



**Missouri Department of Health and Senior Services**

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**Randall W. Williams, MD, FCOG**  
Director



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Governor

The Missouri State Public Health Lab (Newborn Screening) will conduct second tier testing for the following CFTR mutations and 4 variants for Cystic Fibrosis (CF) when an elevated IRT is detected in the primary screening test:

deltaF508(dF508)*	1717-1G>A*	W1282X*	2307insA
deltaI507*	R560T*	1078delT	Y1092X
G542X*	R553X*	394delTT	M1101K
G85E*	G551D*	Y122X	S1255X
R117H*	1898+1G>A*	R347H	3876delA
621+1G>T*	2184delA*	V520F	3905insT
711+1G>T*	2789+5G>A*	A559T	5T/7T/9T
N1303K*	3120+1G>A*	S549N	F508C
R334W *	R1162X*	S549R	I507V
R347P*	3659delC*	1898+5G>T	I506V
A455E*	3849+10kbC>T*	2183AA>G	

This panel includes 39 of the most common CF mutations (including the 23 mutations recommended by the American College of Medical Genetics\*).

[www.health.mo.gov](http://www.health.mo.gov)

**Healthy Missourians for life.**

The Missouri Department of Health and Senior Services will be the leader in promoting, protecting and partnering for health.

AN EQUAL OPPORTUNITY / AFFIRMATIVE ACTION EMPLOYER: Services provided on a nondiscriminatory basis.