London Health Sciences Centre
St JOSEPH's HEALTH CARE LONDON
Pathology and Laboratory Medicine

CYSTIC FIBROSIS

Orderable – E-order/Requisition

Turnaround Time: 6 weeks **STAT: 10 days**

Laboratory:
Molecular Diagnostics Lab

Requisition:
MOLECULAR DIAGNOSTIC REQUISITION

Method of Analysis:
A total of 72 recurrent CFTR mutations, including those recommended by the ACMG, along with the polymorphic poly-pyrimidine intron8/exon9 splice-acceptor site (see attached on page 2 of this report) are screened using Agena iPLEX Pro CFTR Panel v1.0 kit followed by SpectroCHIP® Array detection using the MassARRAY System (Agena).

Alternate Name(s):

CF

Specimen:

Whole blood-2 x 4 mL Lavender EDTA top Vacutainer tube

Collection Information:

Blood samples must be maintained at room temperature.

Reference Ranges:

See report

Interpretive Comments:

CFTR related disorders include cystic fibrosis and absence of the vas deferens. Mutations in the CFTR gene can result in complex multisystem disease with morbidity resulting from pulmonary disease precipitated by lower airway inflammation and chronic endobronchial infection. The F508del mutation is a 3-base pair deletion in exon #10 of the CF gene that is associated with 70% of CF chromosomes in the Caucasian population. The American College of Medical Genetics (ACMG) has recommended (PMID:17394390) that individuals be tested for a further 23 different mutations in the Cystic Fibrosis gene.

Critical Information:

Pedigree required.

Storage and Shipment:

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Must be received in testing laboratory within 5 days of collection, shipped at room temperature by courier/overnight delivery.

Test Schedule:
As required,
Monday to Friday 0800-1600 hours