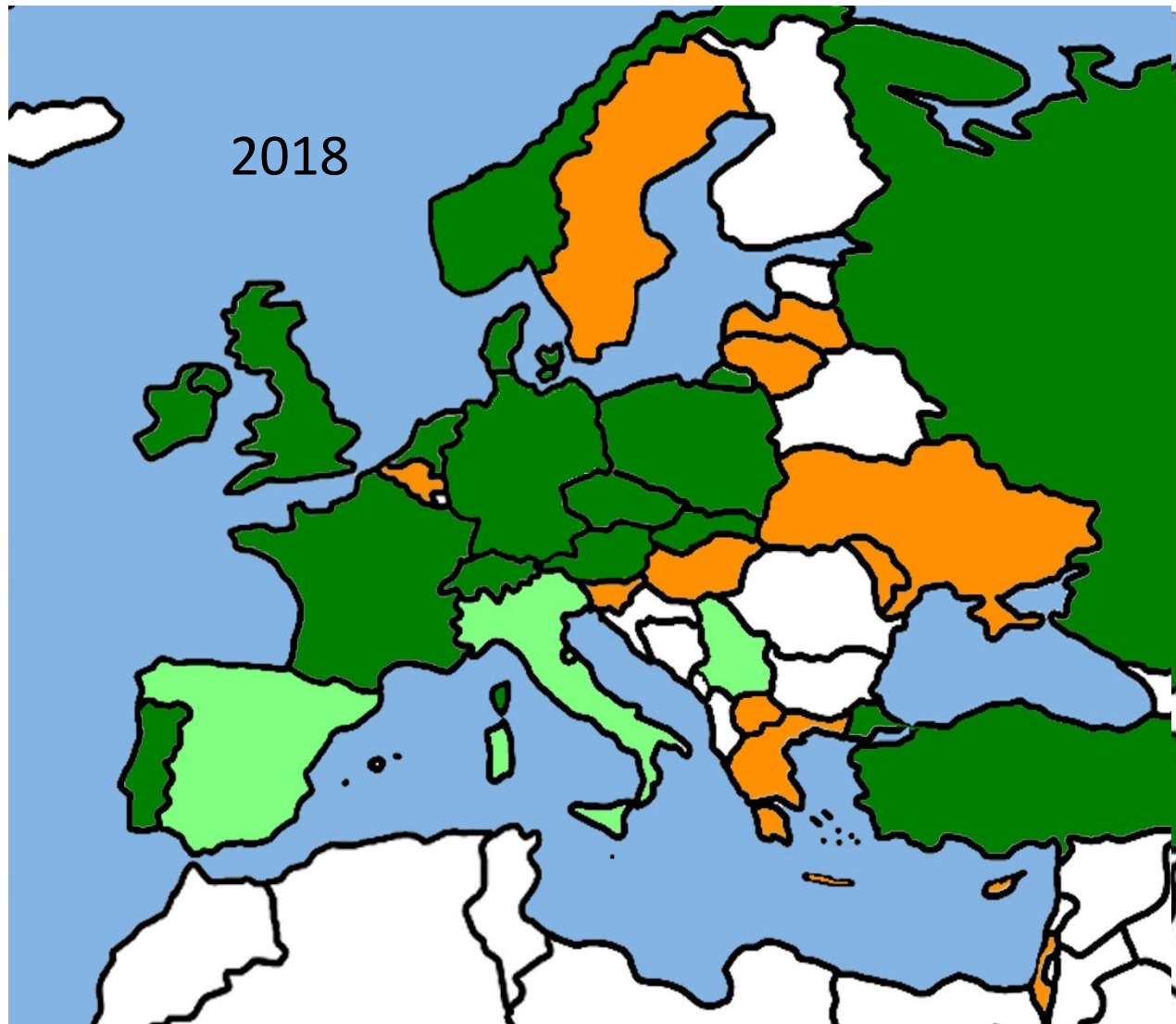
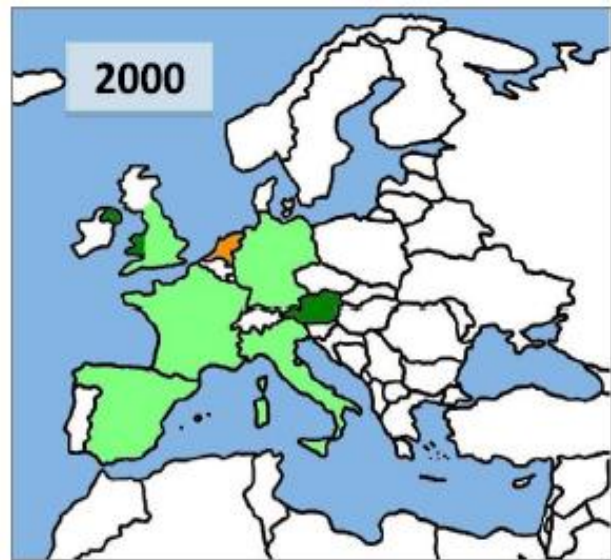


Journée Dépistage Néonatal de la Mucoviscidose
15 novembre 2018

Le dépistage néonatal (DNN) de la mucoviscidose en Europe

Anne Munck, AFDPHE Paris, SFDN





■ National ■ Régional ■ En cours de discussion

Original Article

The expansion and performance of national newborn screening programmes for cystic fibrosis in Europe

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TIR1 puis TIR2 ou PAP ou PAP/ADN et panel restreint /exhaustif avec ou sans TIR2

Année 2014	Northern Ireland	Wales	Austria	France	Scotland	England	Slovakia	Czech Republic	Poland	Ireland	Nether-lands	Switzer-land	Norway
Population (millions)	1.8	3.1	8.2	66.3	5.3	54.3	5.4	10.5	38.5	4.6	16.8	8.2	5.2
Birth per year	24,000	34,000	77,000	828,000	56,000	666,000	55,000	110,000	375,000	68,000	175,000	86,000	60,000
Number of babies with IRT-1 > cut off	120 (0.50%)	170 (0.53%)	500 (0.60%)	3141 (0.39%)	405 (0.72%)	3842 (0.57%)	848 (1.54%)	942 (0.90%)	2558 (0.60%)	670 (1%)	1004 (0.56%)	524 (0.61%)	491 (0.8%)
Number of babies with positive NBS result (referred for sweat test)	16	27	130	409	24	255	181	98	291	72	31	108	16
CF diagnosis	4	9	25	139	18	157	5	15	66	32	21	34	10
CFSPID	0	0	1	17	NA	15	0	2	55	1	2	2	4
Carriers	15	18	NA	190	17	139	NA	80	170	40	6	46	58
PPV													
CF diagnosis only	25%	30%	20%	34%	75%	67%	3%	15%	23%	44%	68%	31%	63%
CF & CFSPID	25%	30%	19%	38%	75%	73%	3%	17%	42%	46%	74%	33%	88%
Ratio CF:CFSPID	0	0	25:1	8:1	NA	10.5:1	0	7.5:1	1.2:1	32:1	10:1	17:1	2.5:1