

Notes (for CMGS labs)

- (1) Activation date is the date on which the sample (or all the required samples) and all information pertaining to the request is received by the testing laboratory. Reporting date is the date on which the result is first issued; by telephone, fax or written report. The reporting time is the difference between the two dates measured in working days.
- (2) The 3 day/2 week turnaround times for urgent tests assumes a minimum period of notification (suggest 5 working days; this could be included in a service specification). In addition that the familial mutation is known and/or informativeness of markers is established.
- (3) Multiplex PCR tests to detect known mutations (eg *CFTR* ARMS PCR or OLA) are included within "specific mutation tests". A separate PCR test (eg. *CFTR* 3199del6 test to investigate a I148T mutation, polyT test or zygosity test for a non- Δ F508 mutation detected by ARMS PCR) would have an additional 2 week reporting time. If samples are pre-screened for Δ F508 prior to a multiplex PCR test, a single 2 week reporting time applies.
- (4) If a mutation screening test report includes both dosage and sequencing (or HRM, CSCE, HPLC etc) analysis, then a reporting time of up to 8 weeks will apply. If a mutation screening test report includes dosage analysis only, then a 2 week reporting time applies.
- (5) A normal Fragile X result would be reported within 2 weeks (specific mutation test), but if Southern blotting was required to confirm an expansion mutation or a homozygous normal result in a female then an 8 week reporting time would apply for the entire test. This also applies to myotonic dystrophy and Huntington disease.
- (6) Laboratories and referrers should reach an agreement on criteria for curtailing/terminating analysis of samples that are not of adequate quality.
- (7) We should be working towards $\geq 95\%$ of tests reported within these times. Reporting time figures should be stated as the absolute percentage of tests within the target.