Cystic Fibrosis European Network

EQA ID 20070999
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Leuven, 07/11/2008

Referring clinician: Doctor X
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Molecular genetic analysis for Cystic Fibrosis (*CFTR* gene)

Last Name:	BRAUN	Ethnic origin:	Mother from Brittany, father from Germany
First name:	Gary		
Date of birth:	20/06/2006	Sample received:	03/06/2007
Gender:	Male	Sample type:	DNA
Place of birth:	Hamburg, Germany	Your reference:	CF06-2
		Our reference:	MUCO-412

Reason for referral: Identification of the *CFTR* mutations responsible for the CF phenotype: failure to thrive, chronic diarrhoea, two episodes of bronchiolitis and a positive sweat test. The mother of Gary Braun has recently become pregnant.

RESULT **Highly likely compound heterozygous for c.1652G>A, p.Gly551Asp (traditional name: G551D) and c.1657C>T, p.Arg553* (traditional name: R553X)**

Genotype in HGVS: c.[1652G>A(;)1657C>T]
Reference Sequence NM_000492.3

INTERPRETATION

Gary Braun is heterozygous for the c.1652G>A, p.Gly551Asp (traditional name: G551D) and c.1657C>T, p.Arg553* (traditional name: R553X) cystic fibrosis mutations. He is highly likely to be a compound heterozygote of these two mutations, which would confirm the diagnosis of cystic fibrosis.

Such confirmation can be obtained by **testing the parents** to establish carrier status and origin of each mutation. **Referral of the parents for genetic counselling** is recommended.
Once carrier detection is confirmed, **this couple has a 25% risk to have an affected child for each pregnancy** and prenatal diagnosis can be offered in this pregnancy and in every subsequent pregnancy.
In addition, **carrier testing** can be offered to their **relatives**, in the framework of a genetic counselling session.

Analysis performed by

Molecular biologist Y

Approved by

Laboratory director Z

The method used: PCR and reverse dot blot (INNO-LIPA CFTR19 & CFTR17+Tn update).
Mutations screened for in traditional nomenclature: 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, 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. **The mutation detection rate is about 87% for the German population (WHO report 2004, http://www.genet.sickkids.on.ca/cftr/).**