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Supp. Table S1. List of dystrophin nonsense mutations used in this study

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IDENTIFIERS AND PHENOTYPE			MUTATION			
Subject ID	Diagnosis	Kindred ID#	Type	Exon	cDNA[a]	Protein
42970	**BMD**	**9**	**UGA**	**1**	**c.9G>A**	**p.Trp3X**
DR43194	**BMD**	**9**	**UGA**	**1**	**c.9G>A**	**p.Trp3X**
DR43043A	**BMD**	**24**	**UGA**	**1**	**c.9G>A**	**p.Trp3X**
DR43640	**BMD**	**24**	**UGA**	**1**	**c.9G>A**	**p.Trp3X**
DR43738	**BMD**	**204**	**UGA**	**1**	**c.9G>A**	**p.Trp3X**
DR43741	**BMD**	**204**	**UGA**	**1**	**c.9G>A**	**p.Trp3X**
DR43800	**BMD**		**UGA**	**1**	**c.9G>A**	**p.Trp3X**
DR43676	**BMD**		**UGA**	**1**	**c.9G>A**	**p.Trp3X**
42969	DMD	61	UAG	5	c.355C>T	p.Gln119X
OSU411	DMD	61	UAG	5	c.355C>T	p.Gln119X
DR43272	DMD		UGA	6	c.429G>A	p.Trp143X
DC0058	DMD		UGA	6	c.433C>T	p.Arg145X
DR43239	DMD		UGA	6	c.433C>T	p.Arg145X
DR43486	DMD		UGA	6	c.433C>T	p.Arg145X
DR43490	DMD		UGA	6	c.440C>G	p.Ser147X
DC0235	DMD		UAG	6	c.457C>T	p.Gln153X
OSU474	DMD		UAG	7	c.565C>T	p.Gln189X
DC0285	DMD		UAG	7	c.568C>T	p.Gln190X
DC0256	DMD		UAA	7	c.580C>T	p.Gln194X
DR43333	DMD		UGA	7	c.583C>T	p.Arg195X
DR43392	DMD		UGA	7	c.583C>T	p.Arg195X
42985	DMD		UAA	8	c.709C>T	p.Gln237X
DC0115	DMD		UAG	8	c.745C>T	p.Gln249X
DC0110	DMD		UAG	8	c.799C>T	p.Gln267X
DR43162	DMD	38	UAG	9	c.903C>G	p.Tyr301X
DR43163	DMD	38	UAG	9	c.903C>G	p.Tyr301X
DR43518	DMD		UAA	10	c.1012G>T	p.Glu338X
DR43396	DMD		UAA	10	c.1093C>T	p.gln365X
DR43144	DMD		UAA	10	c.1117G>T	p.Glu373X
DR43116	DMD		UAA	11	c.1255G>T	p.Glu419X
DC0457	DMD	215	UAG	11	c.1267C>T	p.Gln423X
DC0068	DMD		UAA	11	c.1286C>A	p.Ser429X
DR43362	DMD		UAG	12	c.1388G>A	p.Trp463X
DC0281	DMD		UAG	12	c.1429G>T	p.Glu477X
DC0002	DMD	14	UAA	12	c.1465C>T	p.Gln489X
DR43483	DMD		UGA	14	c.1615C>T	p.Arg539X
DC0001	DMD		UGA	14	c.1615C>T	p.Arg539X
OSU398	DMD		UAG	14	c.1619G>A	p.Trp540X

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IDENTIFIERS AND PHENOTYPE			MUTATION			
Subject ID	Diagnosis	Kindred ID#	Type	Exon	cDNA[a]	Protein
DR43225	DMD		UAA	14	c.1663C>T	p.Gln555X
OSU396	DMD		UAA	14	c.1663C>T	p.Gln555X
DR43689	DMD		UAG	14	c.1702C>T	p.Gln568X
DR43109	DMD	40	UAA	15	c.1783G>T	p.Glu595X
DC0063	DMD		UGA	15	c.1793C>G	p.Ser598X
43018	DMD	12	UAA	16	c.1961T>A	p.Leu654X
DR43759	DMD		UAA	16	c.1966C>T	p.Gln656X
OSU414	DMD		UAG	16	c.1990C>T	p.Gln664X
DC0419	DMD		UAG	17	c.2077C>T	p.Gln693X
OSU353	DMD		UAG	19	c.2299G>T	p.Glu767X
DC0435	DMD	217	UAG	19	c.2317A>T	p.Lys773X
DR43595	DMD		UAA	20	c.2404A>T	p.Lys802X
DR43766	DMD		UAA	20	c.2416G>T	p.Glu806X
DR43607	DMD	213	UAG	20	c.2479G>T	p.Glu827X
43016	DMD		UAG	20	c.2479G>T	p.Glu827X
DR43249	DMD		UAG	20	c.2485C>T	p.Gln829X
DC0149	DMD	108	UAA	20	c.2521C>T	p.Gln841X
DR43721	DMD		UGA	20	c.2582C>G	p.Ser861X
DC0278	DMD	194	UAA	20	c.2611A>T	p.Lys871X
DC0220	DMD		UAA	21	c.2758C>T	p.Gln920X
DR43688	DMD		UAA	21	c.2788A>T	p.Lys930X
43015	DMD		UGA	22	c.2862G>A	p.Trp954X
DR43303	DMD		UAG	23	c.2991C>G	p.Tyr997X
DR43212	DMD	75	UAA	23	c.3121C>T	p.Gln1041X
D46952	DMD		UAG	24	c.3217G>T	p.Glu1073X
OSU364	DMD		UAA	24	c.3220G>T	p.Glu1074X
DR43228	DMD	86	UAG	24	c.3224G>A	p.Trp1075X
43039	DMD		UAG	24	c.3268C>T	p.Gln1090X
DC0257	DMD		UGA	25	c.3281T>G	p.Leu1094X
DR43302	DMD		UAG	25	c.3304C>T	p.Gln1102X
DC0366	**BMD**		**UAG**	**25**	**c.3340A>T**	**p.Lys1114X**
OSU341	DMD		UAG	25	c.3430C>T	p.Gln1144X
43013	DMD	4	UGA	26	c.3500C>G	p.Ser1167X
43014	DMD	4	UGA	26	c.3500C>G	p.Ser1167X
DR43223	IMD		UGA	26	c.3500C>G	p.Ser1167X
42994	DMD	11	UAG	26	c.3544G>T	p.Glu1182X
DR43501	DMD	193	UAG	26	c.3580C>T	p.Gln1194X
DC0153	DMD		UAA	27	c.3622C>T	p.Gln1208X
DR43327	**BMD**		**UAG**	**27**	**c.3700G>T**	**p.Glu1234X**
DR43282	DMD		UGA	28	c.3804G>A	p.Trp1268X
DR43480	IMD	121	UAA	28	c.3822C>A	p.Tyr1274X

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IDENTIFIERS AND PHENOTYPE			MUTATION			
Subject ID	Diagnosis	Kindred ID#	Type	Exon	cDNA[a]	Protein
DR43481	IMD	121	UAA	28	c.3822C>A	p.Tyr1274X
DR43147	DMD		UAA	28	c.3868A>T	p.Lys1290X
DRVH43377	**BMD**	**87**	**UAG**	**29**	**c.3982C>T**	**p.Gln1328X**
DRVH43376	**BMD**	**87**	**UAG**	**29**	**c.3982C>T**	**p.Gln1328X**
43017	**BMD**		**UAG**	**29**	**c.4012G>T**	**p.Glu1338X**
DC0268	DMD		UAG	30	c.4108C>T	p.Gln1370X
43040	DMD		UAG	30	c.4150G>T	p.Glu1384X
DR43174	IMD		UAA	30	c.4213C>T	p.Gln1405X
42719	**BMD**		**UAA**	**31**	**c.4240C>T**	**p.Gln1414X**
42588	**BMD**	**32**	**UAG**	**31**	**c.4250T>A**	**p.Leu1417X**
DR43140	**BMD**	**32**	**UAG**	**31**	**c.4250T>A**	**p.Leu1417X**
DR43139	**BMD**	**32**	**UAG**	**31**	**c.4250T>A**	**p.Leu1417X**
DR43217	**BMD**		**UAA**	**31**	**c.4285A>T**	**p.Lys1429X**
DR43661	DMD		UAA	32	c.4352T>A	p.Leu1451X
DC0201	DMD		UAG	32	c.4405C>T	p.Gln1469X
DC0030	DMD	44	UAA	32	c.4414C>T	p.Gln1472X
DR43735	DMD		UAG	32	c.4483C>T	p.Gln1495X
42964	DMD	8	UAG	34	c.4693C>T	p.Gln1565X
42986	DMD		UAG	34	c.4693C>T	p.Gln1565X
DR43193	DMD		UAG	34	c.4693C>T	p.Gln1565X
DC0455	DMD		UAG	34	c.4697T>A	p.Leu1566X
43021	DMD	174	UGA	34	c.4729C>T	p.Arg1577X
DC0241	DMD		UAA	34	c.4793C>A	p.Ser1598X
DC0302	DMD		UAG	35	c.4864G>T	p.Glu1622X
OSU312	DMD		UAG	35	c.4870C>T	p.Gln1624X
DR43580	DMD		UGA	35	c.4996C>T	p.Arg1666X
DR43587	DMD		UAA	36	c.5134C>T	p.Gln1712X
DR43651	DMD	180	UAA	36	c.5140G>T	p.Glu1714X
DR43650	DMD	180	UAA	36	c.5140G>T	p.Glu1714X
DC0006	DMD	23	UGA	37	c.5255T>G	p.Leu1752X
DR43544	**BMD**	**141**	**UGA**	**37**	**c.5287C>T**	**p.Arg1763X**
DR43543	**BMD**	**141**	**UGA**	**37**	**c.5287C>T**	**p.Arg1763X**
DR43542	**BMD**	**141**	**UGA**	**37**	**c.5287C>T**	**p.Arg1763X**
DC0171	DMD		UAA	38	c.5344G>T	p.Glu1782X
DR43317	**BMD**		**UAA**	**38**	**c.5371C>T**	**p.Gln1791X**
43024	**BMD**	**5**	**UAA**	**38**	**c.5398G>T**	**p.Glu1800X**
43023	**BMD**	**5**	**UAA**	**38**	**c.5398G>T**	**p.Glu1800X**
DR43153	**BMD**		**UAG**	**38**	**c.5404C>T**	**p.Gln1802X**
DR43198	DMD		UGA	39	c.5530C>T	p.Arg1844X
DR43184	DMD	76	UAG	39	c.5563C>T	p.Gln1855X
DC0123	DMD	126	UAA	40	c.5608A>T	p.Lys1870X

IDENTIFIERS AND PHENOTYPE			MUTATION			
Subject ID	Diagnosis	Kindred ID#	Type	Exon	cDNA[a]	Protein
DC0059	DMD	51	UGA	40	c.5637G>A	p.Trp1879X
DC0303	DMD		UAA	40	c.5646C>A	p.Tyr1882X
DC0283	DMD		UAG	40	c.5653C>T	p.Gln1885X
DC0165	**BMD**		**UAA**	**40**	**c.5725G>T**	**p.Glu1909X**
DC0239	DMD	142	UAA	41	c.5752G>T	p.Glu1918X
43003	DMD	36	UAG	41	c.5758C>T	p.Gln1920X
DC0367	DMD	36	UAG	41	c.5758C>T	p.Gln1920X
DR43124	DMD		UAG	41	c.5800G>T	p.Glu1934X
DC0282	DMD	223	UAG	41	c.5867G>A	p.Trp1956X
43005	DMD	17	UGA	41	c.5899C>T	p.Arg1967X
DC0159	DMD		UGA	41	c.5899C>T	p.Arg1967X
OSU423	DMD		UGA	41	c.5899C>T	p.Arg1967X
DR43271	DMD		UGA	41	c.5902A>T	p.Arg1968X
OSU382	DMD		UAA	41	c.5917C>T	p.Gln1973X
DC0166	DMD	122	UAG	42	c.6103G>T	p.Glu2035X
DC0167	DMD	122	UAG	42	c.6103G>T	p.Glu2035X
DC0166	DMD	122	UAG	42	c.6103G>T	p.Glu2035X
OSU391	DMD		UAG	42	c.6103G>T	p.Glu2035X
DC0077	**BMD**		**UGA**	**43**	**c.6255G>A**	**p.Trp2085X**
DR43291	DMD	65	UGA	43	c.6283C>T	p.Arg2095X
DR43408	DMD		UGA	44	c.6292C>T	p.Arg2098X
DC0432	DMD		UAG	44	c.6352C>T	p.Gln2118X
DR43306	DMD	70	UAA	44	c.6391C>T	p.Gln2131X
DR43049A	DMD	70	UAA	44	c.6391C>T	p.Gln2131X
DC0260	DMD		UAG	45	c.6445C>T	p.Gln2149X
DC0019	DMD		UAG	45	c.6460C>T	p.Gln2154X
DR43737	DMD		UAG	45	c.6544C>T	p.Gln2182X
DC0192	DMD		UAA	46	c.6641C>A	p.Ser2214X
DC0377	DMD	212	UAA	46	c.6649C>T	p.Gln2217X
43030	DMD		UAA	47	c.6856G>T	p.Glu2286X
42172	DMD		UAA	47	c.6868A>T	p.Lys2290X
DR43447	DMD		UAA	48	c.7075C>T	p.Gln2359X
DR43441	**BMD**		**UAG**	**51**	**c.7401_7402delGGinsAT**	**p.Glu2468X**
DC0040	DMD	84	UAG	52	c.7576C>T	p.Gln2526X
DC0044	DMD		UGA	52	c.7657C>T	p.Arg2553X
DR43247	DMD		UAG	53	c.7682G>A	p.Trp2561X
DR43723	DMD		UAA	53	c.7693C>T	p.Gln2565X
42962	DMD		UAG	53	c.7720C>T	p.Gln2574X
DR43482	DMD		UGA	53	c.7814C>G	p.Ser2605X
DC0045	DMD		UAG	53	c.7822G>T	p.Glu2608X

IDENTIFIERS AND PHENOTYPE			MUTATION			
Subject ID	Diagnosis	Kindred ID#	Type	Exon	cDNA[a]	Protein
DC0306	DMD		UGA	55	c.8038C>T	p.Arg2680X
DC0194	DMD	158	UAG	55	c.8146C>T	p.Gln2716X
DC0264	DMD		UAG	55	c.8161A>T	p.Lys2721X
DC0234	**BMD**		**UAG**	**56**	**c.8353A>T**	**p.Lys2785X**
DC0114	DMD		UAG	56	c.8357G>A	p.Trp2786X
DR43219	DMD		UAA	56	c.8371A>T	p.Lys2791X
D06332	DMD		UGA	57	c.8460G>A	p.Trp2820X
DR43363	DMD		UAG	57	c.8464C>T	p.Gln2822X
DR43170	DMD		UAG	57	c.8491C>T	p.Gln2831X
DC0183	DMD		UAG	57	c.8527A>T	p.Lys2843X
42968	DMD	188	UGA	58	c.8608C>T	p.Arg2870X
OSU370	DMD		UGA	58	c.8608C>T	p.Arg2870X
DR43692	DMD	197	UGA	59	c.8713C>T	p.Arg2905X
43022	DMD		UGA	59	c.8713C>T	p.Arg2905X
OSU373	DMD		UGA	59	c.8713C>T	p.Arg2905X
43009	DMD		UGA	59	c.8775G>A	p.Trp2925X
DR43493	DMD		UGA	59	c.8872G>T	p.Gly2958X
OSU362	DMD		UGA	60	c.8944C>T	p.Arg2982X
DR43186	DMD	249	UAG	60	c.9082C>T	p.Gln3028X
DR43145	DMD	35	UGA	61	c.9100C>T	p.Arg3034X
DC0083	DMD		UAG	62	c.9182G>A	p.Trp3061X
42953	DMD		UGA	64	c.9337C>T	p.Arg3113X
DC0029	DMD		UGA	64	c.9337C>T	p.Arg3113X
DR43273	DMD		UGA	64	c.9337C>T	p.Arg3113X
OSU363	DMD		UAG	64	c.9346C>T	p.Gln3116X
43007	DMD	62	UGA	65	c.9380C>G	p.Ser3127X
DR43229	DMD	62	UGA	65	c.9380C>G	p.Ser3127X
42992	DMD		UGA	65	c.9380C>G	p.Ser3127X
OSU346	DMD		UAG	65	c.9427C>T	p.Gln3143X
DR43450	DMD		UGA	66	c.9568C>T	p.Arg3190X
D42687	DMD		UGA	66	c.9568C>T	p.Arg3190X
OSU407	DMD		UAG	68	c.9928C>T	p.Gln3310X
DR43492	DMD		UAG	68	c.9952G>T	p.Glu3318X
DR43720	DMD		UAA	69	c.10012C>T	p.Gln3338X
DR43244	DMD	220	UGA	69	c.10033C>T	p.Arg3345X
DC0118	DMD		UAG	69	c.9978C>G	p.Tyr3326X
DC0233	DMD	195	UGA	70	c.10108C>T	p.Arg3370X
DR43004A	DMD	205	UGA	70	c.10108C>T	p.Arg3370X
DR43701	DMD	205	UGA	70	c.10108C>T	p.Arg3370X
DR43263	DMD	71	UGA	70	c.10141C>T	p.Arg3381X
35752	DMD		UGA	70	c.10141C>T	p.Arg3381X

IDENTIFIERS AND PHENOTYPE			**MUTATION**			
Subject ID	**Diagnosis**	**Kindred ID#**	**Type**	**Exon**	**cDNA**[a]	**Protein**
OSU387	DMD	259	UGA	70	c.10141C>T	p.Arg3381X
DC0206	DMD		UGA	70	c.10141C>T	p.Arg3381X
DR43771	DMD	259	UGA	70	c.10141C>T	p.Arg3381X
DR43114	DMD	218	UGA	70	c.10171C>T	p.Arg3391X
DR43397	DMD		UGA	70	c.10171C>T	p.Arg3391X
DR43234	DMD		UGA	70	c.10171C>T	p.Arg3391X
DRVH43502	DMD		UAG	76	c.10801C>T	p.Gln3601X
DC0254	**BMD**		**UGA**	**76**	**c.10888C>T**	**p.Arg3630X**
OSU309	DMD		UAA	76	c.10903C>T	p.Gln3635X

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[a] reference GenBank sequence NM_004006.2. Nucleotide numbering reflects cDNA numbering with +1 corresponding to the A of the ATG translation initiation codon in the reference sequence, according to journal guidelines (www.hgvs.org/mutnomen). The initiation codon is codon 1.

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Supp. Table S2. Disruption of ESE or creation of ESS sites by nonsense mutations for in-frame exons associated with BMD phenotypes

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		Disruption of ESE = 1, no effect = 0						Creation of ESS = 1, no effect = 0			
Mutation[a]	Exon (phenotype)	hexESE	ESE Finder matrices	RESCUE ESE hexamers	PESE Octamers from Zhang & Chasin	EIEs from Zhang et al.	ESE motifs from HSF	hexESS	Silencer motifs from Sironi et al.	PESS Octamers from Zhang & Chasin	hnRNP motifs
c.3328G>T	25 (BMD)	1	0	1	0	1	1	0	0	0	0
c.3337C>T	25 (BMD)	1	1	1	1	1	1	1	0	0	1
c.3340A>T	25 (BMD)	1	1	1	1	1	1	0	0	0	1
c.3631G>T	27 (BMD)	1	0	1	1	1	1	1	0	1	1
c.3700G>T	27 (BMD)	1	1	1	1	1	1	0	0	0	0
c.3935T>A	29 (BMD)	0	0	0	0	0	0	0	1	1	1
c.3940C>T	29 (BMD)	0	1	0	0	1	0	0	0	0	0
c.3982C>T	29 (BMD)	1	1	0	0	1	0	0	1	0	1
c.4012G>T	29 (BMD)	1	0	1	0	1	0	0	0	0	1
c.4240C>T	31 (BMD)	1	0	1	0	0	0	0	0	0	0
c.4250T>A	31 (BMD)	0	0	0	0	1	0	0	0	0	1
c.4285A>T	31 (BMD)	1	0	1	1	1	1	0	0	0	0
c.4294C>T	31 (BMD)	1	1	1	0	1	0	1	1	1	0
c.5260G>T	37 (BMD)	0	1	0	0	1	1	1	1	1	1
c.5287C>T	37 (BMD)	0	0	0	0	1	0	0	0	0	0
c.5371C>T	38 (BMD)	0	0	0	0	1	0	1	0	1	0
c.5398G>T	38 (BMD)	1	0	1	0	1	1	0	0	0	0
c.5404C>T	38 (BMD)	1	1	1	0	1	0	0	0	0	1
c.5407C>T	38 (BMD)	1	1	0	0	0	0	1	0	0	0
c.5480T>A	39 (BMD)	0	0	0	0	1	0	1	0	0	1
c.5725G>T	40 (BMD)	1	1	1	0	1	0	0	0	0	1
c.3281T>G	25 (DMD)	0	0	0	0	0	0	0	0	0	0
c.3304C>T	25 (DMD)	1	1	1	1	1	0	0	0	1	1
c.3397G>T	25 (DMD)	1	0	1	0	1	1	0	0	0	0

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Mutation[a]	Exon (phenotype)	hexESE	ESE Finder matrices	RESCUE ESE hexamers	PESE Octamers from Zhang & Chasin	EIEs from Zhang et al.	ESE motifs from HSF	hexESS	Silencer motifs from Sironi et al.	PESS Octamers from Zhang & Chasin	hnRNP motifs
c.3427C>T	25 (DMD)	1	0	0	0	1	1	0	0	0	0
c.3430C>T	25 (DMD)	1	0	0	0	1	1	0	0	0	0
c.3622C>T	27 (DMD)	1	0	0	1	1	0	0	1	0	0
c.3746G>A	27 (DMD)	0	1	0	0	1	0	0	0	0	1
c.5255T>G	37 (DMD)	0	0	0	0	0	0	1	1	0	0
c.5344G>T	38 (DMD)	1	0	1	1	1	1	0	0	0	0
c.5363C>G	38 (DMD)	0	0	0	0	1	0	0	1	0	0
c.5530C>T	39 (DMD)	1	0	1	0	1	1	0	1	0	0
c.5551C>T	39 (DMD)	1	0	1	1	1	0	1	0	0	1
c.5563C>T	39 (DMD)	1	1	1	0	1	0	1	0	0	1
c.5608A>T	40 (DMD)	1	0	1	0	1	1	0	0	0	0
c.5637G>A	40 (DMD)	0	1	0	0	0	0	0	0	0	0
c.5646C>A	40 (DMD)	0	1	1	0	1	0	0	0	0	0
c.5653C>T	40 (DMD)	0	0	0	0	0	0	1	1	0	0
BMD site change		13/21	9/21	11/21	5/21	17/21	8/21	7/21	4/21	5/21	10/21
DMD site change		10/17	5/17	8/17	4/17	13/17	6/17	4/17	5/17	1/17	4/17
P value (Fisher's exact test)		0.49	0.39	0.45	0.61	0.41	0.51	0.34	0.85	0.13	0.11

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[a] reference GenBank sequence NM_004006.2. Nucleotide numbering reflects cDNA numbering with +1 corresponding to the A of the ATG translation initiation codon in the reference sequence, according to journal guidelines (www.hgvs.org/mutnomen). The initiation codon is codon 1.

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