Statement of Deficiencies	**(X1) Provider/Supplier/CLIA Identification Number**	**(X3) Date Survey Completed**
	21D0951395	11/20/2018
Name of Provider or Supplier	**Street Address, City, State**	
Inflammatory Disease Section, Nhgri	Building 10 East/Room B34129, Bethesda, MD	

For information on the provider's plan to correct this deficiency, please contact the provider or the state survey agency.

(X4) ID Prefix Tag	Summary Statement of Deficiencies
D5403	PROCEDURE MANUAL CFR(s): 493.1251(b) The procedure manual must include the following when applicable to the test procedure: (1) Requirements for patient preparation; specimen collection, labeling, storage, preservation, transportation, processing, and referral; and criteria for specimen acceptability and rejection as described in 493.1242. (2) Microscopic examination, including the detection of inadequately prepared slides. (3) Step-by-step performance of the procedure, including test calculations and interpretation of results. (4) Preparation of slides, solutions, calibrators, controls, reagents, stains, and other materials used in testing. (5) Calibration and calibration verification procedures. (6) The reportable range for test results for the test system as established or verified in 493.1253. (7) Control procedures. (8) Corrective action to take when calibration or control results fail to meet the laboratory's criteria for acceptability. (9) Limitations in the test methodology, including interfering substances. (10) Reference intervals (normal values). (11) Imminently life-threatening test results, or panic or alert values. (12) Pertinent literature references. (13) The laboratory's system for entering results in the patient record and reporting patient results including, when appropriate, the protocol for reporting imminently life threatening results, or panic, or alert values. (14) Description of the course of action to take if a test system becomes inoperable. This STANDARD is not met as evidenced by: A. Based on review of the laboratory procedure manual/testing protocol and interview with the laboratory director, the laboratory failed to ensure the procedure manual for performance of Periodic Fever Genetics (PFG) by PCR included a set of instructions on the labeling of aliqoted samples when those samples are not performed in-house. Findings include: 1. The surveyor reviewed the test protocol as revised on 8-17-18 and noted the policy of the laboratory was that all requests for whole genome testing for PFG was referred to an outside laboratory for testing. 2. There was no written instructions in the procedure manual for testing personnel to follow on: (1) the

	specimen labeling protocol for aliqoting samples and (2) the steps for ordering, packaging and mailing samples to the reference facility. 3. During interview with the laboratory director at approximately 10:20am, there was an admission of not included these instructions in the procedure manual. B. Based on review of the laboratory procedure manual and interview with the laboratory director, the laboratory failed to ensure the procedure manual for performance of Periodic Fever Genetics by PCR included all kits and instruments that are used in the performance of the assay. Findings include: 1. The surveyor reviewed the testing protocol as revised on 8-17-18 and noted, the DNA extraction procedure includes mention of the 'Maxwell 16 DNA Extraction Instrument' and kit: however, there is no step-by-strep instructions provided to testing personnel on the use of the Maxwell 16 DNA Extraction Instrument or kit, within the procedure. 2. During interview with the laboratory director at approximately 10:30am, there was an admission that while the testing personnel following the manufacturer's package insert (PI), there was no instructions included in the protocol, nor was the (PI) signed off as in use by laboratory.
D5417	TEST SYSTEMS, EQUIPMENT, INSTRUMENTS, REAGENT CFR(s): 493.1252(d) Reagents, solutions, culture media, control materials, calibration materials, and other supplies must not be used when they have exceeded their expiration date, have deteriorated, or are of substandard quality. This STANDARD is not met as evidenced by: Based on a tour of the laboratory testing processes, review of run Diagnostic forms (worksheets) and interview with the testing personnel, the laboratory failed to ensure reagents listed as used in the testing protocol (and listed on the worksheets) included both the lot number and expiration date in 10 of 10 runs reviewed in 2018 to show that all necessary reagents, solutions, controls and materials were within their expiration date prior to patient testing. Findings include: 1. The surveyor reviewed run worksheets for PFG by individual exon evaluations. 2. In 10 of 10 runs reviewed during 2018, the worksheets failed to listed the expiration dates for Mawell Extraction and Amplitaq kit lot numbers used in the runs. 3. This information was confirmed by interview with testing personnel at approximately 12:00pm.
D5421	ESTABLISHMENT AND VERIFICATION OF PERFORMANCE CFR(s): 493.1253(b)(1) Each laboratory that introduces an unmodified, FDA-cleared or approved test system must do the following before reporting patient test results: (1)(i) Demonstrate that it can obtain performance specifications comparable to those established by the manufacturer for the following performance characteristics: (1)(i)(A) Accuracy. (1)(i)(B) Precision. (1)(i)(C) Reportable range of test results for the test system. (1)(ii) Verify that the manufacturer's reference intervals (normal values) are appropriate for the laboratory's patient population. This STANDARD is not met as evidenced by: Based on a tour of the laboratory, review of laboratory verification records and interview with the laboratory director, the laboratory failed to have written evidence of precision studies when a new Applied Biosystems (AB) SeqStudio sequencer was introduced in the facility to replace the old ABL 3130XL sequencer in August 2018.

	Findings include: 1. The surveyor reviewed verification of performance of the new AB SeqStudio sequencer that was performed on August 21, 2018 by testing personnel. 2. While, the documentation included an assessment of accuracy, there was no documentation of precision studies that included analysis of run-to-run, day-to-day and within run studies for represenative PFG exons usually tested by the laboratory. 3. During interview with the laboratory director at approximately 11:00am, there was an admission that analysis and documenation of precision was not performed as required under the standard.
D5787	TEST RECORDS CFR(s): 493.1283(a) The laboratory must maintain an information or record system that includes the following: (a)(1) The positive identification of the specimen. (a)(2) The date and time of specimen receipt into the laboratory. (a)(3) The condition and disposition of specimens that do not meet the laboratory's criteria for specimen acceptability. (a)(4) The records and dates of all specimen testing, including the identity of the personnel who performed the test(s). This STANDARD is not met as evidenced by: Based on a tour of the laboratory testing processes, review of run Diagnostic forms (worksheets) and interview with the testing personnel, the laboratory failed to show the identity of testing personnel who performed the assays in 10 of 10 runs reviewed in 2018. Findings include: 1. The surveyor reviewed run worksheets for PFG by individual exon evaluations. 2. In 10 of 10 runs reviewed during 2018, the worksheets failed to list/state the identity of the testing personnel who performed the assay. 3. This information was confirmed by interview with testing personnel at approximately 12:00pm.