

The Value of Genetic Counseling in Duchenne Muscular Dystrophy: An Example of a Personal Case Series

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Abstract

Duchenne muscular dystrophy (DMD) is a hereditary, progressive muscular disorder inherited in an X-linked recessive pattern (Xp21). It typically manifests in childhood and follows a severe, rapid progression. Only males are affected, while females are usually carriers. Given the genetic nature of DMD, genetic counseling is an essential service for individuals affected by or at risk of carrying the disease. This service provides not only crucial medical information but also psychosocial support and ongoing management for both patients and their families. Since the discovery of the dystrophin gene in 1987, advancements in molecular genetics have made it possible to precisely identify the genes responsible for many neuromuscular diseases. These developments have revolutionized diagnosis, prognosis, and most importantly, genetic counseling, offering significant benefits for both patients and their families. To highlight the significance of these advancements, this case report focuses on a 10-year-old boy (Y) diagnosed with DMD. It emphasizes the familial nature of the disease, with Y's two brothers, three cousins, and two maternal uncles also affected, underscoring the inherited pattern of DMD. This reinforces the critical need for early intervention, particularly in regions with high consanguinity, such as North Africa and the Middle East, where genetic counseling and prenatal diagnosis are even more essential. Additionally, the report explores the clinical presentation, diagnostic findings, and promising emerging treatments, including RNA-based therapies, which may play a key role in the future management of DMD. In light of the above, this study underscores the importance of prenatal diagnosis and genetic counseling, particularly in regions like Morocco, where consanguinity rates are notably high. By focusing

on preconception care and early genetic intervention, families can be better informed, leading to more effective disease management and support.

Keywords

Duchenne Muscular Dystrophy, Prenatal Diagnosis, Genetic Counselling

1. Introduction

Duchenne muscular dystrophy (DMD) is the most common form of progressive muscular dystrophy in children, affecting approximately 250,000 individuals worldwide. This hereditary condition is caused by a recessive gene on the X chromosome (Xp21) and typically manifests in early childhood, with rapid progression that often leads to death by the third decade of life.

The prevalence of consanguinity, which is a significant factor in the inheritance of genetic disorders like DMD, varies considerably across regions. Studies on consanguinity in Arab nations have shown alarmingly high rates, ranging from 20% to 50% in the Middle East and North Africa [1]. This widespread consanguinity is strongly linked to an increased incidence of not only DMD but also other genetic disorders, including deaf-mutism and cardiovascular diseases.

Focusing on Morocco, the rate of inbreeding is reported to be between 19% and 23% (2018), with southern regions experiencing a particularly high rate of 28.46%. Notably, first-cousin marriages constitute 16.15% of all unions. Although northern Morocco has lower inbreeding rates, first-cousin marriages still represent 36.18% of marriages [2]. Despite these high rates of consanguinity, the lack of comprehensive awareness programs underscores the need for further research into consanguinity-related diseases. Such research is pivotal for establishing effective premarital screening and genetic counseling strategies aimed at reducing the incidence of genetic disorders.

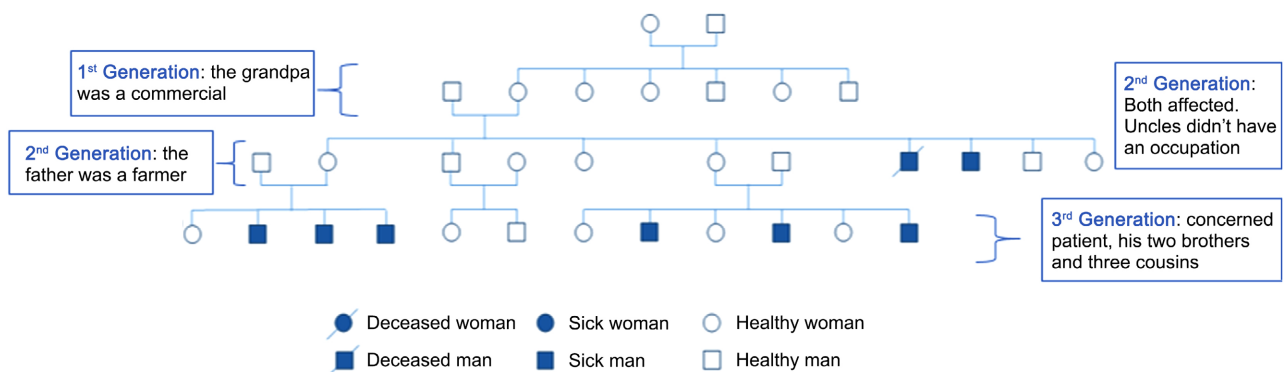


Figure 1. Family tree of concerned patient.

In light of these findings, this study presents a case of familial DMD spanning two generations, which emphasizes the critical importance of prenatal counseling

in regions with high rates of consanguinity. The affected individuals in this case include our patient (Y.B.), his two brothers (A.B. and M.B.), three cousins (C.B., F.B., and S.B.), and two maternal uncles (B.B. and D.B.). By exploring this case, the article highlights how genetic counseling and antenatal diagnosis can play a crucial role in reducing the incidence of DMD, especially in North Africa and the Middle East, where consanguinity remains prevalent (**Figure 1**).

2. Case Presentation

Y's family lives in Benslimane, a rural area near Casablanca. He is a 10-year-old boy, the third of six siblings (three girls and three boys) born to non-consanguineous parents. His father works as a farmer, while his two maternal uncles, both diagnosed with Duchenne muscular dystrophy (DMD) at an early age, have been unable to work. One uncle passed away at 22 due to respiratory failure, and the other, now 20 years old, is bedridden.

Y's family history reveals a significant prevalence of DMD. Two of his brothers are affected: one, aged 14, is bedridden, and the other, aged 8, also has the condition. Additionally, three maternal cousins (aged 3, 7, and 10) from non-consanguineous marriages have been diagnosed with DMD.

Since the age of 5, Youssef has displayed symptoms consistent with Duchenne muscular dystrophy (DMD), including muscular fatigue, difficulty walking, trouble climbing stairs, and frequent falls. Upon clinical examination, a waddling gait, positive Gowers' sign, positive Tabouret sign, scapular winging, lumbar hyperlordosis, and pseudo-hypertrophy of the calves were observed, further supporting the suspicion of a muscular disorder.

Biological assessments revealed significantly elevated levels of CPK (5141 IU/L), LDH (1410 IU/L), and aldolase (90 IU/L). Additionally, liver function tests indicated hepatic cytolysis, with AST and ALT levels of 231/289 IU/L, respectively. An electromyogram (EMG) confirmed a myogenic syndrome, which prompted a muscle biopsy, ultimately diagnosing Duchenne muscular dystrophy (DMD). A cardiac ultrasound revealed early signs of dilated cardiomyopathy, with a left ventricular ejection fraction (LVEF) of 58%. Blood tests showed low vitamin D levels (13.2 µg/ml) and calcium at 98 mg/L.

Respiratory function tests (PFTs) further revealed a severe mixed syndrome, including a restrictive component at 54% of normal capacity and significant irreversible obstruction, as evidenced by a forced expiratory volume (FEV1) of 58% of the theoretical value. Genetic testing confirmed the diagnosis of Duchenne muscular dystrophy, solidifying the clinical findings.

To assess motor function, muscular testing was conducted to obtain analytical results. The primary muscles affected were the Gluteus Maximus (GMax), Gluteus Medius (GMed), Quadriceps, Anterior Hamstrings (A.H.), Rotator Cuff Muscles (R.C.), Abdominals, and Quadratus Lumborum (Q.L.). The results, presented in **Table 1**, provided a measure of disease progression over time according to different scales.

Table 1. Motor function assessment scoring evolution.

DOC/Scoring	Muscular Testing (Mainly Affected Muscles)	MFM 32
2018	GMax = 2–	D1 = 33.33%
	GMed = 3–	D2 = 80.55%
	Quads = 3+	D3 = 76.19%
	A.H. = 3–	
	R.C. = 3+	Total = 60.41%
	Abs = 3–	
	Q.L. = 2+	
2019	GMax = 1+	D1 = 28.2%
	GMed = 2+	D2 = 75%
	Quads = 3–	D3 = 66.6%
	A.H. = 2+	
	R.C. = 3–	Total = 54.16%
	Abs = 2–	
	Q.L. = 1+	
2023	GMax = 0	D1 = 0%
	GMed = 2–	D2 = 83.3%
	Quads = 1+	D3 = 57.14%
	A.H. = 0	
	R.C. = 2–	Total = 43.75%
	Abs = 0	
	Q.L. = 0	

DOC: Date of consultation; MFM: Motor function measurement.

The Motor Function Measurement (MFM) scale revealed significant challenges with unipodal balance, stair climbing and descending, and walking on uneven terrains. Despite these difficulties, upper limb function remained intact, although fine interdigital grips (e.g. writing with a pen) and cylindrical grips were impaired. Pain was reported in the knee and lumbar regions.

In 2018, Y began corticotherapy following a comprehensive muscular and functional assessment. The initial prescription included 20 mg of Cortancyl (three-quarters of a tablet daily for 10 days, followed by a 10-day pause). Additionally, Y was treated with Osteocar (1 cc/day) and D-Cure (25.000 mg every three months for six months). The treatment showed positive results, with muscular assessments conducted every six months until 2020, when Y was lost to follow-up.

After two years, Y returned for consultation and was found to have become

wheelchair-dependent. During this time, he experienced significant personal losses, including the deaths of his bedridden 14-year-old brother and one of his uncles.

3. Discussion

Duchenne muscular dystrophy (DMD) is the most common dystrophinopathy in children, with an X-linked recessive inheritance primarily affecting boys. Globally, it occurs in about 0.5 cases per 10,000 male births. The disease is caused by mutations in the DMD gene, leading to the absence of dystrophin, a key protein in the membrane cytoskeleton. This deficiency results in progressive muscle degeneration, also affecting the heart and brain [3].

3.1. Symptoms

The primary symptoms of Duchenne muscular dystrophy (DMD) include difficulty running, jumping, climbing stairs, frequent falls, and trouble rising from the ground, often referred to as Gower's sign [4]. Initially, the condition affects the proximal muscles, with later involvement of axial and distal muscles [5]. This progression leads to a gradual loss of muscle strength, particularly beginning at the waist, and is typically accompanied by a waddling gait, hyperlordosis, and calf pseudo-hypertrophy [6]. All of these symptoms were evident in our patient.

As the disease progresses, respiratory complications frequently arise, especially during sleep. Nocturnal hypoventilation can cause periodic apnea, leading to morning headaches, nausea, fatigue, loss of appetite, and cognitive issues [7] [8]. Additionally, orthopedic complications, such as scoliosis due to paraspinal muscle weakness, can exacerbate respiratory difficulties and affect up to 90% of patients. In our patient, respiratory function tests (RFTs) revealed a severe mixed syndrome, with a restrictive component at 54% of normal capacity and significant irreversible obstruction, indicated by a forced expiratory volume (FEV1) of 58% of the predicted value [9].

Dilated cardiomyopathy is another common complication, typically leading to a reduced left ventricular ejection fraction (LVEF) and possibly progressing to heart failure. Although cardiac rhythm disturbances, often of a supraventricular nature, are frequent in DMD patients, they rarely result in early mortality [10]. In our patient, asymptomatic dilated cardiomyopathy was detected at the age of 10, though his LVEF remained normal. Respiratory and cardiac complications are the primary causes of death in DMD patients, typically occurring before the age of 25 [11]. This tragic outcome was mirrored in our patient's maternal uncle, who passed away from respiratory failure at the age of 22.

3.2. Treatments

The therapeutic management of Duchenne muscular dystrophy (DMD) involves a multidisciplinary approach, with a focus on symptomatic treatments. Corticosteroids remain a cornerstone of traditional therapy due to their long-term benefits on motor, cardiac, and respiratory functions. Studies have demonstrated that

corticosteroid therapy helps maintain upper limb strength, slows the progression of scoliosis, and delays the onset of cardiac and pulmonary complications when administered consistently. These effects are primarily achieved through the reduction of inflammation, particularly by inhibiting the NF- κ B pathway and minimizing muscle necrosis [12]-[15].

In addition to corticosteroids and supportive treatments, early interventions addressing respiratory, cardiac, orthopedic, psychological, and nutritional challenges, alongside physical therapy, are essential for enhancing patients' quality of life. Such measures have shown promising results in slowing disease progression [16]. For our patient, rehabilitation began immediately following the diagnosis.

Effective rehabilitation management necessitates a comprehensive understanding of DMD pathology, biomechanics, disease progression, and its natural history. Treatment plans should align with the individual's goals and lifestyle to optimize their quality of life. Interventions should address all areas outlined by the International Classification of Functioning, Disability, and Health (ICF) from the time of diagnosis. This includes proactive measures to prevent contractures, deformities, loss of function, compromised skin integrity, pain, and cardiorespiratory decline [17].

3.3. Genetic Counselling

The importance of genetic counseling and preconception diagnosis in managing the patient's condition and its impact on the family cannot be overstated. For example, a study in Australia highlighted innovative strategies to reduce the incidence of Duchenne muscular dystrophy (DMD), including preconception or early pregnancy carrier screening, prenatal exome sequencing, and newborn screening. This approach is particularly relevant to the case of our patient's family, emphasizing the critical role of preconception planning in preventing the transmission of the condition to the third generation [12] [18].

Genetic counseling plays a vital role in managing hereditary neuromuscular disorders, providing tailored guidance based on the mode of inheritance and associated phenotypes. In France, such counseling is strongly recommended for relatives of affected individuals, as some may be at risk of developing late-onset forms of the disease. Establishing an accurate neuromuscular diagnosis in a child not only aids in treatment planning but also encourages parents and relatives to explore preventive options such as prenatal diagnosis or pre-symptomatic testing.

In Morocco, where consanguinity is a significant risk factor, we have begun emphasizing the importance of genetic counseling, prenatal diagnosis, and preconception planning to reduce the prevalence of hereditary diseases. By integrating these preventive measures into familial health care, we aim to replicate successful strategies employed in other countries, such as France, while addressing the unique challenges faced in our context [19].

On the other hand, findings from Canada revealed that genetic counseling alone may not significantly reduce the recurrence of Duchenne muscular dystrophy

(DMD) within families, given the disease's prevalence in the broader population [20]. This underscores the need for additional preventive strategies. For instance, a study conducted in India highlighted the importance of prenatal testing in pregnancies at risk for DMD to mitigate the disease's impact on families. The study further noted that obstetric outcomes for DMD carrier fetuses were comparable to those of DMD-negative female fetuses, supporting the feasibility and safety of such interventions [21].

In China, a 15-year study involving 931 pregnancies highlighted the importance of prenatal diagnosis and genetic counseling. Among these pregnancies, 575 were identified as carriers. The study demonstrated that combining MLPA and Sanger sequencing with STR linkage analyses provides accurate and rapid prenatal diagnoses [17]. Such studies emphasize the crucial role of prenatal diagnosis in mitigating the burden of hereditary diseases and highlight the necessity of adopting similar approaches in regions with high rates of consanguinity, such as North Africa and the Middle East.

In the Middle East and North Africa (MENA) region, muscular dystrophies (MDs) are rare, with a global prevalence of 19.8 to 25.1 per 100,000, although research on the causes and mechanisms of MDs in this population is limited [22]. Consanguinity rates in the Mediterranean, particularly in parts of the Maghreb, are high, reaching about 60%, which increases the risk of recessive hereditary diseases. In northern Morocco, 24.37% of couples are consanguineous, while in the Souss region, the rate is 28.46%, higher than the national average of 23.4%. These high rates of consanguinity, combined with underdiagnoses, limit access to critical diagnostic resources, worsening the prognosis for hereditary diseases like MDs. A comprehensive understanding of these disorders, along with genetic diagnosis, is essential for improving clinical management and patient outcomes. Genetic counseling and screening are crucial for the early detection and management of genetic disorders, especially in consanguineous families. The relationship between consanguinity and socio-economic factors varies by country, underscoring the importance of public awareness of genetic risks associated with inbreeding [1] [2] [23].

4. Conclusions

Genetic counseling should take place in a specialized consultation setting, ensuring that information and support are carefully tailored to the unique circumstances of each case. Early detection and management of cardiac and respiratory complications are crucial for improving survival outcomes in Duchenne muscular dystrophy (DMD). These efforts require a multidisciplinary approach, with interventions designed to address the patient's specific profile and stage of disease progression.

In addition to individual care, family investigation is an essential aspect of managing DMD. This process complements genetic counseling by providing detailed information about the disease, its inheritance patterns, and its clinical variability.

It also emphasizes the importance of preventive measures, such as genetic counseling, for at-risk relatives. By identifying affected individuals and carriers within a family, clinicians can guide relatives toward options like prenatal diagnosis. Family history, clinical evaluation, and creatine phosphokinase (CPK) level assessments are key tools in this investigation. Importantly, two-thirds of DMD cases result from maternal transmission of the mutated gene, although spontaneous mutations during embryogenesis account for approximately one-third [19].

The importance of multidisciplinary care in DMD management cannot be overstated. This approach has been widely adopted and guided by international recommendations, which have significantly improved survival rates and quality of life for patients. Regular monitoring of orthopedic, cardiac, and respiratory health forms the cornerstone of this care [12] [16] [24].

However, despite these advancements, the potential of prenatal diagnosis to prevent new cases remains underutilized, particularly in regions with limited awareness of genetic disorders. This is especially true in rural areas, where high consanguinity rates exacerbate the prevalence of hereditary conditions. Addressing these challenges calls for proactive measures, such as preconception counseling and structured follow-up systems for at-risk families. Such initiatives could reduce the incidence of DMD and improve outcomes for future generations.

In parallel with these preventive efforts, research into therapeutic interventions continues to advance. While a deeper understanding of DMD pathology and the development of new technologies have provided hope, significant obstacles remain. Current treatments are largely palliative, and an effective cure remains elusive.

One promising avenue of research involves RNA-based therapies, particularly the use of small antisense oligonucleotide sequences (AONs). These molecules can target specific messenger RNAs, modulating their maturation to facilitate dystrophin production. Modified AONs, such as 2'-O-methyl phosphorothioates (2'OMe) and morpholinos (PMO), have shown potential in promoting dystrophin synthesis. However, their therapeutic benefit remains limited by challenges such as systemic toxicity, poor distribution throughout the musculature, and an inability to act on cardiac tissue or cross the blood-brain barrier [25].

Given these limitations, the development of therapies that can effectively target skeletal musculature, the heart, and the central nervous system remains an urgent priority. Overcoming these challenges will be critical to transforming the promising results of biotechnological research into meaningful clinical outcomes for DMD patients [26] [27].

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Data Availability

The data supporting the conclusions of this study are available from the corresponding author upon request.

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Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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