

Cystic fibrosis (CFTR) – newborn screening

Panel information

Gene panel	Cystic fibrosis (CFTR) – newborn screening
Version	1
Total variants	12
Activation date	01/01/2026
Publisher	Center for Medical Genetics, Ghent

Variants

c.HGVS	p.HGVS	Legacy
c.1521_1523del	p.Phe508del	F508del
c.3909C>G	p.Asn1303Lys	N1303K
c.3140-26A>G	p.?	3272-26A->G
c.1585-1G>A	p.?	1717-1G->A
c.1624G>T	p.Gly542Ter	G542X
c.2657+5G>A	p.?	2789+5G->A
c.1657C>T	p.Arg553Ter	R553X
c.1364C>A	p.Ala455Glu	A455E
c.3718-2477C>T	p.?	3849+10kbC->T
c.3846G>A	p.Trp1282Ter	W1282X
c.3484C>T	p.Arg1162Ter	R1162X
c.3752G>A	p.Ser1251Asn	S1251N