

# INNO-LiPA™ CFTRiage\*

## A simpler, more powerful concept for CFTR testing



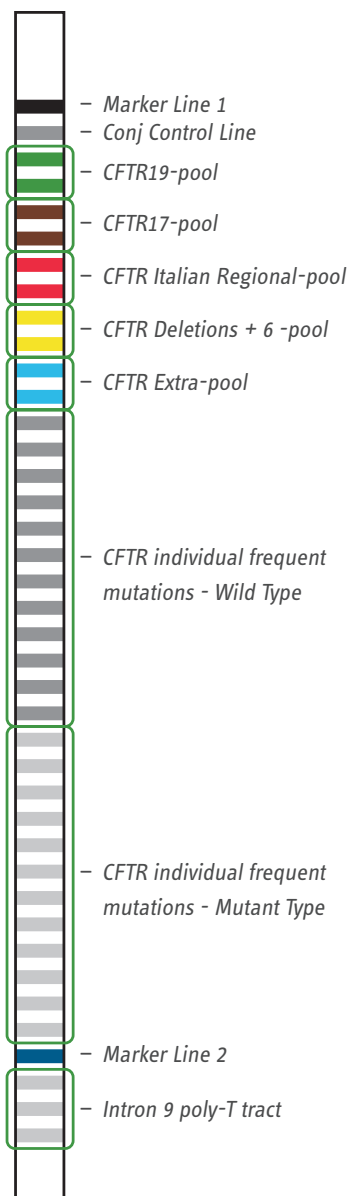
INNO-LiPA CFTRiage has been designed for **easy and efficient** analysis of multiple CFTR mutations. To do so, an amplification product of the CFTR gene is hybridized with ten pools of oligonucleotide probes immobilized on the INNO-LiPA CFTRiage strip (Figure 1).

In addition to the pooled mutation lines, a series of lines corresponding to the mutation and wild type probes of the 12 most frequent CFTR mutations are added (Figure 2). This permits the **differentiation between homozygous and heterozygous results** and thus, immediate discrimination between healthy individuals, healthy carriers, and affected patients.

Finally, **optional poly T testing** has been included. It allows for better genetic counselling as [R117H;(T)5] is considered a mild CF-causing complex allele, whereas [R117H;(T)7] is considered more as a CFTR Related Disorders mutation.<sup>1</sup>

Thanks to this innovative design, the **complete analysis of 88 mutations** can be achieved in more than 95% of samples using a single INNO-LiPA CFTRiage strip. For the remaining samples (less than 5%), the same amplicon can be reused for a second hybridization reaction on the relevant strip(s) (Figure 3).

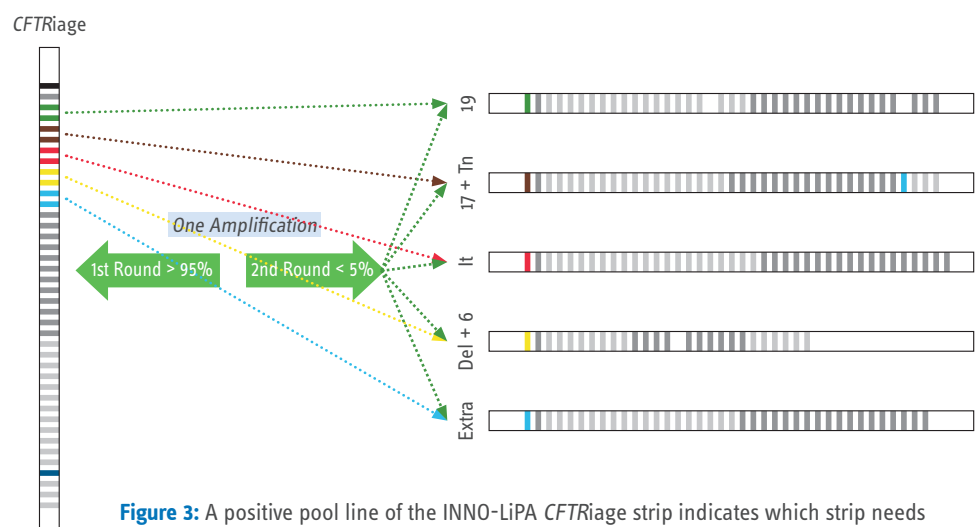
**Figure 1:** INNO-LiPA CFTRiage Strip



CFTR mutation	Frequency
F508del	69,74%
G542X	2,54%
G551D	2,10%
R117H	1,31%
N1303K	1,58%
W1282X	1,22%
621+1G->T	0,93%
1717-1G->A	0,86%
2789+5G->A	0,72%
A455E	0,35%
CFTRdele2,3 (21kb)	0,29%
L927P	0,02%

**Figure 2:** List of the individual mutation lines on the INNO-LiPA CFTRiage strip with the respective allele frequency according to the CFTR2 variants list (March 2019)<sup>2</sup>.

Only when necessitated by a reactive band for one or two of the ten pooled probes (less than 5% of the samples), the same amplicon can be reused for a second hybridization reaction on the relevant INNO-LiPA CFTR strip(s).



**Figure 3:** A positive pool line of the INNO-LiPA CFTRiage strip indicates which strip needs to be performed as a second round in order to complete the genetic analysis of the CFTR mutation.

#### LITERATURE

1. Dequeker et al., Eur J Human Genet. 2009 Jan; 17(1): 51-65

2. CFTR2 database ([www.CFTR2.org](http://www.CFTR2.org))


\* All INNO-LiPA CFTRiage products are not yet available as CE marked products

# INNO-LiPA™ CFTRiage

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### EXAMPLE 1: INNO-LiPA CFTRiage Strip

- Conjugate control = OK
- L12 negative: wild type line for G542X
- L24 positive: mutation G542X
- L37 positive: 9T




Marker line 1
Conj. Control
1 CFTR19 A
2 CFTR19 B
3 CFTR17 A
4 CFTR17 B
5 CFTR Italian A
6 CFTR Italian B
7 CFTR deletions A
8 CFTR deletions B
9 CFTR Extra A
10 CFTR Extra B
11 W.F508del
12 W.G542X
13 W.G551D
14 W.R117H
15 W.N1303K
16 W.W1282X
17 W.621+1G->T
18 W.1717-1G->A
19 W.2789+5G->A
20 W.A455E
21 W.CFTRdele2,3(21kb)
22 W.L927P
23 M.F508del
24 M.G542X
25 M.G551D
26 M.R117H
27 M.N1303K
28 M.W1282X
29 M.621+1G->T
30 M.1717-1G->A
31 M.2789+5G->A
32 M.A455E
33 M.CFTRdele2,3 (21kb)
34 M.L927P
Marker line 2
35 5T
36 7T
37 9T

#### Conclusion:

- Sample with a homozygous G542X mutation
- No further testing required

### EXAMPLE 2: INNO-LiPA CFTRiage Strip

- Conjugate control = OK
- L5 positive: pooled mutations of the CFTR Italian Regional
- L11 positive: wild type line for F508del
- L23 positive: mutation F508del
- L36 positive: 7T
- L37 positive: 9T



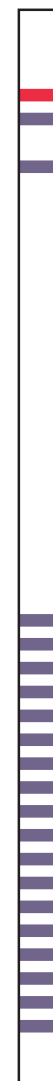
Marker line 1
Conj. Control
1 CFTR19 A
2 CFTR19 B
3 CFTR17 A
4 CFTR17 B
5 CFTR Italian A
6 CFTR Italian B
7 CFTR deletions A
8 CFTR deletions B
9 CFTR Extra A
10 CFTR Extra B
11 W.F508del
12 W.G542X
13 W.G551D
14 W.R117H
15 W.N1303K
16 W.W1282X
17 W.621+1G->T
18 W.1717-1G->A
19 W.2789+5G->A
20 W.A455E
21 W.CFTRdele2,3(21kb)
22 W.L927P
23 M.F508del
24 M.G542X
25 M.G551D
26 M.R117H
27 M.N1303K
28 M.W1282X
29 M.621+1G->T
30 M.1717-1G->A
31 M.2789+5G->A
32 M.A455E
33 M.CFTRdele2,3 (21kb)
34 M.L927P
Marker line 2
35 5T
36 7T
37 9T

#### Conclusion after INNO-LiPA CFTRiage:

- Sample positive for F508del (not homozygous) + a mutation out of CFTR Italian Regional
- Reflex testing with INNO-LiPA CFTR Italian Regional Strip

### INNO-LiPA CFTR Italian Regional Strip

- Conjugate control = OK
- L2 positive: mutation 4016insT
- L22 positive: wild type line for 4016insT



Marker line
Conj. Control
1 M.1259insA
2 M.4016insT
3 M.4382delA
4 M.852del22
5 M.D579G
6 M.G1244E
7 M.G1349D
8 M.I502T
9 M.L1065P
10 M.R1158X
11 M.T338I
12 M.S549R(A>C)
13 M.991del5
14 M.D1152H
15 M.1898+3A>G
16 M.R1070Q
17 M.R1066H
18 M.R347H
19 M.621+3A>G
20 M.E217G + M.R334Q
21 W.1259insA
22 W.4016insT
23 W.4382delA
24 W.825del22
25 W.D579G
26 W.G1244E
27 W.G1349D
28 W.I502T
29 W.L1065P = W.R1066H
30 W.R1158X
31 W.T338I
32 W.S549R(A>C)
33 W.991del5
34 W.D1152H
35 W.1898+3A>G
36 W.R1070Q
37 W.R347H
38 W.621+3A>G + W.E217G + W.R334Q

#### Final conclusion

- Sample is compound heterozygous for mutations F508del and 4016insT
- No further testing required