

Case Report

Duchenne muscular dystrophy in two siblings identified through the family adoption programme: a community-based case report from rural Punjab

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ABSTRACT

Duchenne muscular dystrophy (DMD) is a progressive X-linked disorder that is often recognised late in low-income rural settings. We report two brothers with DMD from a household in rural Ludhiana district, Punjab, encountered through the Family Adoption Programme (FAP), a longitudinal community attachment under the competency-based undergraduate curriculum. The younger brother had been genetically confirmed (hemizygous DMD c.5769delA, likely pathogenic) at a tertiary Centre of Excellence but his rehabilitation had lapsed; the elder brother, wheelchair-dependent since age eight, had never been evaluated because of cost. During a routine FAP home visit, his undiagnosed weakness was recognised, and both siblings were re-linked to structured rehabilitation. We use the household to examine what continuous family-level contact added to established neuromuscular care and where gaps in confirmatory testing, disease-specific therapy and linkage to the National Policy for Rare Diseases persisted. The report is offered as an implementation example, not a novel model of care.

Keywords: Community health services, Duchenne muscular dystrophy, Genetic counselling, Medical education, Rare diseases, Rural health

INTRODUCTION

Duchenne muscular dystrophy (DMD) is the commonest inherited muscular dystrophy of childhood, caused by loss-of-function variants in the DMD gene and marked by early proximal weakness, calf pseudohypertrophy and progressive loss of ambulation.¹ In rural, low-income Indian families, diagnosis is often delayed and long-term care is fragmented by the cost of confirmatory testing, distance from specialist centres and limited awareness of government provisions.² The Family Adoption Programme (FAP) attaches undergraduate students to defined households for repeated visits across the undergraduate years, under the competency-based curriculum of the National Medical Commission and is

best understood as structured longitudinal community attachment rather than a new intervention.^{3,4} We describe two affected siblings encountered through the FAP and ask a narrow question: where referral, physiotherapy and counselling already exist, what did continuous household contact change, and what did it not?

CASE REPORT

The family lives in a village in rural Ludhiana district, Punjab, and is one of the households adopted by the 2023 MBBS batch of the Department of Community Medicine; the village is not named to preserve confidentiality. It comprises the parents and two affected sons, subsists on about ₹13,000 a month (the father drives a hired cab, the

mother is the caregiver), and is lower-middle class on the Modified Kuppaswamy scale. Limited means delayed advanced testing for the elder boy and interrupted both boys' schooling. The household timeline is shown in Figure 1.



Figure 1: Household timeline.

Case 1: Elder sibling (clinical diagnosis)

The elder brother, 13 years, was well until about seven years of age, when he developed insidious gait difficulty with frequent falls and difficulty rising. Within a year he lost independent ambulation and has been wheelchair-dependent since. Confirmatory tests were not performed because of cost, so his diagnosis is clinical, supported by the family history and his brother's confirmed diagnosis. He was born at term (birth weights 4.5 kg) with normal milestones and is in the 9th grade. Examination showed upper-limb hypotonia, knee and ankle flexion contractures, power 3/5 in the upper and 0/5 in the lower limbs, and deep tendon reflexes preserved in the upper limbs but unelicitable in the lower limbs, without cranial-nerve, sensory or spasticity findings.

Case 2: Younger sibling (genetically confirmed)

The younger brother, 10 years, was well until about three years earlier, when he developed progressive difficulty rising from sitting that spread to the upper limbs. He was evaluated at a tertiary Centre of Excellence in 2024, where DMD was confirmed. He sits with occasional support and retains fine-motor skills. Born at term (birth weights 1.5 kg) with neonatal respiratory distress needing about 20 days of intensive care, he had otherwise age-appropriate milestones.

Examination showed power 4/5 (right) and 3/5 (left) in the upper limbs, 0/5 with hypotonia in the lower limbs, absent lower-limb and diminished (1+) upper-limb reflexes with down-going plantars, calf and forearm pseudohypertrophy, a bulky tongue, early neck-flexor weakness and early tendoachilles tightness. Consolidated findings are given in Table 1 and the pedigree in Figure 2.

Investigations

Serum CK-NAC was 3874 U/l (reference 26–308) and CK-MB 168 U/l, indicating skeletal-muscle origin.

SALSA MLPA (probemix P034/P035) showed no DMD deletion or duplication. Whole-exome sequencing identified a hemizygous frameshift variant, DMD c.5769delA (p.Glu1924fs*3) in exon 41 (transcript NM_004006.3; GRCh37 chrX:32360369), predicted to cause nonsense-mediated decay and classified as likely pathogenic (ACMG), confirming the Duchenne phenotype. Two incidental heterozygous carrier variants were also found (CYP11B1 c.1169G>A and SLC26A2 c.1010_1012delTTG), of reproductive-counselling relevance only. The elder brother was not tested; the same variant is the probable cause.

Genetic counselling

Counselling covered X-linked inheritance, recurrence risk and reproductive options. At the 2024 evaluation, the mother, an obligate carrier, was about 20 weeks pregnant, giving the counselling an immediate reproductive dimension: amniocentesis with quantitative-fluorescence PCR and DMD MLPA (with a maternal-cell-contamination check) was offered, a level II anomaly scan advised, and a quadruple screen deemed unnecessary; after counselling, the recorded advice was to continue the pregnancy. Carrier testing was offered to adult at-risk female relatives, with testing of minors deferred to an age of independent consent.

Management and follow-up

Care is anchored in physiotherapy and structured rehabilitation close to home.⁵ At diagnosis the younger boy had been started on calcium and a physiotherapy programme (tendoachilles, hamstring and rectus-femoris stretching; active range-of-motion to all limbs; deep-breathing and thoracic-expansion exercises); this lapsed and was re-established for both boys at a rural outreach hospital after the FAP visit, with multidisciplinary paediatric and neurological review. Current standards also require glucocorticoids and cardiorespiratory surveillance, but neither boy was receiving glucocorticoids; low-dose baclofen was given for contracture-related discomfort.⁶ Functional status is reported descriptively; no standardised functional, quality-of-life or caregiver-burden instruments were applied, so no claim of improved quality of life is made.

DISCUSSION

This household illustrates implementation, not innovation. Case-finding, referral, physiotherapy, counselling and scheme linkage are established components of chronic neuromuscular care; none is novel. What the FAP added was continuity: because the same students returned to the same home, an undiagnosed elder sibling was recognised and a younger sibling whose follow-up had lapsed was re-engaged. Episodic outreach or a single screening camp might have missed both. The FAP does not differ in kind from longitudinal family follow-up already embedded in many curricula; its

contribution here is operational, in that a defined household, repeated contact and a named student-faculty team created the trust and persistence through which a missed diagnosis surfaced.

The family's difficulty was not the absence of provisions but the absence of linkage to them. Under the Rashtriya Bal Swasthya Karyakram, children up to 18 years receive free screening, early intervention and referral.⁷ DMD is a notified rare disease under the National Policy for Rare Diseases, 2021, with support of up to ₹50 lakh per patient at notified Centres of Excellence; the centre where the younger boy was diagnosed is itself such a centre, and eligibility now extends to approximately the poorest 40% identified for the Pradhan Mantri Jan Arogya Yojana.⁸ That a diagnosed, eligible family remained unlinked to this funded route is the gap this report highlights.

Several limitations should be stated plainly: a single household without a comparison group; uncertain sustainability tied to student rotations and faculty time; and scale-up that would require embedding in routine primary-care surveillance rather than academic goodwill.⁹ The care delivered also remained incomplete, lacking confirmatory testing for the elder boy, glucocorticoids, cardiorespiratory surveillance and, above all, linkage to the funded rare-disease route; no standardised outcomes were measured and follow-up was short. The practical lesson is that programmes such as the FAP should carry an explicit protocol for scheme linkage and referral to a Centre of Excellence, so that identification translates into funded, guideline-based care.

CONCLUSION

Two brothers with DMD in a low-income rural household were brought into structured care through repeated FAP visits that surfaced one undiagnosed sibling and re-linked another. The episode is offered not as a new paradigm but as a practical example of how continuous household contact can close identification and linkage gaps for a rare disease, provided it is coupled to confirmatory testing, guideline-based treatment and the financial protection the family was already entitled to.

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