

: 1

1	CFTR: 1677delTA	N / N	
2	CFTR: 2143delT	N / N	
3	CFTR: 2184insA	N / N	
4	CFTR: 3821delT	N / N	
5	CFTR: 3849+10kbC>T	N / N	
6	CFTR: 3944delGT	N / N	
7	CFTR: 394delTT	N / N	
8	CFTR: dele2.3(21kb)	N / N	
9	CFTR: E92K	N / N	
10	CFTR: F508del	N / N	
11	CFTR: G542X	N / N	
12	CFTR: N1303K	N / N	
13	CFTR: R334W	N / N	
14	CFTR: S466X	N / N	
15	CFTR: W1282X	N / N	
16	GALT: K285N	N / N	
17	GALT: N314D	N / N	
18	GALT: Q188R	N / N	
19	GJB2: 35delG	N / N	
20	PAH: D222X	N / N	
21	PAH: E280K	N / N	
22	PAH: IVS10nt54b	N / N	
23	PAH: IVS12+1G>A	N / N	
24	PAH: IVS4+5G>T	N / N	
25	PAH: P281L	N / N	
26	PAH: R158Q	N / N	
27	PAH: R252W	N / N	
28	PAH: R261Q	N / N	
29	PAH: R408W	N / N	
30	PAH: Y414C	N / N	

CFTR(1677delTA,2143delT,2184insA,3821delT,3849+10kbC>T,3944delGT,394delTT,dele2.3(21kb),E92K,F508del,G542X,N1303K,R334W,S466X,W1282X), GALT(K285N,N314D,Q188R), GJB2(35delG), PAH(D222X,E280K,IVS10nt54b,IVS12+1G>A,IVS4+5G>T,P281L,R158Q,R252W,R261Q,R408W,Y414C)



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: 2

1	CFTR: 1677delTA	N / N	
2	CFTR: 2143delT	N / N	
3	CFTR: 2184insA	N / N	
4	CFTR: 3821delT	N / N	
5	CFTR: 3849+10kbC>T	N / N	
6	CFTR: 3944delGT	N / N	
7	CFTR: 394delTT	N / N	
8	CFTR: dele2.3(21kb)	N / N	
9	CFTR: E92K	N / N	
10	CFTR: F508del	N / N	
11	CFTR: G542X	N / N	
12	CFTR: N1303K	N / m	
13	CFTR: R334W	N / N	
14	CFTR: S466X	N / N	
15	CFTR: W1282X	N / N	
16	GALT: K285N	N / N	
17	GALT: N314D	N / N	
18	GALT: Q188R	N / N	
19	GJB2: 35delG	N / N	
20	PAH: D222X	N / N	
21	PAH: E280K	N / N	
22	PAH: IVS10nt54b	N / N	
23	PAH: IVS12+1G>A	N / N	
24	PAH: IVS4+5G>T	N / N	
25	PAH: P281L	N / N	
26	PAH: R158Q	N / N	
27	PAH: R252W	N / N	
28	PAH: R261Q	N / N	
29	PAH: R408W	N / N	
30	PAH: Y414C	N / N	

: CFTR(N1303K).

CFTR(1677delTA,2143delT,2184insA,3821delT,3849+10kbC>T,3944delGT,394delTT,dele2.3(21kb),E92K,F508del,G542X,R334W,S466X,W1282X), GALT(K285N,N314D,Q188R), GJB2(35delG), PAH(D222X,E280K,IVS10nt54b,IVS12+1G>A,IVS4+5G>T,P281L,R158Q,R252W,R261Q,R408W,Y414C)