Statement of Deficiencies	**(X1) Provider/Supplier/CLIA Identification Number**	**(X3) Date Survey Completed**
	05D0862216	01/23/2025
Name of Provider or Supplier	**Street Address, City, State**	
Genetic Disease Laboratory	850 Marina Bay Pkwy Ste G265, Richmond, CA	

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
D5215	EVALUATION OF PROFICIENCY TESTING PERFORMANCE CFR(s): 493.1236(b)(2) The laboratory must verify the accuracy of any analyte, specialty or subspecialty assigned a proficiency testing score that does not reflect laboratory test performance (that is, when the proficiency testing program does not obtain the agreement required for scoring as specified in subpart I of this part, or the laboratory receives a zero score for nonparticipation, or late return or results). This STANDARD is not met as evidenced by: Based on review of laboratory policy, proficiency testing (PT) records (2024), and confirmed in staff interview, the laboratory failed to verify and document the accuracy of alpha fetal protein (AFP) maternal screening results that were not graded by the proficiency testing company for 2 of 3 PT events in 2024. Findings included: 1. The laboratory policy titled "195829.133 GN 111 Proficiency Testing Policy" stated, " ... Any testing events not scored by the PT organization will be evaluated by TS to determine if results successfully demonstrated test accuracy using laboratory QC data and results distribution data supplied by the PT organization ..." 2. Review of the laboratory's College of American Pathologists (CAP) PT records for 2024 revealed the following: CAP FP-A 2024 Maternal Screening "Legend: Exception Reason Codes appearing in this evaluation: [26] = Educational Challenge AFP Interpretation: Specimen FP-01; Your result: Screen Negative: Your Grade See Note [26] Specimen FP-02; Your result: Screen Negative: Your Grade See Note [26] Specimen FP-03; Your result: Screen Negative: Your Grade See Note [26] Specimen FP-04; Your result: Screen Negative: Your Grade See Note [26] Specimen FP-05; Your result: Screen Negative: Your Grade See Note [26]" CAP FP-B 2024 Maternal Screening "Legend: Exception Reason Codes appearing in this evaluation: [26] = Educational Challenge AFP Interpretation: Specimen FP-08; Your result: Screen Negative: Your Grade See Note [26] Specimen FP-09; Your result: Screen Negative: Your Grade See Note [26] Specimen FP-10; Your result: Screen Negative: Your Grade See Note [26]

	Specimen FP-11; Your result: Screen Negative: Your Grade See Note [26] Specimen FP-12; Your result: Screen Negative: Your Grade See Note [26]" Review of the CAP "Survey and Anatomic Pathology Education Programs Maternal Screening Participant Summary" for both FP-A and FP-B stated, " ...Code: 26; Exception Reason Code Description: Education Challenge; Action Required: Review participant summary for comparative results and document performance accordingly ..." The laboratory was asked to provide documentation of self-evaluation for the educational challenges. No documentation was provided. 3. In an interview on 01/22/2025 at 2:05 pm in the conference room, the Laboratory Director confirmed that the testing was not evaluated and documented by the laboratory. Word Key: SOP=Standard Operating Procedure TS=Technical Supervisor
D5311	SPECIMEN SUBMISSION, HANDLING, AND REFERRAL CFR(s): 493.1242(a) (a) The laboratory must establish and follow written policies and procedures for each of the following, if applicable: (a)(1) Patient preparation. (a)(2) Specimen collection. (a)(3) Specimen labeling, including patient name or unique patient identifier and, when appropriate, specimen source. (a)(4) Specimen storage and preservation. (a)(5) Conditions for specimen transportation. (a)(6) Specimen processing. (a)(7) Specimen acceptability and rejection. (a)(8) Specimen referral. This STANDARD is not met as evidenced by: I. Based on a review of procedures, direct observation, and an interview with the technical supervisor, the laboratory failed to follow its written policy for ensuring dried blood spot (DBS) specimens were not stored and transported in plastic bags for two of two specimen bags observed. Findings included: 1. Review of the "Newborn Screening Specimen Collection & Transportation SOP" (GN107, effective date 01/15 /2025) stated, "3. Protocol for Dried Blood Spot Specimen Collection ...Does Not - Enclose TRF/specimen in a plastic bag ...2) Collecting the Specimen ...DO NOT transport in a plastic bag at any time ..." An embedded image of the test request form (TRF) that included filter paper for dried blood spot collection stated, "Instructions for Collecting Adequate Blood Spot Specimen ...*NO COURIER PLASTIC BAGS ...10. DO NOT PUT SPECIMEN IN PLASTIC BAG AT ANY TIME." 2. Review of the online client services manual for specimen collection stated, "Completing the Test Request Forms (TRF) ...Collecting the Specimen ...DO NOT transport in a plastic bag at any time." 3. During an observation of the DBS punching room on 01/23/25 at 2:54 PM, plastic bags of DBS specimens were observed. The DBS specimens were transported in plastic bags with desiccants and received for testing, as follows (random sampling): Bag one labeled "ADB00002M33436", "GDL01232", "012 Sun 2025 Jan-12", "GAMT", "SMA", and "1-355" (500 specimens) Bag two labeled "ADB00002P32178", "GDL 2 1.12.25 SUN JD012", "GAMT", "SMA", and "LSD" (200 specimens) The DBS specimens were punched for Guanidinoacetate Methyltransferase (GAMT), Spinal Muscular Atrophy (SMA), and Lysosomal Storage Disorders (LSD) testing. 4. Review of an additional procedure "CN 002 Ver 11.0 NBS Accession at NAPS" (effective date 11/19/2024) stated, "K. Storage & Shipping of Specimens ...a. ...1) At the end of the day's testing, place all specimens from each tray in a zip lock bag. Note: All bags (mother trays and accession day bags) should be sealed completely with two desiccant packages inside each bag." The procedure served as instructions for contracted laboratories (NAPS - newborn and prenatal screening) that send specimens to the Genetic Disease Laboratory (GDL) for additional testing. The procedure was not aligned with GDL's procedure for not

	putting specimens in plastic bags. 5. During an interview on 01/23/2025 at 3:36 PM, the technical supervisor reviewed the above findings and confirmed the inconsistencies in the procedures for packing and transporting DBS specimens. II. Based on a review of procedures, specimen logs, direct observation, and an interview with public health laboratory technician one (PHL tech-1), the laboratory failed to define storage and transport acceptable temperature requirements for DBS specimens for 6 of 6 tests. Findings included: 1. Review of the "Newborn Screening Specimen Collection & Transportation SOP" (GN107, effective date 01/15/2025) stated, "2. Stability Study ... The stability study demonstrated that all analytes remained stable for 7 days when stored at room temperature. All analytes remained stable for a minimum of 30 days when stored at 4C, -20C, and -80C." The temperature values for storage listed did not include a defined range for LSD, GAMT, SMA, phenylketonuria monitoring, Adrenoleukodystrophy (ALD), and Congenital Adrenal Hyperplasia (CAH) tests. 2. During a tour of the receiving area on 01/23/2025 at 10:12 AM, large white boxes with Styrofoam lining were observed. During an interview on 01/23/2025 at 10:12 AM, PHL tech-1 stated DBS specimens were transported via courier in large white boxes with ice packs from the contracted laboratories. A review of the "Daily Log of Specimens" for January 2025 (01/08/2025 - 01/21/2025) did not include documentation of temperature monitoring and whether ice packs were included. 3. During an interview on 01/23/2025 at 11:43 AM, PHL tech-1 confirmed specimens did not consistently include ice packs. The laboratory failed to define storage and transport temperature ranges in its written procedure to ensure specimen integrity.
D5413	TEST SYSTEMS, EQUIPMENT, INSTRUMENTS, REAGENT CFR(s): 493.1252(b) (b) The laboratory must define criteria for those conditions that are essential for proper storage of reagents and specimens, accurate and reliable test system operation, and test result reporting. The criteria must be consistent with the manufacturer's instructions, if provided. These conditions must be monitored and documented and, if applicable, include the following: (b)(1) Water quality. (b)(2) Temperature. (b)(3) Humidity. (b)(4) Protection of equipment and instruments from fluctuations and interruptions in electrical current that adversely affect patient test results and test reports. This STANDARD is not met as evidenced by: I. Based on review of the Congenital Adrenal Hyperplasia (CAH) 2nd Tier MS/MS Laboratory Protocol, v2.0 (GN 028), the CAH internal standards manufacturer's package insert, the laboratory's Temperature Monitoring Policy, v.1.0 (GN 094), and staff interview, the laboratory failed to define criteria for proper storage conditions of the CAH Internal Standards consistent with manufacturer's instructions for 24 of 24 months from February 2023 to January 2025. Findings included: 1. Review of the CAH 2nd Tier MS/MS Laboratory Protocol, v2.0, p. 9, Table 1B stated, "storage conditions, -20C Protect from light." 2. Review of the Lyso-PC Mix (Lot Number PR-33433), CAH internal standards manufacturer's package insert stated, "Storage Conditions: Store in freezer (-20C). Protect from light." 3. Review of laboratory's Temperature Monitoring Policy, v1.0 (GN 094), p. 37 stated, "the sensor level thresholds in the Thermo Freezer located in room C263 had a Min temperature of -50 C and Max temperature of -35C." 4. In an interview on 01/23/2025 at 3:15 PM, the Testing Personnel number 29 on the Form CMS-209, confirmed that the Thermo Freezer in Room C263 used to store the CAH internal standards had sensor level thresholds of -50C (Min) and -30C (Max), which were not the manufacturer's

specified storage conditions. 43232 II. Based on the review of a procedure, direct observation, review of manufacturer's instructions, and in an interview with staff, the laboratory failed to document temperatures for one of one walk-in freezer where kits, internal standards and quality controls were stored for LSD testing. Findings included: 1. Review of the "Temperature Monitoring Policy" (GN094, version 1.0, effective date 06/01/2024) stated, "Appendix-1: Sensor Locations and Level Threshold ...Walk in Freezer, Room No. C228, Min -25C, Max -10C." 2. During a tour of the laboratory on 01/23/2025 at 2:11 PM, the following was observed in the walk-in freezer C228 (random sampling): Fourteen (14) boxes of Revvity Iduronate 2-sulfatase S&IS (included 5 vials in each box) (Lot 753101). Manufacturer's instructions stated for storage, "Store the I2S S&IS vials in the original package placed inside a sealed plastic bag with desiccant at -30 to -16C." Thirty-nine (39) boxes of Revvity DBS Controls M2 (included 3 cassettes in each box) (Lot 751786). Manufacturer's instructions stated for storage, "The unopened controls are stable at -30 to -16C until expiry date on the bag label." One (1) box (sampling) of Revvity NeoLSD MSMS Kit (Lot 753183). Manufacturer's instructions stated for storage, "The unopened controls are stable at -30 to -16C until expiry date stated on the bag label" and "The unopened substrates and internal standards are stable at -30 to -16C until expiry date stated on the vial label." During an interview on 01/23/2025 at 2:15 PM, the TS stated C228 was not linked to their electronic monitoring system yet but monitored and documented manually. 3. During the exit interview on 01/23/2025 at 4:00 PM, the laboratory did not provide temperature charts, and the staff confirmed there was no documentation of the walk-in freezer temperatures. The defined maximum temperature in policy GN094 of -10C was not within manufacturer's instructions for the maximum temperature of -16C. 46043 III. Based on direct observation, review of manufacturer's instructions, review of laboratory records and policies, and confirmed in staff interview, the laboratory failed to ensure room temperature and refrigerator ranges were within manufacturer's specifications for 6 of 6 months (June - December 2024). Findings included: 1. During a tour of Room C201 on 01/23/2025 at 9:52 am, three BioRad high performance liquid chromatography (HPLC) instruments (Serial numbers 10090, 10092, and 10095) were observed being used for hemoglobinopathy testing. Each BioRad HPLC instrument had the following reagents on board the instrument: One bottle BioRad Hemoglobin Variants System Wash Solution; Lot number 64529034, Expiration date 02/16/2026 One bottle BioRad Hemoglobin Variants System Elution Buffer 1; Lot number 64595301, Expiration date 04/12/2026 One bottle BioRad Hemoglobin Variants System Elution Buffer 2; Lot number 64595300, Expiration date 04/11/2026 Also observed was General Electric refrigerator /freezer. The refrigerator portion was designated by the laboratory as "General Electric Combo Bottom". The following were observed stored inside the refrigerator: Three boxes BioRad Hemoglobin Variants System Calibration Set; Lot number 64614941, Expiration date 07/31/2025 Two boxes BioRad Hemoglobin Variants System Control Set; Lot number 64631366, Expiration date 02/28/2026 2. The manufacturer's instructions for the BioRad HPLC instrument stated," ...3.1 Installation Requirements ...2. Room temperature 15 - 30 C ..." The manufacturer's instructions on the BioRad Hemoglobin Variants System Wash Solution, Elution Buffer 1 and Elution Buffer 2 bottle label specified a storage temperature of 15 - 30 C. The manufacturer's instructions on the BioRad Hemoglobin Variants System Calibration Set and Control Set kit label specified a storage temperature of 2 - 8 C. 3. During a tour of Room C263 on 01/23/2025 at 10:26 am, a Perkin Elmer AutoDelfia immunoassay instrument (Serial Number 323147-1) was observed. The instrument was used for AFP maternal screening. Also observed was an Amana refrigerator /freezer. The refrigerator portion was designated by the laboratory as "Amana Combo Bottom". Observed inside the refrigerator were two AutoDelfia hAFP kits (Lot

	number 753389, expiration date 03/03/2025). 4. The manufacturer's instructions for the Perkin Elmer AutoDelfia instrument stated," ...Working environment Temperature: 15 - 30 C ..." The manufacturer's instructions on the AutoDelfia hAFP kit label specified a storage temperature of 2 - 8 C. 5. Review of laboratory environmental records revealed the laboratory used the Isensix wireless temperature monitoring system. The laboratory policy titled "195829.62 GN 094 Temperature Monitoring Policy", Effective 06/01/2024, revealed the following environmental ranges in use: "Room Temperature; Room C201; Min 20C; Max 40C" The maximum acceptable range (40C) exceeded the manufacturer's specified maximum limit of 30 C for the instrument and the reagents on board the instrument in Room C201. "General Electric Combo bottom; Room C201; Min 5C; Max 12C" The maximum acceptable range (12C) exceeded the manufacturer's specified maximum limit of 8C for the calibration and control sets stored in this refrigerator. "Room Temperature; Room C263; Min 20C; Max 40C" The maximum acceptable range (40C) exceeded the manufacturer's specified maximum limit of 30 C for the instrument in Room C263. "General Electric Combo bottom; Room C263; Min 5C; Max 12C" The maximum acceptable range (12C) exceeded the manufacturer's specified maximum limit of 8C for the hAFP kit stored in this refrigerator. 6. In an interview on 01/25/2025 at 1:30 pm in the conference room, the Laboratory Director confirmed the findings.
D5429	MAINTENANCE AND FUNCTION CHECKS CFR(s): 493.1254(a)(1) (a)(1) Maintenance as defined by the manufacturer and with at least the frequency specified by the manufacturer. This STANDARD is not met as evidenced by: Based on direct observation, review of manufacturer's instructions, Perkin Elmer AutoDelfia maintenance documentation (April 2023 through December 2024), and confirmed in staff interview, the laboratory failed to document daily, weekly, and monthly maintenance for nine of nine months. Findings included: 1. During a tour of Room 263 on 01/23/2025 at 10:26 am, a Perkin Elmer AutoDelfia instrument (Serial Number 323147-1) was observed. The instrument was used for Alpha Fetal Protein (AFP) maternal screening. 2. The manufacturer's instructions for the Perkin Elmer AutoDelfia instrument stated, " ...3 Maintenance Daily maintenance Test washer ... Empty the waste tray of tips, rinse it with water, and dry it ...Check the sample processor tubing for air bubbles during filling ...Check the temperature ...Thoroughly rinse the tubing and washing needles ...Weekly maintenance Sampler probe wash wells ...Wash and Disinfect Sample probes ...Washer and disk remover disinfection ... Check the Enhancement Solution outlet for participation ...Monthly maintenance Empty both the sample processor and the plate processor wash solution bottles and wash them ...Clean the waste bottle liquid sensors and bottle caps ...Rinse the tubing of the sample processor first with a strong wash solution ..., then with distilled water and then with 70% alcohol ...Wipe the plastic cover over the plate conveyor in the plate conveyor in the plate processor and the sample processor cover with 70% alcohol. Wash the standard tray covers ...with tap water and distilled water and dry. Wash the reagent rack and the sample racks. Clean the mirrors on the plate holders ..." 3. Review of the laboratory maintenance documentation from April 2023 through December 2024 revealed the laboratory failed to document performance of daily, weekly, and monthly maintenance. 4. In an interview on 01/23/2025 at 10:30 am in

	Room 263, Testing Person #20 (as listed on the CMS-209 form) was asked to provide documentation of daily, weekly and monthly maintenance. No documentation was provided. This confirmed the finding.
D5775	COMPARISON OF TEST RESULTS CFR(s): 493.1281(a)(c) (a) If a laboratory performs the same test using different methodologies or instruments, or performs the same test at multiple testing sites, the laboratory must have a system that twice a year evaluates and defines the relationship between test results using the different methodologies, instruments, or testing sites. This STANDARD is not met as evidenced by: I. Based on review of the Instrument Correlation Standard Operating Procedure (SOP), correlation studies records review for the Tandem Mass Spectrometry instruments used for testing for Adrenoleukodystrophy (X-ALD) and Congential Adrenal Hyperplasia (CAH), and staff interview, the laboratory failed to compare test results, twice a year, between two Tandem Mass Spectrometry instruments in 2023. Findings included: 1. Review of the California Department of Public Health, Genetic Disease Laboratory Services Branch, Instrument Correlation SOP, v1.0 (GN105), p. 5 stated, "Frequency of instrument correlation studies: if the testing unit uses more than one instrument and method to test for a given analyte, the instruments and methods shall be checked against each other every six months for comparability of results." 2. Review of correlations studies for two Tandem Mass Spectrometry instruments, Xevo TQS Instrument 1 (Serial Number WAA818) and Xevo TQS Instrument 2 (Serial Number WAC1590) used for testing X-ALD and CAH, revealed zero out of two correlations studies were completed in 2023. 3. In an interview on 01/23/2025 at 3:00 PM, the Testing Personnel number 29 on the Form CMS-209, confirmed that no correlation studies were completed in 2023. 46043 II. Based on direct observation, review of the laboratory policy, review of the laboratory's records (2024) and confirmed in staff interview, the laboratory failed to have documentation of performing one of two instrument correlations among three BioRad high performance liquid chromatography instruments. Findings included: 1. During a tour of the laboratory Room 201,oberved were three BioRad high performance liquid chromatography (HPLC) instruments (Serial numbers 10090, 10092, and 10095) used for hemoglobinopathy testing. 2. The laboratory policy titled "195829.123 GN105 Instruments correlation SOP" stated, " ...PROCEDURES 1. Frequency of Instrument Correlation Studies: if the testing unit uses more than one instrument and method to test for a given analyte, the instruments and methods shall be checked against each other every six months for comparability of results ..." 3. Review of laboratory records revealed the laboratory failed to perform two instrument correlations in 2024. 4. In an interview on 01/25/2025 at 1:30 pm in the conference room, the Laboratory Director confirmed the findings.
D6168	TESTING PERSONNEL CFR(s): 493.1487 The laboratory has a sufficient number of individuals who meet the qualification requirements of 493.1489 of this subpart to perform the functions specified in 493. 1495 of this subpart for the volume and complexity of testing performed.

	This CONDITION is not met as evidenced by: Based on review of the Quality Management System Manual, Testing Personnel educational records, and an interview with the Research Scientist Supervisor I, the laboratory failed to ensure two out of 48 Testing Personnel met the required educational qualifications for High Complexity Testing. Findings: refer to D6171
D6171	TESTING PERSONNEL QUALIFICATIONS CFR(s): 493.1489(b) (b) Meet one of the following requirements: (b)(1) Be a doctor of medicine, doctor of osteopathy, or doctor of podiatric medicine licensed to practice medicine, osteopathy, or podiatry in the State in which the laboratory is located; or (b)(2)(i) Have earned a doctoral, master's, or bachelor's degree in a chemical, biological, clinical or medical laboratory science, or medical technology from an accredited institution; or (b)(2)(ii) Be qualified under the requirements of 493.1443(b)(3) or 493.1449(c)(4) or (5); or (b)(3)(i) Have earned an associate degree in a laboratory science or medical laboratory technology from an accredited institution or (b)(3)(ii) Have education and training equivalent to that specified in paragraph (b)(2)(i) of this section that includes (b)(3)(ii)(A) At least 60 semester hours, or equivalent, from an accredited institution that, at a minimum, includes either (b)(3)(ii)(A)(1) 24 semester hours of medical laboratory technology courses; or (b)(3)(ii)(A)(2) 24 semester hours of science courses that include (b)(3)(ii)(A)(2)(i) 6 semester hours of chemistry; (b)(3)(ii)(A)(2)(ii) 6 semester hours of biology; and (b)(3)(ii)(A)(2)(iii) 12 semester hours of chemistry, biology, or medical laboratory technology in any combination; and (b)(3)(ii)(B) Have laboratory training that includes: (b)(3)(ii)(B)(1) Completion of a clinical laboratory training program approved or accredited by the ABHES or the CAAHEP (this training may be included in the 60 semester hours listed in paragraph (b)(3)(ii)(A) of this section); or (b)(3)(ii)(B)(2) At least 3 months documented laboratory training in each specialty in which the individual performs high complexity testing; or (b)(4) Successful completion of an official U.S. military medical laboratory procedures training course of at least 50 weeks duration and having held the military enlisted occupational specialty of Medical Laboratory Specialist (Laboratory Technician); or (b)(5) Notwithstanding any other provision of this section, an individual is considered qualified as a high complexity testing personnel under this section if they were qualified and serving as a high complexity testing personnel in a CLIA-certified laboratory as of December 28, 2024, and have done so continuously since December 28, 2024. (b)(6) For blood gas analysis (b)(6)(i) Be qualified under paragraph (b)(1), (2), (3), (4), or (5) of this section; or (b)(6)(ii) Have earned a bachelor's degree in respiratory therapy or cardiovascular technology from an accredited institution; or (b)(6)(iii) Have earned an associate degree related to pulmonary function from an accredited institution. (b)(7) For histopathology, meet the qualifications of 493.1449 (b) or (f) to perform tissue examinations. This STANDARD is not met as evidenced by: Based on review of the Quality Management System Manual, Testing Personnel educational records, and an interview with the Research Scientist Supervisor I, the laboratory failed to ensure two out of 48 Testing Personnel met the required educational qualifications for High Complexity Testing. Findings included: 1. Review of California Department of Public Health, Gentic Disease Laboratory Services Branch, Quality Management System Manual, v2.0 (GN075), p. 10, B. Personnel, stated, "CLIA regulations mandate that all laboratory personnel, from laboratory director to testing personnel, must meet specific educational qualifications, ..." and on

	p. 14, B.5.b. Testing Personnel, stated, "In California, the specific qualifications for testing personnel in a high complexity testing lab: A bachelor's degree in a chemical, physical, biological, or clinical laboratory science, or medical technology from an accredited institution." 2. Review of educational records for two Testing Personnel revealed the following: a. Testing Personnel number 11 on the Form CMS-209 earned a Bachelor of Science degree in a foreign country and there was no Foreign Equivalency documentation provided to complete evaluation. b. Testing Personnel number 45 on the Form CMS-209 did not have a qualifying degree for a Testing Personnel. 3. In an interview on 01/23/2025 at 3:45 PM, the Research Scientist Supervisor I, confirmed that Testing Personnel numbers 11 and 45 on the Form CMS-209 were not qualified as Testing Personnel based on the credentials provided.