

Doctor X
Street
City, Country

Molecular genetic analysis for Cystic Fibrosis

Name: Gary BRAUN
Date of birth: 20-06-2006
Gender: Male
Place of birth: Hamburg, Germany
Ethnic origin: Caucasian: mother from Brittany, father from Germany
Reason for referral: Failure to thrive, chronic diarrhea, two episodes of bronchiolitis and a positive sweat test
Sample received: 3-06-2007
Sample type: DNA
Sample number: MUCO-412

RESULT: Compound heterozygote of G551D/R553X

INTERPRETATION:

This results shows that Gary Braun is a compound heterozygote of two different Cystic Fibrosis mutations. This result confirms the diagnosis of Cystic Fibrosis in this patient.

COMMENTS:

Cystic Fibrosis is an autosomal recessive disorder and therefore both parents of the affected child should be carriers of the disease. It is thus suggested that the parents' DNA should be tested to establish carrier status and origin of each mutation. Once carrier detection is confirmed, this couple has a 25% risk to have an affected child in each pregnancy. Prenatal diagnosis is therefore recommended in this pregnancy and in every subsequent pregnancy of this couple. Since this is an inherited disease, screening for carrier status of other members of their families is also recommended.

The parents of the patient and their families should consult with the referring doctor and a genetic counselor.

Molecular Biologist



Director



The method used: INNO-LiPA CFTR19 and 17+Tn update (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.