Table 2 (continued): Detection rate of CF-causing *CFTR* mutations in different countries of the world

COUNTRIES	Detection rate (a,b) (n)	Number of mutations (c)	Detection rate of most frequent mutations (b,d)	Number of most frequent mutations (e)	Mutations (proportion) (f)
NORTH AMERICA					
Canada	0.897 (6632)	142	0.3%: 0.847 0.2%: 0.857 0.1%: 0.875	0.3%: 16 0.2%: 21 0.1%: 39	F508del(+I507del) (0.714) / 621+1G>T (0.029) / G542X, G551D (0.015 each) / A455E (0.011) / 711+1G>T (0.010) N1303K (0.009) / M1101K (0.007) / R117H, W1282X (0.006 each) / G85E, I148T (0.005 each) L206W, 1717-1G>A, R553X (0.004 each) / 3849+10kbC>T (0.003) / P67L, R560T, 2789+5G>A, Y1092X, 3659delC (0.002 each) E60X, R75X, 1078delT, R334W, R347H, R347P (0.001 each) / Q493X, V520F, P574H, 1898+1G>A, 2184delA (0.001 each) 2184insA, L1077P, R1162X, 3905insT, 4016insT, Q1313X, CFTRdele2,3(21kb) (0.001 each)
USA	0.880 (37602)	300	0.3%: 0.825 0.2%: 0.855 0.1%: 0.863	0.3%: 15 0.2%: 30 0.1%: 38	F508del (0.686) / G542X (0.024) / G551D (0.021) / 3120+1G>A (0.015) / W1282X (0.014) / N1303K (0.013) 621+1G>T, R553X (0.009 each) / R117H, 1717-1G>A, 3849+10kbC>T (0.007 each) / 1898+1G>T (0.004) G85E, I507del, 2789+5G>A (0.003 each) / R75X, 405+3A>C, 406-1G>A, I148T (0.002 each) F311del, R334W, R347P, A455E, G480C, A559T, R560T, 2307insA, R1162X, 3659delC, S1255X (0.002 each) / 711+1G>T, 935delA, I506T, S549N, 2055del9>A, 2183AA>G, 2184delA, 3199del6 (0.001 each)
LATIN AMERICA					
Argentina	0.724 (832)	20	0.5%: 0.698 0.2%: 0.712	0.5%: 6 0.2%: 11	F508del (0.591) / G542X (0.053) / W1282X (0.025) / N1303K (0.018) / R553X (0.006) / R1162X (0.005) R334W, 1717-1G>A (0.004 each) / 2566insT, 2789+5G>A, 3849+10kbC>T (0.002 each)
Brazil	0.618 (422)	11	0.612	8	F508del (0.436) / G542X (0.055) / R1162X (0.033) / G85E (0.026) / W1282X (0.019) / R334W, N1303K (0.017 each) / R553X (0.009)
Chile	*0.333 (72)*	2	0.333	2	F508del (0.292) / R553X (0.042)
Colombia	0.615 (218)	19	0.615	19	F508del (0.413) / 1811+1.6kbA>G (0.055) / G542X (0.041) / S549R (0.018) / W1282X (0.014) / R1162X, N1303K (0.009 each) / *R334W, 1215delG, 1323insA, A559T, H609R, 2185insC, 2789+5G>A (0.005 each) / 3120+1G>A, R1066C, Y1092X, 3500-2A>G, 3849+1G>A (0.005 each)*
Cuba	0.340 (144)	1	0.340	1	F508del (0.340)
Ecuador	*0.389 (82)*	4	0.389	4	F508del (0.329) / G85E, G542X (0.024 each) / *N1303K (0.012)*
Mexico	0.693 (274)	35	0.615	14	F508del (0.420) / G542X (0.058) / S549N (0.022) / I507del, N1303K (0.018 each) / R75X, 406-1G>A, I148T (0.011 each) / 3849+10kbC>T (0.011) / 935delA, I506T, 2055del9>A, 2183AA>G, 3199del6 (0.007 each)
Uruguay	*0.855 (76)*	12	0.855	12	F508del (0.566) / G542X (0.079) / G85E, R1162X (0.039 each) / R334W, N1303K (0.026 each) *834G>A, I507del, 1717-1G>A, R553X, 2789+5G>A, R1066C (0.013 each)*
Venezuela	*0.333 (54)*	2	0.333	2	F508del (0.296) / G542X (0.037)
MIDDLE EAST					
Bahrain	*0.808 (26)*	8	0.808	8	2043delG (0.308) / H139L (0.192) / F508del, N1303K (0.077 each) / *1161delC, G542X, 3120+1G>A, K1177X (0.038)*
Israel	0.910 (366)	13	0.910	12	W1282X (0.361) / F508del (0.320) / G542X (0.066) / 3849+10kbC>T (0.046) / 405+1G>A (0.036) / N1303K (0.030) Q359K-T360K (0.019) / G85E, D1152H (0.008 each) / 1717-1G>A, S549R(T>G), W1098X(TAG) (0.006 each)
Jordan (g)	0.746 (122)	17	0.746	17	1548delG (0.172) / I1234V (0.123) / F508del (0.115) / 3120+1G>A (0.107) / H139L (0.057) / G115X, 711+1G>A (0.025 each) S549R(T>G) (0.025) / A141D, K1177X, N1303K (0.016 each) / *R75X, Q98H, 1248+1G>A, F533L, R553X, 1811+2T>C (0.008 each)*
Kuwait (g)	0.746 (122)	17	0.746	17	see mutation list from Jordan (g)
Lebanon	*0.975 (40)*	11	0.975	11	F508del (0.350) / W1282X (0.200) / 4010delTAAT, N1303K (0.100 each) / S4X, 4096-3C>G (0.050 each) / *E672del, 2789+5G>A, M952I, 3755delG, 4096-28G>A (0.025 each)*
Oman (g)	0.746 (122)	17	0.746	17	see mutation list from Jordan (g)
Qatar (g)	0.746 (122)	17	0.746	17	see mutation list from Jordan (g)
Saudi Arabia (g)	0.746 (122)	17	0.746	17	see mutation list from Jordan (g)
United Arab Emirates (g)	0.746 (122)	17	0.746	17	see mutation list from Jordan (g)
NORTH-AFRICA					
Algeria	*0.600 (20)*	5	0.600	5	F508del, N1303K (0.200 each) / 711+1G>T (0.100) / *1812-1G>A, V754M (0.050 each)*
Tunisia	*0.603 (78)*	11	0.603	11	F508del (0.180) / G542X (0.090) / 711+1G>T (0.077) / 2766del8, N1303K (0.064 each) G85E, R1066C, D1270N-R74W, W1282X (0.026 each) / *Y122X, T665S (0.013 each)*
AFRICA					
Reunion Island	0.904 (138)	9	0.904	9	F508del (0.520) / Y122X (0.240) / 3120+1G>A (0.080) / A455E (0.022) / G551D (0.014) / *A309G, 1717-1G>A, G542X, 2789+5G>A (0.007 each)*
South Africa	*0.744 (86)*	6	0.744	6	F508del (0.500) / 3120+1G>A (0.174) / G542X, G551D (0.023 each) / *3272-26A>G, R1162X (0.012 each)*
ASIA					
India	*0.523 (48)*	8	0.523	8	F508del (0.292) / 3849+10kbC>T (0.063) / 3601-20T>C, 3622insT (0.042 each) / *1161delC, R560H, Y569H, L997F (0.021 each)*
Japan	*1.000 (22)*	16	1.000	16	1540del10 (0.136) / 125G>C, F508del, T1086I, H1085R (0.091 each) / *R75X, Q98R, M152R, E217G, R347H, R347H-D979A, L441P, 1525-18G>A, 1742delAG, L571S, Q1352H (0.045 each)*
Pakistan (h)	*0.846 (52)*	11	0.846	11	F508del (0.192) / Y569D (0.154) / Q98X (0.115) / 1525-1G>A (0.096) / 296+12T>C, 1161delC (0.077 each) 621+2T>C, 2184insA (0.038 each) / *R560S, 1898+1G>T, R709X (0.019 each)*
OCEANIA					
Australia	0.887 (761)	8	0.887	8	F508del (0.769) / G551D (0.045) / G542X (0.028) / R553X (0.013) / 621+1G>T (0.011) N1303K (0.009) / R117H, W1282X (0.006 each)
New Zealand	0.874 (636)	5	0.874	5	F508del (0.780) / G551D (0.044) / G542X (0.020) / N1303K (0.019) / 621+1G>T (0.011)

Contributors of the mutation data are given in the references of the Bobadilla et al. study [28], however updated data contributed by Aulehla-Scholz C., Bal J.,Baranov V., Bartolo C., Barton D., Bauer I., Benga G., Bergsteinsson H., Bienvenu T.,Bieth E., Blavau M., Bonizzato A., Bozon D., Casals T., Castaldo G., Cheadle J., Chiba-Falek O., Claustres M., Cuppens H.,Deltas C., Desgeorges M., Dörk T., Efremov G., Eiklid C., Fekete G., Férec C., Froster U., Garami M., Gillfilan A., Ginter E., Glaeser D., Glavac D., Graham C., Hammer U., Harris A., Heinritz W., Huber K.,Hughes D., Iron A., Kaahre T., Kadasi L., Kere J.,Kerem B-S., Keyeux G., Kirdar B., Koceva S., Kouvatsi A., Krumina A., Kucinskas V., Liechti S., Livshits L., Macek M. Jr., Meredith L., Mittre H., Monnier N., Morgutti M., Naehrlich L., Novelli G., Onay T., Ozcelik U., Pacheco P., Padoan R., Pivetta O., Radivojevic D., Radojkovic D., Ravnik-Glavac M., Reis A., Restagno G., Rosatelli C., Sanguiolo F., Santostasi T., Savov A.,Scheffer H., Schwartz M., Schwarz M., Seia M., Sertic J., Sobczynska A., Stuhrmann M.,Teder M.,Telleria J.,Torricelli F.,Tsukermann G.,Tzetis M., Vouk K., Wagner K., Witt M.and Zielenski J. were used (Milan Macek, personal communication).

(a) Proportion of *CFTR* alleles derived from CF patients on which a mutation could be identified. These data are illustrated in figure 2.(n) denotes the total number of *CFTR* alleles that were studied. This detection rate for each country is the maximum detection rate obtained so far, irrespective of the sensitivity of the screening assays used. In some countries, lower detection rates are indeed obtained because of the use of screening assays that have low sensitivities. The data given in 'red/italics/underlined' refer to studies in which less than 100 *CFTR* alleles were studied (either because of an incomplete study, or because they deal with small populations in which less than 50 CF patients (100 *CFTR* alleles) exist), and each mutation therefore contributes for 1% or more.

(b) For the detection rate, a color code is used as given in figure 2.

(c) The total number of *CFTR* mutations that have been found in CF patients of the respective population.

(d) Proportion of *CFTR* alleles derived from CF patients on which a mutation could be identified if one only screens for the most common mutations. In general, a common mutation is defined as a mutation having a frequency of 0.5%, or higher. If not specified, mutations having a frequency of 0.5%, or higher, are only considered. When the total number of studied *CFTR* alleles exceeds 475, and if the total proportion of *CFTR* alleles on which a mutation can be identified is higher than 95%, all mutations found at least twice are also included, and the sensitivity of these is then specified (e.g. Czech Republic). For populations in which more than 950 *CFTR* alleles were studied, all mutations having a frequency of 0.3%, or higher, are included. When a relatively low number of mutations is observed in a given population, mutations having a frequency lower than 0.3%-0.5% may be included, and the sensitivity rate is then specified (e.g. Denmark). For the populations in which more than 5000 *CFTR* alleles were studied, all mutations having a frequency of 0.1-0.15%, or higher, are included. For some regions, more than one sensitivity rate may be given in order to allow better comparison between data (e.g. Canada). For Latin America, given the smaller number of *CFTR* alleles that were screened, all mutations found at least twice are included. For South Africa, Asia and some countries of the Middle East, were even a lower number of *CFTR* alleles have been studied, all mutations are given.

(e) The total number of common mutations that are found in the respective population. See also the remarks given in (d).

(f) The mutations given in 'red/italics/underlined' are only found once, but given the low number of mutations that were studied, they contribute for 0.5%, or more than 0.5%, of all *CFTR* alleles. They might still be private mutations in that given population.

(g) Data obtained from one study including patients from Jordan, Kuwait, Oman, Qatar, Saudi Arabia and the United Arab Emirates; therefore not all these mutations may be found in each country of that study.

(h) Data are derived from respective individuals living in another country, i.e. people of Pakistani origin living in the United Kingdom.