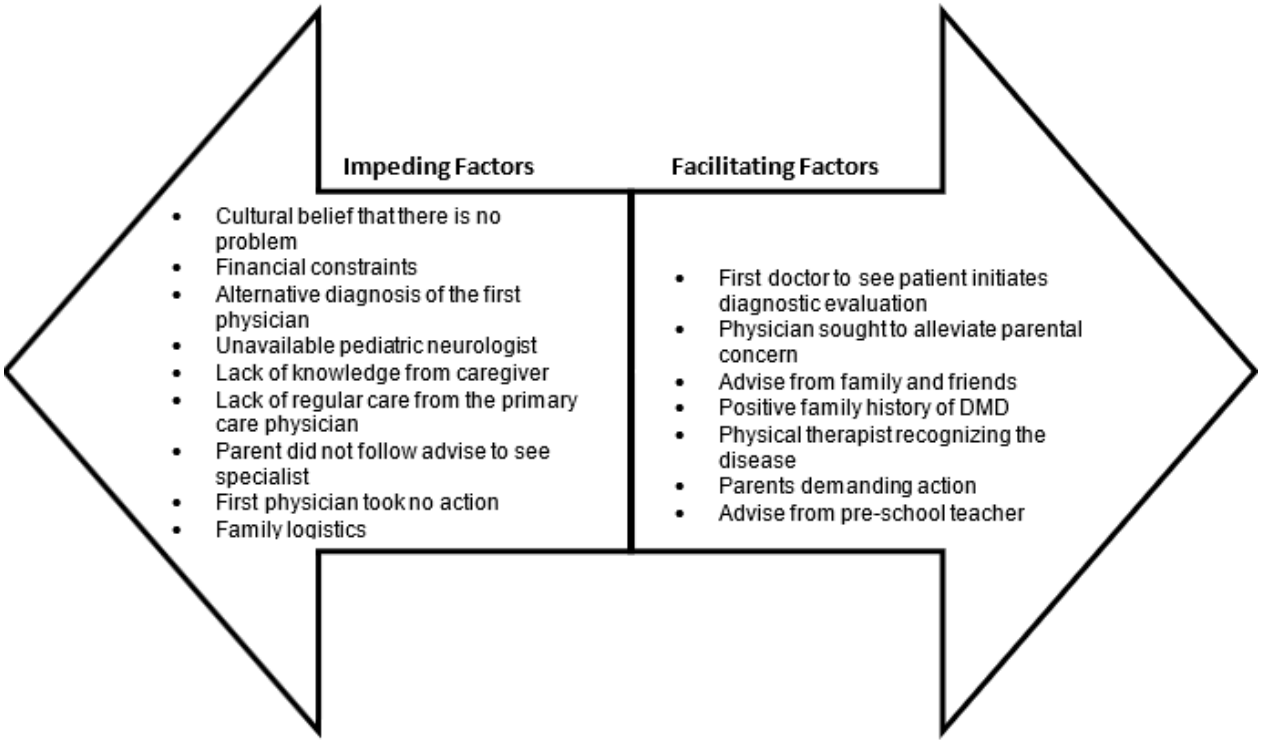
Among the study participants, 42% opted to observe when they noticed the early signs and symptoms of DMD but 75% eventually decided to seek medical consult upon worsening of these manifestations. Twelve out of 55 parents have waited a minimum interval of less than 1 year before bringing the patient to medical consultation. The reasons for the delay in medical consult are multifactorial. These included hesitancy (78.18%), financial constraints (41.82%), difficulty finding time to schedule a physician (12.73%), lack of knowledge (7.27%) and preference to go to a traditional healer (1.82%).

Approximately 40% of the patients were initially seen by a general pediatrician but only half of this group were eventually referred to pediatric neurologists.

Factors Affecting Service Selection

Service selection is affected by several factors. In this cohort, half of the first physicians to see their respective patients already initiated a diagnostic evaluation eventually arriving at the proper diagnosis. Thirteen patients were identified to have a family history of DMD hence the medical diagnostics were immediately facilitated after the first medical consult. Other factors influencing facilitation of the medical diagnostics include 1) physician alleviating the caregiver concern, 2) physical therapists identifying the problem and 3) advice from family, friends and pre-school teacher. (**Figure 1**)

Figure 1. Factors affecting arrival to diagnosis



On the contrary, multiple variables have been identified that impeded moving towards the correct diagnosis. These include 1) cultural belief that there is no problem, 2) financial constraints, 3) misdiagnosis by the initial medical officer, and 4) unavailability of a pediatric neurologist. (**Figure 1**)

DISCUSSION

Boys with DMD typically present at two to five years old with abnormal gait, often as toe walking and in some as delayed gait onset.[9,10] In our study, the mean age when the caregivers first note of symptoms is at 4 (SD$\pm$ 2.3) years old. This finding is consistent with our cohort where the most commonly noted initial presentation is abnormal gait. The other observed signs and symptoms include Gower sign, gross motor delay, calf pseudohypertrophy, inability to jump, decreased endurance, decreased head control when pulled to sit, difficulty

climbing stairs, flat feet, frequent falling or clumsiness, hypotonia, inability to keep up with peers, loss of motor skills, muscle pain or cramping, toe walking and difficulty running or climbing. Non-motor symptoms have also been described and include behavioral issues, cognitive delay, failure to thrive or poor weight gain, learning and attentional issues and speech delay.[3,11] These were also noted in our patients (**Table 3**).

The natural history of DMD has been described to be composed of 5 stages. Upon note of suggestive symptoms, a prompt referral to a child neurologist or a neuromuscular specialist should be done. A markedly elevated serum CK increases the index of suspicion. Pre-genetic counseling[4] before confirmatory testing with multiplex ligation-dependent probe amplification (MLPA)[12] or comparative genomic hybridization assay[13] should be done. In our study, the mean age of confirmation of diagnosis is 7.4 ($\pm$ 2.7) years old. This is an approximately three-year delay from the time the symptoms were first noted.

At the time they get the proper medical attention for DMD, the physicians and the caregivers are now faced with multiple complications from the chronic illness. Significant complications include respiratory insufficiency, scoliosis, and secondary cardiomyopathy.

At present, the overall mean age for respiratory death is 17.7 years for those without ventilatory support but may extend to 27.9 years for patients with ventilatory support. Case reports of survival up to the third decade of life are possible but uncommon.[14]

About 85-90% develops dilated cardiomyopathy at 18 years of age. Low levels of pulmonary function and skeletal-muscle impairment make them poor candidates for heart transplant.[15,16] This progression highlights the cost of illness leading to a 16-fold increase of expenses from direct and indirect costs.[5] [17]

The mainstay of treatment includes physiotherapy and glucocorticoid administration yielding the best outcome if started at an early age. Prednisone is often prescribed however some experts routinely use deflazacort for DMD owing for its more favorable side effect profile compared with prednisone particularly weight gain.[17] Benefits for both types of steroid include prolonged ambulation, preserved cardiac, respiratory and upper limb function and delay in the need of scoliosis surgery. Mutation-specific drugs (e.g. ataluren,[18] eteplirsen,[19] and viltolarsen[20]) have been studied and showed proven efficacy.

Care for patients with DMD will require a multidisciplinary approach involving specialists from the fields of pediatric neurology, neuromuscular, clinical genetics, cardiology, pulmonology, rehabilitation medicine, orthopedics, gastroenterology, nutrition and endocrinology.[4,6,7]

To achieve these benefits, early diagnosis is essential. In a developing country like the Philippines, certain challenges are present even before these patients arrive to the proper medical specialists. From the end of the caregiver, multiple aspects affect early and prompt diagnosis.

The first action taken by the caregivers in our study upon note of the symptoms was not an immediate medical attention. Yet the majority opted for observation and traditional medicine. Only 2 out of 10 of the caregivers in the study immediately sought medical help upon notice of the symptoms. Despite regulation for traditional medicines in the Philippines, there are no approved therapeutic options available for this disease.[21] Yet, in our study, a good number of caregivers would still choose these as the first step in addressing a noted problem with

their children. We also noted that around 40% of caregivers chose to initially observe their patient before taking other actions. Four of 10 caregivers in the cohort eventually sought medical help with progression of the symptoms. However, it is noteworthy that a few are already familiar with norms of child development and waited for their children to improve, some came across multiple media platforms (e.g. internet, television, prints), have a positive family history, had an incidental finding of markedly elevated serum creatine kinase and others as suggested by relatives.

Eighty percent consulted at least a year after the first symptom. Reasons like hesitancy, financial constraints, difficulty to schedule a physician consultation, distance of the nearby health facility and lack of knowledge were among their responses.

Upon initial consultation, only 10% get to see a pediatric neurologist and about 50% of the DMD patients were evaluated by a neurologist on their second consultation. Most of the caregivers originally sought consultation with a general pediatrician before being referred to a pediatric neurologist. Other specialists that were sought in the cohort include an orthopedic surgeon, a physiatrist and a general physician. The subsequent increase of pediatric neurologists seeing the patient reflects knowledge of the first physician that the patient has a neurologic problem.

Despite having a correct clinical impression or a differential diagnosis that could have arrived at the proper diagnosis, several factors would influence this.

Upon the first medical consultation, a diagnostic process is initiated by the physician in half of the cohorts. However, around 25% of the physicians started the diagnostic process only to alleviate the concern of the caregiver. Other facilitating factors include a positive family history, parents demanded the action, and the physiatrists were the ones who recognized the symptoms. In a study by Daak-Hirsch et. al. the effect of a positive family history for DMD has delayed the process of seeking help due to parent's readiness to face the diagnosis while others perceived it as stigma.[9]

On the other hand, around 25% of the first physicians who saw the patient were reported to initially have an alternative diagnosis. Only 5% of the first physician who saw the patient took no action. However, several other reasons that impeded facilitating the medical service include parents not following through the referral done, unavailable pediatric neurologist, cultural belief that there is no problem, financial constraints and a pregnant mother.

CONCLUSION & RECOMMENDATIONS

Our study provides insights on the healthcare-seeking behavior of parents in a developing country which affects timely diagnosis of DMD. In the problem-recognition stage, a significant delay is contributed by parents opting to observe the patient. This is influenced by cultural beliefs that traditional medicine is an option in treating medical illnesses. Disease worsening is the major reason in seeking medical consultation. This gap in healthcare should stimulate exploring other possibilities for early diagnosis of DMD such as newborn screening and

possibly genetic testing for suspected female carriers. Furthermore, improving the knowledge on this chronic neuromuscular condition would translate better identification at the primary care level and prompt referral to a specialist.

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