



Original Research Article

Health Inequalities in Connection with Socioeconomic Position of Duchenne / Becker Muscular Dystrophy Patients, Gujarat, India

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ABSTRACT

Background: The exploration of socioeconomic position concerning inequalities in health of Duchenne/Becker muscular dystrophy (D/BMD) families has hardly been assessed till today. We have investigated burden of D/BMD across Gujarat by approaching molecular as well biochemical indices. Moreover, we too correlate the same with socioeconomic position.

Subjects and methods: In this pilot study, a total 101 Gujarati D/BMD patients were reviewed on basis of their clinical characteristics, confirmed by genetic analysis. Socioeconomic position was determined based on income, localities of the residence, characteristics of housing, education, and family history.

Results: The study data revealed that development of most dreadful stances are associated with SEP of families. It was also observed that patients with low SEP have more severe condition compare to upper class than would be expected by chance alone.

Conclusions: Study data support the hypothesis that better conditions enable higher SEP groups to maintain patients regarding their special requirements. In-depth research is necessary to elucidate various factors that will be responsible regarding health, pathology, and management of proband.

Key words: Duchenne/Becker muscular dystrophy, socioeconomic position (SEP), dystrophin, exon.

INTRODUCTION

Muscular dystrophy (MD) is a group of inherited genetic disorders characterized through weakness, and progressive deterioration of muscle. In this genomic epoch more than 30 different types, and subtypes of MDs have been classified, each of which is caused by mutation/s in a special gene. [1] To date, the mechanisms responsible for divergence in pathophysiology have not been identified in detail for the same. MD is ubiquitous

worldwide disease that suffers no exception. Majority of MDs are caused by bouleversement of different components of the dystrophin glycoprotein complex (DGC) that is allied with integrity of skeletal muscle cells. [2,3] Dystrophin protein is one of the largest component of the DGC which is absent or drastic reduced in a DMD (Duchenne Muscular Dystrophy; OMIM# 310200), while decreased levels in allelic form of BMD (Becker Muscular Dystrophy; OMIM# 300376). DMD, and BMD are the

most common forms of muscular dystrophy in humans, and together termed as dystrophinopathies. [4] DMD alone accounts for approximately 80% of all the myopathies, with an incidence of around 1 in 3500 males. [5] In DMD probands, carry gene mutation/s, which causes premature translation termination (OUT-frame mutation); affected boys are usually wheelchair-bound by the age of 13, and die early in their third decade of life. On the other hand milder allelic BMD is associated with a later age of onset, and in a slower clinical progression. [6,7] The incidence of BMD is around 1 in 18,500 live births of males. [8]

Socioeconomic position (SEP) of an individual is an environmental matter which has been identified as a potent factor for determinant of human health. [9] The primary point of contention lies in whether, SEP is useful to consider as vulnerable factor for various genetic conditions opening out, and management their burden in various populations. Though, this observation can be taken as inconsistent with the evidence, and have been an irritant to geneticists who deem in population variation. [10]

So far no population based study has been done in Gujarat to comprehend the gist of this condition. By considering above ground, the aim of the study is to examine the burden of Duchenne/Becker muscular dystrophy (D/BMD) in Gujarat. In addition, we have attempted to investigate SEP on the subject matter of inequalities of health in D/BMD patients first time in Gujarat, India.

MATERIALS AND METHODS

Sample Collection

The study was approved by the Institutional ethics board. All boys with D/BMD were reviewed who has consulted at Gujarat Genetic Diagnostic Center (GenDiCe), and Indian Muscular Dystrophy Society (IMDS) Ahmedabad, Gujarat, India

between the periods of 2011 - 13 for their clinicopathological condition. Informed consent was given in writing before sample collection by the subject or the parents of individuals <18years.

Genetic analysis

Patients genomic DNA was extracted from peripheral blood lymphocytes by phenol chloroform extraction protocol. [11] PCR for deletion detection were performed on ABI Veriti® thermal cycler by 3 sets of Multiplex PCR (M-PCR) reaction sets, allow screening for exon/s deletions.

Measurements of SEP

Information on patients familial SEP was obtained directly from parents. Residence in particular localities of the city, characteristics of housing, parents' education, family income, and family history were also taken into account. The income according to the profession was also utilized to classify the subjects.

RESULTS

In study total of 101 patients (89 cases of DMD, and 12 with BMD phenotype) presented with complaining of repeated fall particularly with lower limbs muscle weakness, and calf hypertrophy were included. Of 101 cases clinically suspected D/BMD boys, the diagnoses of D/BMD were confirmed by multiplex PCR in 71 (70.30%) patients. Out of 71 probands, 66 (92.96%) DMD, and 5 (7.04%) BMD were identified. In study, 16 cases had a positive family history with 20 (19.80%) of innate cases.

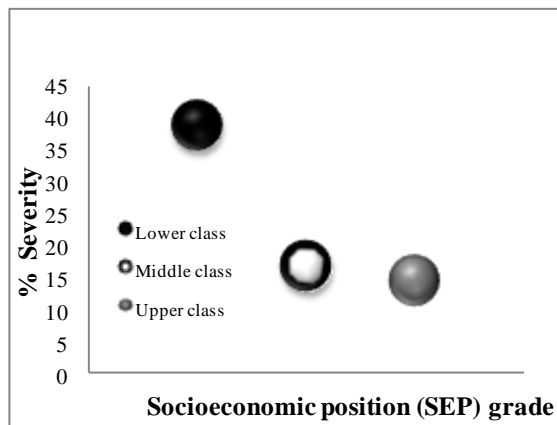
Severity, state of being graveness was considered using various stances like onset of awkward movement, wheelchair bound, and degree of scoliosis [Table 1]. The highest percent of severity was observed in lower class (38.71%) followed by (16.67%) middleclass families [Graph 1]. We found that brutality of disorders were more likely to be found among families with

low SEP than would be expected by chance alone. The well documented relationship between SEP, and D/BMD severity reveal in

our study. In familial cases severity was observed higher than sporadic cases.

TABLE 1: Clinical characteristics of D/BMD patients, Gujarat, India

Characteristics	Patients (n=101)	
	DMD (n=89)	BMD (n=12)
Mean age of onset	04.2 ± 0.2	11.0 ± 2.2
Mean age of presentation	11.1 ± 0.4	18.3 ± 2.9
Consanguinity	1	0
Familial cases	16	0
Scoliosis	23	0
State of ambulation	Supported	49
	Wheelchair bound	40
Religion	Hindu	73
	Muslim	16
Socioeconomic position (SEP)	Lower class	26
	Middle class	37
	Upper class	26
Education	Less than high school	45
	High school	18
	Bachelor's degree	18
	Master's degree	8



GRAPH 1: SEP and percent severity in D/BMD.

DISCUSSION

The populace of human is not identical regarding risk of disease, and its severity. However, in this DNA era with help of large information on genetic constituents one can cogitate that every human being has a uniquely health or risk of disease, based on inherited genetic material in addition to environmental exposure. [12] This includes a variety of factors one of them is socioeconomic position (SEP) also. SEP is a critical social determinant for human health, affecting outcomes through

various mechanisms. [13] The present study addresses inequalities in health of D/BMD patients in connection with SEP. In this study, individual SEP was considered on base of their income, and assets information given by the parents.

In our study of 101 Gujarati patients from Gujarat, India the deletion rate was 70.30% (71/101) in agreement to the frequency reported earlier in western region of India. [14,15] Our observation also concluded that exon 50 (53.52%), and del 45-52 (9.86%) at the central deletion hot spot region of the dystrophin gene is more deletion prone in Gujarat population. Hence, the present study suggested that this part of the DMD or dystrophin gene is more deletion prone in Gujarat population. [15]

The recognition of various stances like awkward movement, wheelchair bound, and degree of scoliosis are very important aspects of medical, and rehabilitative care of D/BMD probands [Fig 1]. In D/BMD patients, the onset, and the evolution of the abnormal postures are related to the development of condition, and eloquent

indices for severity of disease. Study data revealed that development of these stances are associated with SEP of families [Graph 1]. It might be due to lack of special requirements for the disease which not been

competent to the families with low SEP. Also other factors like diet, medication, development to stress, depression, and less education may partly explain the variation in health of D/BMD; bred by SEP.



FIG 1: The photo graphs portray various degree of scoliosis in the studied DMD probands.

In the study parameters, family history is one of the most important indices. Out of 101 cases, 16 cases had a positive family history with 20 (19.80%) of innate cases. A positive family history, in particular a maternal history is very useful and inexpensive tool to identify individual at high risk who may require specific consideration. Study showed that the SEP was related to having a positive family history of D/BMD which itself is a risk factor in developing the disease, and its severity. Moreover, education is also a fundamental factor in development of SEP. Education is theoretically, and empirically grants better human health amid superior SEP. Study data revealed severity of condition is lesser compare to less educate for the reason that awareness or understanding regarding the condition. There are many questions that still remain to

be answered related to inequalities in health of D/BMD patients apart from SEP. Other factor including genetic deletion (e.g. in or out-frame mutation) make up may be significant aspects of health for individuals with D/BMD.

CONCLUSION

In sum, this research is thus a preliminary contribution for these kinds of disorders first time in Gujarat population, India. There is an urgent need to have further investigation into the various aspects of D/BMD condition in order to provide scientifically valid answer to many unsolved questions to certain extent.

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