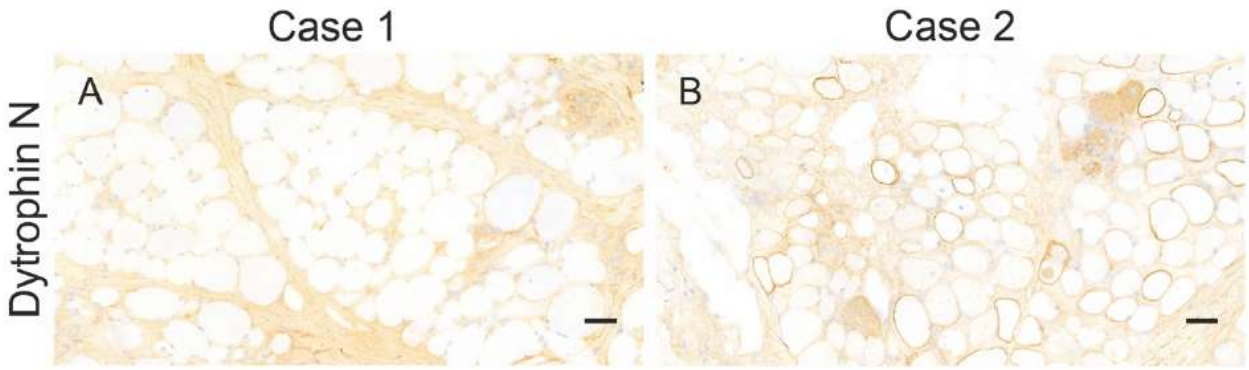

Supplementary Figure 1 Dystrophin N-terminal immunostaining in Case 1 (A) and Case 2 (B). A: Negative staining for dystrophin N-terminal in most muscle fibers; B: Biphasic dystrophin staining pattern. The dystrophin N-terminal image in Case 2 (B) was obtained from the same region shown in Figure 1E–H, although not from consecutive sections, as the staining procedure was performed separately.
Note: Dys-3, dystrophin N-terminal. Original magnification ×200; scale bar = 20 μm.

Supplementary Table 1 Summary of Published Case Reports Describing the Coexistence of Duchene muscular dystrophy/ Becker muscular dystrophy and Down Syndrome

	Sex	Age (y)	AAO (y)	Phenotype	Clinical features	Serum CK	EMG study	Muscle biopsy	Genetic testing for DMD
Moser H, 1971 [1]	M	6	3	DMD	- Unable to walk independently - Calf hypertrophy - Known family with DMD	Elevated	Myopathic pattern	Advance stage muscular dystrophy	N/A
Sakai K *et al*, 1993 [2]	M	22	12	BMD	Progressive muscle wasting	545 IU/L	N/A	Marked dystrophic change Immunohistochemical study: patchy immunostaining for dystrophin	MLPA for dystrophin: deletion in exon 45 – 47 of DMD gene
Lerario A *et al*, 2016 [3]	M	8	6	BMD	- No muscle weakness or fatigability over 6-month follow up period - Incidentally elevated serum CK	1775 IU/L	Myopathic pattern	Moderate dystrophic pattern Immunohistochemical study: normal DYS-1, DYS-2 staining with faint DYS-3 staining and few dystrophin negative fibers	- MLPA for dystrophin: negative - Sequence analysis: splice-site mutation c.1812+1G>A in intron 15
Cabradilla R *et al*, 2017 [4]		9	2	DMD	- Progressive proximal muscle weakness - Gowers sign - Calf hypertrophy	7655 IU/L	N/A	N/A	MLPA for dystrophin: hemizygous deletion in exon 51-54 of DMD gene
Our case	M	8	5	DMD	- Gross motor developmental delayed - Calf hypertrophy - Incidentally elevated serum transaminase and serum CK	13710 IU/L	N/A	Dystrophic pattern Immunohistochemical study: negative DysN and DysC, faintly positive DysRD, diffuse positivity Utrophin	- MLPA for dystrophin: negative - Sequence analysis: Hemizygous pathogenic variant of the DMD gene: c.3917dup(p.Asp1307Argfs*4)

AAO, Age at onset; BMD, Becker muscular dystrophy; CK, creatine kinase; DMD, Duchenne muscular dystrophy; EMG, electromyography; IU/L, international units per liter; N/A, not available; y, year(s).

Supplementary Table 2 Summary of Published Case Reports Describing the Coexistence of Duchene muscular dystrophy and Turner Syndrome

	Age at diagnosis (y)		AAO (y)	Age at loss of ambulation (y)	Karyotype	% 45/X	TS features	DMD features	Serum CK	Muscle biopsy	Genetic testing for DMD
	TS	DMD	DMD								
Ferrier *et al*, 1965 [5]	7.5	6	6	N/A	45,X / 46,XX	38	-Facial dysmorphism -Pigmented nevi	-Walking difficulties -Gowers sign -Family DMD history	Elevated	Muscle fiber necrosis	N/A
Bortolini *et al*, 1986 [6]	4.4	4.4	3	Not yet at 7	45,X/ 46,XX/ 47,XXX	36	-Facial dysmorphism -Short stature	-Proximal muscle weakness -Calf hypertrophy -Gowers sign -Family DMD history	Elevated (485 SU)	Degenerating fibers with proliferation of connective tissue	N/A
Chelly *et al*, 1986 [7]	2.2	2.2	1	9	45, X	-	-Facial dysmorphism -Short stature -Pigmented nevi -No sign of puberty	-Delay motor milestones -Walking difficulty	Elevated	Compatible with dystrophino-pathy	Deletion in DNA region hybridizing Pert 87 probes (87.1, 87.8, 87.15)
Sano *et al*, 1987 [8]	23	6	5	Not yet at 21	45,X/ 46,XX	1	-Short stature -Normal external and internal genitalia	-Walking difficulty -Calf hypertrophy -Gowers sign -Family DMD history	4130 IU/L	Necrotic and regenerating fibers	N/A
Kinoshita, *et al*. 1990 [9]	53	53	52	Not yet at 53	45,X/ 46,XX/ 47,XXX	N/A	N/A	-Muscle weakness -Family DMD history	Elevated	Compatible with muscular dystrophy	N/A
Ou *et al*, 2010 [10]	4	4	10 m	N/A	46, X, i(X)(q10)	-	No abnormalities	-Delay motor milestones -Walking difficulty -Calf hypertrophy -Proximal muscle weakness	9487 IU/L	N/A	Deletion of exons 46, 47 in DMD gene
Kaczorowska *et al*, 2016 [11]	Neonatal period	1.5	6 m	Not yet at 4.5	45, X	-	-Facial dysmorphism -Wide nipples -Feet lymphedema -Aortic coarctation -Moderate cognitive impaired	-Delay motor milestones -Walking difficulties -Gowers sign -Calf hypertrophy	20451 IU/L	Myopathic pattern with significant decrease of dystrophin expression	Frameshift variant c.9055delG (p.D3019Tfs*2) of exon 60 in *DMD* gene
Verma *et al*, 2017 [12]	0.33	5	5	9	45, X	-	-Facial dysmorphism -Short stature -Broad chest with wide nipples	-Walking difficulty -Gowers sign -Calf hypertrophy	4504 IU/L	N/A	Deletion of exon 51 in *DMD* gene

	Age at diagnosis (y)		AAO (y)	Age at loss of ambulation (y)	Karyotype	% 45/X	TS features	DMD features	Serum CK	Muscle biopsy	Genetic testing for DMD
	TS	DMD	DMD								
							-Lymphedema of hands and feet	-Proximal muscle weakness			
Wu *et al*, 2019 [13]	8	4	N/A	N/A	46, X, i(X)(q10)	-	-Cubitus valgus	-Walking difficulties	N/A	N/A	N/A
Kesavan *et al*, 2019 [14]	10	10	3	9	45, X	-	-Short stature -Short neck -Low posterior hair line	-Walking difficulties -Calf hypertrophy	Elevated	N/A	Deletion of exon 45 - 50 in *DMD* gene
Chen *et al*, 2020 [15]	9	9	9	N/A	45,X (50)	-	-Facial dysmorphism -Pigmented nevi -Shield chest -Inverted nipple -Strabismus -Cubitus valgus -Nail hypoplasia -Short 5th metacarpal -Lymphedema of hands and feet -Short stature -Low posterior hair line -Mild Cognitive impaired	-Walking difficulties -Gowers sign -Calf hypertrophy	6566 IU/L	N/A	Frameshift variant c.10273delT (p.S3245Pfs*20) of exon 72 in *DMD* gene
Patil *et al*, 2021 [16]	5	8	N/A	N/A	45 X/46XrX	-	-Short stature -Short neck -Low posterior hair line -Strabismus -Scoliosis	-Walking difficulties -Proximal muscle weakness -Gowers sign -Calf hypertrophy	16012 IU/L	N/A	Deletion of exons 46-52 in *DMD* gene
Our case	6	6	3	Not yet at 8	45,X/ 46,XX	29	-Short stature -Pectus excavatum	-Delay motor milestones -Walking difficulties -Gowers sign -Calf hypertrophy	15147 IU/L	Necrotic and regenerating fibers biphasic pattern of strongly positive and negative	Delins variant c.9121delinsAGTCCCACATGCAGGGACCGAGTCAGGCAGCTGCAAGT (p.A3041Sfs*69) of exon 61 in *DMD* gene

	Age at diagnosis (y)		AAO (y)	Age at loss of ambulation (y)	Karyotype	% 45/X	TS features	DMD features	Serum CK	Muscle biopsy	Genetic testing for DMD
	TS	DMD	DMD								
										dystrophin fibers	

AAO, Age at onset; CK, creatine kinase; DMD, Duchenne muscular dystrophy; international units per liter, IU/L; m, month(s); N/A, not available; SU, Somogyi units; TS, Turner syndrome; y, year(s).

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