

## Missouri Department of Health and Senior Services

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Jeremiah W. (Jay) Nixon Governor

Gail Vasterling Acting Director

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

A455E*	R117H*	V520F	1898+1G>A*
deltaF508*	R334W*	W1282X*	2183AA>G
deltal507*	R347H	Y122X	2184delA*
D1152H	R347P*	Y1092X C>A	2789+5G>A*
E60X	R553X*	Y1092X C>G	3120+1G>A*
F508C	R560T*	394delTT	3659delC*
G85E*	R1162X*	621+1G>T*	3849+4A>G
G542X*	S549N	711+1G>T*	3849+10kbC>T*
G551D*	S549R A>C	1078delT	3876delA
N1303K*	S549R T>G	1717-1G>A*	3905insT
Q493X			

This panel includes 40 of the most common CF mutations (including the 23 mutations recommended by the American College of Medical Genetics\*).