

Leuven, 07-04-2008

Referring clinician: Doctor X
Street
City, Country

Molecular genetic analysis for Cystic Fibrosis

Last Name: LINDGREN
First name: Lasse
Date of birth: 18/02/1972
Gender: Male
Place of birth: Bullerbyn, Sweden
Ethnic origin: Caucasian (based on place of birth)
Reason for referral: Azoospermia and CBAVD. Lasse's partner, Lotta Langstromp (24/12/80) is a carrier of 394delTT (information supplied with referral). The couple wish to undergo assisted reproduction.
Sample received: 15-02-2008
Sample type: DNA
Your reference: CF07-2
Our reference: 158/45-5

RESULT: Compound heterozygote F508del/R117H; IVS8 variant: 7T/9T

INTERPRETATION:

This result is compatible with CBAVD in Lasse Lindgren.

COMMENTS:

- Patients with this genotype have a risk to develop other symptoms found in atypical CF such as pancreatitis and lung disease. Therefore, **further clinical evaluation** of the patient is recommended.
- Confirmation of the genotype of the patient's partner**, Lotta Langstromp (believed to carry the 394TT mutation) is needed (re-test) for accurate assessment of the risk for *CFTR* related disorders in their offspring.
- If the genotype for Lotta Langstromp is confirmed and assisted reproduction is performed, their child will have a 50% probability of having CF (including CBAVD), **25% of being affected** (F508del/394delTT) and 25% of having a mild form of CF (R117H/394delTT, with the possibility of male infertility in the case of a son).
- The relatives of Lasse Lindgren and his partner Lotta Langstromp should be offered **cascade screening**.
- Genetic counseling** is strongly recommended for this couple.

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.