

26th February 2015

Re: Genetic Analysis for Cystic Fibrosis – TAT of 5 days

Dear Colleague,

We are offering an updated Cystic fibrosis mutation panel (test code: **CFS**) while maintaining the very efficient turnaround time of **5 working days**. The Illumina MiSeqDx Cystic Fibrosis Carrier Screening Assay is a CE marked in vitro diagnostic (IVD) next-generation sequencing test designed to detect **139** CFTR variants as defined in the CFTR database <http://www.cftr2.org/>. This replaces the 32 mutation assay. Other carrier screening methods test only for those variants most commonly found in Caucasians, potentially missing CF causative variants in a wider demographic. The MiSeqDx Cystic Fibrosis Carrier Screening Assay offers the largest CF screening panel available to overcome this bias and provide comprehensive detection across diverse demographics. This results in the most complete answer the first time, reducing residual risk.

The Illumina MiSeqDx Cystic Fibrosis Carrier Screening Assay is an in vitro diagnostic system used to simultaneously detect 139 clinically relevant variants of the cystic fibrosis transmembrane conductance regulator (CFTR) gene, including those currently recommended for carrier screening purposes by the American College of Medical Genetics 2004 (ACMG) and the American College of Obstetricians and Gynecologists 2011 (ACOG), in genomic DNA samples isolated from human peripheral whole blood specimens. The test is intended for carrier screening in adults of reproductive age, in confirmatory diagnostic testing of newborns and children, and as an initial test to aid in the diagnosis of individuals with suspected cystic fibrosis. The results of this test are interpreted by our clinical scientists and results are used in conjunction with other available clinical information.

Please do not hesitate to contact me with any further queries you may have.

Kind regards,



Dr Colin Clelland, MD, FRCPath
Medical Director