Result SummaryPositive. /.br//.br/ Result- The following heterozygous sequence change was identified. Amino Acid: p.F508del (Phe508del), DNA change: c.1521_1523delCTT (g. 117199646_117199648), Classification: Pathogenic. /.br//.br/ Interpretation- This result indicates that this individual is a carrier of cystic fibrosis (CF). This interpretation assumes that this individual is not clinically affected with CF. Since a mutation has been identified, genetic testing of at risk family members could be considered. If appropriate, genetic testing should be offered to this individual's reproductive partner to further clarify their risk of having a child with CF. /.br//.br/ Method- A multiplex PCR based was used to detect 106 mutations, including the 23 mutations specified in the American College of Medical Genetics (ACMG) standards for population based carrier screening...Poly T determination and confirmatory testing of homozygous results are performed as reflex tests when appropriate.