



## Duchenne muscular dystrophy coexisting with Down syndrome or Turner syndrome: Two case reports

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### Abstract

#### BACKGROUND

Dystrophinopathies are X-linked recessive neuromuscular disorders caused by pathogenic variants in the dystrophin gene (*DMD*). Down syndrome (DS) and Turner syndrome (TS) are well-characterized chromosomal conditions; however, their co-occurrence with monogenic disorders such as Duchenne muscular dystrophy (DMD) is rare and presents unique diagnostic and management challenges.

## CASE SUMMARY

We report two rare cases of DMD coexisting with chromosomal abnormalities, both followed at Siriraj Hospital. The first case involved a 6-year-old boy with DS who presented with incidentally detected elevated serum transaminase levels, calf pseudohypertrophy, and gait difficulty; gene panel testing identified a hemizygous c.3917dup (p.Asp1307Argfs\*4) variant in *DMD*. The second case involved a 6-year-old girl with proximal muscle weakness and calf pseudohypertrophy; genetic studies revealed a heterozygous delins variant in the *DMD* (p.Ala3041Serfs\*69), skewed X-chromosome inactivation, and mosaic TS [45,X(29%)/46,XX].

## CONCLUSION

The coexisting of DMD with DS or TS produces overlapping phenotypes that can complicate diagnosis; careful genetic evaluation and multidisciplinary management are therefore essential.

**Key Words:** Duchenne muscular dystrophy; Dystrophinopathy; Down syndrome; Turner syndrome; Case report

**Core Tip:** This report describes two unprecedented cases of Duchenne muscular dystrophy coexisting with Down syndrome (DS) and Turner syndrome (TS), highlighting the diagnostic challenges at the intersection of chromosomal and monogenic disease. In patient with DS, hypotonia and developmental delay may mask progressive weakness; in females, a heterozygous variant in the dystrophin gene should prompt evaluation for mosaic TS. Next-generation sequencing, guided by clinical suspicion, enabled recognition of these dual genetic diagnoses. These findings broaden the phenotypic spectrum of Duchenne muscular dystrophy and underscore the need for individualized multidisciplinary care.

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## INTRODUCTION

Dystrophinopathies are X-linked recessive neuromuscular disorders caused by pathogenic variants in the dystrophin gene (*DMD*) on chromosome Xp21.2. The phenotypic spectrum comprises two principal forms: Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy. DMD manifests in early childhood with progressive muscle weakness, loss of ambulation, and cardiopulmonary complications. It affects approximately 1 in 5000-6000 live-born males and is the most common inherited muscular dystrophy in children[1]. Because females possess a second functional X chromosome, fully manifesting DMD in females is extremely rare[2-5].

Chromosomal disorders are among the most frequently encountered genetic conditions in clinical practice. Down syndrome (DS), caused by trisomy 21, occurs in approximately 1 in 700 live births and is the most common chromosomal disorder; it is characterized by distinctive craniofacial features, global developmental delay, and comorbidities such as congenital heart defects and hypothyroidism[6]. Turner syndrome (TS), affecting approximately 1 in 2000-2500 live-born females, results from complete or partial monosomy of the X chromosome and typically presents with short stature, gonadal dysgenesis, and characteristic physical stigmata[7].

Although DS and TS are well characterized, their coexistence with a monogenic disorder such as DMD is exceptionally rare and poses unique diagnostic challenges. Only a few cases of DMD in individuals with DS have been reported worldwide[8-11]. Similarly, DMD in individuals with TS is rare, with only a limited number of cases reported in the literature, particularly in those with mosaic TS[12-15]. In this report, we present two rare cases of DMD with underlying chromosomal abnormalities and highlight the diagnostic complexity, genetic implications, and multidisciplinary care requirements.

## CASE PRESENTATION

### Chief complaints

**Case 1:** A 6-year-old boy with DS (47,XY,+21) was referred to the pediatric neurology clinic for persistently elevated serum creatine kinase levels.

**Case 2:** A 6-year-old girl was referred to the pediatric neurology clinic for progressive muscle weakness and elevated serum creatine kinase levels.

### History of present illness

**Case 1:** During hospitalization for symptomatic coronavirus disease 2019 at 5.9 years of age, incidental laboratory testing revealed elevated transaminases [aspartate aminotransferase (AST) 350 U/L; alanine aminotransferase (ALT) 484 U/L]. At follow-up, values remained elevated (AST: 191-698 U/L; ALT: 328-532 U/L). A comprehensive hepatitis workup, including hepatitis B and C serologies, was negative. Abdominal ultrasonography demonstrated mild hepatomegaly with a smooth surface and homogeneous parenchymal echogenicity; liver biopsy findings were normal. Creatine kinase was markedly elevated at 13710 U/L, prompting evaluation for an underlying neuromuscular disorder.

**Case 2:** Gross motor delay was noted at 2 years of age, when she could not walk independently; she achieved independent ambulation at 3 years, but by 6 years she remained unable to climb stairs without assistance. Language and social milestones were unremarkable.

### History of past illness

**Case 1:** DS was diagnosed at birth; comorbidities included global developmental delay with severe intellectual disability, hyperopia, astigmatism, and obstructive sleep apnea.

**Case 2:** No previous systemic illnesses, perinatal complications, or chronic medical conditions were reported.

### Personal and family history

**Case 1:** Two maternal uncles reportedly had childhood-onset muscle weakness of unclear etiology and died at approximately 20 years of age, suggesting a hereditary neuromuscular disorder.

**Case 2:** There was no family history of neuromuscular disease; both nonconsanguineous parents were clinically healthy.

### Physical examination

**Case 1:** Examination revealed typical DS facies, generalized hypotonia, and calf pseudohypertrophy. He moved symmetrically against gravity and exhibited a waddling gait with a positive Gowers sign, indicating proximal muscle weakness. Deep tendon reflexes were 1+, and the Babinski sign was negative.

**Case 2:** Height was 103 cm (below the third percentile), consistent with short stature. She had pectus excavatum but no dysmorphic facial features, webbed neck, or lymphedema. Calf pseudohypertrophy and proximal weakness were present, with a waddling gait and a positive Gowers sign. Deep tendon reflexes were symmetric at 1+, and the Babinski sign was negative.

### Laboratory examinations

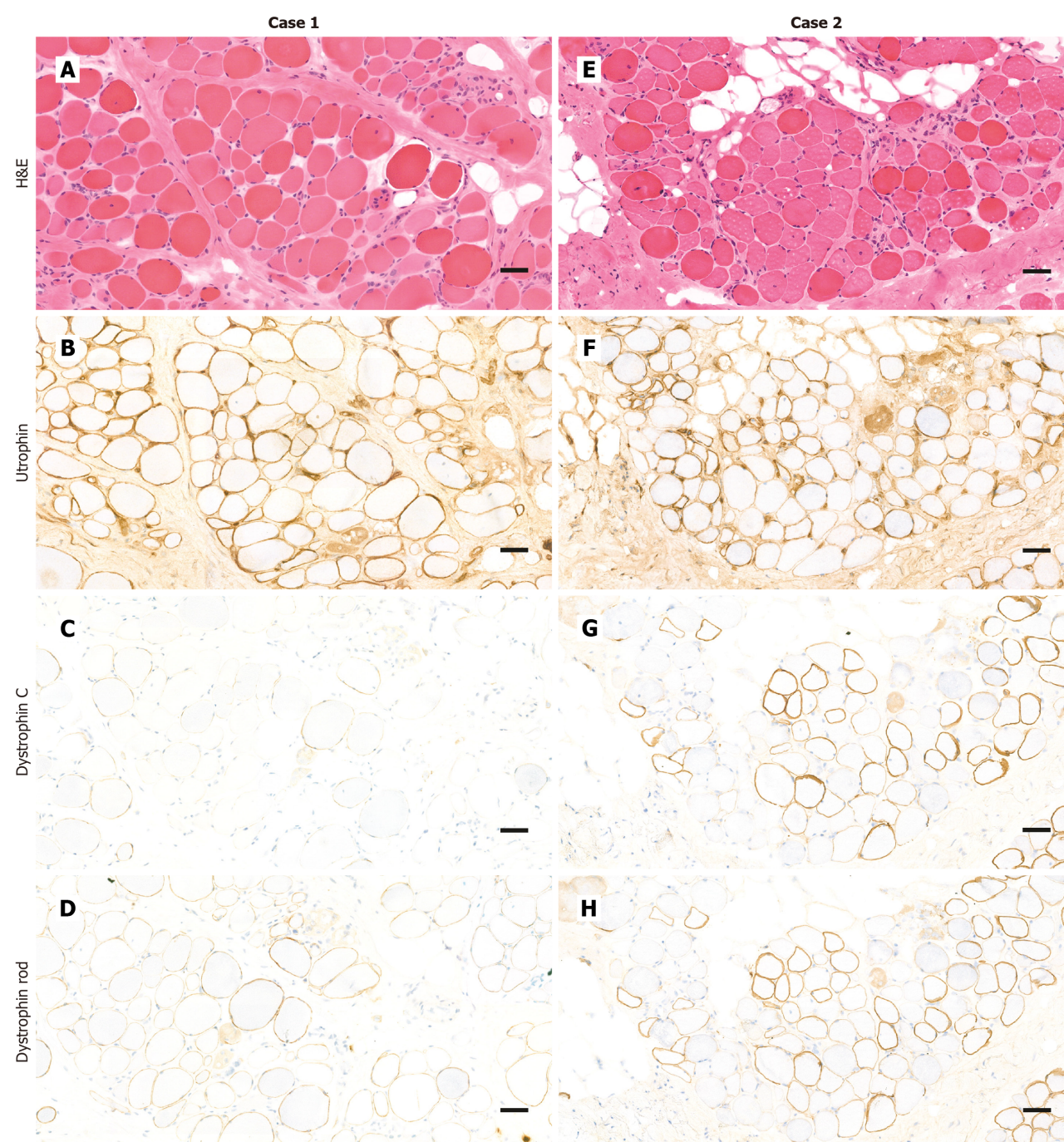
**Case 1:** Multiplex ligation-dependent probe amplification (MLPA) analysis of the *DMD* gene was negative. A comprehensive neuromuscular gene panel identified a hemizygous pathogenic *DMD* variant: c.3917dup (p.Asp1307Argfs\*4). Right quadriceps muscle biopsy demonstrated myopathic changes, including fiber size variation, scattered necrotic and regenerating fibers, and endomysial fibrosis. Immunohistochemistry showed diffuse utrophin positivity and faint-to-negative dystrophin staining (Figure 1A-D, Supplementary Figure 1A), confirming dystrophinopathy consistent with DMD. Subsequent maternal testing revealed heterozygous carrier status for the same *DMD* variant.

**Case 2:** At the referring hospital, creatine kinase was markedly elevated at 15147 U/L. MLPA detected a heterozygous deletion of exon 61 in the *DMD* gene, which was initially not recognized as causative because heterozygous findings in females are typically interpreted as carrier status. She was subsequently referred to Siriraj Hospital for further evaluation. Given her clinical features strongly compatible with DMD, additional investigations were undertaken to confirm the diagnosis in this female patient and to exclude other myopathies that may mimic DMD. Trio whole-genome sequencing, performed to search for other neuromuscular causes, identified a c.9121delinsAGTCCCACATGCAGGGACCGAGTCAG-GCAGCTGCAAGT (p.Ala3041Serfs\*69) variant in *DMD*. Sanger sequencing confirmed the variant in the patient but not in either parent, indicating a *de novo* event. This discordance arose because the variant disrupts the MLPA probe-binding site for exon 61, generating an artifactual deletion signal. Left quadriceps muscle biopsy showed dystrophic changes. Utrophin staining was strongly positive with reciprocal loss of dystrophin, indicating mosaic expression and supporting a diagnosis of DMD consistent with the genetic findings (Figure 1E-H, Supplementary Figure 1B).

To investigate DMD manifestation in a female, X-chromosome inactivation was assessed using the human androgen receptor gene assay targeting the polymorphic CAG repeat in the androgen receptor gene. This methylation-sensitive polymerase chain reaction used *HpaII*, which cleaves unmethylated DNA on the active X chromosome while leaving methylated inactive-X DNA uncleaved[16]. After parental phasing, the patient showed extremely skewed X-chromosome inactivation at 92.53%, with preferential activation of the maternal X chromosome (Figure 2). Peripheral blood karyotyping demonstrated mosaic TS: 45,X[5]/46,XX[45]. Fluorescence in situ hybridization confirmed mosaicism, identifying 45,X.ish X (DXZ1 × 1, SRY × 0) in 29% of cells. Echocardiography and renal ultrasonography were unremarkable.

### Imaging examinations

Not applicable.



**Figure 1 Histopathological and immunohistochemical findings from quadriceps muscle biopsies confirming dystrophinopathy.** A-D: A 6-year-old boy with Duchenne muscular dystrophy (DMD) and Down syndrome (Case 1); E-H: A 6-year-old girl with DMD and mosaic Turner syndrome (Case 2). A: Hematoxylin and eosin (H&E) staining demonstrating myopathic changes, including mild-to-moderate fiber size variation, scattered necrotic and regenerating fibers with clustered regeneration, and mild-to-moderate endomysial fibrosis with adipose tissue infiltration; B: Diffuse utrophin positivity, indicating compensatory upregulation secondary to dystrophin deficiency; C: Absent dystrophin C-terminal (Dys-2) staining in most fibers; D: Faint dystrophin rod domain (Dys-1) staining; E: H&E staining demonstrating dystrophic changes similar to Case 1; F: Biphasic utrophin staining pattern consistent with mosaic dystrophin expression; G: Biphasic Dys-2 staining: Fibers with weak utrophin staining demonstrate dystrophin positivity, whereas fibers with strong utrophin staining are dystrophin-negative; H: Biphasic Dys-1 staining showing a reciprocal pattern to utrophin expression, reflecting mosaic X-chromosome inactivation. Original magnification  $\times 200$ ; scale bar = 20  $\mu\text{m}$ .

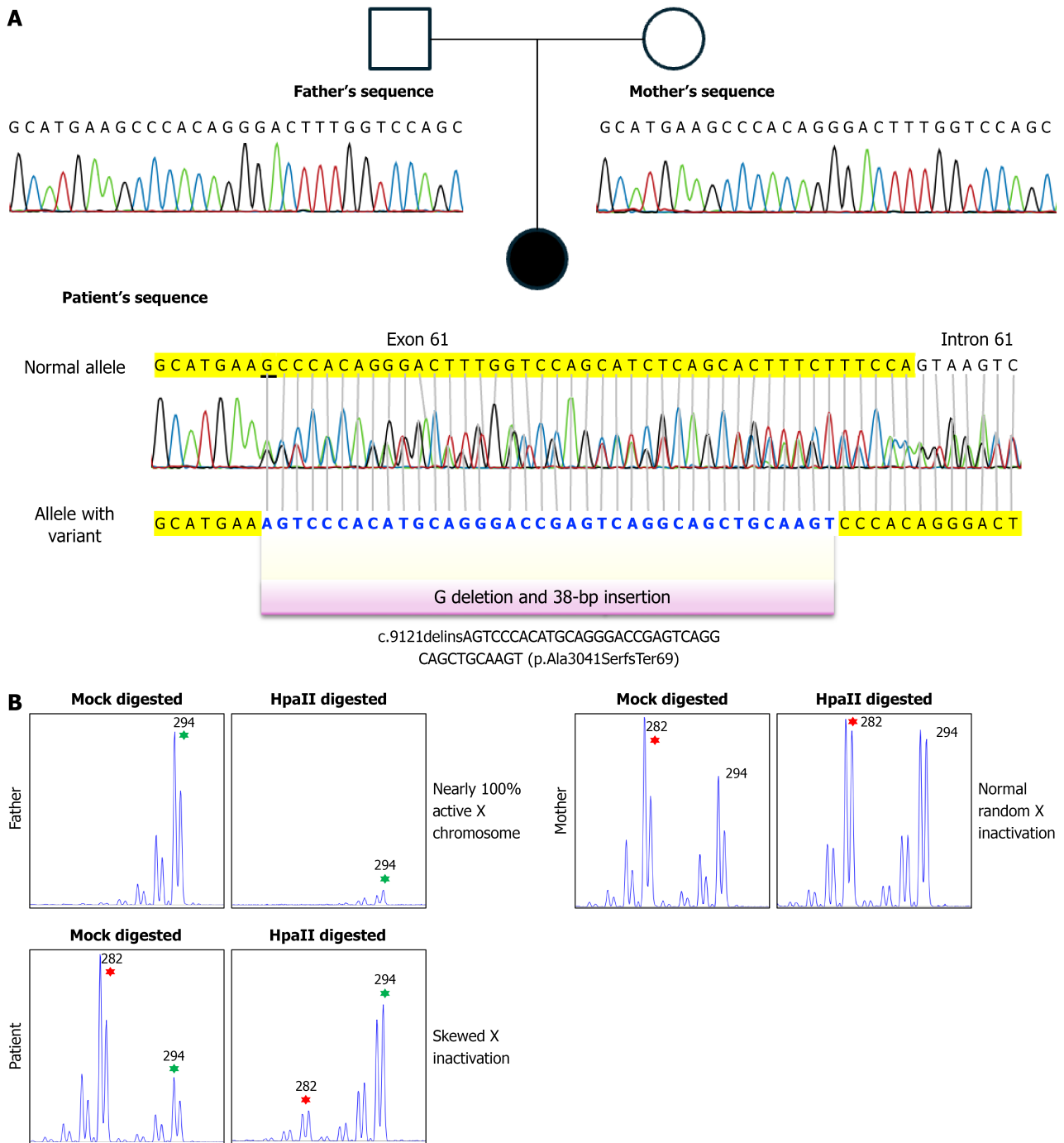
## FINAL DIAGNOSIS

### Case 1

The clinical, histopathological, and molecular findings established a diagnosis of DMD in a child with DS.

### Case 2

A diagnosis of DMD with mosaic TS was established based on clinical, histopathological, and molecular findings.



**Figure 2 Molecular characterization of Case 2 Sanger sequencing confirmation of the dystrophin gene variant and X-chromosome inactivation analysis demonstrating extreme skewing.** A: Sanger sequencing of dystrophin gene exon 61. The patient's sequence (bottom) compared with the reference sequence (top) reveals a complex deletion-insertion (delins) variant, c.9121delinsAGTCCCACATGCAGGGACCGAGTCAGGCAGCTGCAAGT. This variant involves deletion of a single guanine nucleotide at position c.9121 with concomitant insertion of a 38-bp fragment (blue sequence). The resulting frameshift is predicted to generate a premature termination codon, p.Ala3041Serfs\*69. Parental testing confirmed de novo occurrence; B: X-chromosome inactivation (XCI) analysis using the androgen receptor (AR) gene methylation assay. Capillary electrophoresis profiles show HpaII-digested (methylation-sensitive; right panels) and mock-digested (left panels) DNA samples from family members.

Father: Single allele (294 bp) with nearly 100% active X chromosome, as expected for a male. Mother: Balanced representation of both alleles (282 bp and 294 bp) in HpaII-digested samples, indicating normal random X-inactivation. Patient: Extremely skewed XCI (92.53%), with preferential inactivation of the paternal X chromosome. This is evidenced by predominant amplification of the 294-bp paternal allele (green star) and minimal amplification of the 282-bp maternal allele (red star) in HpaII-digested samples. The AR gene methylation assay exploits the polymorphic CAG repeat in exon 1 of the androgen receptor gene. HpaII cleaves unmethylated CpG sites on the active X chromosome, leaving methylated (inactive X) DNA intact for PCR amplification.

## TREATMENT

### Case 1

He has received prednisolone 0.75 mg/kg/day since 6 years of age. He receives multidisciplinary care through a neuromuscular clinic, including regular physical therapy, and follow-up at a genetic clinic for surveillance of disease-related comorbidities.

### Case 2

Prednisolone 0.75 mg/kg/day was initiated after diagnostic confirmation and family counseling. She is followed in a multidisciplinary neuromuscular clinic, with additional follow-up at a genetic clinic for surveillance of disease-related comorbidities.

## OUTCOME AND FOLLOW-UP

### Case 1

At the most recent evaluation at 9.3 years of age, his gait had progressively worsened. He remained ambulatory but fell more frequently and could no longer climb stairs independently.

### Case 2

At the most recent follow-up at 8 years of age, she remained ambulatory and could ascend stairs with handrail support. Height was 110 cm (below the third percentile), consistent with persistent short stature. Cardiac evaluation demonstrated normal function, with a left ventricular ejection fraction of 55% and no congenital heart disease.

## DISCUSSION

We report two rare cases of DMD coexisting with chromosomal abnormalities. DS and TS are well characterized; however, when combined with DMD, clinical presentation becomes more complex, creating diagnostic challenges and implications for long-term management. To date, only four dystrophinopathy cases in individuals with DS have been reported globally (Supplementary Table 1)[8-11], including two with DMD and two with Becker muscular dystrophy. Co-occurrence of TS and DMD is also rare, with only 13 cases reported since 1965, encompassing classic 45,X and mosaic forms (Supplementary Table 2)[5,12-15,17-23].

A review of reported cases reveals a broad spectrum of DMD mutations, including deletions, duplications, and nonsense variants, without a single mutational hotspot, suggesting random co-occurrence rather than a shared mechanism. Our report adds a frameshift variant in a boy with DS (p.Asp1307Argfs\*4) and a delins variant in a girl with mosaic TS (p.Ala3041Serfs\*69), thereby expanding the mutational spectrum associated with these dual diagnoses.

Diagnosis may be delayed when DMD coexists with a chromosomal disorder because clinical features overlap. In our first case, hypotonia and developmental delay, typical of DS, initially obscured the neuromuscular disease. Persistent transaminase elevation, often attributed to autoimmune hepatitis in DS, diverted attention toward hepatic causes and delayed creatine kinase testing. Unlike the generally nonprogressive hypotonia of DS, DMD produces progressive weakness, a positive Gowers sign, and calf hypertrophy. Therefore, any boy with DS who demonstrates motor regression, frequent falls, or a positive Gowers sign with markedly elevated creatine kinase warrants prompt evaluation for dystrophinopathy.

In our second case, diagnosing DMD in a girl required particular care because X-linked inheritance typically affects males. In girls presenting with progressive weakness, calf pseudohypertrophy, and markedly elevated creatine kinase, limb-girdle muscular dystrophies, such as sarcoglycanopathies, are often considered first, as females are usually regarded as carriers rather than affected individuals in DMD. Moreover, when MLPA identifies a single-exon deletion, confirmation with a secondary testing method is essential to exclude a false-positive result caused by probe-binding failure due to a small sequence variant[1].

Nevertheless, DMD can occur in females with X-chromosome monosomy, skewed X-chromosome inactivation, or balanced X-autosomal translocations. Assessment for sex chromosome abnormalities, such as karyotyping, may be considered in female patients with a DMD phenotype (so-called “DMD-like” girls), particularly when clinical features are strongly suggestive of DMD, as chromosomal abnormalities-most notably balanced X-autosome translocations-can lead to complete skewing of X-inactivation of the normal X chromosome and result in a DMD phenotype[24].

In classic TS (45,X), short stature and characteristic features accompanied by progressive weakness and calf pseudohypertrophy may prompt early consideration of DMD. Mosaic TS may present with subtler findings, delaying diagnosis. The phenotype in mosaic TS appears to be influenced by both mosaicism level and X-chromosome inactivation pattern. Our patient, with 29% mosaicism, had markedly elevated creatine kinase, whereas a previously reported case with 1% mosaicism showed lower levels[14]. Although higher degrees of mosaicism may contribute to higher enzyme levels, variability across tissues and X-inactivation skewing limits the utility of mosaicism percentage and creatine kinase levels as predictors of severity.

DMD in a child with DS introduces management complexity. Glucocorticoids remain foundational therapy for DMD; however, in DS, steroid-associated weight gain can exacerbate preexisting obstructive sleep apnea. Intellectual disability, communication difficulties, and behavioral issues may impair treatment adherence and limit engagement in physical therapy, potentially affecting outcomes.

Management of TS with coexisting DMD also requires tailored strategies. Short stature, common in TS, may be exacerbated by long-term glucocorticoid therapy, and obesity risk may be further increased. Cardiomyopathy attributable to DMD can compound congenital heart defects frequently seen in TS, including bicuspid aortic valve, aortic root dilatation, and coarctation of the aorta. These overlapping cardiovascular risks underscore the need for individualized care and vigilant cardiac surveillance. Although reported TS-DMD cases remain few, current data suggest that proximal weakness and creatine kinase elevation do not differ substantially between classic and mosaic TS; however, long-term outcomes, such as age at loss of ambulation may be more favorable in mosaic TS.

## CONCLUSION

Although the co-occurrence of DMD with chromosomal abnormalities such as DS and TS is exceedingly rare, overlapping clinical manifestations can obscure the diagnosis. Advances in diagnostic technologies, combined with heightened clinical suspicion, now enable earlier recognition of such dual genetic conditions. This report underscores the importance of considering dual diagnoses in children with atypical clinical trajectories and of integrating genomic testing early to improve diagnostic accuracy, refine prognostic assessment, and guide individualized multidisciplinary management.

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## FOOTNOTES

**Specialty type:** Pediatrics

**Country of origin:** Thailand

**Author contributions:** Pongsakornkullachart P and Kulsirichawaroj P equally contributed to conceptualization; Pongsakornkullachart P and Kulsirichawaroj P collected clinical data and managed patients; Pho-Iam T, Yongwattana N, and Dhachpramuk D performed genetic analysis and interpreted data; Tanboon I supervised the pathological studies; Pongsakornkullachart P curated the data, conducted literature review and drafted the manuscript; Dhachpramuk D, Pho-Iam T, Chanvanichtrakool M, Kulsirichawaroj P, and Sanmaneechai O critically reviewed the manuscript; Kulsirichawaroj P supervised the study, provided clinical oversight; All authors have read and approved the final manuscript.

**AI contribution statement:** ChatGPT was used only for minor language editing to improve grammar, clarity and readability. The intellectual content, structure and interpretation remained entirely the work of the authors. No portion of the manuscript (abstract, introduction, material and methods, results, discussion and conclusion) was generated by AI. All scientific content and writing were originally produced by authors. Also, AI tools were not involved in the study design, data analysis or interpretation of results.

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