Duchenne muscular dystrophy: a case series report from a tertiary referral hospital of Midwest Brazil

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INTRODUCTION

Duchenne muscular dystrophy (DMD; OMIM#310200) is the most common hereditary neuromuscular disease. It is caused by mutation of the dystrophin gene (*DMD*) on chromosome Xp21, presenting X-linked recessive inheritance. The prevalence is estimated at 4 – 5 cases per 100,000 males. In females, it is even rarer: <1 case per million.

CASE REPORT

Seven patients, all male, were referred to the Genetics Outpatient Clinic of Hospital Universitário Júlio Müller (HUJM, Cuiabá, Brazil) for evaluation due to symptom presentation suggestive of DMD or positive familial history. All patients were born and reside in Mato Grosso (MT) state; only one resides in the metropolitan area of Cuiabá. Current age ranges from 3 years, 3 months old (mo) up to 19 years old (yo). The mean age of onset was 3.5 yo, ranging from 20 mo up to 6 years, 10 mo. Four patients, 2 of whom are brothers, present a positive familial history of DMD. All cases were confirmed by molecular testing: nonsense point mutations were the most common (4 cases; 2 of whom are brothers), followed by large deletions spanning several exons (2 cases), and single nucleotide splice donor variant (1 case). Five patients are currently on glucocorticoid treatment (3 on deflazacort and 2 on prednisolone). Four patients are taking D vitamin supplements. Only one patient, carrying a nonsense mutation, was treated with ataluren for 2 years. Despite positive results regarding muscle function, medication had to be discontinued due to side effects.

Table 1. Clinical findings

	n
Toe Walking	4
Positive Gowers sign	3
Hypertrophy of calves	2
Frequent falling	2
Attention-deficit/hyperactivity disorder	2
Scoliosis	1
Generalized muscle hypertrophy	1
Bone deformity	1
Loss of ambulation	1
Dilated cardiomyopathy	1

n = absolute number of cases.

DISCUSSION

Even though all DMD cases reported here are confirmed by both clinical symptoms and molecular testing, it is interesting to note that there was no single phenotypic feature that was common to all patients. This observation reinforces the importance of a robust, specialized clinical assessment and molecular testing as crucial tools for diagnosis.

CONCLUSION

DMD is a rare genetic disease with well-defined cause and symptomatology. However, access to specialists such as medical geneticists, molecular testing, and treatment is still limited in the Brazilian Midwest and an obstacle to diagnosis and building a clearer profile of those residing in the MT state. Such epidemiological profile is essential to the public health system, especially when considering symptom severity, the need for constant multidisciplinary care, and the emergence of high-cost drugs such as ataluren.

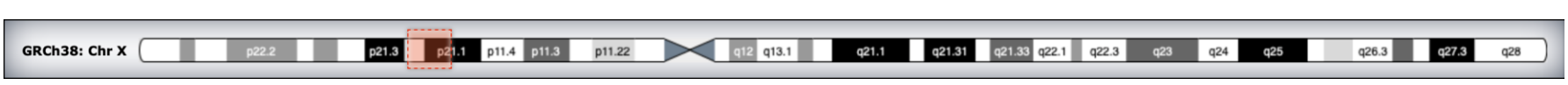

Figure 1: Schematic representation of chromosome X showing *DMD* gene location (in red) according to DECIPHER.