CASE REPORT

# A 10-year follow-up of therapeutic rehabilitation in a child with LMNA-associated congenital muscular dystrophy: a case report

Zhimei Liu1, Shuo Zhao2, Ziheng Wang3, Minhua Chen4, Kelong Cai5, Kang Yang6

## Abstract

Lamin A/C (LMNA)-associated congenital muscular dystrophy (L-CMD) is a rare neuromuscular disorder caused by mutations in the LMNA gene, characterised by droopy head syndrome with motor developmental delays and weakness of the spinal axial and proximal muscles of the extremities. This case report describes the 10-year therapeutic rehabilitation and follow-up of an 18-month-old girl whose parents complained of slender neck and muscle weakness at the Rehabilitation Medicine Department, Northern Jiangsu People's Hospital, Yangzhou, China. The purpose of this case report is to demonstrate that early, continuous, and systematic motor rehabilitation training proved to be conducive to the development of motor function in this L-CMD child.

**Keywords:** LMNA-related congenital muscular dystrophy, Genetic deficiency, Therapeutic rehabilitation, Motor function.

**DOI: https://doi.org/10.47391/JPMA.30406**

## Introduction

LMNA-associated congenital muscular dystrophy (L-CMD) is a rare form of congenital muscular dystrophy, typically appearing at birth or within the first few months of life.[1] It is marked by droopy head syndrome, motor developmental delays, and weakness in the spinal axial and proximal muscles. Some patients may need early respiratory and nutritional support or show early signs of heart disease. L-CMD has a prevalence of less than one in one million children and follows an autosomal dominant inheritance pattern. Most affected children cannot sit or walk unaided, and the condition affects both cardiac and skeletal muscles. Cardiomyopathy is the leading cause of death, contributing to a poor prognosis, while hypokinesia and developmental delays are common.[2] Although therapeutic rehabilitation benefits neuromuscular disorder patients, its effects on L-CMD remain under-researched, with existing studies focussing on clinical symptoms, muscle pathology, and genetics rather than exercise rehabilitation and long-term outcomes.[3] We conducted a ten-year rehabilitation and follow-up study on a child with L-CMD to assess its impact on motor function development.

## Case Report

An 18-month-old girl visited the Rehabilitation Medicine Department at Northern Jiangsu People's Hospital, China, on May 26, 2013, where rehabilitation was started. Her parents reported a slender neck, weakness, and difficulty lifting her head. She was born full-term via vaginal delivery, weighing 2,750 gm. She showed delayed motor development: unstable head control at five months, inability to sit alone at 10 months, inability to crawl at one year, and inability to walk alone at 18 months. Her cognitive and language development were normal. Her parents were healthy, unrelated, and had no similar family history.

She demonstrated preserved horizontal visual and auditory tracking in supine position. Prone positioning revealed sustained 90° head elevation with bilateral upper limb support. Independent axial rotation (rollover) was achieved, though truncal instability was noted during seated three-level postural control testing. Quadrupedal locomotion presented with characteristic lumbar lordosis. She could roll over independently, but had trunk instability during seated posture control tests. Crawling revealed a typical lumbar curve. She could kneel on all fours for three to five minutes and transition between positions with help. She couldn't walk independently, but with Knee-Ankle-Foot Orthoses, she could stand with an anterior weight shift, trunk/pelvic tilt, and hip flexion. Assisted walking allowed for 10-12 steps with noticeable ataxia and a waddling gait. Examination showed bilateral calf muscle tightness (Modified Ashworth Scale 1+)2 and ankle joint contractures limiting upward foot movement

[1]Department of Rehabilitation Medicine, College of Physical Education, Yangzhou University, Yangzhou, China. [2,3]Department of Children Rehabilitation, Yangzhou Maternal and Child Health Hospital, Yangzhou, China. [4]Developmental Behavioural Child Health unit, Affiliated Hospital of Yangzhou University, Yangzhou, China. [5]Sports Psychology Laboratory of College of Physical Education, Yangzhou University, Yangzhou, China.[6] Department of Rehabilitation Medicine, Northern Jiangsu People's Hospital, Yangzhou, China.

**Correspondence:** Kang Yang. **Email:** yangkanglc@yzu.edu.cn

**ORCID ID:** 0000-0002-8669-3105

**Submission complete:** 17-04-2025 **First Revision received:** 12-09-2025

**Acceptance:** 18-03-2026 **Last Revision received:** 17-03-2026

**Table:** List of abnormal values

| Inspection | Exceptional item | Result | Report | Reference | Unit |
|---|---|---|---|---|---|
| biochemical test | Glutamine transaminase (AST) | 48 | ↑ | 13-35 | IU/L |
| | Prealbumin (PA) | 132.0 | ↓ | 200-400 | mg/L |
| | Calcium (CA) | 2.58 | ↑ | 2.11-2.52 | mmol/L |
| | Carbon dioxide (CO2) | 20.23 | ↓ | 22-30 | mmol/L |
| | Creatine kinase (CK) | 370 | ↑ | 25-170 | IU/L |
| | Lactate dehydrogenase (LDH) | 448 | ↑ | 100-240 | IU/L |
| | Hydroxybutyrate dehydrogenase (HBDH) | 398 | ↑ | 90-220 | IU/L |
| | CK-MB mass (CK-MB) | 47.6 | ↑ | <5 | ng/ml |
| gas chromatography mass spectrometry | Oxalic acid-2 | 3.35 | ↑ | 00-1.10.7-3.70.8-2.30000-13.800 | |
| | 3-Hydroxy-propionic acid-2 | 2.2 | ↑ | | |
| | 3-Hydroxy-butyric acid-2 | 774.8 | ↑ | | |
| | 3-Hydroxy-isovaleric acid-2 | 21.64 | ↑ | | |
| | Acetoacetic acid-2 | 7.36 | ↑ | | |
| | Glycerol | 9.89 | ↑ | | |
| | Vanillic acid-2 | 182.21 | ↑ | | |
| | Palmitic acid-1 | 32.75 | ↑ | | |
| | MGA | 100 | ↑ | | |
| | (3-Hydroxy-phenyl)-3-hydroxy-propionic acid-3 | 6.88 | ↑ | | |
| amino acid and carnitine spectrum test | Suanic acid (Thr) | 16.4 | ↓ | 22-200 | umol/L |
| | Free carnosic acid (C0) | 16.43 | ↓ | 20-60 | umol/L |
| | Propionyl carnitine (C3) | 0.57 | ↓ | 1-4. | umol/L |
| | Tetradecadienoyl carnitine (C14:2) | 0.07 | ↑ | 0-0.04 | umol/L |

(equinus deformity). Muscle strength was grade IV in the upper limbs and grade IV- in the lower limbs, with core strength graded III-IV. Pathological reflexes, including Hoffmann's, were present.

At age two, she underwent survival motor neuron 1 (SMN1) gene exon 7 deletion test and duchenne muscular dystrophy (DMD) gene 79 exon deletion/duplication test, that ruled out spinal muscular atrophy and pseudohypertrophic muscular dystrophy. Neuromuscular disease V3 panel gene test confirmed the diagnosis of L-CMD at age four. Details of other medical examinations like cardiac colour ultrasound, cranial magnetic resonance imaging (MRI), frontal X-ray of the hip joints, electromyography (EMG) and rehabilitation interventions from the patient's birth are presented in time lines in Figure 1A. The results of gene sequencing map are shown in Figure 1B&C.

The child's rehabilitation programme began with gross and fine motor training. Her motor skills improved steadily before age five, following typical development patterns, but showed regression or stagnation after age five. Consequently, the training goals were adjusted to suit her developmental stage.[4, 5]

The motor training programme for under five years of age aimed to improve head control, rolling, sitting, crawling, standing, and walking. After age five, it focussed on muscle strength, endurance, core stability, and cardiorespiratory fitness, using assistive devices. Sessions were 40 minutes, five times a week.[6] The fine motor programme enhanced upper limb skills like grasping, bending, and rotating through games for children under five. After age five, it emphasised grasping, copying, and self-care skills like writing, eating, and dressing, with the same session frequency and duration.[7] (Figure 1D&E). The publication of this case report has been read and approved by her guardian.

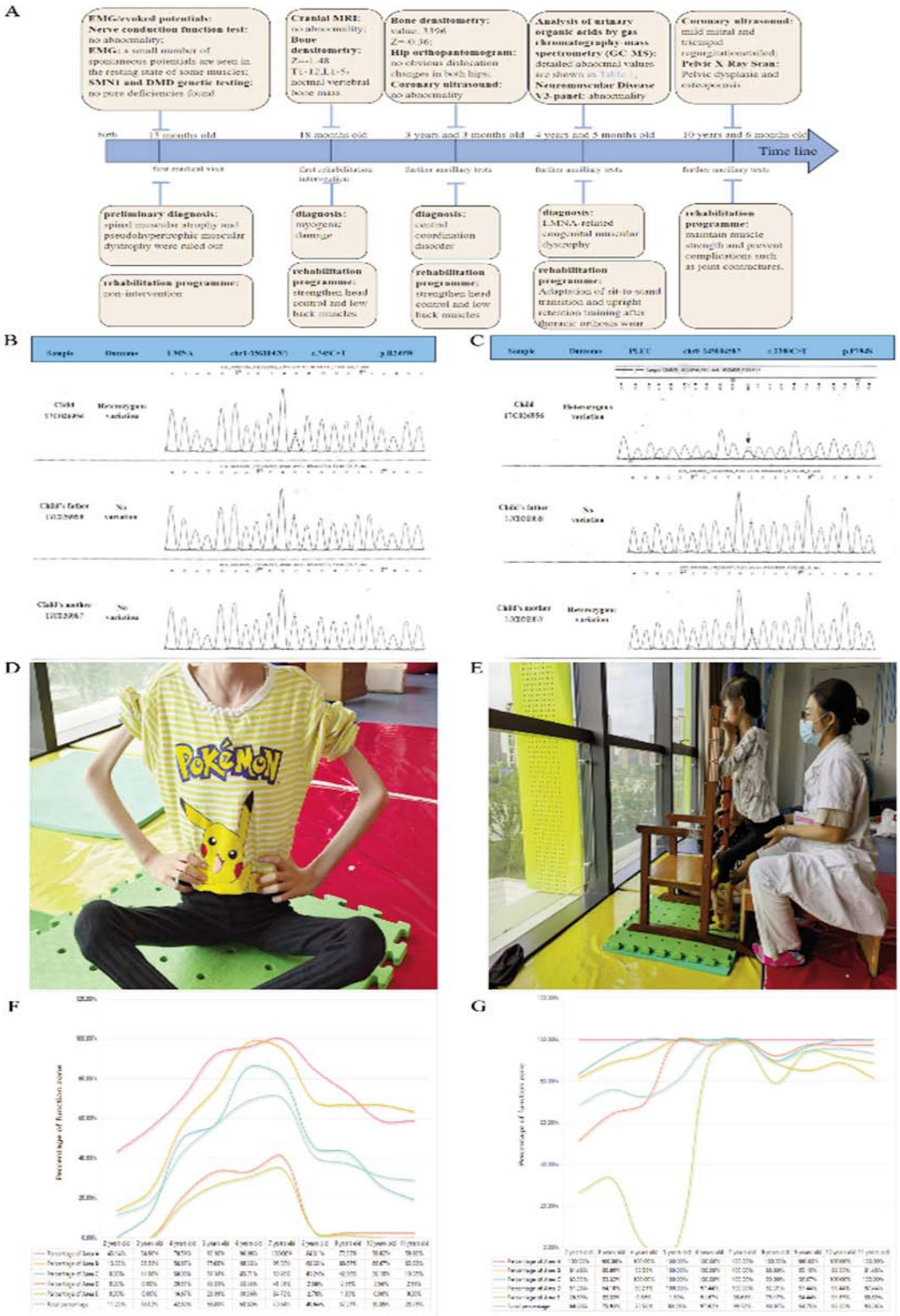


**Figure:** A time line of details of medical examinations and rehabilitation interventions; B&C gene sequencing map; D&E rehabilitation training programme; F trends in the GMFM-88 assessment results and progression (Zone A: supine and prone ability; Zone B: sitting ability; Zone C: crawling and kneeling ability; Zone D: standing ability; and Zone E: walking, running and jumping ability); G Trends in FMFM assessment results and progression (Zone A: visual and auditory tracking; Zone B: upper extremity joint mobility; Zone C: grasping ability; Zone D: manipulative ability; and Zone E: hand—eye coordination)

## Discussion

This case presents the 10-year rehabilitative follow-up of a girl with L-CMD, harbouring a classic pathogenic LMNA mutation (c.745C>T, p.R249W). Her clinical course underscores both the potential benefits of sustained rehabilitation and the profound impact of environmental factors on motor function in this progressive disorder.

The core finding is that a structured, dual-model ('hospital + home') rehabilitation programme was associated with significant motor improvement in early childhood. Before age seven, the patient achieved milestones like sitting, crawling, and brief assisted walking with orthoses. This aligns with evidence from other neuromuscular diseases, where intensive, early physical therapy is known to enhance motor performance and delay functional decline.[8,9] For instance, structured training has been shown to improve strength and motor scores in children with various forms of congenital muscular dystrophy.[9]

However, a critical observation was the rapid decline in gross motor skills between ages seven and eight, coinciding with school admission and a reduction in dedicated training time. This regression highlights the relentless nature of L-CMD and the necessity of consistent, lifelong intervention to maintain function. This nuance offers a partial contrast to some natural history studies, which often depict a steady, linear decline in L-CMD regardless of therapy.[10] The current case suggests that while the underlying disease is progressive, functional trajectories can be significantly modulated—both positively and negatively—by the intensity of rehabilitative support.

Furthermore, the patient's fine motor and hand-eye coordination also demonstrated improvement with training, regression with reduced focus, and subsequent stabilisation. The recovery of upper limb function was facilitated not only by direct training but also by increased manual activities at school. This points to a potential role for cognitive engagement and enriched environments in motor learning, a concept supported by rehabilitation research in other paediatric neurological conditions.[7] It implies that rehabilitative strategies for L-CMD could benefit from integrating cognitive-motor tasks, moving beyond pure physical training.

## Conclusion

Although L-CMD remains a severe condition, this long-term case study indicates that a proactive, intensive, and adaptable rehabilitation regimen is crucial. It can help maximise functional potential and quality of life. Future work should focus on establishing standardised, personalised rehabilitation protocols and further exploring the synergy between cognitive and motor interventions in this population.

**Consent:** Approval for publishing this report was acquired from the parents of the child.

**Acknowledgement:** The authors are grateful to the patient and her family for contributing to this research.

**Disclaimer:** This article has not been presented or published before.

**Conflict of Interest:** None.

**Source of Funding:** This research was funded by the Postgraduate Research & Practice Innovation Programme of Jiangsu Province (KYCX21_3161 & KYCX22_3419) and the Scientific Research Fund of Northern Jiangsu People's Hospital (SBKY21010)..

## References

1. Maggi L, Mavroidis M, Psarras S, Capetanaki Y, Lattanzi G. Skeletal and Cardiac Muscle Disorders Caused by Mutations in Genes Encoding Intermediate Filament Proteins. Int J Mol Sci 2021;22:4256. doi: 10.3390/ijms22084256.
2. Harb A, Margetis K, Kishner S. Modified Ashworth Scale. Treasure Island, FL: StatPearls Publishing; 2026.
3. Hammer S, Toussaint M, Vollsæter M, Nesbjørg Tvedt M, Drange Røksund O, Reychler G, et al. Exercise Training in Duchenne Muscular Dystrophy: A Systematic Review and Meta-Analysis. J Rehabil Med 2022;54:jrm00250. doi: 10.2340/jrm.v53.985.
4. Liu X, Fu X, Dai G, Wang X, Zhang Z, Cheng H, et al. Comparative analysis of curative effect of bone marrow mesenchymal stem cell and bone marrow mononuclear cell transplantation for spastic cerebral palsy. J Transl Med 2017;15:48. doi: 10.1186/s12967-017-1149-0.
5. Rivera D, García A, Ortega JE, Alarcos B, van der Meulen K, Velasco JR, et al. Intraindividual Variability Measurement of Fine Manual Motor Skills in Children Using an Electronic Pegboard: Cohort Study. JMIR Mhealth Uhealth 2019;7:e12434. doi: 10.2196/12434.
6. Wang X, Zhou B. Motor development-focused exercise training enhances gross motor skills more effectively than ordinary physical activity in healthy preschool children: an updated meta-analysis. Front Public Health 2024;12:1414152. doi: 10.3389/fpubh.2024.1414152.
7. Scholtes VA, Becher JG, Comuth A, Dekkers H, Van Dijk L, Dallmeijer AJ. Effectiveness of functional progressive resistance exercise strength training on muscle strength and mobility in children with cerebral palsy: a randomized controlled trial. Dev Med Child Neurol 2010;52:e107-13. doi: 10.1111/j.1469-8749.2009.03604.x.
8. Chatzifrangkeskou M, Le Dour C, Muchir A. Modulation of cytoskeleton in cardiomyopathy caused by mutations in LMNA gene. Am J Physiol Cell Physiol 2023;324:C1223-35. doi: 10.1152/ajpcell.00471.2022.
9. Gómez-Domínguez D, Epifano C, Miguel F, Castaño AG, Vilaplana-Martí B, Martín A, et al. Consequences of Lmna Exon 4 Mutations in Myoblast Function. Cells 2020;9:1286. doi: 10.3390/cells9051286.

10. Salavati M, Krijnen WP, Rameckers EA, Looijestijn PL, Maathuis CG, van der Schans CP, et al. Reliability of the modified Gross Motor Function Measure-88 (GMFM-88) for children with both Spastic Cerebral Palsy and Cerebral Visual Impairment: A preliminary study. Res Dev Disabil 2015;45-46:32-48. doi: 10.1016/j.ridd.2015.07.013.

---

**AUTHORS' CONTRIBUTIONS:**

**ZL, SZ, ZF, MC, KC & KY:** Concept, design, data acquisition, analysis, interpretation, drafting, revision, final approval and agreement to be accountable for all aspects of the work.