Traditional and Human Genome Variation Society (HGVS) Standard Nomenclature for Cystic Fibrosis Transmembrane Conductance Regulator (*CFTR)* Mutations and Variants Included on the Michigan Newborn Screening Panel

American College of Medical Genetics (ACMG) Recommended Mutations		
Legacy Name, (xTAG CF Kit Name)	**HGVS Nomenclature**	
	DNA Name	**Protein Name**
deltaF508, (dF508)	c.1521_1523delCTT	p.Phe508del
R117H	c.350G>A	p.Arg117His
3120+1G>A	c.2988+1G>A	no protein name
G551D	c.1652G>A	p.Gly551Asp
G542X	c.1624G>T	p.Gly542X
N1303K	c.3909C>G	p.Asn1303Lys
R553X	c.1657C>T	p.Arg553X
W1282X	c.3846G>A	p.Trp1282X
1898+1G>A	c.1766+1G>A	no protein name
1717-1G>A	c.1585-1G>A	no protein name
621+1G>T	c.489+1G>T	no protein name
A455E	c.1364C>A	p.Ala455Glu
3849+10kbC>T	c.3717+12191C>T	no protein name
2184delA	c.2052delA	p.Lys684AsnfsX38
R347P	c.1040G>C	p.Arg347Pro
2789+5G>A	c.2657+5G>A	no protein name
R334W	c.1000C>T	p.Arg334Trp
R1162X	c.3484C>T	p.Arg1162X
deltaI507, (dI507)	c.1519_1521delATC	p.Ile507del
G85E	c.254G>A	p.Gly85Glu
R560T	c.1679G>C	p.Arg560Thr
3659delC	c.3528delC	p.Lys1177SerfsX15
711+1G>T	c.579+1G>T	no protein name
5T	*c.1210-12[5]*	*no protein name*
7T	*c.1210-12[7]*	*no protein name*
*9T**	*not available*	*not available*
F508C	*c.1523T>G*	*p.Phe508Cys*
I507V	*c.1519A>G*	*p.Ile507Val*
I506V	*c.1516A>G*	*p.Ile506Val*

Italics refers to ACMG recommended variants/polymorphisms identified in the CFTR gene.

*Not found in CFTR databases, but to be included in testing according to the ACMG guidelines. Based on HGVS nomenclature of the 5T and 7T variants, the HGVS name for the 9T variant would be c.1210-12[9].

Most Common Additional Mutations

Legacy Name, (xTAG CF Kit Name)	HGVS Nomenclature	
	DNA Name	Protein Name
1078delT	c.948delT	p.Phe316LeufsX12
394delTT	c.262_263delTT	p.Leu88IlefsX22
Y122X	c.366T>A	p.Tyr122X
R347H	c.1040G>A	p.Arg347His
V520F	c.1558G>T	p.Val520Phe
A559T	c.1675G>A	p.Ala559Thr
S549R T>G	c.1647T>G	p.Ser549Arg
S549N	c.1646G>A	p.Ser549Asn
1898+5G>T	c.1766+5G>T	no protein name
2183AA>G	c.2051_2052delAAinsG	p.Lys684SerfsX38
2307insA	c.2175_2176insA	p.Glu726ArgfsX4
Y1092X C>A	c.3276C>A	p.Tyr1092X
Y1092X C>G	c.3276C>G	p.Tyr1092X
M1101K	c.3302T>A	p.Met1101Lys
S1255X	c.3764C>A	p.Ser1255X
3876delA	c.3744delA	p.Lys1250ArgfsX9
3905insT	c.3773_3774insT	p.Leu1258PhefsX7

Broad Ethnic Coverage

Legacy Name, (xTAG CF Kit Name)	HGVS Nomenclature	
	DNA Name	Protein Name
CFTRdele2,3 ,(del e2e3)	c.54-5940_273+10250del21kb	p.Ser18ArgfsX16
W1089X	c.3266G>A	p.Trp1089X
1677delTA	c.1545_1546delTA	p.Tyr515X
D1152H	c.3454G>C	p.Asp1152His
R1158X	c.3472C>T	p.Arg1158X
G178R	c.532G>A	p.Gly178Arg
3791delC	c.3659delC	p.Thr1220LysfsX8
L206W	c.617T>G	p.Leu206Trp
E60X	c.178G>T	p.Glu60X
R75X	c.223C>T	p.Arg75X
Q493X	c.1477C>T	p.Gln493X
2055del9>A	c.1923_1931del9insA	p.Ser641ArgfsX5
S1196X	c.3587C>G	p.Ser1196X
935delA	c.803delA	p.Asn268IlefsX17
2143delT	c.2012delT	p.Leu671X
K710X	c.2128A>T	p.Lys710X
G330X	c.988G>T	p.Gly330X
Q890X	c.2668C>T	p.Gln890X
R1066C	c.3196C>T	p.Arg1066Cys
3199del6	c.3067_3072delATAGTG	p.Ile1023_Val1024del
406-1G>A	c.274-1G>A	no protein name

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3) Den Dunnen, JT, Antonarakis, SE. Mutation nomenclature extensions and suggestions to describe complex mutations: A discussion. Human Mutation 2000;15(1): 7-12.

4) Nomenclature of Cystic Fibrosis Sequence Variants in Luminex xTAG Cystic Fibrosis Kits [White Paper WP612.0616]. Luminex Corporation 2013-2016. Cited 2016 Aug. Available from https://www.luminexcorp.com/clinical/genetic-testing/cystic-fibrosis/resources/

5) Luminex, The Leader in Cystic Fibrosis Testing, The test choice for more laboratories providing broad coverage from the trusted leader in CF testing. [SS_496.06_0114]. Luminex Corporation 2010-2014. Cited 2016 Aug. Available from https://www.luminexcorp.com/clinical/genetic-testing/cystic-fibrosis/resources/