

## Severe Bulbar Dysfunction Without Cardiac Involvement In a Pediatric Congenital Titin Myopathy: A Rare Phenotype In an Irish Traveller Family with Complex Genetic, Clinical, and Sociomedical Implications: A Case Report

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### ABSTRACT

Congenital titinopathies, arising from biallelic truncating, splice-altering, or metatranscript-only variants in the TTN gene, typically present with hypotonia, arthrogryposis, feeding difficulty, and respiratory insufficiency. Cardiac risk in this group is variant-position-dependent and age-penetrant rather than uniform, and presentations dominated by bulbar dysfunction without cardiac involvement remain sparsely documented. We describe a 12-year-old girl of Irish Traveller descent with a clinical diagnosis of congenital titinopathy and arthrogryposis multiplex congenita (AMC); biallelic TTN truncating variants were reported in early childhood, but the primary molecular report was not retained and could not be recovered, leaving the variant position and its predicted effect on cardiac isoforms unknown. Her phenotype is marked by severe bulbar impairment manifesting as persistent oropharyngeal dysphagia, recurrent aspiration, and gastrointestinal dysmotility, in the absence of cardiac or pharyngeal anatomical abnormalities. She is BiPAP-dependent and PEG-fed, with only intermittent oral intake due to aspiration risk, and has undergone multiple hospitalisations for aspiration pneumonia, faecal loading, and suspected sepsis. Despite repeated episodes of respiratory decompensation, cardiac structure and function have remained normal on serial echocardiography and ECG across more than a decade, and surveillance is continuing. A sibling reported to share the same TTN genotype demonstrates only distal weakness, underscoring notable intrafamilial phenotypic variability. This case documents a bulbar-dominant congenital titinopathy without cardiomyopathy to date, while illustrating that the loss of molecular records precludes genotype-based cardiac risk stratification and therefore does not permit surveillance to be relaxed. The patient's sociomedical context, including caregiver burden and structural barriers to care, further emphasises the need for culturally contextualised genomic services and longitudinal multidisciplinary support in underserved populations.

**Categories:** Family/General Practice, Genetics, Pediatrics

**Keywords:** Arthrogryposis; Caregiver burden; Congenital myopathy; Genomic inequity; Health equity; Intrafamilial variability; Irish traveller; Peg feeding; Titinopathy; ttn gene

## INTRODUCTION

Titin, encoded by the TTN gene, is the largest human protein and forms the third myofilament system of striated muscle, spanning the sarcomere from the Z-disk to the M-band, where it contributes to sarcomeric assembly, passive elasticity, and mechanosensing.<sup>[1]</sup> Pathogenic TTN variants cause a heterogeneous group of disorders collectively termed titinopathies, which vary in mode of inheritance, age at onset, pattern of muscle involvement, severity, and rate of progression.<sup>[1,2]</sup>

The dominant, adult-onset end of this spectrum includes tibial muscular dystrophy and hereditary myopathy with early respiratory failure (HMERF), the latter caused by missense variants clustering in the Abundant fibronectin type III 119 domain and characterised by proximal and axial weakness with ventilatory failure in ambulant adults.<sup>[3,4]</sup> By contrast, congenital titinopathies are recessively inherited and typically arise from biallelic truncating, splice-altering, or metatranscript-only variants, presenting with arthrogryposis multiplex congenita, severe neonatal hypotonia, feeding difficulty, and respiratory insufficiency.<sup>[5,6,7]</sup> Histopathology in these congenital cases is variable and has included centronuclear features, leading some to be initially classified as centronuclear myopathy.<sup>[6]</sup>

Cardiac risk in TTN-related disease is variant-dependent rather than uniformly age-dependent.

Heterozygous truncating variants affecting constitutively expressed cardiac exons confer an increased risk of adult-onset dilated cardiomyopathy (DCM),<sup>[8]</sup> while biallelic truncating variants impacting the N2B and N2BA cardiac isoforms are associated with congenital cardiac anomalies and early-onset cardiac involvement, including the fatal childhood DCM described in the original reports of recessive M-line titinopathy.<sup>[7,9,10]</sup> Conversely, patients carrying at least one pathogenic variant in metatranscript-only exons appear less likely to develop cardiac abnormalities, and arthrogryposis with congenital myopathy in the absence of cardiac involvement has been documented in this subgroup.<sup>[7,11]</sup> Importantly, cardiomyopathy in titinopathy shows age-related penetrance, and normal cardiac assessment in childhood does not exclude later cardiac disease.<sup>[5,7]</sup>

Genotype-phenotype correlation therefore remains incomplete. In the largest recessive titinopathy cohort assembled to date, the position of premature termination codons within the M-band correlated inversely with clinical severity, yet marked phenotypic variability persisted between individuals carrying comparable variants; the authors proposed a contribution from the second allele rather than invoking an established modifier mechanism.<sup>[7]</sup> Functional validation and longitudinal natural history data remain limited.

This report describes a child with a reported biallelic TTN truncating genotype who exhibits severe neuromuscular and bulbar dysfunction with no cardiac involvement across more than a decade of serial screening. Her presentation is further contextualised within the Irish Traveller community, an ethnic minority group underrepresented in genomic research, subject to well-documented health inequities, and in which consanguineous union is more frequent than in the settled Irish population, plausibly increasing population-specific risk for autosomal recessive disorders.<sup>[12,13]</sup>

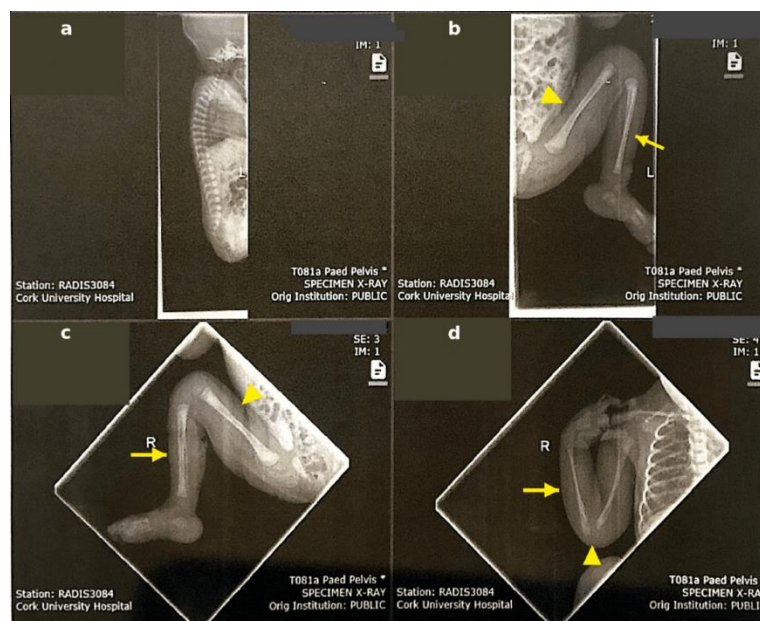
## CASE PRESENTATION

### Patient Information

The patient is a 12-year-old female of Irish Traveller descent with a clinical diagnosis of congenital titin myopathy and arthrogryposis multiplex congenita. Biallelic TTN truncating variants were reported following genetic testing in early childhood; the primary molecular report is no longer available for review (see Limitations). She also has mild intellectual disability. Her family history is notable for one sibling reported to carry the same TTN genotype who manifests a milder distal myopathy, while another sibling remains unaffected; the sibling's molecular report was likewise unavailable for direct review. Socially, the patient resides in overcrowded accommodation that lacks adaptations for wheelchair accessibility. Her care is provided entirely by her mother, who has been the sole caregiver since infancy. The family has limited access to consistent home-based nursing or community health support services.

### Clinical Findings

In the neonatal period, a skeletal survey was performed to characterize the contractures and skeletal abnormalities present at birth (**Figure 1**). This demonstrated fixed flexion deformities of the hips (arrowheads in 1b and 1c), and elbows (arrowhead in 1d), with thinned, gracile long bones (arrows in 1b, 1c, and 1d), most pronounced in the upper limbs, together with nonspecific symmetric metaphyseal lucency. No skeletal dysplasia was identified (1a).



**FIGURE 1:** Neonatal skeletal survey showing fixed flexion deformities of the hips (arrowheads in b and c) and elbows (arrowhead in d) with thinned, gracile long bones (arrows in b, c, and d; most pronounced in the upper limbs) and symmetric metaphyseal lucency. No skeletal dysplasia identified (a).

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On clinical examination at the age of 12 years, the patient exhibits generalised hypotonia, marked muscle atrophy, and fixed flexion contractures affecting both the upper and lower limbs, consistent with arthrogryposis multiplex congenita and with the deformities first documented on the neonatal skeletal survey (Figure 1). She is non-ambulatory and uses a wheelchair for all mobility. Bulbar dysfunction is prominent and includes impaired lingual propulsion, reduced laryngeal elevation, partial epiglottic inversion, and a weak cough reflex. These findings contribute to persistent oropharyngeal dysphagia, with near-complete dependency on PEG feeding. Despite cautious reintroduction of limited oral intake, she continues to experience recurrent respiratory infections requiring hospitalisation. Cognitive function is within the mild intellectual disability range on clinical assessment, but expressive and receptive language skills are well preserved. Cardiovascular examination and imaging have shown no evidence of cardiac involvement to date.

Clinical Timeline

A summary of the key clinical events across the patient's life is provided in Table 1.

| Age        | Key Clinical Event                                                                                                                                                                                                                                                 |
|------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Infancy    | Presentation with severe hypotonia and arthrogryposis multiplex congenita; PEG feeding initiated; nocturnal BiPAP commenced. Genetic testing and muscle biopsy performed confirming biallelic TTN truncating variants; results subsequently not retained by family |
| ~3–5 years | Ongoing hospitalizations for recurrent aspiration pneumonia and respiratory decompensation; long-term antibiotic courses initiated                                                                                                                                 |
| ~6–7 years | Progressive scoliosis identified; physiotherapy and supportive care intensified                                                                                                                                                                                    |
| ~8 years   | MAGEC spinal rod insertion for progressive scoliosis                                                                                                                                                                                                               |
| ~10 years  | Cautious reintroduction of IDDSI Level 6 oral diet trialed under SLT guidance; PEG remains primary nutrition route                                                                                                                                                 |
| ~12 years  | Sleep study performed; satisfactory ventilatory control with nocturnal BiPAP confirmed; recent hospitalization for fecal loading and suspected sepsis                                                                                                              |
| ~12 years  | Most recent serial echocardiogram and 12-lead ECG: normal cardiac function confirmed; sibling genetic data reviewed confirming same TTN genotype with milder phenotype                                                                                             |

TABLE 1: Clinical timeline of key diagnostic and therapeutic events.

Diagnostic Assessment

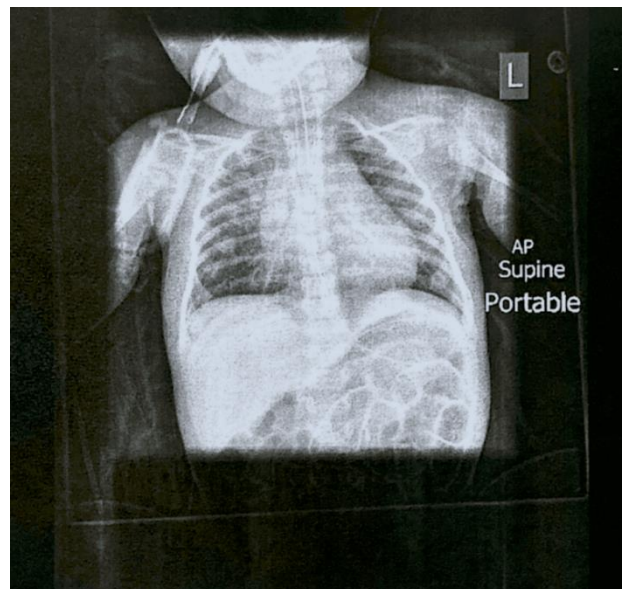
Genetic testing and muscle biopsy were reportedly completed in early childhood and were understood by the family to have confirmed a titin-related congenital myopathy. The primary reports were not retained, and no copies were available from the originating service at the time of this review. In the absence of these documents, the diagnosis rests on the clinical phenotype, the pattern of familial segregation, and supportive imaging; the precise variants, their location within TTN, and their predicted effect on cardiac isoforms are therefore unknown.

MRI of skeletal muscle demonstrated widespread fatty infiltration of the limb musculature(only report available). This finding is consistent with, but not specific to, congenital titinopathy; selective patterns of involvement, particularly semitendinosus and medial gastrocnemius, have been described as more

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discriminating features in this group.<sup>[7,14]</sup> A sleep study performed at age 12 was satisfactory with the use of nocturnal BiPAP. Serial echocardiograms and ECGs, including the most recent, have remained normal. Swallowing assessments by speech and language therapy revealed severe oropharyngeal dysfunction, confirming the patient's high risk of aspiration. Transient sinus tachycardia was observed on ECG but was not associated with structural cardiac abnormalities.

Radiological investigations obtained during acute admissions reflected her chronic gastrointestinal and respiratory burden. Radiographs obtained during an acute admission with persistent vomiting demonstrated prominent colonic faecal residue with rectal loading, without obstruction or pneumoperitoneum, while chest radiography confirmed severe thoracolumbar scoliosis with intact spinal instrumentation and a gastrostomy tube in situ, without acute consolidation on that occasion (Figure 2). Recurrent right lower lobe consolidation has, however, been documented on chest radiographs obtained during separate episodes of aspiration pneumonia.



**FIGURE 2:** Radiographs obtained for persistent vomiting. Chest: severe thoracolumbar scoliosis with intact bilateral spinal rod and transpedicular screw fixation and a gastrostomy tube in situ; no new consolidation on this specific occasion. Abdomen: prominent colonic faecal residue with rectal loading, no obstruction or pneumoperitoneum.

*Therapeutic intervention*

Nutritional management has centred on long-term PEG feeding, initiated in infancy and continued to the present. Since age 10, cautious reintroduction of IDDSI Level 6 oral intake has been trialled, although PEG remains the primary route of nutrition due to persistent aspiration risk. Respiratory support includes nocturnal BiPAP, in place since early childhood.

The patient has required frequent courses of antibiotics, typically intravenous co-amoxiclav or cefotaxime, during episodes of suspected aspiration pneumonia. She is maintained on regular osmotic laxatives (macrogol) for chronic constipation and receives antiemetics (cyclizine and ondansetron) as needed. Supportive therapies

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include saline nebulisers and regular chest physiotherapy. Surgically, she underwent insertion of MAGEC spinal rods at age 8 for progressive scoliosis.

*Follow-up and Outcomes*

The patient remains medically fragile with persistent reliance on BiPAP and PEG feeding. She continues to experience intermittent hospital admissions for aspiration pneumonia and gastrointestinal dysmotility. Despite high aspiration risk, carefully titrated oral intake continues under speech and language therapy guidance. There has been no evidence of cardiac involvement over a decade of follow-up, and annual cardiac surveillance is continuing. Functional status is stable but limited. Her care depends heavily on maternal support, complicated by significant environmental and systemic challenges. Cognitive function allows for effective communication despite low speech volume and occasional intelligibility issues.

**DISCUSSION**

This case illustrates a congenital titinopathy phenotype dominated by bulbar dysfunction, gastrointestinal dysmotility, and respiratory vulnerability, in which cardiac structure and function have remained normal across more than a decade of surveillance. Such a trajectory is recognised but under-represented in the literature: arthrogryposis and congenital myopathy without cardiac involvement have been reported in association with variants in prenatally expressed, metatranscript-only TTN exons,<sup>[11]</sup> and cohort data indicate that patients carrying at least one metatranscript-only pathogenic variant are less likely to develop cardiac abnormalities than those with biallelic variants affecting the N2B and N2BA cardiac isoforms.<sup>[5,7]</sup>

This observation should not be generalised into an expectation that congenital titinopathy spares the heart. The first description of a purely recessive congenital titinopathy, arising from homozygous C-terminal Mline truncations, was defined by congenital muscle weakness followed by childhood-onset fatal dilated cardiomyopathy,<sup>[9]</sup> and recessive truncating variants have subsequently been shown to define forms of core myopathy with heart disease.<sup>[10]</sup> Cardiac penetrance is also age-dependent, such that a child with normal serial imaging may still develop cardiomyopathy in later years.<sup>[5,7]</sup> In the present case the variant position is unknown, and the patient therefore cannot be assigned to a low-risk genotypic category on molecular grounds. The clinically defensible inference is not that cardiac involvement can be assumed absent, but that a bulbar- and respiratory-dominant presentation may occur without concurrent cardiomyopathy, and that lifelong cardiac surveillance remains indicated regardless of the prevailing skeletal muscle phenotype.

Complicating the diagnostic process was the unavailability of genetic reports and histopathological results. Although muscle biopsy and genetic testing were performed in early childhood, the records were not retained. The patient's parents, who come from a marginalised Irish Traveller background with limited formal education, discarded the materials without an understanding of their medical significance. This reflects a communication failure: no structured genetic counselling or follow-up support was offered, and the family received little explanation about the long-term value of these results. Histological findings in TTN-related myopathies are heterogeneous but follow recognisable patterns, with prominent nuclear internalisation and defects in oxidative staining characterising congenital recessive phenotypes, and ultrastructural sarcomeric disorganisation with M-band and A-band disruption representing a common baseline lesion.<sup>[15]</sup> The absence of these records therefore

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represents a substantive missed diagnostic opportunity, both for phenotypic classification and for cardiac risk stratification, and underscores the importance of retained documentation in chronic paediatric conditions.

This is not an isolated issue. It points to systemic gaps in care that disproportionately affect historically underserved communities. Irish Travellers experience well-documented disparities across all levels of healthcare, including markedly elevated mortality relative to the general population, alongside fragmented service contact, implicit bias, and logistical barriers.<sup>[13]</sup> Where consanguineous union is more common, the case for structured, culturally appropriate genetic counselling is correspondingly stronger, yet service models designed around such families remain underdeveloped in Ireland.<sup>[12]</sup> The prevailing attitude within some clinical environments remains *laissez-faire*, with limited effort made to contextualise complex diagnoses in an accessible way or to coordinate multidisciplinary care that includes social determinants of health.

The recognised phenotypic spectrum of TTN truncating variants continues to broaden. Ceyhan-Birsoy et al. identified recessive truncating TTN mutations in patients whose biopsies had led to a diagnosis of centronuclear myopathy, concluding that TTN analysis should be considered in suspected centronuclear myopathy without mutations in the classic genes.<sup>[6]</sup> Our case similarly underscores the importance of recognising non-cardiac-dominant trajectories within congenital titinopathy. The observed intrafamilial variability, in which a sibling reported to share the same genotype remains only mildly affected, is consistent with the incomplete genotype-phenotype correlation described in large cohorts, where phenotypic differences persisted between individuals with comparable variants and were attributed in part to the contribution of the second allele.<sup>[7]</sup> This variability strengthens the case for functional variant validation and for longitudinal natural history registries.

Beyond its clinical features, this case calls attention to the structural inequities that limit diagnostic yield and care coordination for patients in marginalised settings. Addressing these disparities requires not only clinical expertise but investment in culturally competent communication, sustained follow-up, and infrastructure that meets families where they are.

**Differential Diagnosis**

The presentation of severe neonatal hypotonia, arthrogryposis, bulbar dysfunction, and respiratory compromise necessitated a broad differential. Spinal muscular atrophy type I was an early consideration given the degree of motor involvement and respiratory compromise; however, the absence of tongue fasciculations, the presence of congenital upper limb contractures, and survival with a stable, nonprogressive course beyond a decade argued strongly against it. Congenital myasthenic syndrome was considered given the bulbar and respiratory predominance, but weakness has been fixed rather than fatigable, and there has been no clinical response to a trial of pyridostigmine. Congenital muscular dystrophies, including merosin-deficient and LMNA-related forms, were considered given the fatty infiltration on muscle MRI; the static course, the absence of the marked creatine kinase elevation typical of merosin deficiency, and the reported TTN result argued against these, although targeted exclusion could not be re-confirmed from primary records. MTM1-related myotubular myopathy was considered given the centronuclear histopathology that may occur in titinopathy, but X-linked inheritance and the absence of ptosis or ophthalmoplegia make this unlikely in this female patient. The reported biallelic TTN

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truncating variants, together with the clinical and imaging profile and the familial segregation pattern, remain the most parsimonious explanation for the phenotype observed.

**Limitations**

Several limitations merit acknowledgement. Most significantly, the original genetic sequencing data and muscle biopsy histopathology from early childhood were not retained by the family and could not be recovered from the originating service, precluding confirmation of the precise variants, their position within TTN, and their predicted impact on cardiac isoforms. This constrains the report in two specific ways: the diagnosis of congenital titinopathy is clinical rather than molecularly re-confirmed, and the patient cannot be placed within the genotype-based cardiac risk categories described in the literature. Resequencing was not available at the time of this report and remains outstanding. Second, the muscle MRI finding of generalised fatty infiltration is supportive but non-specific, and no formal comparison against the described selective involvement patterns was undertaken. Third, this is a single-centre report; the observations regarding healthcare inequity, while consistent with published literature on Irish Traveller health outcomes, reflect a singular clinical encounter and cannot be generalised without caution. Fourth, the intrafamilial phenotypic comparison is limited by the absence of formal neurological and functional assessment of the sibling and by the unavailability of the sibling's molecular report, so genotypic concordance is family-reported rather than verified. Finally, the absence of a formal neuropsychological battery means the characterisation of mild intellectual disability rests on clinical impression rather than standardised testing. These limitations notwithstanding, the longitudinal cardiac data are robust: serial echocardiography and ECG over more than a decade support the core observation of a bulbar-dominant titinopathy phenotype without cardiomyopathy to date.

**Patient Perspective**

The patient's mother is the sole primary caregiver and has spent nearly two decades managing the care of two children with complex special needs. Having left school early, she has learned to navigate the medical system through lived experience rather than formal instruction. Her responsibilities include coordinating PEG feeding, BiPAP support, and numerous hospital admissions, often without external assistance. She reports significant difficulties accessing home care nursing, transport, and social supports. Despite these barriers, she maintains a high level of engagement with her daughter's care and draws strength from her extended family and community, which provide emotional support and moments of respite. The patient communicates effectively, with good expressive and receptive language skills, though speech volume is occasionally low. Despite her physical limitations and mild intellectual disability, she remains socially interactive and well-integrated into her immediate support network.

**CONCLUSIONS**

This report describes a bulbar-dominant congenital titinopathy with preserved cardiac function over more than a decade of surveillance, a trajectory that is recognised but sparsely documented. Three learning points emerge. First, bulbar and respiratory morbidity can dominate the clinical picture in congenital TTN-related disease and may occur without concurrent cardiomyopathy; equally, because cardiac risk in titinopathy is both variant-position-dependent and age-penetrant, the absence of cardiac features in childhood does not remove the need for

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lifelong surveillance, particularly where the variant position is unknown. Second, intrafamilial phenotypic variability within the same reported genotype illustrates the limits of current genotype-phenotype models and argues for functional variant validation and longitudinal natural history registries in titinopathy. Third, this case demonstrates the compounding effect of structural healthcare inequity: the loss of diagnostic records, inadequate genetic counselling, and fragmented community support have each had measurable consequences for this patient's care, including the inability to stratify her cardiac risk.

These findings support the need for culturally adapted genomic counselling, particularly in communities where consanguineous union is more frequent and access to genetic services is poorer. Multidisciplinary involvement spanning neurology, respiratory medicine, cardiology, gastroenterology, speech and language therapy, and social work remains essential for optimising outcomes in complex congenital myopathies.

**REFERENCES**

1. Kellermayer D, Smith JE 3rd, Granzier H. Titin mutations and muscle disease. Pflugers Arch. 2019; 471:673-682.
2. Savarese M, Sarparanta J, Vihola A, Udd B, Hackman P. Increasing role of titin mutations in neuromuscular disorders. J Neuromuscul Dis. 2016;3:293-308.
3. Pfeffer G, Elliott HR, Griffin H, et al. Titin mutation segregates with hereditary myopathy with early respiratory failure. Brain. 2012;135:1695-1713.
4. Toro C, Olivé M, Dalakas MC, et al. Exome sequencing identifies titin mutations causing hereditary myopathy with early respiratory failure (HMERF) in families of diverse ethnic origins. BMC Neurol. 2013;13:29.
5. Oates EC, Jones KJ, Donkervoort S, et al. Congenital titinopathy: comprehensive characterization and pathogenic insights. Ann Neurol. 2018;83:1105-1124.
6. Ceyhan-Birsoy O, Agrawal PB, Hidalgo C, et al. Recessive truncating titin gene, TTN, mutations presenting as centronuclear myopathy. Neurology. 2013; 81:1205-14.
7. Savarese M, Vihola A, Oates EC, et al. Genotype-phenotype correlations in recessive titinopathies. Genet Med. 2020;22:2029-2040.
8. Ware JS, Cook SA. Role of titin in cardiomyopathy: from DNA variants to patient stratification. Nat Rev Cardiol. 2018;15:241-252.
9. Carmignac V, Salih MAM, Quijano-Roy S, et al. C-terminal titin deletions cause a novel early-onset myopathy with fatal cardiomyopathy. Ann Neurol. 2007;61:340-351.
10. Chauveau C, Bonnemann CG, Julien C, et al. Recessive TTN truncating mutations define novel forms of core myopathy with heart disease. Hum Mol Genet. 2014;23:980-991.
11. Fernandez-Marmiesse A, Carrascosa-Romero MC, Alfaro Ponce B, et al. Homozygous truncating mutation in prenatally expressed skeletal isoform of TTN gene results in arthrogryposis multiplex congenita and myopathy without cardiac involvement. Neuromuscul Disord. 2017;27:188-192.
12. Barrett P: A review of consanguinity in Ireland - estimation of frequency and approaches to mitigate risks. Ir J Med Sci. 2016;185:17-28.

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13. Report All Ireland Traveller Health Study Team, School of Public Health, Physiotherapy and Population Science, University College Dublin: All Ireland Traveller Health Study - Our Geels: Summary of Findings. Department of Health and Children. Dublin, 2010.
14. Yu M, Zhu Y, Xie Z, et al. Novel TTN mutations and muscle imaging characteristics in congenital titinopathy. Ann Clin Transl Neurol. 2019;6:1311-1318.
15. Ávila-Polo R, Malfatti E, Lornage X, et al. Loss of sarcomeric scaffolding as a common baseline histopathologic lesion in titin-related myopathies. J Neuropathol Exp Neurol. 2018;77:1101-1114.