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Phenotypic and Genetic Characterization of a Patient with Isolated Exon 50 Deletion in the *DMD* Gene: A Case Report from Kinshasa

Caractérisation phénotypique et génétique d'un patient porteur d'une délétion isolée de l'exon 50 du gène DMD : rapport d'un cas à Kinshasa

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Résumé

La dystrophie musculaire de Duchenne (DMD) est une dystrophinopathie sévère liée à l'X, généralement causée par des mutations hors cadre de lecture dans le gène DMD. Une délétion isolée de l'exon 50 est généralement prédite comme perturbant le cadre de lecture et entraînant un phénotype classique de DMD. Nous décrivons un homme congolais de 22 ans présentant une délétion isolée de l'exon 50 identifiée par Multiplex Ligation-dependent Probe Amplification (MLPA). Le patient a présenté des troubles de la marche à l'âge de 9 ans, a reçu un diagnostic génétique confirmé à l'adolescence et a été traité par déflazacort. À l'âge de 20 ans, il était dépendant du fauteuil roulant la plupart du temps, mais pouvait encore marcher sur de courtes distances avec assistance. L'examen clinique a révélé une hypotonie des membres supérieurs, une pseudohypertrophie des mollets, une scoliose et des rétractions tendineuses. L'exploration fonctionnelle respiratoire a montré un trouble restrictif modéré (FVC %pred <64) ainsi qu'une obstruction des petites voies aériennes. L'échocardiographie a ultérieurement mis en évidence une altération de la fonction systolique ventriculaire gauche. Les examens biologiques ont montré une élévation marquée de la créatine kinase, une élévation des transaminases, une hypocalcémie et une carence en vitamine D. Ce phénotype apparaît

Summary

Duchenne muscular dystrophy (DMD) is a severe X-linked dystrophinopathy usually caused by out-of-frame mutations in the *DMD* gene. Isolated exon 50 deletion is usually predicted to disrupt the reading frame and cause a classical DMD phenotype. We describe a 22-year-old Congolese male with an isolated exon 50 deletion identified by Multiplex Ligation-dependent Probe Amplification (MLPA). The patient presented with gait disturbances at age 9, received a confirmed genetic diagnosis during adolescence, and was treated with deflazacort. At the age of 20, he was wheelchair-dependent most of the time but was still able to walk short distances with support. Clinical examination revealed hypotonia of the upper limbs, pseudohypertrophy of the calves, scoliosis, and tendon contractures. Pulmonary function testing revealed moderate restrictive impairment (FVC %pred <64) and small airway obstruction. Echocardiography later demonstrated impaired left ventricular systolic function. Laboratory findings showed markedly elevated creatine kinase, elevated transaminases, low calcium levels, and vitamin D deficiency. This phenotype appears milder than classical DMD, suggesting a possible molecular rescue mechanism through endogenous exon skipping (RNA-level studies were not performed). These findings contribute to documenting genotype-phenotype variability within the distal DMD hotspot region and may illustrate the natural rescue observed in some patients with expected out-of-frame

plus modéré que la DMD classique, suggérant un possible mécanisme de sauvetage moléculaire par saut d'exon endogène (aucune étude au niveau de l'ARN n'a été réalisée). Ces observations contribuent à documenter la variabilité génotype-phénotype au sein du hotspot distal du gène DMD et pourraient illustrer le mécanisme de sauvetage naturel observé chez certains patients porteurs de délétions théoriquement hors cadre de lecture.

Mots-clés : Dystrophie musculaire de Duchenne, dystrophinopathie, corrélation génotype-phénotype, RD Congo

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Introduction

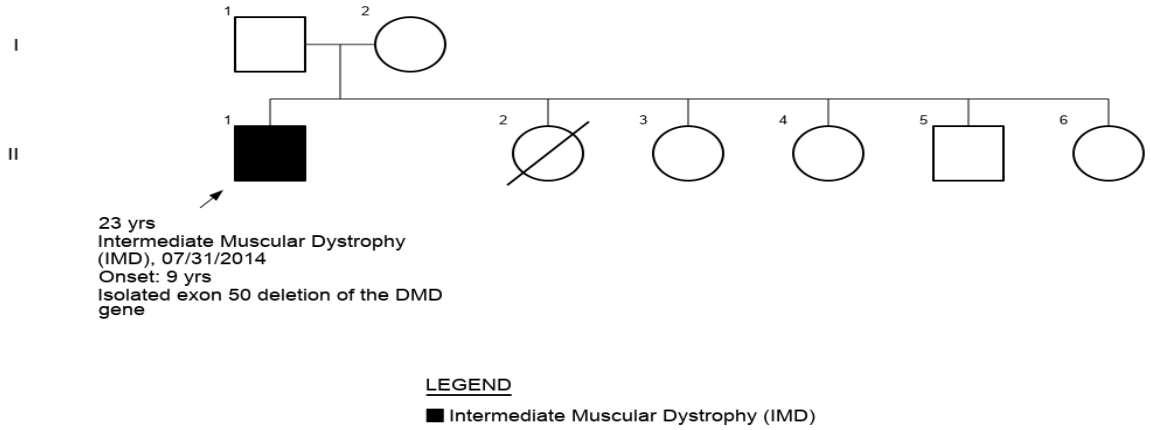
Duchenne muscular dystrophy (DMD) is the most common and severe childhood muscular dystrophy, caused by X-linked mutations in the DMD gene encoding dystrophin, a protein essential for muscle membrane stability (1). Out-of-frame mutations typically result in classical DMD, characterized by progressive muscle weakness, loss of ambulation during early adolescence, and cardiopulmonary complications, whereas in-frame mutations generally produce milder phenotypes such as Becker muscular dystrophy (1). Deletions within the exon 45–55 hotspot represent the most frequent mutational pattern. Although isolated exon 50 deletion is usually predicted to produce a severe DMD phenotype through reading-frame disruption, phenotypic variability has been reported (2–5). Data from African populations remain scarce, restricting the understanding of genotype–phenotype correlations in this setting (6).

This report presents a molecularly confirmed DMD patient from the Democratic Republic of the Congo (DRC) and illustrates clinical variability in a Central African patient. The patient was recruited as part of the "African Rare Disease Initiative (ARDI)" research project, which was approved by the Internal Review Board of the Kinshasa School of Public Health (ESP/CE/086/2022).

Clinical observation

A 22-year-old man was referred to our consultation for reassessment and follow-up of progressive muscle weakness previously diagnosed as Duchenne muscular dystrophy. He was born at term by normal vaginal delivery, showed normal adaptation to extrauterine life, and had growth parameters within the normal range at birth. He was the eldest of six children, including one who died 9 hours after birth due to asphyxia in the context of maternal malaria, while the others are reportedly healthy (Fig. 1). Both parents were unrelated, healthy, and young

at the time of conception. No similarly affected individual has been reported in the family up to three generations above.



Legends: □ = male, ○ = female, ■ = affected individual, ↗ = proband, ∅ = deceased, (I, II) = generation numbering, (1, 2, 3, 4, 5, 6) = Numbering or order of individual in a generation from left to right.

Figure 1. Pedigree of the family
The pedigree is consistent with an X-linked recessive inheritance pattern, but no other affected family members were reported. (Carrier status is not indicated, as genetic testing was not performed yet in all female relatives).

Psychomotor development was considered satisfactory, apart from irritability, depressive behavior, and suicidal thoughts at the age of 17. He was hospitalized three times before the age of one and underwent surgery at the age of 13 for pathological fractures of the leg bones. At the age of 9, he was found to have difficulty standing, an unstable gait, and frequent falls while walking. The possibility of Duchenne muscular dystrophy was considered by a neurologist at the age of 10, and a muscle enzyme assay revealed markedly elevated creatine kinase (CK) levels and elevated lactate dehydrogenase (LDH) (Table 1), supporting the suspicion of dystrophinopathy.

Table 1. Summary of Investigations and Clinical Assessments

Table 1. Summary of Investigations, Clinical Assessments and Interventions

Domain	Parameters / reference values	Results by age (years old)							Interpretation
		10	11	15	16	20	21	22	
Biochemical assays	Creatine Kinase (CK) (25-195) *U/L*	3,568						12,078	Markedly elevated, consistent with dystrophinopathy
	Lactate Deshydrogenase (LDH) *U/L*	1131							Elevated, consistent with dystrophinopathy
	AST (5‑34) *U/L*							91	Elevated, muscle-related
	ALT (0‑55) *U/L*							129	Elevated, muscle-related
	Calcium (8.5-10.1) *mg/dl*							8.2	Mild hypocalcemia
	25-OH Vitamin D (>20) *ng/ml*							18.76	Vitamin D deficiency
	Creatinine (0.70-1.30)						0.27		Low, reflecting reduced muscle mass

	mg/dl				
Bone assessment	Limb X-ray	Osteoporosis			Consistent with lack of calcium and Vitamin D
	DXA scan	Osteopenia			
Pulmonary function tests	FVC (% predicted)			<64%	Moderate restrictive ventilatory defect
	FEV1/FVC (% predicted)			>95%	
	FEF25‑75 or PEFR (% predicted)			<70%	Early small airway obstruction
Cardiac evaluation	ECG		Normal		No conduction abnormalities
	Echocardiography		Normal		No morphological abnormalities
	Echocardiography			Impaired LV systolic function	Early cardiomyopathy, no chamber dilation
Neuromuscular exam	Ambulation			Partial, with support	Preserved beyond 20 years
	Muscle strength			UL: 3/5; LL: 2/5	Progressive proximal weakness
	Other findings			Calf	Typical DMD

						pseudohypertrophy, scoliosis, contractures	features
Genetic analysis	DMD MLPA			Isolated exon 50 deletion			Predicted out-of-frame mutation
	DMD Multiplex PCR	Isolated exon 50 deletion					
Therapeutic interventions	Corticosteroids		Deflazacort (60 mg/day)		Deflazacort (30 mg/day)		Long-term treatment
	Bone health	Vitamin D, calcifediol					Secondary prevention
	Cardiac prevention					ACE inhibitor initiated	Cardiomyopathy prophylaxis
	Supportive care			Wheelchair, physiotherapy			Functional support

Legends: AST = Aspartate aminotransferase, ALT = Alanine aminotransferase, DXA scan = Dual-energy X-ray absorptiometry, FVC = Forced vital capacity, FEV1 = Forced expiratory volume in one second, FEF25-72 = Forced expiratory flow between 25% and 75% of forced vital capacity, PEFR = Peak expiratory flow rate, ECG = Electrocardiogram, DMD = Duchenne muscular dystrophy, MLPA = Multiplex ligation-dependent probe amplification, PCR = Polymerase chain reaction, U/L = Units per liter, mg/dl = Milligrams per deciliter, ng/m l= Nanograms per milliliter, LV = Left ventricle, UL = Upper limbs, LL = Lower limbs, ACE = Angiotensin-converting enzyme.

A blood sample was drawn for *DMD* gene analysis abroad, which confirmed the diagnosis. At the age of 11, limb X-ray revealed osteoporosis, and calcifediol and vitamin D treatment were implemented for two years, with annual reassessment in India. Corticosteroid therapy with deflazacort was started at the age of 15 at 60 mg per day, with gradual dose de-escalation. When he came to our consultation, the patient was still receiving deflazacort at a dose of 30 mg per day and demonstrated difficulty walking on all fours.

Furthermore, throughout his medical journey, he consulted an orthopedic surgeon and a neurologist in the DRC. He also visited four different neurologists in India. Standard radiographs were performed on seven occasions. Genetic analyses were conducted in the proband in South Africa and Belgium at the age of 10 and later repeated in the USA at the age of 18. These genetic tests consistently confirmed DMD. In addition, the proband was reassessed and retested twice in India at ages 11 and 13. Despite repeated testing, the parents did not find a local physician to interpret and provide counseling on the genetic results. The interval between the initial genetic confirmation and the start of local follow-up initiated by the Centre for Rare and Undiagnosed Diseases (CRMRND) was 11 years (Fig. 2).

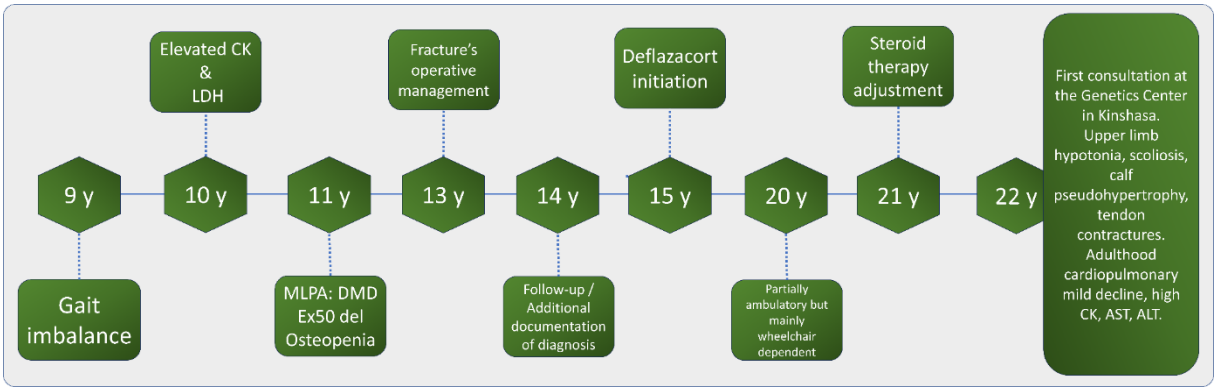


Figure 2. Patient's timeline

Chronological overview of the patient's disease course from symptom onset to adulthood. The timeline summarizes the age at first motor difficulties, initial clinical suspicion of Duchenne muscular dystrophy, genetic confirmation, initiation of corticosteroid therapy, progression of motor impairment, respiratory and cardiac evaluations, and current functional status. This longitudinal representation highlights the atypical preservation of ambulation beyond 20 years of age and the delayed onset of cardiopulmonary involvement, supporting an intermediate dystrophinopathy phenotype.

When the patient was brought to our consultation, he was in a wheelchair, although he was still able to take a few steps with support. We recorded normal growth parameters, but chest expansion and cough reflex were moderately decreased. The abdominal examination was unremarkable, and cognition was unaffected. Cervical muscle tone was rated at 5/5. The patient had hypotonia of the arm muscles, amyotrophy of the hand muscles, very significant pseudohypertrophy of the calves, fasciculation of the tongue, deformation of the lower limbs secondary to moderate tendon retractions, subtle macroglossia, and partially reducible, slightly disabling kyphoscoliosis.

His medical tests, most of which were performed at the age of 21, were normal. These included Doppler echocardiography, ECG, CBC, and measurements of bilirubin, ALP, GGT, total protein (albumin, globulin, A/G ratio), urea, uric acid, and other serum electrolytes (PO4, Na+, K+, Cl−). The main findings are summarized in Table 1. The family carried the reports from previous DMD gene analyses performed abroad using the MLPA technique, which revealed an in-frame deletion of exon 50 of the gene. This deletion is known to be pathogenic and causative of classical DMD. This interstitial deletion is known to disrupt the reading frame and result in lack of protein due to nonsense-mediated decay (NMD) of the mutated mRNA.

During a follow-up assessment at the age of 22, he reported feelings of fear and sadness and expressed uncertainty about his future.

Segmental muscle strength was rated at 3 in the upper limbs and 2 in the lower limbs. We observed fasciculation of the tongue, amyotrophy of the shoulder girdle, hypotonia of the limbs more pronounced in the lower limbs, tenderness of the spinous processes along the entire length of the spine on palpation, and tenderness in the epigastric region and lower abdomen.

Based on the clinical evaluation, this patient's phenotype is reminiscent of a milder form of muscular dystrophy, an Intermediate Muscular Dystrophy (IMD), which is not consistent with the expected DMD phenotype due to exon 50 deletion. The following care was implemented: introduction of ACE inhibitors to delay the onset of cardiomyopathy, a full respiratory assessment, and initiation of mobility assistance. The mother was offered genetic counseling. At the time of this report, *DMD* gene analysis has been prescribed for the unaffected sisters to assess the risk to their male offspring. In addition, contacts were initiated for compassionate administration of an antisense oligonucleotide (ASO) treatment; however, these were not successful given the patient's age and the already established loss of ambulation.

Discussion

We report the case of a 22-year-old Congolese patient with genetically confirmed DMD, presenting an atypical and attenuated clinical course. Although the overall phenotype suggests DMD, notable features such as preservation of walking ability, even with support, beyond the age of 20 and the relatively late onset of cardiomyopathy are more consistent with intermediate muscular dystrophy (IMD) than with classical Duchenne phenotypes (Fig. 2). In classical DMD, loss of walking ability occurs before the age of 13. The prolonged walking ability in our patient represents significant evidence of an intermediate phenotype.

The isolated deletion of exon 50 identified in our patient has been previously reported and is typically linked with severe DMD (2,3). This exon is part of a mutational hotspot (exons 45-55), where deletions often disrupt the reading frame, leading to out-of-frame transcripts that usually result in absent dystrophin and a classical DMD phenotype with early cardiopulmonary involvement (3,4). In contrast, in-frame deletions maintain partial dystrophin expression, typically associated with milder or intermediate phenotypes (1,4,6).

Out-of-frame deletions are generally associated with classical DMD, but phenotypic variability is well documented within the exon 45–55 hotspot, where delayed loss of ambulation has been reported, particularly in the context of presumed residual dystrophin expression, as indicated by the Canadian Neuromuscular Disease Registry (4). Cohort studies also support this heterogeneity, with significant intra-group variability reported by Vengalil *et al*. (2017) and a broad phenotypic spectrum highlighted by Amr *et al*. (2024), despite exon 50 deletions being usually linked to severe DMD (2,3). Waldrop *et al*. (2020) further described a spectrum of severity among exon 51 skip–equivalent deletions, ranging from classical DMD to intermediate phenotypes with delayed cardiopulmonary decline (5). ASO and gene-editing strategies have been developed to restore the reading frame, but their implementation in resource-limited settings remains constrained by high cost and limited availability (Fig. 3) (5).

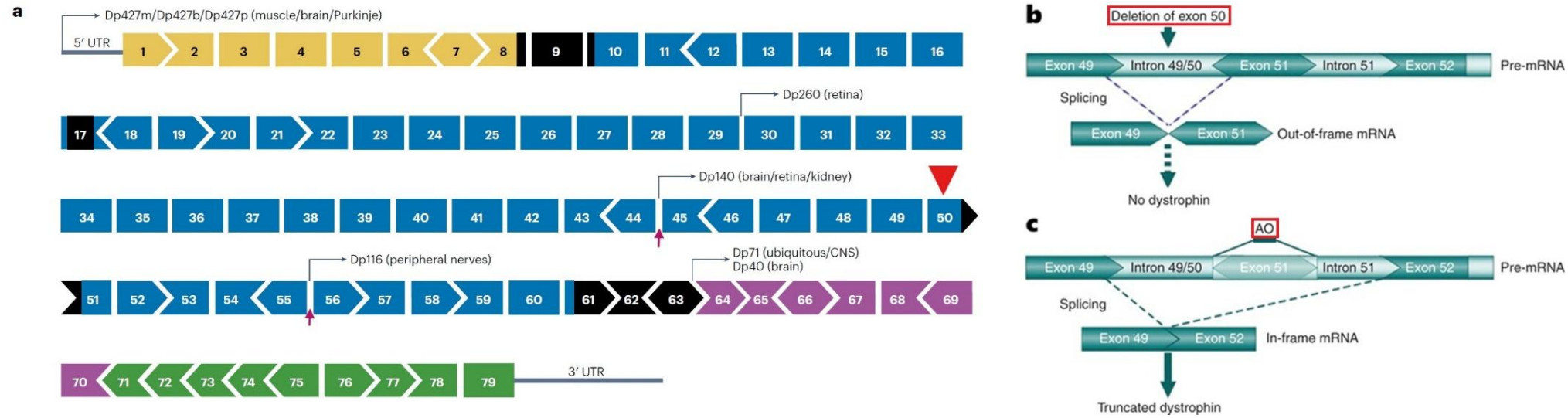


Figure 3. Schematic representation of exon 50 deletion and potential endogenous exon skipping in the *DMD* gene (*adapted and modified with written permission from the original authors and publishers of Roberts et al., 2023, Nature Reviews Drug Discovery* (9), *and Pichavant et al., 2011, Molecular Therapy.* (10)*)*

(a) Schematic of the DMD gene. The color of each exon represents the protein domain it encodes: the actin binding domain (ABD) in brown, the hinge regions in black, the central rod domain in blue, the cysteine-rich region (CR) in purple and the C-terminal domain (CT) in green. Exon shapes indicate how the triplet base code is distributed across the exons, such that they fit together to generate an in-frame mature DMD transcript. The locations of the transcription start sites for the various dystrophin protein isoforms are indicated by black arrows. The location of the distal hotspot is indicated by 2 purple arrows. Exon 50 is indicated by a red arrow.
(b) Schematic of a single exon 50 deletion at distal hotspot. The absence of exon 50 in the dystrophin gene leads to an out-of-frame mRNA creating a premature stop codon in exon 51, thus aborting dystrophin synthesis during translation.
(c) Schematic of an exon skipping mechanism. Using an antisense oligonucleotide (AO) targeting exon 51, this exon is skipped during splicing. This restores the open reading frame of the transcript and allows the synthesis of an internally deleted dystrophin.

Interestingly, spontaneous endogenous exon skipping has been reported in some out-of-frame DMD mutations, generating alternative transcripts (e.g., del50-51 or del45-55) and residual dystrophin expression that may contribute to intermediate phenotypes (4,5). Importantly, an isolated exon 50 deletion is not a skip-equivalent deletion. While endogenous skipping has been described for isolated exon 45 deletions, it has not been documented for isolated exon 50 deletions. Nevertheless, the intermediate clinical course observed in our patient raises the possibility of a similar splicing rescue mechanism, warranting further investigation.

Besides exon skipping mechanism, a multitude of other factors could account for our patient's prolonged ambulation. Genetic variability within the *DMD* locus, especially in the central rod domain, might affect transcript stability or dystrophin function, even with mutations expected to be out-of-frame. In addition, modifier genes playing roles in muscle regeneration, fibrosis or inflammation could also influence disease severity (2,4). Early implementation of therapies such as long-term corticosteroid treatment are known to extend ambulation and delay cardiopulmonary decline. Early initiation of ASO-based treatment results in significant clinical improvement. Also, access to orthopedic interventions and physical therapies may also improve clinical outcome.

However, this report carries several limitations. Despite the clinical presentation suggesting an intermediate dystrophinopathy, RNA-level analyses (e.g., RT-PCR or transcript sequencing) were not performed to confirm endogenous exon skipping or alternative splicing related to exon 50. Dystrophin protein quantification via immunohistochemistry or Western blot was also unavailable. Thus, the speculation regarding endogenous exon skipping remains hypothetical and should be interpreted with caution. Future studies incorporating transcriptomic and proteomic analyses are needed to elucidate the molecular basis for the observed attenuated phenotype.

Psychosocially, the patient reported symptoms of anxiety, depression, and hopelessness, common in DMD and detrimental to quality of life (8). Unfortunately, these symptoms were unquantified by standardized tools, prompting the need for integrating validated instruments like the Patient Health Questionnaire (PHQ-9) and Generalized Anxiety Disorder 7-item scale (GAD-7) into routine follow-up of patients with chronic neuromuscular conditions. Psychological support, therapeutic education, measures enhancing social and educational inclusion are crucial for comprehensive care, particularly in resource-limited settings.

Moreover, this report illustrates the systemic challenge of inadequate local capacity for genetic testing. The confirmatory molecular diagnosis relied on analyses conducted abroad, resulting in more than ten years of diagnostic delay before the initiation of local follow-up. Addressing this issue requires coordinated capacity-building measures, including training healthcare professionals and establishing national or regional centers for rare diseases, which could centralize expertise, standardize care pathways, and improve equitable access to diagnostics and therapies.

Overall, this case highlights the complexity of genotype–phenotype correlations in DMD, especially in the exon 45–55 hotspot. It underscores the limitations of relying solely on predicted reading-frame disruption for

prognostic assessment and reinforces the need to integrate molecular data, longitudinal observations, functional outcomes, and psychosocial dimensions to guide patient care. This also identifies a potential candidate for gene therapy in Africa.

Disclosure of conflict of interests

The authors declare no conflict of interest.

Authors' contributions

Jocelyn Nonga was actively involved in study design and data collection. Jocelyn Nonga, Prince Makay, Andoni Urtizberea, and Aimé Lumaka were actively involved in clinical evaluation, genotype–phenotype analysis, patient counseling, and follow-up. Guy Bumoko and Aimé Lumaka provided overall supervision. All authors contributed to manuscript drafting, critical revision, and approved the final version.

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Data availability

Clinical data leading to this publication are identifiable personal information and cannot be fully disclosed. However, reasonable request for information may be addressed to the corresponding author.

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