Indiana University Health Molecular Pathology Laboratory

NEXT GENERATION SEQUENCING FOR CYSTIC FIBROSIS

CLINICAL INFORMATION
IU Health Molecular Pathology Laboratory offers next generation sequencing to detect 139 clinically relevant CFTR variants. The variants reported by this assay were specifically chosen because they represent the full set of clinically validated variants classified as CF-causing in the CFTR2 database at Johns Hopkins University, a product of the CFTR2 initiative. The assay tests for 134 CF causing variants, one ACMG recommended panel variant (R117H, classified as a Mutation of varying Clinical Consequence, MVCC, by CFTR2); one conditionally reported modifying variant (PolyTG/PolyT); and three conditionally reported benign variants (I506V, I507V, F508C). 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.

CERNER ORDERABLE
Using IU Health Pathology requisition; Order through CoPath please call 317.491.6654.

CPT CODES
81220

METHODOLOGY
The assay uses next generation sequencing for detecting139 clinically relevant CFTR variants.

SPECIMENS
1 mL Peripheral Blood in EDTA

SPECIMEN STABILITY and SHIPPING
Transport peripheral blood refrigerated.

CAUSES FOR REJECTION
Peripheral blood collected in Heparin.

SPECIFICITY
Assay detects these CFTR variants:
M1V, T338I, Q552X, 3121-1G>A, CFTR dele2,3, 1154insTC, R553X, 3272-26A>G, Q39X, S341P, A559T, L1065P,E60X, R347H, R560T, R1066C, P67L, R347P, R560K,R1066H, R75X, R352Q, 1811+1.6kb A>G, L1077P, G85E, 1213delT, 1812-1 G>A, W1089X, 394delTT, 1248+1G>A, E585X, Y1092X(C>A), 405+1 G>A, 1259insA, 1898+1G>A, Y1092X(C>G,406-1G>A, W401X (c.1202G>A, 1898+3A>G, M1101K, E92X, W401X (c.1203G>A), 2143delT, E1104X, E92K, 1341+1G>A, R709X, R1158X, Q98X, PolyTG/PolyT, K710X, R1162X, 457TAT>G, 1461ins4, 2183delAA>G, 3659delC, D110H, A455E, 2184insA, S1196X, R117C, 1525-1G>A, 2184delA, W1204X (c.3611G>A), R117H, S466X (C>A), 2307insA, W1204X (c.3612G>A), Y122X, S466X (C>G), L732X, 3791delC, 574delA, L467P, 2347delG, 3849+10kbC>T, 621+1G>T, 1548delG†, R764X, G1244E, 663delT, S489X, 2585delT, 3876delA, G178R, S492F, E822X, S1251N, 711+1G>T, Q493X, 622+1G>A, 3905insT, 711+3A>G, I507del, E831X, W1282X, 711+5 G>A, F508del, W846X, 4005+1G>A, 712-1 G>T, 1677delTA, R851X, N1303K, H199Y, V520F, 2711delT, 4016insT, P205S, Q525X, 2789+5G>A, Q1313X, L206W, 1717-8G>A, Q890X, 4209TGTT>AA, Q220X, 1717-1G>A, L927P, CFTRdele22,23, 852del22, G542X, S945L, 4382delA, 1078delT, S549R (c.1645A>C), 3007delG, I506V, G330X, S549R (c.1647T>G), G970R, I507V, R334W, S549N, 3120G>A, F508C, I336K, G551D, 3120+1G>A

ASSAY RANGE
Mutation not detected. No clinically actionable mutation was found.
Mutation detected. Results will specify which clinically actionable mutation was found.

TURNAROUND TIME
14-17 Working days