

Leuven, 07-04-2008

Referring clinician: Doctor X  
Street  
City, Country

## Molecular genetic analysis for Cystic Fibrosis

Last Name: CONTI  
First name: Dante  
Date of birth: 17/11/1975  
Gender: Male  
Place of birth: Milano, Italy  
Ethnic origin: Northern Italy  
Reason for referral: Dante Conti is a healthy individual from Northern Italy with no family history of CF. His partner is Anna Batini, whose brother is affected with CF. Dante Conti has informed you that Anna is a CF carrier, heterozygous for 1717-1G>A. Dante wants to know his carrier status and the risk of having a CF child with Anna.  
Sample received: 03/06/2007  
Sample type: DNA  
Your reference: CF08-2  
Our reference: MUCO-413

**RESULT: Heterozygous carrier of F508del mutation: F508del/normal or according to HGVS c.[1520\_1522delCTT]+[=]**

### INTERPRETATION:

This result confirms that Dante Conti is a carrier of the F508del (c.1520\_1522delCTT, p.Phe508del) cystic fibrosis mutation.

Dante's partner Anna Batini is said to be a carrier of 1717-1G>A (c.1585-1G>A) mutation. We would be happy to confirm this result. If Dante and Anna are both carriers of cystic fibrosis, they have a combined risk of 25% for having a child with cystic fibrosis. They are a couple-in-risk for having an affected child, so they can be offered prenatal diagnosis. We would recommend cascade screening for the relatives in the family of Dante and Anna. We would also recommend genetic counseling to discuss this further.

**Analysis performed by**



Molecular biologist Y

**Approved by**



Laboratory director Z

The method used: PCR and reverse dot blot.

Mutations screened for: F508del, I507del, G542X, 1717-1G>A, G551D, R553X, R560T, Q552X, W1282X, S1251N, 3905insT, N1303K, CFTRdele2,3, 711+1G>T, 3272-26A>G, 1898+1G>A, 1148T, 3199del6, 3120+1G>A, 394delTT, G85E, E60X, 621+1G>T, R117H, 1078delT, R347P, R334W, 2143delT, 2183AA>G, 2184delA, 711+5G>A, R1162X, 3659delC, 3849+10kbC>A, A455E, 5T, 7T, 9T

**The mutation detection rate is about 90% for the Caucasian population.**