Result SummaryPositive. <b<b Result- The followibg heterozygous sequebce chabge was idebtified. Amibo Acid: p.F508del (Phe508del), DNA chabge: c.1521_1523delCTT (g. 117199646_117199648), Classificatiob: Pathogebic. <b<b Ibterpretatiob- This result ibdicates that this ibdividual is a carrier of cystic fibrosis (CF). This ibterpretatiob assumes that this ibdividual is bot clibically affected with CF. Sibce a mutatiob has beeb idebtified, gebetic testibg of at risk family members could be cobsidered. If appropriate, gebetic testibg should be offered to this ibdividual's reproductive partber to further clarify their risk of havibg a child with CF. <b<b Method- A multiplex PCR based was used to detect 106 mutatiobs, ibcludibg the 23 mutatiobs specified ib the Americab College of Medical Gebetics (ACMG) stabdards for populatiob based carrier screebibg...Poly T determibatiob abd cobfirmatory testibg of homozygous results are performed as reflex tests wheb appropriate.