Abstract
Abstract Background: The demand for prenatal diagnosis (PD) of cystic fibrosis (CF) is increasing. Methods: We performed pre-test multidisciplinary counselling for 192 couples at CF reproductive risk. In 11/192 (5.7%) cases PD was not performed mainly because counselling revealed a reproductive risk for atypical (mild) CF, while 181 PDs were performed in couples revealed at high risk for CF mainly because they already had a CF child (148/181, 81.8%) or had been identified through cascade screening (28/181, 15.5%). Results: In 167/181 (92.3%) cases (including two dichorionic twin pregnancies), PD was performed on chorionic villi, and in 14 on amniocyte DNA. Only 1/181 PD was unsuccessful. In all other cases, single tandem repeat analysis excluded maternal contamination, and PD was made within 7 days of sampling. In total 116/180 (64.4%) PDs were made with dot-blot analysis; 40 (22.2%) required gene sequencing; in 4/180 cases we tested the gene for large rearrangements; in 23/180 (12.8%) cases linkage analysis was necessary because parental mutation(s) were unknown. Forty-two out of 180 (23.3%) PDs revealed an affected foetus. All couples but one interrupted pregnancy. The first twin PD revealed the absence (1 foetus) and the presence of one mutation (the other foetus); the second twin PD revealed one parental mutation (1 foetus) and both parental mutations (the other foetus); the couple planned selective interruption. Conclusions: PD for CF should be performed in reference laboratories equipped for gene scanning and linkage analysis, with a multidisciplinary staff able to offer counselling to couples during all phases of PD.