Statement of Deficiencies	**(X1) Provider/Supplier/CLIA Identification Number**	**(X3) Date Survey Completed**
	21D2093330	02/02/2018
Name of Provider or Supplier	**Street Address, City, State**	
Nih Undiagnosed Diseases Program Clinical Lab	50 South Drive Room 5525, 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
D5217	EVALUATION OF PROFICIENCY TESTING PERFORMANCE CFR(s): 493.1236(c)(1) At least twice annually, the laboratory must verify the accuracy of any test or procedure it performs that is not included in subpart I of this part. This STANDARD is not met as evidenced by: Based on review of quality assessment records and interview with the laboratory director, the laboratory failed to put a system in place to verify the accuracy of the sequencing data analysis twice annually. Findings include: 1. Quality assessment records for 2017 did not include verification of accuracy of the sequencing analysis portion of the laboratory's testing. 2. The laboratory director confirmed at approximately 10:35 am on the day of the survey that he was the only person who performed the analysis and interpretation of the sequencing data and that no system was in place to verify the accuracy of that portion of the testing process.
D5391	PREANALYTIC SYSTEMS QUALITY ASSESSMENT CFR(s): 493.1249(a) The laboratory must establish and follow written policies and procedures for an ongoing mechanism to monitor, assess, and when indicated, correct problems identified in the preanalytic systems specified at 493.1241 through 493.1242. This STANDARD is not met as evidenced by: Based on review of standard operating procedures, review of electronic patient testing records and interview with the laboratory director, the laboratory failed to establish a written policy for monitoring the status of samples referred to other laboratories for DNA sequencing. Findings include: 1. The laboratory's Standard Operating Procedure entitled Interpretation, Validation and Returning of Results to Patients dated 09/12/16

	states that, "The expected turn-around-time currently from the day the sequencing centers received the clinical specimens to when all data is 12 weeks or less." 2. During review of electronic patient testing records 1 of 1 record that was traced from start to finish showed that the sequencing results file had not been received, yet further investigation revealed in fact that the final result had been reported and that the sequencing data had been received. 3. The laboratory director stated at approximately 10:10 am on February 2, 2018 that the laboratory did not have a system in place for monitoring pending sequencing results. The laboratory director confirmed that the standard operating procedure did not address a mechanism for keeping track of samples that had been sent for sequencing but for which data had not been received.