Traditional and Human Genome Variation Society (HGVS) Standard Nomenclature for Cystic Fibrosis Transmembrane Conductance Regulator (*CFTR;* NM_000492.3*)* Mutations and Variants Included on the Michigan Newborn Screening Panel.

Michigan Cystic Fibrosis DNA Mutation Panel		
Legacy Name, (Illumina 139-variant CF Kit Name)	**HGVS Nomenclature**	
	DNA Name	**Protein Name**
M1V	c.1A>G	no protein name
CFTRdele2,3	c.54-5940_273+10250 del21kb	p.Ser18ArgfsX16
Q39X	c.115C>T	p.Gln39X
E60X	c.178G>T	p.Glu60X
P67L	c.200C>T	p.Pro67Leu
R75X	c.223C>T	p.Arg75X
G85E	**c.254G>A**	**p.Gly85Glu**
394delTT	c.262_263delTT	p.Leu88IlefsX22
405+1 G>A	c.273+1G>A	no protein name
406-1G>A	c.274-1G>A	no protein name
E92K	c.274G>A	p.Glu92Lys
E92X	c.274G>T	p.Glu92X
Q98X	c.292C>T	p.Gln98X
457TAT>G	c.325_327delTATinsG	p.Tyr109GlyfsX4
D110H	c.328G>C	p.Asp110His
R117C	c.349C>T	p.Arg117Cys
R117H	**c.350G>A**	**p.Arg117His**
Y122X	c.366T>A	p.Tyr122X
574delA	c.442delA	p.Ile148LeufsX5
621+1G>T	**c.489+1G>T**	**no protein name**
663delT	c.531delT	p.Ile177MetfsX12
G178R	c.532G>A	p.Gly178Arg
711+1G>T	**c.579+1G>T**	**no protein name**
711+3A>G	c.579+3A>G	no protein name
711+5G>A	c.579+5G>A	no protein name
712-1G>T	c.580-1G>T	no protein name
H199Y	c.595C>T	p.His199Tyr
P205S	c.613C>T	p.Pro205Ser
L206W	c.617T>G	p.Leu206Trp
Q220X	c.658C>T	p.Gln220X
852del22	c.720_741del22	p.Gly241GlufsX13
1078delT	c.948delT	p.Phe316LeufsX12
G330X	c.988G>T	p.Gly330X
R334W	**c.1000C>T**	**p.Arg334Trp**
I336K	c.1007T>A	p.Ile336Lys
T338I	c.1013C>T	p.Thr338Ile

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	DNA Name	**Protein Name**
S341P	c.1021T>C	p.Ser341Pro
1154insTC	c.1022_1023insTC	p.Phe342HisfsX28
R347P	**c.1040G>C**	**p.Arg347Pro**
R347H	c.1040G>A	p.Arg347His
R352Q	c.1055G>A	p.Arg352Gln
1213delT	c.1081delT	p.Trp361GlyfsX8
1248+1G>A	c.1116+1G>A	no protein name
1259insA	c.1127_1128insA	p.Gln378AlafsX4
W401X(c.1202G>A)	c.1202G>A	p.Trp401X
W401X(c.1203G>A)	c.1203G>A	p.Trp401X
1341+1G>A	c.1209+1G>A	no protein name
5T*	c.1210-12T[5]	no protein name
7T*	c.1210-12T[7]	no protein name
9T*	c.1210-12T[9]	no protein name
1461ins4	c.1329_1330insAGAT	p.Ile444ArgfsX3
A455E	**c.1364C>A**	**p.Ala455Glu**
1525-1G>A	c.1393-1G>A	no protein name
S466X(C>A)	c.1397C>A	p.Ser466X
S466X(C>G)	c.1397C>G	p.Ser466X
L467P	c.1400T>C	p.Leu467Pro
1548delG	c.1418delG	p.Gly473GlufsX54
S489X	c.1466C>A	p.Ser489X
S492F	c.1475C>T	p.Ser492Phe
Q493X	c.1477C>T	p.Gln493X
I506V**	**c.1516A>G**	**p.Ile506Val**
I507del	**c.1519_1521delATC**	**p.Ile507del**
I507V**	**c.1519A>G**	**p.Ile507Val**
F508del	**c.1521_1523delCTT**	**p.Phe508del**
F508C**	**c.1523T>G**	**p.Phe508Cys**
1677delTA	c.1545_1546delTA	p.Tyr515X
V520F	c.1558G>T	p.Val520Phe
Q525X	c.1573C>T	p.Gln525X
1717-8G>A	c.1585-8G>A	no protein name
1717-1G>A	**c.1585-1G>A**	**no protein name**
G542X	**c.1624G>T**	**p.Gly542X**
S549R(c.1645A>C)	c.1645A>C	p.Ser549Arg
S549N	c.1646G>A	p.Ser549Asn
S549R(c.1647T>G)	c.1647T>G	p.Ser549Asn
G551D	**c.1652G>A**	**p.Gly551Asp**

Michigan Cystic Fibrosis DNA Mutation Panel		
Legacy Name, (Illumina 139-variant CF Kit Name)	**HGVS Nomenclature**	
	DNA Name	**Protein Name**
Q552X	c.1654C>T	p.Gln552X
R553X	**c.1657C>T**	**p.Arg553X**
A559T	c.1675G>A	p.Ala559Thr
R560K	c.1679G>A	p.Arg560Lys
R560T	**c.1679G>C**	**p.Arg560Thr**
1811+1.6kbA>G	c.1679+1.6kbA>G	no protein name
1812-1 G>A	c.1680-1G>A	no protein name
E585X	c.1753G>T	p.Glu585X
1898+1G>A	**c.1766+1G>A**	**no protein name**
1898+3A>G	c.1766+3A>G	no protein name
2143delT	c.2012delT	p.Leu671X
2183AA>G	c.2051_2052delAAinsG	p.Lys684SerfsX38
2184insA	c.2052_2053insA	p.Gln685ThrfsX4
2184delA	**c.2052delA**	**p.Lys684AsnfsX38**
R709X	c.2125C>T	p.Arg709X
K710X	c.2128A>T	p.Lys710X
2307insA	c.2175_2176insA	p.Glu726ArgfsX4
L732X	c.2195T>G	p.Leu732X
2347delG	c.2215delG	p.Val739TyrfsX16
R764X	c.2290C>T	p.Arg764X
2585delT	c.2453delT	p.Leu818TrpfsX3
E822X	c.2464G>T	p.Glu822X
2622+1G>A	c.2490+1G>A	no protein name
E831X	c.2491G>T	p.Glu831X
W846X	c.2537G>A	p.Trp846X
R851X	c.2551C>T	p.Arg851X
2711delT	c.2583delT	p.Phe861LeufsX3
2789+5G>A	**c.2657+5G>A**	**no protein name**
Q890X	c.2668C>T	p.Gln890X
L927P	c.2780T>C	p.Leu927Pro
S945L	c.2834C>T	p.Ser945Leu
3007delG	c.2875delG	p.Ala959HisfsX9
G970R	c.2908G>C	p.Gly970Arg
3120G>A	c.2988G>A	no protein name
3120+1G>A	**c.2988+1G>A**	**no protein name**
3121-1G>A	c.2989-1G>A	no protein name
3272-26A>G	c.3140-26A>G	no protein name
L1065P	c.3194T>C	p.Leu1065Pro
R1066C	c.3196C>T	p.Arg1066Cys

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Legacy Name, (Illumina 139-variant CF Kit Name)	**HGVS Nomenclature**	
	DNA Name	**Protein Name**
R1066H	c.3197G>A	p.Arg1066His
L1077P	c.3230T>C	p.Leu1077Pro
W1089X	c.3266G>A	p.Trp1089X
Y1092X(C>A)	c.3276C>A	p.Tyr1092X
Y1092X(C>G)	c.3276C>G	p.Tyr1092X
M1101K	c.3302T>A	p.Met1101Lys
E1104X	c.3310G>T	p.Glu1104X
R1158X	c.3472C>T	p.Arg1158X
R1162X	**c.3484C>T**	**p.Arg1162X**
3659delC	**c.3528delC**	**p.Lys1177SerfsX15**
S1196X	c.3587C>G	p.Ser1196X
W1204X(c.3611G>A)	c.3611G>A	p.Trp1204X
W1204X(c.3612G>A)	c.3612G>A	p.Trp1204X
3791delC	c.3659delC	p.Thr1220LysfsX8
3849+10kbC>T	**c.3717+12191C>T**	**no protein name**
G1244E	c.3731G>A	p.Gly1244Glu
3876delA	c.3744delA	p.Lys1250ArgfsX9
S1251N	c.3752G>A	p.Ser1251Asn
3905insT	c.3773_3774insT	p.Leu1258PhefsX7
W1282X	**c.3846G>A**	**p.Trp1282X**
4005+1G>A	c.3873+1G>A	no protein name
4016insT	c.3884_3885insT	p.Ser1297PhefsX5
N1303K	**c.3909C>G**	**p.Asn1303Lys**
Q1313X	c.3937C>T	p.Gln1313X
CFTRdele22,23	c.3964-78_4242+577del	no protein name
4209TGTT>AA	c.4077_4080delTGTTinsAA	no protein name
4382delA	c.4251delA	p.Glu1418ArgfsX14

Bold indicates American College of Medical Genetics (ACMG) recommended 23 mutations and 6 variants/polymorphisms identified in the CFTR gene.

* Conditionally reported when R117H Present

** Benign variant, conditionally reported with either dI507 or dF508 homozygous result

Blue highlight indicates newly added *CFTR* variants to the MI NBS 2nd tier panel as of March of 2025.

REFERENCES

1) Watson, MS, et al. Cystic fibrosis population carrier screening: 2004 revision of American College of Medical Genetics mutation panel. Genetics in Medicine 2004; 6(5): 387-391.

2) Illumina TruSight Cystic Fibrosis *CFTR* 139-variant package insert. Document# 1000000097720 v04 (Oct 2023)