

\*cDNA Reference sequence **NM\_000492.3** with the exception of c.1408A instead of G

\*\*Protein Reference sequence **NP\_000483.3** with the exception of p.Met470 instead of p.Val470

#exon/intron (#usual)	Usual name (CFMD)	c.DNA nomenclature UMD*	Predicted protein UMD**
prom (1)	48C/G	c.1-85C>G	p.Met1?
prom (1)	125G/C	c.1-8G>C	p.Met1?
E1 (1)	M1V	c.1A>G	p.Met1?
E1 (1)	M1K	c.2T>A	p.Met1?
E1 (1)	M1I(ATA)	c.3G>A	p.Met1?
E1 (1)	S4X	c.11C>A	p.Ser4X
E1 (1)	P5L	c.14C>T	p.Pro5Leu
E1 (1)		c.49_50dup	p.Trp19AlafsX7
E2 (2)	CFTRdele2 ; CFTR- dele2	c.54_164del	p.Ser18_Glu54del
E2-3 (2-3)	CFTRdele2, 3 ; Del exon 2- 3	c.54_273del	p.Ser18ArgfsX16
E2 (2)		c.95T>C	p.Leu32Pro
E2 (2)	S50P	c.148T>C	p.Ser50Pro
IVS1 (1)	185+ 4A- >T	c.53+4A>T	
IVS2 (2)		c.165-2A>C	
IVS2 (2)	297- 3C- >T	c.165-3C>T	
E3 (3)	300delA	c.168delA	p.Glu56AspfsX35
E3 (3)	W57G	c.169T>G	p.Trp57Gly
E3 (3)	D58G	c.173A>G	p.Asp58Gly
E3 (3)	306insA	c.175dup	p.Arg59LysfsX10
E3 (3)	E60K	c.178G>A	p.Glu60Lys
E3 (3)	E60X	c.178G>T	p.Glu60X
E3 (3)	P67L	c.200C>T	p.Pro67Leu
E3 (3)	R74W	c.220C>T	p.Arg74Trp
E3 (3)	R75X	c.223C>T	p.Arg75X
E3 (3)	R75Q	c.224G>A	p.Arg75Gln
E3 (3)		c.228T>G	p.Cys76Trp
E3 (3)	359insT	c.233dup	p.Trp79LeufsX32
E3 (3)	G85E	c.254G>A	p.Gly85Glu
E3 (3)	F87L	c.259T>C	p.Phe87Leu
E3 (3)	394delTT	c.262_263delTT	p.Leu88lfefsX22
IVS3 (3)	405+ 46G/T	c.273+46T>G	
IVS3 (3)	406- 1G- >C	c.274-1G>C	
E4 (4)	Q98R	c.293A>G	p.Gln98Arg
E4 (4)	I105N	c.314T>A	p.Ile105Asn
E4 (4)	D110E	c.330C>A	p.Asp110Glu
E4 (4)	P111L	c.332C>T	p.Pro111Leu

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E4 (4)	R117C	c.349C>T	p.Arg117Cys
E4 (4)	R117H	c.350G>A	p.Arg117His
E4 (4)	Y122X	c.366T>A	p.Tyr122X
E4 (4)	A141D	c.422C>A	p.Ala141Asp
E4 (4)	574delA	c.442delA	p.Ile148LeufsX5
E4 (4)	I148T	c.443T>C	p.Ile148Thr
E4 (4)	M152V	c.454A>G	p.Met152Val
E4 (4)	Y161N	c.481T>A	p.Tyr161Asn
IVS4 (4)	621+ 1G- >T	c.489+1G>T	
IVS4 (4)	621+ 2T- >G	c.489+2T>G	
IVS4 (4)	621+ 3A- >G	c.489+3A>G	
E5 (5)	L165S	c.494T>C	p.Leu165Ser
E5 (5)	R170H	c.509G>A	p.Arg170His
E5 (5)	I175V	c.523A>G	p.Ile175Val
E5 (5)	G178R	c.532G>A	p.Gly178Arg
E5 (5)	N186K	c.558C>A	p.Asn186Lys
IVS5 (5)	711+ 1G- >T	c.579+1G>T	
IVS5 (5)	711+ 3A- >G	c.579+3A>G	
IVS5 (5)	712- 1G- >T	c.580-1G>T	
E6 (6a)	G194V	c.581G>T	p.Gly194Val
E6 (6a)	F200I	c.598T>A	p.Phe200Ile
E6 (6a)	V201M	c.601G>A	p.Val201Met
E6 (6a)	733delG	c.601delG	p.Val201CysfsX14
E6 (6a)	I203M	c.609C>G	p.Ile203Met
E6 (6a)	L206W	c.617T>G	p.Leu206Trp
E6 (6a)	Q220X	c.658C>T	p.Gln220X
E6 (6a)	L227R	c.680T>G	p.Leu227Arg
E6 (6a)	V232D	c.695T>A	p.Val232Asp
E6 (6a)	Q237E	c.709C>G	p.Gln237Glu
E6 (6a)	852del22	c.720_741del	p.Gly241GlufsX13
E6 (6a)	M244K	c.731T>A	p.Met244Lys
IVS6 (6a)	875+ 40A/G	c.743+40A>G	
IVS6 (6a)		c.744-33GATT[4]	
IVS6 (6a)	TTGA repeats	c.744-33GATT[6]	
IVS6 (6a)	TTGA repeats	c.744-33GATT[7]	
IVS6 (6a)	TTGA repeats	c.744-33GATT[8]	

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#exon/intron (#usual)	Usual name (CFMD)	c.DNA nomenclature UMD*	Predicted protein UMD**
E7 (6b)	R258G	c.772A>G	p.Arg258Gly
E7 (6b)	N287Y	c.859A>T	p.Asn287Tyr
E7 (6b)	991del5	c.861_865delCTTAA	p.Asn287LysfsX19
IVS7 (6b)		c.869+5G>A	
IVS7 (6b)	1001+ 11C/T	c.869+11C>T	
E8 (7)	1078delT	c.948delT	p.Phe316LeufsX12
E8 (7)	R334W	c.1000C>T	p.Arg334Trp
E8 (7)	I336K	c.1007T>A	p.Ile336Lys
E8 (7)	T338I	c.1013C>T	p.Thr338Ile
E8 (7)	R347P	c.1040G>C	p.Arg347Pro
E8 (7)	R347H	c.1040G>A	p.Arg347His
E8 (7)	R352Q	c.1055G>A	p.Arg352Gln
E8 (7)	1215delG	c.1083delG	p.Trp361CysfsX8
IVS8 (7)	1248+ 1G- >A	c.1116+1G>A	
E9 (8)	E379K	c.1135G>A	p.Glu379Lys
IVS9 (8)	1341+ 18A- >C	c.1209+18A>C	
IVS9 (8)		c.1210-12T[3]	
IVS9 (8)	poly- T tract variations	c.1210-12T[5]	
IVS9 (8)		c.1210-12T[6]	
IVS9 (8)	poly- T tract variations	c.1210-12T[7]	
IVS9 (8)	poly- T tract variations	c.1210-12T[9]	
IVS9 (8)	1342- 12(GT)n	c.1210-34TG[9]	
IVS9 (8)	1342- 12(GT)n	c.1210-34TG[10]	
IVS9 (8)	1342- 12(GT)n	c.1210-34TG[11]	
IVS9 (8)	1342- 12(GT)n	c.1210-34TG[12]	
IVS9 (8)		c.1210-34TG[13]	
E10 (9)	D443Y	c.1327G>T	p.Asp443Tyr
E10 (9)	Q452P	c.1355A>C	p.Gln452Pro
E10 (9)	A455E	c.1364C>A	p.Ala455Glu
IVS10 (9)		c.1392+52T[10]	
IVS10 (9)	1525- 61A/G	c.1393-61A>G	
E11 (10)	1531C/T (L467F)	c.1399C>T	p.Leu467Phe
E11 (10)	M470V	c.1408A>G	p.Met470Val
E11 (10)	1548delG	c.1418delG	p.Gly473GlufsX54
E11 (10)	S489X	c.1466C>A	p.Ser489X
E11 (10)	Q493X	c.1477C>T	p.Gln493X

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E11 (10)	1609delCA	c.1477_1478delCA	p.Gln493ValfsX10
E11 (10)	T501A	c.1501A>G	p.Thr501Ala
E11 (10)	I506V (1648A/G)	c.1516A>G	p.Ile506Val
E11 (10)	I506T	c.1517T>C	p.Ile506Thr
E11 (10)	[delta]I507	c.1519_1521delATC	p.Ile507del
E11 (10)	[delta]F508	c.1521_1523delCTT	p.Phe508del
E11 (10)	F508C	c.1523T>G	p.Phe508Cys
E11 (10)	1677delTA	c.1545_1546delTA	p.Tyr515X
E11 (10)	1706del17	c.1574_1590del	p.Gln525LeufsX37
E11 (10)	1716G/A	c.1584G>A	p.Glu528Glu
IVS11 (10)	1716+ 2T- >C	c.1584+2T>C	
IVS11 (10)	1717- 8G- >A	c.1585-8G>A	
IVS11 (10)	1717- 1G- >A	c.1585-1G>A	
E12 (11)	G542X	c.1624G>T	p.Gly542X
E12 (11)	G544V	c.1631G>T	p.Gly544Val
E12 (11)	S549R(T- >G)	c.1647T>G	p.Ser549Arg
E12 (11)	G551D	c.1652G>A	p.Gly551Asp
E12 (11)	R553X	c.1657C>T	p.Arg553X
E12 (11)	1802delC	c.1670delC	p.Ser557PhefsX2
E12 (11)	L558S	c.1673T>C	p.Leu558Ser
E12 (11)	R560K	c.1679G>A	p.Arg560Lys
IVS12 (11)	1811+ 1.6kbA- >G	c.1680-886A>G	
IVS12 (11)	1811 + 1650 T>A	c.1680-870T>A	
IVS12 (11)	1812- 1G- >A	c.1680-1G>A	
E13 (12)	V562I	c.1684G>A	p.Val562Ile
E13 (12)	G576A	c.1727G>C	p.Gly576Ala
E13 (12)	T582I	c.1745C>T	p.Thr582Ile
E13 (12)	E585X	c.1753G>T	p.Glu585X
IVS13 (12)	1898+ 1G- >A	c.1766+1G>A	
IVS13 (12)	1898+ 1G- >C	c.1766+1G>C	
IVS13 (12)	1898+ 5G- >A	c.1766+5G>A	
IVS13 (12)	1898+ 152T/A	c.1766+152T>A	
E14 (13)	I601F	c.1801A>T	p.Ile601Phe
E14 (13)	1949del84	c.1820_1903del	p.Met607_Gln634del
E14 (13)	H609L	c.1826A>T	p.His609Leu
E14 (13)		c.1920T>C	p.Phe640Phe

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E14 (13)	E656X	c.1966G>T	p.Glu656X
E14 (13)	2118del4	c.1986_1989delAACT	p.Thr663ArgfsX8
E14 (13)	E664X	c.1990G>T	p.Glu664X
E14 (13)	R668C	c.2002C>T	p.Arg668Cys
E14 (13)	2143delT	c.2012delT	p.Leu671X
E14 (13)	2183AA- >G	c.2051_2052delinsG	p.Lys684SerfsX38
E14 (13)	2183delAA	c.2051_2052delAA	p.Lys684ThrfsX4
E14 (13)	2184insA	c.2052dup	p.Gln685ThrfsX4
E14 (13)	R709X	c.2125C>T	p.Arg709X
E14 (13)	K710X	c.2128A>T	p.Lys710X
E14 (13)	E725K	c.2173G>A	p.Glu725Lys
E14 (13)	L732X	c.2195T>G	p.Leu732X
E14 (13)	S737F	c.2210C>T	p.Ser737Phe
E14 (13)	2377C/T	c.2245C>T	p.Leu749Leu
E14 (13)	2380_2387del	c.2248_2255del	p.Pro750GlnfsX26
E14 (13)	2391 C/T	c.2259C>T	p.Ser753Ser
E14 (13)	R764X	c.2290C>T	p.Arg764X
E14 (13)	2423delG	c.2291delG	p.Arg764GlnfsX7
E14 (13)	2456delAC	c.2324_2325delAC	p.His775LeufsX3
E14 (13)	S776X	c.2327C>G	p.Ser776X
E14 (13)	R792X	c.2374C>T	p.Arg792X
E14 (13)	K830X	c.2488A>T	p.Lys830X
IVS14 (13)	2622+ 1G- >A	c.2490+1G>A	
E15 (14a)		c.2496C>A	p.Cys832X
E15 (14a)	2634delT	c.2502delT	p.Phe834LeufsX10
E15 (14a)	2634insT	c.2502dup	p.Asp835X
E15 (14a)	2640delT	c.2508delT	p.Asp836GlufsX8
E15 (14a)	W846X (2670TGG>TGA)	c.2538G>A	p.Trp846X
E15 (14a)	R851X	c.2551C>T	p.Arg851X
E15 (14a)	2694T/G	c.2562T>G	p.Thr854Thr
E15 (14a)	2711delT	c.2583delT	p.Phe861LeufsX3
E15 (14a)	C866R	c.2596T>C	p.Cys866Arg
IVS15 (14a)		c.2620-17G>T	
IVS16 (14b)	2789+ 5G- >A	c.2657+5G>A	
IVS16 (14b)	2790- 1G- >C	c.2658-1G>C	
E17 (15)	Q890X	c.2668C>T	p.Gln890X

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E17 (15)	S902R	c.2706C>G	p.Ser902Arg
E17 (15)	2851A/G	c.2719A>G	p.Ile907Val
E17 (15)	2869insG	c.2736dup	p.Tyr913ValfsX62
E17 (15)	2896insAG	c.2763_2764dup	p.Val922GlufsX2
E17 (15)	D924N	c.2770G>A	p.Asp924Asn
E17 (15)	S945L	c.2834C>T	p.Ser945Leu
E17 (15)	M952I	c.2856G>C	p.Met952Ile
E17 (15)	3007delG	c.2875delG	p.Ala959HisfsX9
E17 (15)	3030G/A	c.2898G>A	p.Thr966Thr
E17 (15)	L967S	c.2900T>C	p.Leu967Ser
IVS17 (15)	CFTRdele16- 17b ; CFTR- dele 16- 17a- 17b	c.2909_3367del	p.Gly971_Gly1123del
IVS17 (15)	3041- 15T- >G	c.2909-15T>G	
IVS17 (15)	3041- 92G/A	c.2909-92G>A	
IVS17 (15)	3041- 71G/C	c.2909-71G>C	
E18 (16)	S977F	c.2930C>T	p.Ser977Phe
E18 (16)	I980K	c.2939T>A	p.Ile980Lys
E18 (16)	D985H	c.2953G>C	p.Asp985His
E18 (16)	D993Y	c.2977G>T	p.Asp993Tyr
E18 (16)	F994C	c.2981T>G	p.Phe994Cys
E18 (16)	3120G- >A	c.2988G>A	p.Gln996Gln
IVS18 (16)	3120+ 1G- >A	c.2988+1G>A	
E19 (17a)	L997F	c.2991G>C	p.Leu997Phe
E19 (17a)		c.2989_3139del	p.Leu997AlafsX13
E19 (17a)	3121- 977_3499+ 248del2515	c.2989_3367del	p.Leu997GlufsX11
E19 (17a)	Del exon 17a- 17b	c.2989_3468del	p.Leu997_Leu1156del
IVS18 (16)	3121- 92A12/13	c.2989-93A[12]	
IVS18 (16)	3121- 1G- >A	c.2989-1G>A	
E19 (17a)	3129del4	c.2997_3000delAATT	p.Ile1000X
E19 (17a)	I1005R	c.3014T>G	p.Ile1005Arg
E19 (17a)	3196del54	c.3064_3117del	p.Val1022_Gln1039del
E19 (17a)	3195del6 ; 3199del6	c.3067_3072del	p.Ile1023_Val1024del
E19 (17a)	3200_3204delTAGTG	c.3068_3072delTAGTG	p.Ile1023SerfsX22
E19 (17a)	I1027T	c.3080T>C	p.Ile1027Thr
E19 (17a)	M1028R	c.3083T>G	p.Met1028Arg
E19 (17a)	Y1032C	c.3095A>G	p.Tyr1032Cys

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E19 (17a)	Q1042X	c.3124C>T	p.Gln1042X
E19 (17a)		c.3131A>G	p.Glu1044Gly
IVS19 (17a)	3271+ 1G- >A	c.3139+1G>A	
IVS19 (17a)	3271+ 18C/T	c.3139+18C>T	
IVS19 (17a)	3272- 93T/C	c.3140-92T>C	
IVS19 (17a)	3272- 26A- >G	c.3140-26A>G	
E20 (17b)	W1063X	c.3189G>A	p.Trp1063X
E20 (17b)	L1065P	c.3194T>C	p.Leu1065Pro
E20 (17b)	R1066C	c.3196C>T	p.Arg1066Cys
E20 (17b)	A1067T	c.3199G>A	p.Ala1067Thr
E20 (17b)	R1070W	c.3208C>T	p.Arg1070Trp
E20 (17b)	F1074L	c.3222T>A	p.Phe1074Leu
E20 (17b)		c.3263dup	p.Asn1088LysfsX68
E20 (17b)	Y1092X(C- >A)	c.3276C>A	p.Tyr1092X
E20 (17b)	3417A/T	c.3285A>T	p.Thr1095Thr
E20 (17b)	R1102X	c.3304A>T	p.Arg1102X
E20 (17b)	E1104X	c.3310G>T	p.Glu1104X
IVS20 (17b)	3499+ 37G/A	c.3367+37G>A	
IVS20 (17b)	3500- 140A/C	c.3368-140A>C	
E21 (18)	3532AC- >GTA	c.3400_3401delinsGTA	p.Thr1134ValfsX22
E21 (18)	D1152H	c.3454G>C	p.Asp1152His
E21 (18)	V1153E	c.3458T>A	p.Val1153Glu
E21 (18)	3600G- >A	c.3468G>A	p.Leu1156Leu
IVS21 (18)	3601- 65C/A	c.3469-65C>A	
E22 (19)	CFTRdele19	c.3469_3717del	p.Met1157_Arg1239del
E22 (19)	R1158X	c.3472C>T	p.Arg1158X
E22 (19)	R1162X	c.3484C>T	p.Arg1162X
E22 (19)	3617G/T	c.3485G>T	p.Arg1162Leu
E22 (19)	D1168G	c.3503A>G	p.Asp1168Gly
E22 (19)	3659delC	c.3528delC	p.Lys1177SerfsX15
E22 (19)	3737delA	c.3605delA	p.Asp1202AlafsX9
E22 (19)	S1206X(C>A)	c.3617C>A	p.Ser1206X
E22 (19)	3750delAG	c.3618_3619delAG	p.Gly1208ProfsX56
E22 (19)	3755delG	c.3623delG	p.Gly1208AlafsX3
E22 (19)	I1234V	c.3700A>G	p.Ile1234Val
E22 (19)	S1235R	c.3705T>G	p.Ser1235Arg

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IVS22 (19)	3849+ 40A- >G	c.3717+40A>G	
IVS22 (19)	3849+ 10kbC- >T	c.3718-2477C>T	
IVS22 (19)	3850- 1G- >A	c.3718-1G>A	
E23 (20)	G1244E	c.3731G>A	p.Gly1244Glu
E23 (20)	G1249E	c.3746G>A	p.Gly1249Glu
E23 (20)	S1251N	c.3752G>A	p.Ser1251Asn
E23 (20)	3905insT	c.3773dup	p.Leu1258PhefsX7
E23 (20)	D1270N	c.3808G>A	p.Asp1270Asn
E23 (20)	W1274X	c.3822G>A	p.Trp1274X
E23 (20)	W1282X	c.3846G>A	p.Trp1282X
E23 (20)	P1290P (4002A/G)	c.3870A>G	p.Pro1290Pro
IVS23 (20)	4005+ 1G- >A	c.3873+1G>A	
IVS23 (20)	4006- 200G/A	c.3874-200G>A	
E24 (21)	4010delI	c.3883_3886delATTT	p.Ile1295PhefsX32
E24 (21)	4015delA	c.3883delA	p.Ile1295PhefsX33
E24 (21)	4016insT	c.3889dup	p.Ser1297PhefsX5
E24 (21)	4029A/G	c.3897A>G	p.Thr1299Thr
E24 (21)	N1303H	c.3907A>C	p.Asn1303His
E24 (21)	N1303K	c.3909C>G	p.Asn1303Lys
E24 (21)	D1305E	c.3915T>A	p.Asp1305Glu
E24 (21)	Q1313X	c.3937C>T	p.Gln1313X
IVS24 (21)		c.3963+69A>G	
IVS24 (21)	4096- 283T/C	c.3964-283T>C	
IVS24 (21)	4096- 3C- >G	c.3964-3C>G	
E25 (22)	CFTRdele22, 23	c.3964_4242del	p.Val1322_Leu1414del
E25 (22)	Del exon 22- 24	c.3964_4440del	p.Val1322_Leu1480del
E25 (22)	G1349D	c.4046G>A	p.Gly1349Asp
E25 (22)	Q1352H(G- >C)	c.4056G>C	p.Gln1352His
E25 (22)	4218insT	c.4086dup	p.Lys1363X
E25 (22)	A1364V	c.4091C>T	p.Ala1364Val
E25 (22)	I1366T	c.4097T>C	p.Ile1366Thr
E25 (22)	D1377H	c.4129G>C	p.Asp1377His
IVS25 (22)	4269- 139G/A	c.4137-139G>A	
E26 (23)	Q1382X	c.4144C>T	p.Gln1382X
E26 (23)	4279insA	c.4147dup	p.Ile1383AsnfsX3
E26 (23)	Q1390X	c.4168C>T	p.Gln1390X

\*cDNA Reference sequence **NM\_000492.3** with the exception of c.1408A instead of G

\*\*Protein Reference sequence **NP\_000483.3** with the exception of p.Met470 instead of p.Val470

#exon/intron (#usual)	Usual name (CFMD)	c.DNA nomenclature UMD*	Predicted protein UMD**
E26 (23)	4332delTG	c.4200_4201delTG	p.Cys1400X
IVS26 (23)	4374+ 1G- >A	c.4242+1G>A	
IVS26 (23)	4374+ 13A/G	c.4242+13A>G	
IVS26 (23)	4374_4374+ 1GG>TT	c.4242_4242+1delinsTT	
E27 (24)	4382delA	c.4251delA	p.Glu1418ArgfsX14
E27 (24)	R1422W	c.4264C>T	p.Arg1422Trp
E27 (24)	4404C/T	c.4272C>T	p.Tyr1424Tyr
E27 (24)	S1426P	c.4276T>C	p.Ser1426Pro
E27 (24)	4521G/A	c.4389G>A	p.Gln1463Gln
E27 (24)	Q1476X	c.4426C>T	p.Gln1476X