

Supplementary Table S1

Authors	Phenotype	Patients	Genomic Breakpoints(hg19/GRCH37)	Notes
Banerjee et al.	DMD	2	Unknown	
Giliberto et al.	DMD	1	Unknown	Dystrophin Absent on WB
Mital et al.	DMD	3, (DG, KS, RI)	Unknown	we note for KS and RI that southern blots were normal
Zamani et al.	DMD	1	Unknown	
Deepha et al.	DMD	3 (205,206,210)	Unknown	
	BMD	1 (209)		
UMD-DMD	Unknown	3 (ID :17760,17686, 17572)	Unknown	Origins : China
Brisson et al.	Likely DMD	Family ID: 6	g.(?_31,854,529)_(31,877,889_ ?)	NIPS then verified by CGH array in a male foetus
Marey et al.	BMD	1 (Patient #5272)	g.31839857_31892375del	Only deletion with precise genomic breakpoints
Yun et al.	BMD	2 (ID: 15, 15-1)	Unknown	
Clinvar (Baylor Genetics)	BMD	Variant ID : 625643	Unknown	Clinvar (Baylor Genetics)
Clinvar (invitae)	DMD	Variant ID : 833063	Unknown	Clinvar (invitae)
LOVD	BMD	Individual: 00135563		
	Unknown (Pathogenic?)	Individuals: 00137264,00239877,00297283,00297696,00287744,00297304,00297717		

List of reported isolated deletion of exon 49 in literature, in total : 28 isolated deletion of exon 49