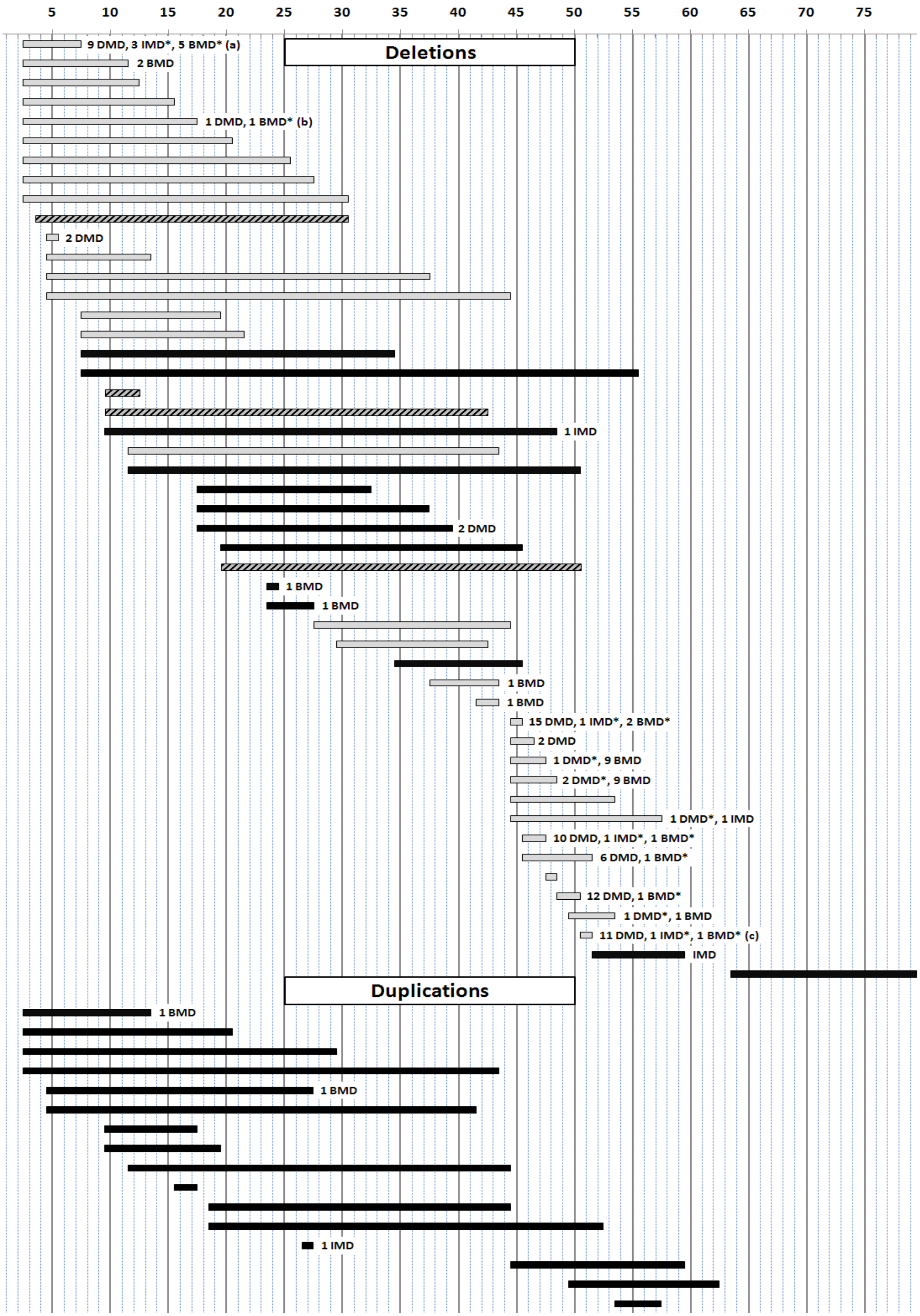
Supp. Figure S1. (next page) Exonic distribution of newly identified deletions and simple duplications as well as deletions that do not conform to the reading-frame rule. Tick marks are exon numbers and each horizontal bar indicates the range of exons involved in the mutation; black bars represent new mutations; gray bars show deletions associated with at least one occurrence of a phenotype not following the reading-frame rule; hatched bars are previously unreported mutations that do not follow the reading-frame rule. Except for those labeled “a”, “b”, and “c”, bar labels indicate the phenotype(s) and the number(s) of unrelated patients sharing the same exonic range; bars without any label represent mutations associated with a single independent occurrence of Duchenne muscular dystrophy. In mutations with more than one phenotype, the non-rule-conforming clinical diagnosis is marked with an asterisk (*). Three deletions were associated with more than one phenotype in the same family, marked as follows: (a) both IMD and BMD phenotypes (kindred 7); (b) both DMD and BMD phenotypes (kindred 54); (c) both DMD and IMD phenotypes (kindred 15).


5
10
15
20
25
30
35
40
45
50
55
60
65
70
75
Deletions
9 DMD, 3 IMD*, 5 BMD* (a)
2 BMD
1 DMD, 1 BMD* (b)
2 DMD
1 IMD
2 DMD
1 BMD
1 BMD
1 BMD
1 BMD
15 DMD, 1 IMD*, 2 BMD*
2 DMD
1 DMD*, 9 BMD
2 DMD*, 9 BMD
1 DMD*, 1 IMD
10 DMD, 1 IMD*, 1 BMD*
6 DMD, 1 BMD*
12 DMD, 1 BMD*
1 DMD*, 1 BMD
11 DMD, 1 IMD*, 1 BMD* (c)
IMD
Duplications
1 BMD
1 BMD
1 IMD

Supp. Table S1 is included as a separate Excel file.

Supp. Table S2. Mutations detected among 858 subjects enrolled in the United Dystrophinopathy Project.

The category of “No Mutation Detected” includes both patients for whom muscle biopsy tissue was not analyzed by RT-PCR (n=37) and patients in whom exhaustive mutation analysis, including sequencing of mRNA, revealed no mutation (n=3). For further discussion see text.

UDP (Phenotyped) SUMMARY

	DMD	**IMD**	**BMD**	**Manifesting Carrier**	**Carrier**	**Unknown**	**Total**	**%**
DELETION	282	15	55	3	13	0	368	48.9%
in	*30*	*2*	*36*	*1*	*2*	*0*	*71*	
out	*242*	*13*	*18*	*1*	*11*	*0*	*285*	
other	*10*	*0*	*1*	*1*	*0*	*0*	*12*	
STOP	113	4	25	2	13	0	157	20.8%
UGA	*48*	*1*	*11*	*2*	*8*	*0*	*70*	
UAG	*36*	*0*	*9*	*0*	*0*	*0*	*45*	
UAA	*29*	*3*	*5*	*0*	*5*	*0*	*42*	
SUBEXONIC	48	0	10	1	5	0	64	8.5%
FS Ins	*15*	*0*	*1*	*1*	*2*	*0*	*19*	
FS Del	*32*	*0*	*4*	*0*	*3*	*0*	*39*	
FS Ins/Del	*1*	*0*	*2*	*0*	*0*	*0*	*3*	
in-frame deletion	*0*	*0*	*3*	*0*	*0*	*0*	*3*	
DUPLICATION	87	7	10	5	5	0	114	15.1%
SPLICE	19	3	7	2	4	0	35	4.6%
MISSENSE	2	1	6	0	0	0	9	1.2%
PSEUDOEXON	0	2	2	0	0	0	4	0.5%
POTENTIAL	2	0	0	0	0	0	2	0.3%
OTHER	0	0	0	0	0	0	0	0.0%
TOTAL MUTATIONS	**553**	**32**	**115**	**13**	**40**	**0**	**753**	**100.0%**
No mutation detected *	25	0	15	0	0	0	40	
Not dystrophinopathy							14	
TOTAL SYMPTOMATIC SUBJECTS							**807**	
Negative Carrier Testing							51	
TOTAL SUBJECTS							**858**	

Supp. Table S3. Distribution of nonsense mutations by target size and target mutability.

Expected Mutations = the target mutability x (number of total nonsense mutations / number of total target codons). The total number of nonsense mutations observed was 243, and the target size of NM_004006 is 1500 codons. **Target codons** = number of codons per exon that can be mutated to a nonsense codon via a single base substitution. **Target mutability** = the exon mutability calculated from the summation of the observed relative target codon mutabilities (see Table 4 for mutability values)

Exon	Observed Mutations	Expected Mutations	Target Codons	Target Mutability	Mutations	CGA codons
1	5	1.0	5	6	p.3-TGG(5)	
2	0	1.2	11	7		
3	0	1.1	7	7		
4	0	0.6	8	4		
5	1	1.3	8	8	p.119-CAG(1)	
6	7	4.5	16	27	p.145-CGA(4) p.147-TCA(1) p.143-TGG(1) p.153-CAG(1)	1
7	7	4.3	14	26	p.190-CAG(2) p.195-CGA(2) p.189-CAG(1) p.194-CAA(1) p.183-TGG(1)	1
8	4	3.4	25	21	p.229-TTA(1) p.267-CAG(1) p.237-CAA(1) p.249-CAG(1)	
9	1	3.9	14	24	p.301-TAC(1)	1
10	4	3.2	26	19	p.373-GAA(1) p.365-CAA(1) p.338-GAA(1) p.354-TGG(1)	
11	4	3.4	26	21	p.423-CAG(2) p.429-TCA(1) p.419-GAA(1)	
12	3	2.9	25	17	p.489-CAA(1) p.477-GAG(1)	

Exon	Observed Mutations	Expected Mutations	Target Codons	Target Mutability	Mutations	CGA codons
					p.463-TGG(1)	
13	0	1.4	13	9		
14	8	4.8	16	29	p.539-CGA(3) p.555-CAA(3) p.568-CAG(1) p.540-TGG(1)	1
15	3	1.9	13	11	p.598-TCA(1) p.576-TCA(1) p.595-GAA(1)	
16	3	3.7	27	22	p.656-CAA(1) p.664-CAG(1) p.654-TTA(1)	
17	2	3.0	18	18	p.702-GAA(1) p.693-CAG(1)	
18	0	1.9	15	11		
19	2	3.8	14	23	p.767-GAG(1) p.773-AAG(1)	1
20	12	5.5	35	33	p.827-GAG(2) p.807-CAA(2) p.802-AAA(1) p.841-CAA(1) p.806-GAA(1) p.871-AAA(1) p.829-CAG(1) p.814-GAA(1) p.861-TCA(1) p.812-TGG(1)	
21	2	5.0	23	30	p.920-CAA(1) p.930-AAA(1)	1
22	1	3.0	19	18	p.954-TGG(1)	
23	6	6.8	36	41	p.1041-CAA(1) p.1046-CAA(1) p.1018-TAT(1) p.1029-TGG(1) p.997-TAC(1)	1

Exon	Observed Mutations	Expected Mutations	Target Codons	Target Mutability	Mutations	CGA codons
					p.990-CAA(1)	
24	4	2.4	17	15	p.1075-TGG(1) p.1073-GAG(1) p.1090-CAG(1) p.1074-GAA(1)	
25	6	2.9	19	18	p.1144-CAG(2) p.1113-CAG(1) p.1102-CAG(1) p.1114-AAG(1) p.1094-TTA(1)	
26	4	3.2	33	20	p.1167-TCA(2) p.1182-GAG(1) p.1194-CAG(1)	
27	3	3.1	26	19	p.1208-CAA(1) p.1248-CAG(1) p.1234-GAG(1)	
28	3	2.8	22	17	p.1274-TAC(1) p.1268-TGG(1) p.1290-AAA(1)	
29	2	4.6	18	28	p.1338-GAG(1) p.1328-CAG(1)	1
30	4	3.4	23	20	p.1370-CAG(2) p.1384-GAG(1) p.1405-CAA(1)	
31	3	2.0	16	12	p.1429-AAA(1) p.1414-CAA(1) p.1417-TTG(1)	
32	4	5.5	27	33	p.1495-CAG(1) p.1472-CAA(1) p.1469-CAG(1) p.1451-TTA(1)	1
33	1	2.1	26	13	p.1540-GAA(1)	
34	9	4.8	30	29	p.1577-CGA(4) p.1565-CAG(3) p.1598-TCA(1) p.1566-TTG(1)	1

Exon	Observed Mutations	Expected Mutations	Target Codons	Target Mutability	Mutations	CGA codons
35	4	4.9	28	29	p.1666-CGA(2) p.1624-CAG(1) p.1622-GAG(1)	1
36	2	3.1	21	18	p.1712-CAA(1) p.1714-GAA(1)	
37	2	3.9	18	23	p.1752-TTA(1) p.1763-CGA(1)	1
38	6	2.3	19	14	p.1791-CAA(3) p.1800-GAA(1) p.1802-CAG(1) p.1782-GAA(1)	
39	2	4.9	27	29	p.1855-CAG(1) p.1844-CGA(1)	1
40	5	2.7	25	16	p.1885-CAG(1) p.1882-TAC(1) p.1879-TGG(1) p.1870-AAA(1) p.1909-GAA(1)	
41	10	5.3	25	32	p.1967-CGA(4) p.1918-GAA(1) p.1968-AGA(1) p.1956-TGG(1) p.1973-CAA(1) p.1920-CAG(1) p.1934-GAG(1)	1
42	3	2.1	22	13	p.2035-GAG(2) p.2013-GAA(1)	
43	5	5.3	22	32	p.2095-CGA(3) p.2085-TGG(2)	1
44	4	5.5	24	33	p.2131-CAA(1) p.2118-CAG(1) p.2098-CGA(1) p.2106-TGG(1)	1
45	4	3.9	26	23	p.2198-CAG(1) p.2182-CAG(1) p.2154-CAG(1) p.2149-CAG(1)	

Exon	Observed Mutations	Expected Mutations	Target Codons	Target Mutability	Mutations	CGA codons
46	3	3.2	28	19	p.2214-TCA(1) p.2241-GGA(1) p.2217-CAA(1)	
47	4	2.8	23	17	p.2286-GAA(2) p.2290-AAA(1) p.2302-TGG(1)	
48	2	3.5	29	21	p.2359-CAA(1) p.2336-CAG(1)	
49	1	2.3	18	14	p.2369-GAA(1)	
50	0	1.5	13	9		
51	1	3.1	21	19	p.2468-GAG(1)	
52	2	4.0	16	24	p.2553-CGA(1) p.2526-CAG(1)	1
53	5	5.8	40	35	p.2574-CAG(1) p.2608-GAG(1) p.2565-CAA(1) p.2561-TGG(1) p.2605-TCA(1)	
54	0	2.1	15	13		
55	3	5.4	27	33	p.2721-AAG(1) p.2680-CGA(1) p.2716-CAG(1)	1
56	4	1.8	18	11	p.2791-AAA(2) p.2786-TGG(1) p.2785-AAG(1)	
57	4	3.0	17	18	p.2831-CAG(1) p.2822-CAG(1) p.2820-TGG(1) p.2843-AAG(1)	
58	4	4.0	19	24	p.2870-CGA(4)	1
59	7	7.2	38	44	p.2905-CGA(4) p.2925-TGG(1) p.2894-GAG(1) p.2958-GGA(1)	1

Exon	Observed Mutations	Expected Mutations	Target Codons	Target Mutability	Mutations	CGA codons
60	2	4.1	15	25	p.3028-CAG(1) p.2982-CGA(1)	1
61	3	2.9	5	18	p.3034-CGA(3)	1
62	1	1.1	7	7	p.3061-TGG(1)	
63	0	1.5	8	9		
64	4	2.9	8	17	p.3113-CGA(3) p.3116-CAG(1)	1
65	4	2.4	17	15	p.3127-TCA(3) p.3143-CAG(1)	
66	3	2.7	9	17	p.3190-CGA(3)	1
67	0	2.1	13	12		
68	2	2.0	16	12	p.3318-GAG(1) p.3310-CAG(1)	
69	4	3.0	10	18	p.3345-CGA(1) p.3334-TAT(1) p.3338-CAA(1) p.3326-TAC(1)	1
70	16	8.0	17	48	p.3381-CGA(7) p.3370-CGA(5) p.3391-CGA(4)	3
71	0	0.4	1	2		
72	0	0.9	5	6		
73	0	0.3	5	2		
74	1	2.8	22	17	p.3493-CAG(1)	
75	0	4.1	27	25		
76	3	3.4	7	21	p.3635-CAA(1) p.3630-CGA(1) p.3601-CAG(1)	1
77	0	1.4	10	8		
78	0	0.3	6	2		

Supp.Table S4. Single Nucleotide Polymorphisms in the DMD gene observed by SCAIP analysis.

All variants listed here are considered polymorphisms, not disease-causing mutations. Exonic (coding-region) polymorphisms are in bold.

dbSNP rs/ss#		**Exon**	**cDNA/Peptide**	**minor allele (-)**	**major allele (-)**	**minor allele count**	**total allele count**	**Ancestral Allele (chimp)**
rs72470543	ss102660540	1[1]	c.-545_-547delAG[1]	-	AG	1	606	AG
rs66718538	ss102660539	1[1]	c.-457A>C[1]	C	A	67	637	C
rs5927163	ss102660538	1[1]	c.-210A>G[1]	G	A	36	692	A
rs72470542	ss102660537	1	c.-114dupT	T	-	4	680	-
rs72470541	ss102660536	1[2]	c.-427G>T[2]	T	G	2	577	T
rs72470540	ss102660535	1[2]	c.-348G>A[2]	A	G	1	683	G
rs72470539	ss102660534	1[2]	c.-219C>T[2]	T	C	1	685	C
rs72470538	ss102660533	1[2]	c.-87T>C[2]	C	T	1	686	T
rs72470537	ss102660532	2	c.32-99A>T	T	A	1	654	T
rs72470536	ss102660531	2	c.32-78G>T	T	G	3	683	G
rs72470535	ss102660530	2	c.93+98T>C	C	T	1	685	T
rs72470534	ss102660529	2	c.93+183A>G	G	A	2	687	A
rs16990838	ss102660528	2	c.93+240A>G	G	A	10	670	A
rs2025666	ss102660527	2	c.93+303T>A	A	T	39	574	T
rs72470533	ss102660526	3	c.94-160dupT	T	-	12	641	-
rs3764763	ss102660525	3	c.94-133G>A	A	G	176	668	A
rs72470532	ss102660524	3	c.94-112A>T	T	A	1	683	A
rs66800250	ss102660523	3	c.94-9dupT	T	-	69	690	-
rs72470531	ss102660522	3	c.186+35A>T	T	A	11	692	A
rs989355	ss102660521	3	c.186+243T>C	C	T	65	549	T

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[1] NM_000109.2 , Dp427c Isoform

[2] NM_004009.2, Dp427p1 Isoform

dbSNP rs/ss#		**Exon**	**cDNA/Peptide**	**minor allele (-)**	**major allele (-)**	**minor allele count**	**total allele count**	**Ancestral Allele (chimp)**
rs72470530	ss102660520	4	c.187-14T>G	G	T	1	697	T
rs72470529	ss102660518	5	c.357+42T>C	C	T	3	686	T
rs72470528	ss102660517	5	c.357+298dupA	A	-	2	528	?[3]
rs72470527	ss102660516	5	c.357+351_357+352insC	C	-	224	488	-
rs72470526	ss102660515	6	c.358-428C>T	T	C	35	675	C
rs6631649	ss102660514	6	c.358-139G>A	A	G	9	665	G
rs17338849	ss102660513	6	c.358-80T>G	G	T	3	690	T
rs72470525	ss102660512	6	c.358-73C>T	T	C	1	688	T
rs72470524	ss102660511	6	c.358-51G>C	C	G	1	689	G
rs72470523	ss102660510	6	c.530+7A>T	T	A	1	688	A
rs72470522	ss102660509	6	c.530+86A>G	G	A	1	677	A
rs72470521	ss102660508	7	c.649+76A>G	G	A	1	684	A
rs1015509	ss102660507	7	c.649+145C>T	T	C	321	659	T
rs72470520	ss102660506	7	c.649+206T>C	C	T	9	600	T
rs72470519	ss102660505	8	c.650-374T>G	G	T	17	677	T
rs1800264	ss102660504	8	**c.802T>C p.Leu268**	C	T	3	692	T
rs72470518	ss102660503	8	c.831+95A>T	T	A	2	692	A
rs72470517	ss102660502	8	c.831+148_831+149dup	GA	-	21	629	GA
rs41303183	ss102660501	9	c.832-54A>G	G	A	26	692	A
rs72470516	ss102660500	9	c.832-44T>A	A	T	1	692	T
rs72470515	ss102660499	9	c.832-18C>G	G	C	16	692	C
rs72470514	ss102660498	9	c.832-17C>A	A	C	17	692	C
rs72470513	ss102660497	9	c.832-15A>G	G	A	1	692	A
rs1800265	ss102660496	9	**c.837G>A p.Thr279**	A	G	21	692	A

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[3] No Human Chimp Alignment

dbSNP rs/ss#		Exon	cDNA/Peptide	minor allele (-)	major allele (-)	minor allele count	total allele count	Ancestral Allele (chimp)
rs72470512	ss102660495	9	c.960+50del	-	G	7	683	G
rs6631629	ss102660494	9	c.960+312T>G	G	T	21	676	G
rs6631628	ss102660493	9	c.960+330G>A	A	G	35	676	A
rs6631627	ss102660492	9	c.960+388G>A	A	G	21	672	A
rs6631626	ss102660491	9	c.960+412C>T	T	C	25	670	T
rs72470511	ss102660490	10	c.961-148C>T	T	C	13	640	C
rs72470510	ss102660489	10	c.961-124G>C	C	G	1	692	G
rs16990527	ss102660488	10	c.961-109A>G	G	A	16	693	A
rs72470509	ss102660487	10	c.961-39A>G	G	A	1	692	A
rs72470508	ss102660486	10	c.961-36T>G	G	T	1	694	T
rs1800266	ss102660485	10	**c.1095A>C p.Gln365His**	C	A	1	695	A
rs72470507	ss102660484	10	**c.1098A>T p.Gly366**	T	A	1	696	A
rs72470506	ss102660483	10	c.1149+118G>A	A	G	1	674	G
rs72470505	ss102660482	11	c.1150-254dupG	G	-	25	614	-
rs34155804	ss102660481	11	**c.1225A>T p.Thr409Ser**	T	A	1	695	A
rs41303189	ss102660480	11	c.1331+26A>G	G	A	6	695	A
rs6628728	ss102660479	11	c.1331+55A>G	G	A	22	694	A
rs5928065	ss102660478	11	c.1331+127G>A	A	G	217	691	G
rs72470504	ss102660477	11	c.1331+131G>C	C	G	5	691	C
rs72470503	ss102660476	11	c.1331+145del	-	T	49	654	T
rs72468702	ss102660475	12	c.1332-293C>A	A	C	14	593	C
rs808513	ss102660474	12	c.1332-286T>C	C	T	263	599	T
rs72468701	ss102660473	12	c.1332-189A>G	G	A	2	669	A
rs72468700	ss102660472	12	c.1332-9A>G	G	A	1	692	A
rs72468699	ss102660471	12	**c.1337A>G p.His446Arg**	G	A	2	695	A

dbSNP rs/ss#		**Exon**	**cDNA/Peptide**	**minor allele (-)**	**major allele (-)**	**minor allele count**	**total allele count**	**Ancestral Allele (chimp)**
rs72468698	ss102660470	12	c.1482+25T>A	A	T	1	695	T
rs72468697	ss102660469	13	c.1483-125A>G	G	A	4	681	A
rs808542	ss102660468	13	c.1483-123G>T	T	G	288	681	T
rs808543	ss102660467	13	c.1483-110G>A	A	G	287	682	A
rs17309542	ss102660466	13	c.1483-72T>C	C	T	69	685	T
rs1435727	ss102660465	13	c.1483-67A>T	T	A	31	685	A
rs61733587	ss102660464	13	**c.1554T>A p.Asp518Glu**	A	T	1	694	T
rs72468696	ss102660463	13	c.1602+308delA	-	A	18	604	A
rs72468695	ss102660462	14	c.1603-144T>G	G	T	2	667	T
rs5927083	ss102660461	14	**c.1635A>G p.Arg545**	G	A	98	692	?[3]
rs5927082	ss102660460	14	c.1704+51T>C	C	T	90	692	?[3]
rs5972599	ss102660459	15	**c.1718C>T p.Ala573>Val**	T	C	1	689	?[3]
rs66481382	ss102660458	15	c.1812+118A>G	G	A	6	661	A
rs72468694	ss102660457	15	c.1812+169C>A	A	C	1	637	C
rs5927081	ss102660456	15	c.1812+329C>A	A	C	61	582	A
rs72468693	ss102660455	16	c.1813-68C>T	T	C	1	651	C
rs1800267	ss102660454	16	**c.1869C>T p.Leu623**	T	C	3	688	C
rs72468692	ss102660453	16	**c.1888A>G p.Thr630Ala**	G	A	2	687	A
rs72468691	ss102660452	16	c.1992+116T>G	G	T	2	652	T
rs6653877	ss102660451	16	c.1992+195A>T	T	A	18	617	A
rs115571	ss102660450	17	c.1993-37G>T	T	G	213	688	G
rs228373	ss102660449	17	c.2168+13T>C	C	T	199	692	T
rs1006211	ss102660448	17	c.2168+191G>C	C	G	272	654	C
rs228372	ss102660447	17	c.2168+222G>A	A	G	116	611	A
rs228371	ss102660446	17	c.2168+278A>G	G	A	85	548	A

dbSNP rs/ss#		**Exon**	**cDNA/Peptide**	**minor allele (-)**	**major allele (-)**	**minor allele count**	**total allele count**	**Ancestral Allele (chimp)**
rs72468690	ss102660445	18	c.2169-306G>A	A	G	4	621	G
rs228333	ss102660444	18	c.2169-204T>C	C	T	129	611	C
rs72468689	ss102660443	18	c.2169-96T>C	C	T	6	666	T
rs72468688	ss102660442	19	c.2293-517del	-	G	160	432	G
rs72468687	ss102660441	19	c.2293-185C>G	G	C	1	645	C
rs72468686	ss102660440	19	c.2293-183C>T	T	C	1	657	C
rs72468685	ss102660439	19	c.2293-182A>G	G	A	1	659	A
rs67750310	ss102660438	19	c.2380+47A>G	G	A	2	690	A
rs72468684	ss102660437	19	c.2380+322del	-	C	1	609	C
rs72468683	ss102660436	20	c.2381-278_2381-277del	-	AG	4	620	AG
rs72468682	ss102660435	20	c.2381-199A>G	G	A	1	624	T
rs72468681	ss102660434	20	**c.2391T>G p.Asn797Lys**	G	T	9	693	T
rs72468680	ss102660433	20	**c.2457A>C p.Leu819**	C	A	1	694	A
rs72468679	ss102660432	20	**c.2490C>T p.Asn830**	T	C	1	694	C
rs72468678	ss102660431	20	c.2622+33A>G	G	A	3	690	A
rs1545078	ss102660430	21	c.2623-503A>G	G	A	20	632	A
rs72468677	ss102660429	21	c.2623-67C>T	T	C	1	689	C
rs1028360	ss102660428	21	c.2623-11C>G	G	C	5	693	C
rs228406	ss102660427	21	**c.2645G>A p.Gly882Asp**	A	G	222	693	G
rs1028359	ss102660426	21	c.2803+167G>T	T	G	16	645	T
rs72468676	ss102660425	22	c.2804-320G>T	T	G	4	638	G
rs228396	ss102660424	22	c.2804-282G>C	C	G	189	652	G
rs72468675	ss102660423	22	c.2804-87A>C	C	A	1	684	A
rs72468674	ss102660422	22	c.2949+37G>A	A	G	1	689	G
rs72468673	ss102660421	22	c.2949+127T>G	G	T	3	670	T

dbSNP rs/ss#		**Exon**	**cDNA/Peptide**	**minor allele (-)**	**major allele (-)**	**minor allele count**	**total allele count**	**Ancestral Allele (chimp)**
rs66592193	ss102660420	22	c.2949+137A>G	G	A	3	667	A
rs72468672	ss102660419	22	c.2949+161T>C	C	T	4	667	T
rs72468671	ss102660418	22	c.2949+169_2949+172del	-	CTAT	1	660	CTAT
rs72468670	ss102660417	22	c.2949+254G>C	C	G	2	622	G
rs72468669	ss102660416	23	c.2950-209A>G	G	A	1	547	A
rs72468668	ss102660415	23	c.2950-90A>C	C	A	4	692	A
rs72468667	ss102660414	23	**c.2971G>C p.Glu991Gln**	C	G	1	691	G
rs1800268	ss102660413	23	**c.3021G>A p.Ser1007**	A	G	14	693	G
rs72468666	ss102660412	23	**c.3030G>T p.Ala1010**	T	G	1	691	G
rs56061889	ss102660411	23	c.3162+254T>G	G	T	20	521	T
rs72468665	ss102660410	23	c.3162+310_3162+311dupT	T	-	70	490	-
rs72468664	ss102660409	23	c.3162+339_3162+340insT	T	-	68	409	-
rs228391	ss102660408	23	c.3162+364C>T	T	C	204	653	T
rs72468663	ss102660407	24	c.3163-59T>A	A	T	1	675	T
rs72468662	ss102660406	24	c.3163-18T>C	C	T	1	690	T
rs72468661	ss102660405	24	c.3276+14_3276+15del	-	AT	1	688	AT
rs61354466	ss102660404	24	c.3276+228T>C	C	T	7	683	T
rs228388	ss102660403	24	c.3276+427C>T	T	C	115	475	C
rs228387	ss102660402	25	c.3277-345A>G	G	A	91	322	A
rs3761608	ss102660401	25	c.3277-231T>G	G	T	9	646	T
rs170608	ss102660400	25	c.3277-152G>A	A	G	154	615	A
rs72468660	ss102660399	25	c.3277-30C>T	T	C	3	688	C
rs3827462	ss102660398	25	**c.3406A>T p.Thr1136Ser**	T	A	4	689	A

dbSNP rs/ss#		Exon	cDNA/Peptide	minor allele (-)	major allele (-)	minor allele count	total allele count	Ancestral Allele (chimp)
rs72468659	ss102660397	25	c.3432+36C>T	T	C	1	690	C
rs72468658	ss102660396	25	c.3432+37G>A	A	G	1	687	G
rs1800269	ss102660395	27	**c.3734C>T p.Thr1245Ile**	T	C	15	690	C
rs72468657	ss102660394	27	c.3786+183T>C	C	T	3	684	T
rs5972569	ss102660393	27	c.3786+296T>C	C	T	259	541	T
rs72468656	ss102660392	28	c.3787-18T>C	C	T	4	696	T
rs41303185	ss102660391	28	c.3921+128C>T	T	C	232	680	C
rs66596198	ss102660390	28	c.3921+146C>G	G	C	16	659	C
rs66809536	ss102660389	28	c.3921+271G>C	C	G	13	617	C
rs2076366	ss102660388	29	c.3922-204C>G	G	C	255	643	C
rs66703963	ss102660387	29	c.4071+409T>G	G	T	47	280	T
rs10521995	ss102660386	30	c.4072-737G>A	A	G	67	630	G
rs7063700	ss102660385	30	c.4072-710T>C	C	T	4	685	T
rs72468655	ss102660384	30	c.4072-668T>C	C	T	1	688	C
rs72468654	ss102660383	30	c.4072-582A>T	T	A	1	685	A
rs67778598	ss102660382	30	c.4072-541A>G	G	A	3	690	A
rs56094071	ss102660381	30	c.4072-473T>A	A	T	82	691	T
rs72468653	ss102660380	30	c.4072-407T>C	C	T	4	691	T
rs16990320	ss102660379	30	c.4072-396T>A	A	T	2	689	T
rs16990318	ss102660378	30	c.4072-389G>T	T	G	3	689	G
rs72468652	ss102660377	30	c.4072-356A>C	C	A	1	682	G
rs72468651	ss102660376	30	c.4072-319C>T	T	C	1	679	C
rs72468650	ss102660375	30	c.4072-185T>A	A	T	1	693	T
rs72468649	ss102660374	30	c.4072-136C>T	T	C	1	696	C
rs72468648	ss102660373	30	c.4072-65C>T	T	C	2	696	C

dbSNP rs/ss#		Exon	cDNA/Peptide	minor allele (-)	major allele (-)	minor allele count	total allele count	Ancestral Allele (chimp)
rs28715870	ss102660372	30	**c.4162T>G p.Phe1388Val**	G	T	4	696	T
rs41303181	ss102660371	31	c.4234-13A>G	G	A	43	689	A
rs72468647	ss102660370	31	**c.4275A>G p.Glu1425**	G	A	2	690	A
rs72468646	ss102660369	31	c.4344+17A>T	T	A	2	688	A
rs72468645	ss102660368	31	c.4344+29G>A	A	G	1	690	G
rs72468644	ss102660367	31	c.4344+39T>A	A	T	1	690	T
rs72468643	ss102660366	32	c.4345-39G>C	C	G	2	693	G
rs72468642	ss102660365	32	c.4518+78_4518+79dup	(AG)2	(AG)3	1	653	(AG)3
rs67653684	ss102660364	33	c.4519-319A>G	G	A	78	661	A
rs72468641	ss102660363	33	c.4519-264_4519-263dup	(AT)7	(AT)6	256	526	(AT)6
rs16990296	ss102660362	33	c.4519-136C>T	T	C	74	635	C
rs72468640	ss102660361	33	c.4519-95A>C	C	A	2	648	A
rs72468639	ss102660360	33	c.4519-34T>A	A	T	5	696	T
rs72468638	ss102660359	33	**c.4529A>G p.Lys1510Arg**	G	A	1	697	A
rs66989299	ss102660358	33	c.4674+71A>G	G	A	1	694	A
rs12837503	ss102660357	33	c.4674+178T>C	C	T	12	694	T
rs57969988	ss102660356	33	c.4674+270A>G	G	A	81	684	A
rs16990293	ss102660355	33	c.4674+275C>G	G	C	80	684	T
rs16998283	ss102660354	34	c.4675-370T>C	C	T	33	476	?[3]
rs16998281	ss102660353	34	c.4675-342G>A	A	G	52	547	?[3]
rs72468637	ss102660352	34	c.4675-79A>G	G	A	1	688	?[3]
rs72468636	ss102660351	34	c.4675-53G>T	T	G	5	693	?[3]
rs72468635	ss102660350	34	c.4845+69G>A	A	G	2	688	T
rs5927976	ss102660349	35	c.4846-153A>G	G	A	217	638	A

dbSNP rs/ss#		**Exon**	**cDNA/Peptide**	**minor allele (-)**	**major allele (-)**	**minor allele count**	**total allele count**	**Ancestral Allele (chimp)**
rs72468634	ss102660348	35	**c.4877T>A p.Val1626Glu**	A	T	1	697	T
rs61733574	ss102660347	35	**c.4878G>T p.Val1626**	T	G	2	697	G
rs16990264	ss102660346	35	**c.5016T>A p.Asn1672Lys**	A	T	2	697	T
rs5927975	ss102660345	35	c.5025+103G>A	A	G	331	678	A
rs55676629	ss102660344	36	c.5026-93A>G	G	A	19	681	A
rs72468633	ss102660343	36	c.5026-59A>G	G	A	1	688	A
rs72468632	ss102660342	36	**c.5040C>T p.His1680**	T	C	1	692	C
rs5972544	ss102660341	37	c.5155-412C>A	A	C	268	662	C
rs12387861	ss102660340	37	c.5155-63T>A	A	T	9	698	A
rs72468631	ss102660339	37	c.5155-16T>A	A	T	1	695	T
rs72468630	ss102660338	37	**c.5163G>C p.Lys1721Asn**	C	G	1	697	G
rs34102501	ss102660337	37	**c.5182C>T p.Arg1728Cys**	T	C	1	696	C
rs1801187	ss102660336	37	**c.5234G>A p.Arg1745His**	A	G	329	695	G
rs72468629	ss102660335	37	c.5325+206C>T	T	C	1	666	T
rs72468628	ss102660334	38	c.5326-258A>C	C	A	16	664	A
rs41309607	ss102660333	38	c.5326-54A>C	C	A	11	693	A
rs72468627	ss102660332	38	c.5326-22G>T	T	G	6	693	T
rs72468626	ss102660331	38	c.5448+67A>G	G	A	1	690	A
rs72468625	ss102660330	38	c.5448+148G>T	T	G	4	664	A
rs331309	ss102660329	38	c.5448+169T>A	A	T	171	663	A
rs926953	ss102660328	39	c.5449-463G>A	A	G	308	664	A
rs72468624	ss102660327	39	c.5449-117T>C	C	T	6	695	T

dbSNP rs/ss#		**Exon**	**cDNA/Peptide**	**minor allele (-)**	**major allele (-)**	**minor allele count**	**total allele count**	**Ancestral Allele (chimp)**
rs72468623	ss102660326	39	c.5586+30G>A	A	G	4	695	G
rs72468622	ss102660325	39	c.5586+52A>G	G	A	2	695	A
rs72468621	ss102660324	39	c.5586+96_5586+97del	-	TC	159	687	-
rs1325579	ss102660323	39	c.5586+238A>G	G	A	226	601	A
rs1456733	ss102660322	40	c.5587-402C>T	T	C	199	685	T
rs72468620	ss102660321	40	c.5587-340T>G	G	T	3	620	T
rs1456732	ss102660320	40	c.5587-308T>G	G	T	170	613	T
rs1456731	ss102660319	40	c.5587-306C>A	A	C	210	573	A
rs72468619	ss102660318	40	c.5587-229A>G	G	A	6	650	A
rs72468618	ss102660317	40	c.5587-85C>T	T	C	1	690	C
rs72468617	ss102660316	40	c.5739+39T>A	A	T	3	688	T
rs72468616	ss102660315	40	c.5739+162_5739+1165del	-	AAGT	1	632	AAGT
rs7891425	ss102660314	40	c.5739+218G>A	A	G	99	639	A
rs6527184	ss102660313	41	c.5740-67G>T	T	G	57	630	G
rs72468615	ss102660312	41	c.5922+16C>G	G	C	1	679	C
rs72468614	ss102660311	41	c.5922+76_5922+77dup	(A)10	(A)8	286	672	(A)8
rs58019410	ss102660310	42	c.5923-212A>G	G	A	5	627	A
rs6631543	ss102660309	42	c.5923-179A>G	G	A	246	643	G
rs72468613	ss102660308	42	**c.6028G>T** **p.Ala2010Ser**	T	G	1	697	G
rs5927072	ss102660307	42	c.6117+383C>T	T	C	23	104	T
rs72468612	ss102660306	43	c.6118-63A>T	T	A	9	680	T
rs3788896	ss102660304	43	c.6290+27T>A	A	T	43	698	T
rs72468610	ss102660303	43	c.6290+92dupA	A	-	8	689	-
rs72468609	ss102660302	44	c.6291-179G>A	A	G	8	626	G

dbSNP rs/ss#		**Exon**	**cDNA/Peptide**	**minor allele (-)**	**major allele (-)**	**minor allele count**	**total allele count**	**Ancestral Allele (chimp)**
rs3747400	ss102660301	44	c.6291-115G>A	A	G	119	634	A
rs72468608	ss102660300	44	c.6291-102G>A	A	G	7	655	G
rs16990169	ss102660299	44	**c.6322C>T p.Arg2108Cys**	T	C	1	693	C
rs72468607	ss102660298	44	c.6438+120_6438+122dup	(CAT)1	(CAT)2	1	675	?[3]
rs1718050	ss102660296	1[4]	c.1-381658A>G[4]	G	A	232	677	A
rs72468605	ss102660295	1[4]	c.1-381564T>C[4]	C	T	1	684	T
rs55672607	ss102660294	1[4]	c.1-381429A>G[4]	G	A	50	684	A
rs72468604	ss102660293	1[4]	c.1-381220C>T[4]	T	C	1	650	C
rs3761605	ss102660292	45	c.6439-143G>A	A	G	189	624	G
rs67729860	ss102660291	45	c.6439-38G>A	A	G	2	688	G
rs1800273	ss102660290	45	**c.6463C>T p.Arg2155Trp**	T	C	35	685	C
rs72468603	ss102660289	45	c.6614+13A>T	T	A	1	685	A
rs3761604	ss102660288	45	c.6614+26G>T	T	G	210	684	G
rs72466602	ss102660287	45	c.6614+119A>G	G	A	1	683	A
rs72466601	ss102660286	45	c.6614+166delT	-	T	1	674	T
rs72466600	ss102660285	45	c.6614+186T>C	C	T	1	659	T
rs72466599	ss102660284	45	c.6614+237_6614+240del	-	AAAT	2	651	AAAT
rs72466598	ss102660283	46	c.6615-58A>C	C	A	5	684	?[3]
rs16989970	ss102660282	46	c.6762+141C>T	T	C	33	675	?[3]
rs72466597	ss102660281	46	c.6762+184T>C	C	T	30	672	T
rs72466596	ss102660280	47	c.6763-97C>T	T	C	2	678	C
rs72466595	ss102660279	47	**c.6828C>T p.Pro2276**	T	C	1	694	C

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[4] NM_004013.1, Dp140 Isoform

dbSNP rs/ss#		**Exon**	**cDNA/Peptide**	**minor allele (-)**	**major allele (-)**	**minor allele count**	**total allele count**	**Ancestral Allele (chimp)**
rs72466594	ss102660278	47	c.6912+128_6912+132dup	(GTTTT)4	(GTTTT)3	21	602	(GTTTT)3
rs72466593	ss102660277	47	c.6912+141G>A	A	G	1	624	G
rs72466592	ss102660276	47	c.6912+366del	-	A	33	537	A
rs45618538	ss102660275	48	c.6913-114A>T	T	A	86	671	A
rs1800275	ss102660274	48	**c.7096A>C p.Lys2366Gln**	C	A	121	678	A
rs5972454	ss102660273	48	c.7098+156G>C	C	G	225	656	G
rs7880185	ss102660272	49	c.7099-373T>G	G	T	73	494	T
rs72466591	ss102660271	49	c.7099-20A>G	G	A	2	682	A
rs72466590	ss102660270	49	**c.7183G>A p.Ala2395Thr**	A	G	2	678	G
rs72466589	ss102660269	49	c.7200+26C>T	T	C	1	682	C
rs72466588	ss102660268	49	c.7200+51T>A	A	T	1	681	T
rs1379871	ss102660267	49	c.7200+53C>G	G	C	230	675	C
rs5927044	ss102660266	50	c.7201-159A>C	C	A	83	644	A
rs72466587	ss102660265	50	c.7309+134+7309+135del	-	TG	1	611	TG
rs17341004	ss102660264	50	c.7309+176T>C	C	T	84	563	T
rs5927855	ss102660263	51	c.7310-250C>T	T	C	69	566	C
rs72466586	ss102660262	51	c.7310-36C>T	T	C	10	675	C
rs72466585	ss102660261	51	c.7542+13A>G	G	A	6	438	A
rs72466584	ss102660260	52	c.7543-159T>G	G	T	1	640	T
rs5927823	ss102660259	52	c.7543-156C>G	G	C	95	653	C
rs72466583	ss102660258	52	c.7660+125del	-	A	1	672	A
rs72466582	ss102660257	53	c.7661-61T>A	A	T	12	687	T
rs1801188	ss102660256	53	**c.7728T>C p.Asn2576**	C	T	137	688	T

dbSNP rs/ss#		Exon	cDNA/Peptide	minor allele (-)	major allele (-)	minor allele count	total allele count	Ancestral Allele (chimp)
rs72466581	ss102660255	53	**c.7820A>T p.Lys2607Met**	T	A	1	686	A
rs5927803	ss102660254	53	c.7872+324G>A	A	G	219	570	A
rs72466580	ss102660253	54	c.8027+7A>T	T	A	1	685	A
rs2270672	ss102660252	54	c.8027+11C>T	T	C	225	685	C
rs72466579	ss102660251	55	c.8028-114C>A	A	C	1	678	C
rs72466578	ss102660250	55	c.8028-42C>G	G	C	1	684	C
rs16989681	ss102660249	1[5]	c.1-285G>A[5]	A	G	113	683	A
rs72466577	ss102660248	1[5]	c.1-272delA[5]	-	A	1	683	A
rs72466576	ss102660247	1[5]	c.1-187T>A[5]	A	T	1	684	T
rs3788893	ss102660246	1[5]	c.30+125G>A[5]	A	G	98	609	A
rs72466575	ss102660245	56	**c.8322G>T p.Leu2774**	T	G	2	693	G
rs3788892	ss102660244	56	c.8390+174T>C	C	T	7	690	T
rs7058685	ss102660243	56	c.8390+422C>T	T	C	132	625	T
rs62588956	ss102660242	56	c.8390+443G>A	A	G	34	618	G
rs5927759	ss102660241	57	c.8391-287T>C	C	T	168	545	C
rs5972392	ss102660240	57	c.8391-109A>G	G	A	3	658	A
rs72466574	ss102660239	57	**c.8496A>C p.Ala2832**	C	A	1	689	A
rs6418632	ss102660238	57	c.8547+153C>T	T	C	185	680	T
rs72466573	ss102660237	57	c.8547+225A>T	T	A	3	660	A
rs72466572	ss102660236	58	c.8548-250A>G	G	A	26	673	A
rs72466571	ss102660235	58	c.8548-249T>C	C	T	17	673	T
rs17338590	ss102660234	58	c.8548-149A>G	G	A	12	655	A
rs72466570	ss102660233	58	**c.8571T>C p.Thr2857**	C	T	3	696	T
rs72466569	ss102660232	58	**c.8579C>A**	A	C	1	696	C

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[5] NM_004014 .1, Dp116 Isoform

dbSNP rs/ss#		Exon	cDNA/Peptide	minor allele (-)	major allele (-)	minor allele count	total allele count	Ancestral Allele (chimp)
			p.Pro2860His					
rs66517855	ss102660231	58	c.8668+209C>T	T	C	42	683	T
rs72466568	ss102660230	59	c.8669-85C>T	T	C	2	678	C
rs17338583	ss102660229	59	c.8669-75C>G	G	C	57	690	C
rs41305353	ss102660228	59	**c.8729A>T p.Glu2910Val**	T	A	11	696	A
rs1800278	ss102660227	59	**c.8734A>G p.Asn2912Asp**	G	A	11	696	A
rs1800279	ss102660226	59	**c.8762A>G p.His2921Arg**	G	A	24	696	A
rs1800280	ss102660225	59	**c.8810A>G p.Gln2937Arg**	G	A	49	694	A
rs72466567	ss102660224	59	**c.8852G>A p.Arg2951His**	A	G	1	693	G
rs72466566	ss102660223	59	c.8937+198T>C	C	T	23	625	T
rs67957153	ss102660222	60	c.8938-431G>A	A	G	15	335	G
rs72466565	ss102660221	60	c.9084+66A>C	C	A	1	691	A
rs6631295	ss102660220	61	c.9085-431T>C	C	T	177	669	T
rs72466564	ss102660219	61	c.9085-70G>T	T	G	1	693	G
rs72466563	ss102660218	61	**c.9093C>T p.Val3031**	T	C	1	692	C
rs72466562	ss102660217	61	**c.9094G>A p.Glu3032Lys**	A	G	1	692	G
rs72466561	ss102660216	61	c.9163+38C>A	A	C	1	690	C
rs6628596	ss102660215	61	c.9163+189G>T	T	G	157	644	G
rs7059188	ss102660214	62	c.9164-145A>G	G	A	10	644	A
rs72466560	ss102660213	62	c.9224+151C>A	A	C	1	696	C
rs7053818	ss102660212	62	c.9224+264T>A	A	T	7	636	T
rs72466559	ss102660211	62	c.9224+265A>G	G	A	1	605	A

dbSNP rs/ss#		Exon	cDNA/Peptide	minor allele (-)	major allele (-)	minor allele count	total allele count	Ancestral Allele (chimp)
rs17338423	ss102660210	63	c.9225-138A>G	G	A	120	668	A
rs72466558	ss102660209	63	c.9286+52A>T	T	A	1	694	A
rs72466557	ss102660208	64	c.9287-119A>G	G	A	1	672	A
rs10127360	ss102660207	64	c.9361+138T>C	C	T	221	639	T
rs16989449	ss102660206	64	c.9361+259G>T	T	G	61	610	G
rs5972341	ss102660205	64	c.9361+364A>G	G	A	217	635	A
rs72466556	ss102660204	64	c.9361+383C>T	T	C	1	471	C
rs72466555	ss102660203	65	c.9362-114T>C	C	T	1	657	T
rs72466554	ss102660202	65	c.9563+27G>A	A	G	2	694	G
rs72466553	ss102660201	65	c.9563+68G>A	A	G	1	695	G
rs5927704	ss102660200	66	c.9564-287C>T	T	C	75	664	T
rs72466552	ss102660199	66	c.9564-286G>A	A	G	22	667	G
rs72466551	ss102660198	66	c.9564-267A>G	G	A	2	665	A
rs2293666	ss102660197	66	c.9564-210C>T	T	C	73	673	T
rs2293667	ss102660196	66	c.9564-97T>C	C	T	73	690	T
rs2293668	ss102660195	66	c.9649+15C>T	T	C	72	690	C
rs72466550	ss102660194	66	c.9649+86A>T	T	A	1	683	A
rs5927701	ss102660193	67	c.9650-342G>A	A	G	131	423	A
rs67403730	ss102660192	67	c.9650-158A>C	C	A	5	618	A
rs72466549	ss102660191	67	c.9650-64C>T	T	C	3	688	C
rs72466548	ss102660190	67	c.9807+63A>G	G	A	1	696	A
rs72466547	ss102660189	67	c.9807+139G>T	T	G	1	689	G
rs5926992	ss102660188	67	c.9807+218C>G	G	C	27	690	C
rs72466546	ss102660187	67	c.9807+423delT	-	T	168	451	-
rs72466544	ss102660185	69	c.9975-460A>G	G	A	14	676	G

dbSNP rs/ss#		Exon	cDNA/Peptide	minor allele (-)	major allele (-)	minor allele count	total allele count	Ancestral Allele (chimp)
rs72466543	ss102660184	69	c.9975-111A>G	G	A	1	688	A
rs12558839	ss102660183	69	c.9975-79G>A	A	G	4	696	G
rs5927693	ss102660182	69	c.10086+283C>T	T	C	246	573	T
rs72466542	ss102660181	70	c.10087-139C>G	G	C	2	695	C
rs72466541	ss102660180	70	c.10087-24A>G	G	A	1	695	A
rs41303187	ss102660179	70	c.10087-20C>T	T	C	3	695	C
rs16989418	ss102660178	71	c.10224-101T>C	C	T	1	696	T
rs12391957	ss102660177	72	c.10263-411T>C	C	T	122	624	T
rs2404496	ss102660176	72	c.10328+67A>G	G	A	102	691	A
rs72466540	ss102660175	72	c.10328+158A>G	G	A	1	668	A
rs2404495	ss102660174	72	c.10328+379T>C	C	T	81	497	C
rs3788884	ss102660173	72	c.10328+501T>C	C	T	6	323	T
rs2896876	ss102660172	74	c.10395-178A>G	G	A	97	640	A
rs72466539	ss102660171	75	c.10554-37_10554-34ins	CTTT	-	124	670	-
rs72466538	ss102660170	75	**c.10620G>A p.Pro3540**	A	G	1	681	G
rs1800281	ss102660169	75	**c.10789C>T p.Leu3597**	T	C	7	683	C
rs72466537	ss102660168	75	c.10797+42C>G	G	C	8	647	C
rs1484852	ss102660167	75	c.10797+82G>A	A	G	94	611	G
rs3816796	ss102660166	75	c.10797+135A>G	G	A	21	642	A
rs61037625	ss102660165	76	c.10798-171T>A	A	T	8	636	A
rs59170794	ss102660164	76	c.10798-110A>C	C	A	8	670	A
rs17340575	ss102660163	76	c.10798-100G>C	C	G	6	674	G
rs72466536	ss102660162	76	c.10921+133C>T	T	C	3	688	T
rs7066986	ss102660161	76	c.10921+183A>G	G	A	16	602	A
rs72466535	ss102660160	76	c.10921+236C>T	T	C	9	554	C

dbSNP rs/ss#		Exon	cDNA/Peptide	minor allele (-)	major allele (-)	minor allele count	total allele count	Ancestral Allele (chimp)
rs72466534	ss102660159	77	c.11014+65A>G	G	A	2	680	A
rs72466532	ss102660157	78	c.11046+119A>G	G	A	3	651	A
rs436628	ss102660156	79	c.11047-249A>T	T	A	150	663	A
rs72466531	ss102660155	79	c.11058+924_11058+925dup	ACAA	-	1	697	-
rs72466530	ss102660154	79	c.11058+968dupT	T	-	2	697	?[3]
rs72466529	ss102660153	79	c.11058+1048_11058+1046dup	GAT	-	3	697	-
rs72466527	ss102660151	79	c.11058+1245_11058+1248dup	CTTT	-	1	697	-
rs3361	ss102660150	79	c.11058+1447A>G	G	A	166	697	G
rs72466526	ss102660149	79	c.11058+1565_11058+1566insAAAG	AAAG	-	25	697	-
rs72466525	ss102660148	79	c.11058+1591_11058+1594dup	TGTT	-	4	697	-
rs72466524	ss102660147	79	c.11058+1758_11058+1762del	-	TTCAT	5	696	TTCAT
rs7886658	ss102660146	79	c.11058+1783A>G	G	A	6	696	A
rs72466523	ss102660145	79	c.11058+2039C>A	A	C	4	697	?[3]
rs72466522	ss102660144	79	c.11058+2153_11058+2154insATGTGACGCTGGACCTT	ATGTGACGCTGGACCTT	-	3	696	-
rs72466521	ss102660143	79	c.11058+2237_11058+2240dup	AAGT	-	11	696	-
rs72466520	ss102660142	79	c.11058+2581_11058+2584dup	ACTT	-	1	695	-
rs72466519	ss102660141	79	c.11058+2760_11058+2763del	-	ACTT	77	695	ACTT
rs72466518	ss102660140	79	c.11058+2771_11058+2774dup	AATA	-	4	695	-

Supp. Table S5. Variable Nucleotide Repeats in the DMD gene observed by SCAIP analysis.

dbSNP ss#	Relative Exon	cDNA	Allele (Unit)Count	Allele Count	Allele Frequency
ss102660519	4	c.264+361			
			(TTTTC)5	391	75.0%
			(TTTTC)4	126	24.2%
			(TTTTC)6	4	0.8%
ss102660305	43	c.6118-61			
			(AT)7	549	82.7%
			(AT)8	104	15.7%
			(AT)6	9	1.4%
			(AT)9	2	0.3%
ss102660297	44	c.6438+60942			
			(GT)22	297	90.3%
			(GT)17	20	6.1%
			(GT)14	9	2.7%
			(GT)21	2	0.6%
			(GT)18	1	0.3%
ss102660186	68	c.9974+23			
			(A)11	333	49.8%
			(A)10	287	42.9%
			(A)9	49	7.3%
ss102660158	78	c.11015-44		683	
			(T)8	674	98.7%
			(T)7	8	1.2%
			(T)9	1	0.1%

dbSNP ss#	Relative Exon	cDNA	Allele (Unit)Count	Allele Count	Allele Frequency
-	79	c.11058+463			
			(CA)8(TACA)1(CA)6	471	77.5%
			(CA)8(TACA)2(CA)6	106	17.4%
			(CA)12	14	2.3%
			(CA)9	4	0.7%
			(CA)8(TACA)1(CA)5	2	0.3%
			(CA)10	2	0.3%
			(CA)8(TACA)1(CA)4(TACA)1	2	0.3%
			(CA)3(TACA)1(CA)6	2	0.3%
			(CA)16	1	0.2%
			(CA)3(TACA)1(CA)11	1	0.2%
			(CA)8(TACA)2(CA)7	1	0.2%
			(CA)8(TACA)1(CA)8	1	0.2%
			(CA)13	1	0.2%
ss102660152	79	c.11058+1068			
			TGAT(5)	588	84.4%
			TGAT(6)	100	14.3%
			TGAT(4)	9	1.3%

Supp. Table S6. Mono Exon Skipping Summary by Mutation Class and Patient.

Number of patients predicted to benefit from skipping an individual exon are noted in parentheses.

	Mutation Class (# of Patients)		
Exon Skipped	**Deletion**	**Duplication**	**Point**
Ex1			
Ex2	ex03ex07del(26)	ex02dup(15)	
Ex3			ex03pt(3)
Ex4			ex04pt(2)
Ex5			ex05pt(5)
Ex6			
Ex7	ex08ex11del(5) ex08ex43del(4) ex08ex45del(1)	ex07dup(1)	
Ex8	ex03ex07del(26) ex05ex07del(5) ex06ex07del(1)	ex08dup(1)	
Ex9			ex09pt(2)
Ex10			ex10pt(6)
Ex11	ex12ex25del(2) ex12ex41del(1) ex12del(1)		
Ex12	ex10ex11del(5) ex03ex11del(2) ex06ex11del(1)	ex12dup(1)	
Ex13			ex13pt(2)
Ex14			ex14pt(10)
Ex15			ex15pt(4)
Ex16			ex16pt(6)

	Mutation Class (# of Patients)		
Exon Skipped	**Deletion**	**Duplication**	**Point**
Ex17	ex18del(5) ex18ex39del(3) ex18ex20del(1) ex18ex41del(1) ex18ex44del(1) ex18ex37del(1) ex18ex32del(1) ex18ex22del(1)	ex17dup(1)	
Ex18	ex03ex17del(2) ex14ex17del(2) ex19ex45del(1) ex19ex43del(1)	ex18dup(1)	
Ex19			
Ex20	ex21del(1) ex19del(1) ex16ex19del(1)		
Ex21	ex22ex26del(1) ex22ex47del(1) ex22ex31del(1)		
Ex22	ex21del(1) ex19ex21del(1)	ex22dup(1)	
Ex23			ex23pt(8)
Ex24			ex24pt(8)
Ex25			ex25pt(10)
Ex26			ex26pt(5)
Ex27			ex27pt(6)
Ex28			ex28pt(8)
Ex29			ex29pt(4)
Ex30			
Ex31			
Ex32			ex32pt(6)
Ex33			ex33pt(2)

	Mutation Class (# of Patients)		
Exon Skipped	**Deletion**	**Duplication**	**Point**
Ex34			ex34pt(9)
Ex35			ex35pt(5)
Ex36			ex36pt(5)
Ex37			ex37pt(4)
Ex38			ex38pt(9)
Ex39			ex39pt(4)
Ex40	ex18ex39del(3)		
Ex41	ex42ex43del(1)		
Ex42			ex42pt(5)
Ex43	ex44del(12) ex44ex49del(1) ex44ex48del(1)	ex43dup(1)	
Ex44	ex45del(21) ex45ex54del(7) ex43ex43del(3) ex10ex43del(1) ex38ex43del(1) ex19ex43del(1) ex14ex43del(1) ex42ex43del(1)		
Ex45	ex46ex47del(17) ex44del(12) ex46ex51del(9) ex46ex48del(7) ex46ex46del(2) ex46ex60del(2) ex46ex53del(2) ex46ex55del(1) ex18ex44del(1)	ex45dup(1)	
Ex46	ex45del(21) ex35ex45del(1) ex19ex45del(1)		
Ex47			ex47pt(6)

	Mutation Class (# of Patients)		
Exon Skipped	**Deletion**	**Duplication**	**Point**
Ex48			ex48pt(5)
Ex49			ex49pt(1)
Ex50	ex51del(17) ex51ex55del(3)	ex50dup(1)	
Ex51	ex45ex50del(27) ex49ex50del(15) ex52del(12) ex48ex50del(11) ex50del(4) ex47ex50del(1)	ex51dup(1)	
Ex52	ex51del(17) ex53ex55del(5)		
Ex53	ex45ex52del(21) ex52del(12) ex49ex52del(7) ex48ex52del(6) ex50ex52del(4)	ex53dup(1)	
Ex54	ex46ex53del(2) ex55del(2)	ex54dup(1)	
Ex55	ex45ex54del(7) ex49ex54del(4) ex52ex54del(3) ex48ex54del(3) ex54del(1)		
Ex56	ex55del(2) ex46ex55del(1)		
Ex57			
Ex58			
Ex59			
Ex60			ex60pt(3)
Ex61		ex61dup(1)	
Ex62	ex63del(2)		
Ex63			

	Mutation Class (# of Patients)		
Exon Skipped	**Deletion**	**Duplication**	**Point**
Ex64			ex64pt(5)
Ex65			
Ex66			
Ex67			
Ex68			
Ex69			
Ex70			
Ex71			ex71pt(2)
Ex72			
Ex73			
Ex74			ex74pt(8)
Ex75			
Ex76			
Ex77			
Ex78			
Ex79			