Day 5 blood spot samples: IRT assay

Assay in singlicate

IRT < cut-off 0
IRT > cut-off 0
Re-assay in duplicate

IRT < cut-off 1
IRT > cut-off 1
DNA analysis - 4 mutations

CF Not Suspected

Two CF mutations
One CF mutation
No mutation detected

DNA analysis - 29 or 31 panel

IRT on 2nd blood spot sample

IRT >99.9th centile

CF SUSPECTED
Yes
IRT on 2nd blood spot

IRT > cut-off 2
IRT < cut-off 2

CF SUSPECTED
Probable carrier
CF Not Suspected

Cut-off 0 ≈ 60 ng/ml
Cut-off 1 ≡ 99.5th centile
Cut-off 2 is 10 ng/ml below cut-off 1
99.9th centile ≈ 120 ng/ml

Note: CF infants with meconium ileus may show a normal IRT. They, and babies at high risk due to family history or who have shown echogenic bowel, should be investigated according to clinical circumstances as well as being screened.