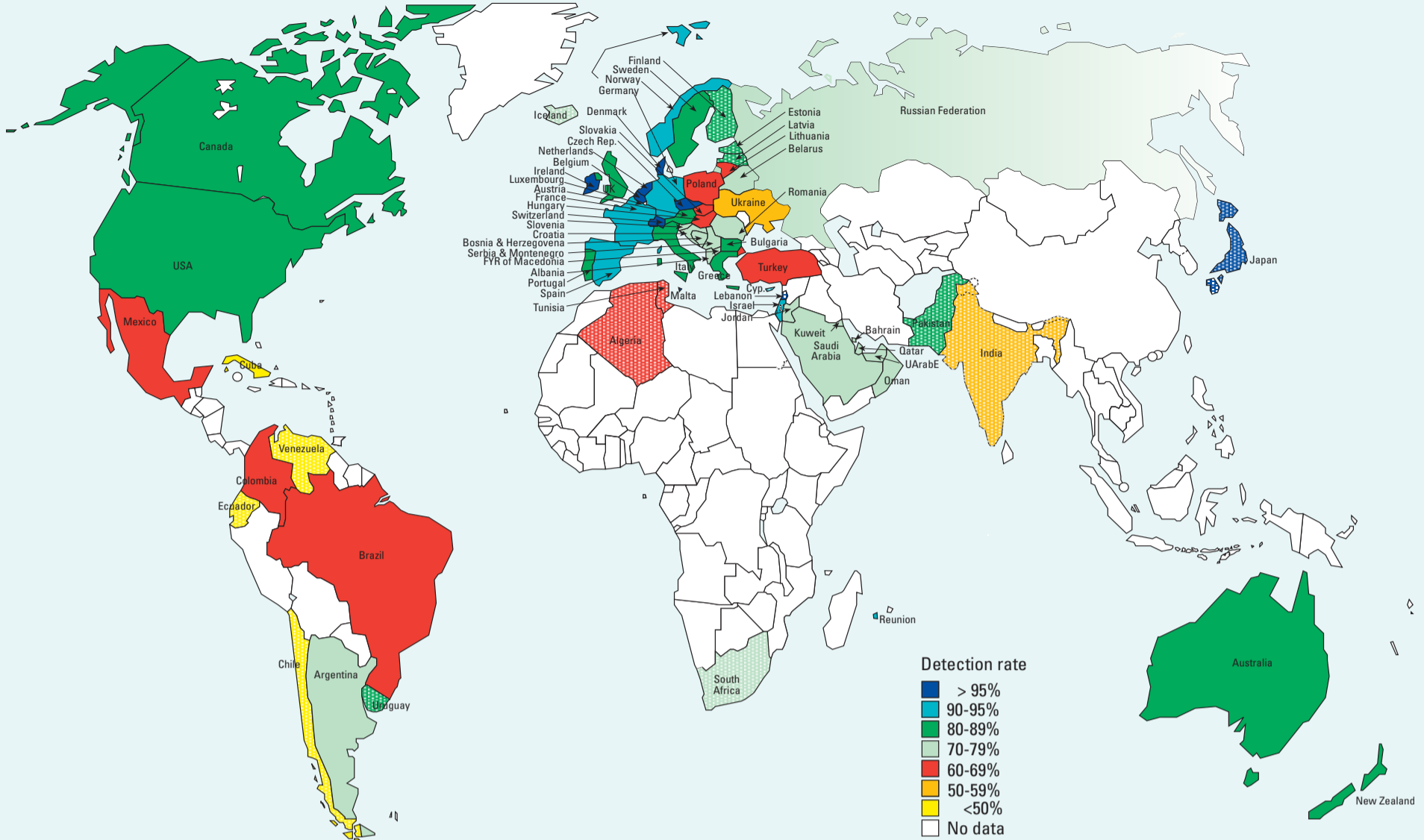


The molecular genetic epidemiology of cystic fibrosis

Figure 2: Detection rate of CF-causing *CFTR* mutations



The detection rate of CF-causing *CFTR* mutations, i.e. the proportion of *CFTR* alleles derived from CF patients on which a mutation can be identified, are given for the different countries of the world. This detection rate for each country is the maximum detection rate obtained so far, irrespective of the sensitivity of the screening assays used. A color code is used for different detection rates, as shown in the inset. Detailed numbers of the detection rates are given in table 2. The countries marked with a grid pattern in the map refer to studies in which less than 100 *CFTR* genes were studied. They might, therefore, be less representative. For the regions coloured in white, no data are available.

The designations employed and the presentation of material on this map do not imply the expression of any opinion whatsoever on the part of the World Health Organization concerning the legal status of any country, territory, city or area or of its authorities, or concerning the delimitation of its frontiers or boundaries. Dashed lines represent approximate border lines for which there may not yet be full agreement.

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Table 2: 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)
EUROPE					
Albania	0.719 (146)	4	0.719	4	F508del (0.699) / <i>G85E, C276X, R1070Q (0.007 each)</i>
Austria	0.851 (824)	35	0.812	13	F508del (0.652) / G542X (0.042) / R1162X (0.027) / G551D, CFTRdele2,3(21kb) (0.015 each) / R553X (0.012) / 457 / TAT>G (0.011) / R347P, 2183AA>G (0.008 each) / R117H (0.007) / 1148T, 621+1G>T, 3849+10kbC>T (0.005 each)
Belarus	0.731 (238)	9	0.714	5	F508del (0.609) / CFTRdele2,3(21kb) (0.059) / N1303K (0.025) / G542X (0.013) / W1282X (0.008)
Belgium	0.982 (392)	38	0.913	11	F508del (0.760) / G542X (0.038) / N1303K (0.028) / 1717-1G>A (0.018) / E60X, S1251N (0.015 each) / 2183AA>G (0.013) / W1282X (0.010) / W401X(TGA), A455E, 3272-26A>G (0.005 each)
Bulgaria	0.889 (506)	31	0.852	15	F508del (0.638) / N1303K (0.047) / G542X (0.038) / R347P (0.020) / 1677delTA (0.018) / R1070Q (0.016) / Q220X, 3849+10kbC>T (0.012 each) / W1282X, G1244V(+S912L) (0.010 each) / 2184insA (0.008) / G85E, 2183AA>G, 2789+5G>A, 4374+1G>A (0.006 each)
Croatia	0.739 (276)	9	0.725	5	F508del (0.645) / G542X (0.033) / N1303K (0.029) / G551D (0.011) / 3849+10kbC>T (0.007)
Cyprus	0.909 (44)	11	0.909	11	F508del (0.455) / L364P (0.136) / 1677delTA (0.068) / S549N, 3849+10kbC>T, N1303K (0.046 each) / <i>621+1G>T, Y301C, L346P, M348K, 2789+5G>A (0.023 each)</i>
Czech Republic	0.978 (494)	38	0.4%: 0.929	0.4%: 15	F508del (0.719) / CFTRdele2,3(21kb) (0.047) / G551D (0.041) / N1303K (0.030) / G542X, 1898+1G>A (0.022 each) / 2143delIT (0.012) / R347P (0.008) / W1282X (0.006) / E92X, 711+1G>T, 1717-1G>A, R1162X, 3849+10kbC>T, 4374+1G>A (0.004 each)
Denmark	0.989 (825)	39	0.25%: 0.962	0.25%: 17	F508del (0.867) / 394delITT (0.019) / N1303K (0.013) / 3659delC (0.010) / 621+1G>T (0.007) / G542X (0.006) / E60X, R117H, 1717-1G>A, W1282X (0.005 each) / 2790-1G>C, 3849+10kbC>T, CFTRdele2,3(21kb) (0.004 each) / R117C, R334W, L558S, 3056delGA (0.0025 each)
Estonia	0.800 (60)	10	0.800	10	F508del (0.517) / 394delITT (0.133) / S1235R (0.033) / <i>359insT, R117C, E217G, I1005R, R1066H, 3659delC, S1196X (0.017 each)</i>
Finland	0.800 (40)	4	0.800	4	F508del (0.450) / 394delITT (0.300) / <i>G542X, 3732delA (0.025 each)</i>
France	0.942 (7420)	359	0.15%: 0.856	0.15%: 36	F508del (0.672) / G542X (0.029) / N1303K (0.021) / 1717-1G>A (0.013) / G551D, 2789+5G>A (0.010 each) / R553X, W1282X (0.009 each) / 1078delIT, I507del, 2183AA>G (0.007 each) / 711+1G>T, Y1092X(C>A), R1162X (0.004 each) / 3659delC, 3849+10kbC>T (0.004 each) / G85E, 621+1G>T, R334W, R347P, W846X1, 3272-26A>G (0.003 each) / 394delITT, L206W, R347H, 1811+1.6kbA>G, S945L, R1066C, 3905insT, 4005+1G>A (0.002 each) / E60X, R117H, Y122X, 2184insA, 2711delIT, S1251N (0.0015 each)
Germany	0.941 (2324)	97	0.15%: 0.906	0.15%: 28	F508del (0.702) / N1303K (0.025) / G542X (0.021) / G551D (0.020) / R553X, CFTRdele2,3(21kb) (0.015 each) / R347P (0.014) / 1717-1G>A (0.012) / 3849+10kbC>T (0.011) / R117H (0.009) / 2789+5G>A (0.008) / 2143delIT, 2183AA>G (0.006 each) / 3272-26A>G (0.005) / W1282X, 3659delC (0.004 each) / Q39X, 1078delIT, R334W, I336K, Y1092X(C>A) (0.003 each) / M1101K, R1162X (0.003 each) / 1342-2A>C, I507del (0.002 each) / 394delITT, 621+1G>T, (0.0015 each)
2184insA					
Greece	0.895 (1006)	86	0.3%: 0.831	0.3%: 31	F508del (0.538) / 621+1G>T (0.066) / G542X (0.041) / N1303K (0.025) / 2789+5G>A (0.015) / E822X (0.014) / 2183AA>G (0.012) / R1158X (0.009) / R334W, 1677delTA, R1070Q (0.008 each) / G85E, W496X, 3272-26A>G (0.007 each) / 711+3A>G, I507del, W1282X (0.006 each) / 574delA, 621+3A>G, 3120+1G>A (0.005 each) / D110H, R553X (0.004 each) / A46D(+R3W), D110E, C276X, 1525-1G>A, G551D, 2184insA, 3120+1kb del8.6kb, 4382delA, CFTRdele2,3(21kb) (0.003 each)
Hungary	0.601 (1022)	14	0.3%: 0.594	0.3%: 7	F508del (0.527) / G542X (0.022) / CFTRdele2,3(21kb) (0.018) / N1303K (0.011) / R553X (0.009) / W1282X (0.005) / 2184insA (0.003)
Iceland	0.797 (59)	3	0.797	3	F508del (0.373) / N1303K (0.339) / 1078delIT (0.085)
Ireland	0.965 (1318)	35	0.15%: 0.956	0.15%: 23	F508del (0.764) / G551D (0.067) / R117H (0.030) / R560T (0.024) / 621+1G>T (0.016) / I507del (0.008) / 1717-1G>A (0.007) / G542X (0.006) / 1461insA, N1303K (0.004 each) / E60X, R352Q, V520F, 3007delG (0.003 each) / G85E, 3849+10kbC>T (0.002 each) / 1154insTC, 1471delA, 2622+1G>A, 2752-26A>G, R1162X, 3659delC, L1335P (0.0015 each)
Italy	0.879 (3873)	116	0.3%: 0.824	0.3%: 28	F508del (0.520) / N1303K (0.061) / G542X (0.052) / 2183AA>G (0.023) / 1717-1G>A (0.022) / R1162X (0.017) / W1282X (0.015) / 2789+5G>A (0.014) / R553X (0.011) / R347P (0.008) / G85E, 711+5G>A, D1152H (0.007 each) / 3849+10kbC>T (0.006) / L1065P, L1077P, R1158X (0.005 each) / R117H, 621+1G>T, 852del22, 1259insA, 4016insT, 4382delA (0.004 each) / R334W, T338I, D579G, R1066C, R1066H (0.003 each)
Latvia	0.806 (36)	6	0.806	6	F508del (0.583) / 3849+10kbC>T (0.083) / N1303K (0.056) / <i>394delITT, W1282X, CFTRdele2,3(21kb) (0.028 each)</i>
Lithuania	0.644 (118)	6	0.644	6	F508del (0.559) / R553X (0.034) / N1303K, CFTRdele2,3(21kb) (0.017 each) / <i>G314R, W1282X (0.008 each)</i>
Malta	1.000 (20)	3	1.000	3	F508del (0.800) / G542X (0.150) / <i>E831X (0.050)</i>
Macedonia	0.765 (226)	13	0.752	10	F508del (0.611) / G542X (0.049) / N1303K (0.022) / 621+1G>T, CFTRdele2,3(21kb) (0.013 each) / 457TAT>G, 711+3A>G, 2184insA, 3849G>A, V1397E (0.009 each)
Netherlands	0.975 (804)	38	0.3%: 0.950	0.3%: 18	F508del (0.792) / A455E (0.026) / G542X (0.016) / 1717-1G>A, S1251N (0.015 each) / R553X (0.014) / R1162X (0.011) / 3272-26A>G, W1282X (0.009 each) / E60X, 711+1G>T, 2789+5G>A, N1303K (0.008 each) / Y1092X(C>A) (0.004) / 1078delIT, 1898+5G>T, R1070Q, 3659delC (0.003 each)
Norway	0.901 (282)	18	0.869	9	F508del (0.709) / 394delITT (0.039) / R117C (0.028) / 4005+2T>C (0.025) / R117H (0.021) / 3659delC (0.018) / G551D (0.014) / G542X, N1303K (0.007 each)
Poland	0.691 (1726)	35	0.3%: 0.671	0.3%: 13	F508del (0.530) / 3849+10kbC>T (0.027) / G542X (0.025) / CFTRdele2,3(21kb) (0.020) / 1717-1G>A, N1303K (0.017 each) / R553X (0.010) / W1282X (0.006) / R560T, 2184insA (0.005 each) / R334W, G551D, 2143delIT (0.003 each)
Portugal	0.893 (494)	46	0.816	15	F508del (0.593) / A561E (0.032) / R334W, R1066C (0.028 each) / G542X (0.026) / G85E (0.022) / N1303K (0.018) / G576A (0.014) / 711+1G>T, 3272-26A>G (0.010 each) / Y1092X (0.008) / R74W, 621+1G>T, D1100P, I1234V (0.006 each)
Romania	0.758 (153)	19	0.758	19	F508del (0.588) / G542X, W1282X (0.026 each) / CFTRdele2,3(21kb) (0.020) / <i>457TAT>G, R117H, 621+1G>T, R334W (0.007 each)</i> / <i>1677delTA, 1717-2A>G, 2183AA>G, R735K, R785X, E831X, 3272-26A>G, R1162X, 3849+10kbC>T, N1303K (0.007 each)</i>
Russian Federation	0.743 (668)	21	0.731	13	F508del (0.534) / CFTRdele2,3(21kb) (0.057) / N1303K (0.027) / 2143delIT, 2184insA (0.020 each) / G542X, W1282X (0.017 each) / 3849+10kbC>T (0.011) / R334W (0.008) / 394delITT, 1677delTA, S1196X (0.006 each) / 3732delA (0.005)
Serbia-Montenegro	0.746 (622)	22	0.3%: 0.725	0.3%: 9	F508del (0.661) / G542X (0.034) / 621+1G>T (0.010) / S466X(TAA) (0.005) / 525delIT, G551D, 2789+5G>A, R1070Q, N1303K (0.003 each)
Slovakia	0.654 (410)	15	0.639	9	F508del (0.485) / G542X (0.037) / N1303K (0.029) / 3849+10kbC>T (0.027) / R553X, CFTRdele2,3(21kb) (0.020 each) / R347P (0.012) / 2143delIT, W1282X (0.005 each)
Slovenia	0.720 (132)	20	0.720	20	F508del (0.523) / R1162X (0.030) / Q552Q, Q685X, 3905insT, CFTRdele2,3(21kb) (0.015 each) / <i>S4X, 457TAT>G (0.008 each)</i> / <i>458delAT, D192G, R347P, G542X, S549L, R553X, A559T, R766M, R792G, 2907delIT, 3667insA, W1282X (0.008 each)</i>
Spain	0.920 (2006)	112	0.3%: 0.849	0.3%: 32	F508del (0.506) / G542X (0.084) / N1303K (0.026) / R334W (0.025) / R1162X (0.016) / 711+1G>T, 1811+1.6kbA>G (0.015 each) / 2789+5G>A (0.014) / Q890X (0.012) / R1066C (0.011) / I507del (0.010) / L206W (0.009) / W1282X (0.008) / G85E (0.007) / 712-1G>T, 1609delCA, 2183AA>G, 2869insG (0.007 each) / 3272-26A>G (0.006) / V232D, 2184insA, K710X, A1006E (0.005 each) / R117H, 621+1G>T, 1078delIT, 3849+10kbC>T (0.004 each) / G85V, R347H, R553X, 1812-1G>A, R709X (0.003 each)
Sweden	0.872 (920)	39	0.3%: 0.831	0.3%: 9	F508del (0.666) / 394delITT (0.075) / 3659delC (0.062) / R117C (0.008) / E60X, 1112delIT, R764X (0.004 each) / R117H, 621+1G>T (0.003 each) / F508del (0.658) / 3905insT (0.048) / R553X (0.044) / 1717-1G>A (0.034) / N1303K (0.020) / G542X (0.017) / W1282X (0.016) / 2347delIG (0.010) / R1162X (0.006) / R117H, R347P, Q525X, 3849+10kbC>T (0.005 each) / R334W (0.004) / 621+1G>T, E585X, 2789+5G>A, M1101K, 3732delA, S1235R (0.003 each)
Switzerland	0.957 (952)	66	0.3%: 0.897	0.3%: 20	F508del (0.220) / 1677delTA (0.045) / G542X, 2183AA>G (0.029 each) / N1303K (0.024) / E92K, 2789+5G>A (0.020 each) / R347H, 2043delIG (0.016 each) / G85E, 2181delA (0.012 each) / 296+9A>T, D110H, I148T, M152V, L571S, F1052V, R1158X, W1282X (0.008 each)
Turkey	0.606 (246)	43	0.508	19	F508del (0.754) / G551D (0.031) / G542X (0.017) / 621+1G>T (0.009) / 1717-1G>A (0.006) / R117H, R553X (0.005 each) / 1898+1G>A, N1303K (0.005 each) / R560T (0.004) / I507del (0.003) / E60X, G85E, 1154insTC, V520F, W1282X (0.002 each) / 1078delIT, R347P, Q493X, S549N, S589N, 3659delC, 3849+10kbC>T, R1283M (0.001 each)
United Kingdom	0.862 (9804)	61	0.1%: 0.857	0.1%: 24	F508del (0.500) / N1303K (0.024) / CFTRdele2,3(21kb) (0.010) / W1282X (0.005)
Ukraine	0.560 (580)	11	0.540	4	