IN THE UNITED STATES DISTRICT COURT

FOR THE DISTRICT OF SOUTH CAROLINA

COLUMBIA DIVISION

H. Freeman Belser, Esquire, as Personal Representative of the Estate [redacted]; and Amy Williams, individually,	)	C/A No. 3:16-0972-MBS
Plaintiffs,	)	
vs.	)	**ORDER AND OPINION**
Quest Diagnostics, Inc.; Athena Diagnostics, Inc.; and ADI Holdings, Inc.,	)	
Defendants.	)	

Plaintiffs Amy Elizabeth Williams, as the Personal Representative the Estate [redacted],[1] and Amy Elizabeth Williams, individually (together, "Plaintiff"), filed this action in the Court of Common Pleas for Richland County, South Carolina, on February 24, 2016 against Defendants Quest Diagnostics, Inc. ("Quest"); Athena Diagnostics, Inc. ("Athena"); and ADI Holdings, Inc. (together, "Defendants").[2] Plaintiff asserted claims of negligence/gross negligence resulting in the wrongful death of her minor son, [redacted] ("Decedent"). Plaintiff also asserted a survival action based on the suffering Decedent experienced before death, as well as claims for negligent misrepresentation and/or fraud, civil conspiracy, and violation of the South

[1]The current personal representative, H. Freeman Belser, Esquire, was substituted by order filed October 1, 2020.

[2]Quest Diagnostics is the parent company of ADI Holding Company, Inc., and owns 100% of the shares of ADI Holding Company. ECF No. 3. ADI Holding Company, Inc., is the parent corporation of Athena Diagnostics, Inc. and owns 100% of the shares of Athena Diagnostics, Inc. ECF No. 4.

Carolina Unfair Trade Practices Act ("SCUTPA"). Defendants removed the complaint on March 28, 2016 on the grounds of diversity jurisdiction.

I. FACTS

Decedent began suffering febrile focal motor seizures in December 2005, when he was approximately four months old. Decedent's pediatrician, Deborah Greenhouse, M.D., referred Decedent to a neurologist, Timothy Scott Livingston, M.D. Dr. Livingston opined in his follow-up patient notes dated August 24, 2006, that Decedent's history was consistent with severe myclonic epilepsy of infancy, or SMEI (also known as Dravet Syndrome). Dr. Livingston noted that the differential also should include mitochondrial disorders and several other metabolic disorders. Decedent current prescriptions included Keppra, clonazepam, and Vitamin B6. ECF No. 165-4. In November 2006, Dr. Livingston referred Decedent for a neurometabolic subspecialty evaluation at Horizon Molecular Institute in Atlanta, Georgia.

On December 15, 2006, Dr. Livingston's opinion was requested by Dr. McDonald/Pediatric Intensive Care. Decedent was receiving Keppra and fosphenytoin. Dr. Livingston again observed that Decedent likely had SMEI disorder. Dr. Livingston noted that studies had indicated a large majority of children with this disorder have a genetic defect in brain sodium channels. Dr. Livingston indicated that he was considering the possibility of a beta-oxidation defect, mitochondrial disorder, Rett's disorder, and other conditions. Dr. Livingston noted that a formal evaluation with a neurometabolic subspecialist in Atlanta, Dr. Shoffner, was pending. Decedent was continued on Keppra, clonazepam, fosphenytoin, and started on carbamazepine, a sodium channel blocking medication. ECF No. 165-5. Although carbamazepine is contraindicated for patients suffering from SMEI, Dr. Livingston was of the opinion that carbamazepine was the best treatment, given that

nonsodium channel blocking medications had not been successful and Decedent's diagnosis was uncertain. ECF No. 165-3, 14.

Decedent was first seen by John McKinley Shoffner, M.D., a clinical geneticist at Horizon Molecular Institute in Atlanta, Georgia, on January 17, 2007. Dr. Shoffner prepared the same day a summary of his impressions from his examination and recommended testing for mitochondrial disease, creatine deficiency syndromes, Angelman syndrome, adenylosuccinate lyase deficiency, SCN1A mutations, as well a chromosome microarray analysis. ECF No. 165-7.

In March 2007, Dr. Livingston increased Decedent's dose of carbamazepine. On April 29, 2007, Dr. Shoffner drafted a summary letter to Dr. Livingston and Dr. Greenhouse setting out the results of testing performed since the January 17, 2007 examination. While most tests were returned "unremarkable," the tests did show Complex I and Complex III defects from the skeletal muscle Oxidative phosphorylation enzymology. ECF No. 165-8, 2. The letter indicated that SCN1A sequencing was pending and the results would be forwarded when received. Dr. Shoffner made a preliminary diagnosis of mitochondrial encephalomyopathy and recommended that Decedent be treated with CoQ10 and creatine monohydrate. Dr. Shoffner indicated mitochondrial DNA sequencing would be performed to differentiate between a nuclear DNA mutation and mitochondrial DNA mutation. ECF No. 165-8.

Dr. Livingston testified that he did not receive the April 29, 2007 letter, and the letter was not found in Decedent's file. On May 7, 2007, however, Plaintiff informed Dr. Livingston that Decedent had been diagnosed with mitrochondrial encephalomyopathy pursuant to testing done by Dr. Shoffner. ECF No. 167-6, 55. Dr. Livingston indicated he was concerned about Decedent's risk for seizures and progressive problems with his new diagnosis of mitrochondrial disorder. Id. at 56.

Decedent was continued on his medications and started on L-carnitine, thiamine, and riboflavin.

Decedent's DNA was provided to Athena in May 2007 for an SCN1A DNA Sequencing Clinical Diagnostic Test. ECF No. 165-11. An SCN1A DNA Sequencing Clinical Report (the "2007 Report") was issued by Athena on June 30, 2007. ECF No. 165-12. The 2007 Report noted that Decedent possessed a DNA mutation in the SCN1A gene classified as a "#4: variant of unknown significance" ("VUS").[3] On June 30, 2007, the SCN1A report was faxed and mailed to Dr. Shoffner. It does not appear, however, that the 2007 Report was forwarded to Dr. Livingston, or that Dr. Shoffner reviewed the Report. ECF No. 165-3, 7; 165-9, 20.

On July 11, 2007, Decedent was seen by Dr. Frances Kendell, Dr. Shoffner's associate at Horizon Molecular Medicine. She memorialized the meeting in a letter to Plaintiff the same day. Dr. Kendell indicated that Decedent's work up "identified an OXPHOS defect as the cause for his problems, but, to date, the actual gene that is causing his difficulties has not been identified." ECF No. 165-15. She opined that Decedent had a primary mitochondrial defect and that he should continue on CoQ10. On August 20, 2007, Dr. Kendall corresponded with Plaintiff and indicated that Decedent's dose of CoQ10 should be doubled. ECF No. 165-16. Dr. Kendall did not mention the SCN1A test or 2007 Report. Dr. Kendall denies being aware of the 2007 Report. ECF No. 165-10, 7.

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[3] According to the 2007 Report, "Since these types of sequence variants are similar to those observed in both disease-associated mutations at other nucleotide positions and in benign polymorphisms, the nature of this variation precluded clear interpretation. These DNA sequence variants may or may not alter the functional aspects of the SCN1A gene and/or its protein product. While methodologically accurate, the results of this analysis cannot be definitively interpreted due to the absence of published studies correlating these variant(s) with clinical presentation and/or pathology. Therefore, based on this single analysis, it is not possible to conclude with any reasonable degree of clinical certainty at this time whether or not this variant is associated with the phenotype in question." ECF No. 165-12, 2

Dr. Livingston continued treating Decedent with increasing doses carbamazepine. On September 17, 2007, Dr. Livingston noted that Decedent's development had improved substantially, but that he continued to experience seizures. ECF No. 167-6, 47. Tragically, Decedent died on January 5, 2008 following a traumatic seizure.

In September 2014, Plaintiff contacted Amy Dobson, M.S., C.G.C., a genetic counselor at Greenwood Generic Center in Greenwood, South Carolina, to obtain a copy of Decedent's SCN1A Sequencing Clinical Diagnostic Report, after determining to undergo genetic testing before considering a second pregnancy. Ms. Dobson obtained Dr. Livingston's records, which indicated that an SCN1A test had been ordered, but no copy of the report was in his file. Ms. Dobson requested a copy of the 2007 Report directly from Athena. ECF No. 165-18, 5. Ms. Dobson received the 2007 Report on September 29, 2014. The same day, Ms. Dobson called Athena and left a message asking for an official reinterpretation of the result. She learned from Dr. Carol Hoffman, a genetic counselor at Athena, that the "variant of unknown significance" had since been reported as pathogenic. Id. at 11. At Dr. Hoffman's request, Michelle McCarthy, a variant scientist at Athena, analyzed Decedent's variant and determined that Decedent's variant would still classify as pathogenic because by that time, October 6, 2014, the variant had been reported in multiple de novo SMEI cases and was absent from 13,374 chromosomes of the general population. ECF No. 165-24. A revised report was issued on January 30, 2015 (the "2015 Report."). The 2015 Report indicated that "[a]nalysis of this individual's SCN1A gene identified a DNA sequence variant that has been reported in the literature to be associated with SMEI or SMEB, the severe phenotypes associated with SCN1A mutations." ECF No. 165-27, 2.

Plaintiff's expert witness, pediatric neurologist Max Winitzer, M.D., opined in his deposition

that the VUS should have been reported as pathogenic in the 2007 Report. ECF No. 153-9, 7. Dr. Winitzer stated that, had Dr. Livingston been aware that the mutation was a pathogenic variant, he would not have managed Decedent the way he did. Id. at 9. Dr. Winitzer further testified that, had Decedent not been prescribed carbamazepine, he would have been more resistant to seizures and therefore, more likely than not, he would not have experienced the severe seizure that caused his death. ECF No. 153-9, 9-10.

II. PROCEDURAL HISTORY

Plaintiff filed an amended complaint on June 2, 2016, in which she dropped the fraud claim in favor of a negligent misrepresentation claim and added a constructive fraud claim. Defendants filed a motion to dismiss the amended complaint on June 24, 2016. Defendants argued (1) that the applicable statutes of limitation operated to bar each claim because Plaintiff had constructive notice of her potential claims from warnings and recommendations included in the 2007 Report; (2) that the wrongful death and survivorship actions and claims for negligent misrepresentation and constructive fraud were predicated on the alleged misdiagnosis of Decedent's medical condition in 2007, and were subject to the six-year statute of repose for medical malpractice claims as set forth in S.C. Code Ann. § 15-3-545(A), regardless of when Plaintiff had constructive notice;[4] (3) that the

[4]Section 15-3-545(A) provides:

> (A) In any action, other than actions controlled by subsection (B), to recover damages for injury to the person arising out of any medical, surgical, or dental treatment, omission, or operation by any licensed health care provider as defined in Article 5, Chapter 79, Title 38 acting within the scope of his profession must be commenced within three years from the date of the treatment, omission, or operation giving rise to the cause of action or three years from date of discovery or when it reasonably ought to have been discovered, not to exceed six years from date of occurrence, or as tolled by this section.

claims for negligent misrepresentation and constructive fraud failed to sufficiently allege justifiable reliance on the alleged false statement; (4) that by virtue of their unitary ownership Defendants were incapable as a matter of law of conspiring with themselves, and the conspiracy claim did not set forth additional facts or special damages unique to the alleged conspiracy; and (5) that Plaintiff failed to adequately plead Defendants' wrongful acts affect the public interest so as to state a claim under the SCUTPA. In order to determine whether Defendants, as a testing facility, could be liable for medical malpractice, on March 31, 2017 the court entered an Order of Certification to the South Carolina Supreme Court as follows:

> Is a federally licensed genetic testing laboratory acting as a "licensed health care provider" as defined by S.C. Code. Ann. § 38-79-410 when, at the request of a patient's treating physician, the laboratory performs genetic testing to detect an existing disease or disorder?

ECF No. 40.

On June 17, 2018, the South Carolina Supreme Court answered in the affirmative. ECF No. 59. On October 18, 2018, the court issued an order on Defendants' motion to dismiss. ECF No. 62. The court noted that, according to Defendants, Plaintiff's claims sound in medical malpractice and thus are barred by the six-year statute of repose set forth in section 15-3-545. Plaintiff asserted that, to the contrary, the complaint states ordinary negligence claims "of a nonmedical, administrative, or ministerial type" that results "from a lack of routine care surrounding the publishing of test results." ECF No. 28, 23-24. The court determined that the matter should proceed to discovery for the purpose of determining what caused Athena's laboratory staff to misclassify the gene mutation. The court further found that it could not, at that time, determine whether Plaintiff's claims are barred by the applicable statute of limitations based on Plaintiff's contention that she only

discovered the alleged negligence in 2015, upon receipt of the 2015 Report. The court dismissed Plaintiff's civil conspiracy claim. Thus, remaining before the court are Plaintiff's claims for wrongful death, survival, negligent misrepresentation, constructive fraud, and violation of the South Carolina Unfair Trade Practices Act.

This matter now is before the court on Defendants' motion for summary judgment, which motion was filed on February 2, 2020. Plaintiff filed a response in opposition on March 26, 2020, to which Defendants filed a reply on April 2, 2020. The court held a hearing on October 20, 2020. Defendants make the following three arguments: Defendants' actions are not the proximate cause of Decedent's death and Plaintiff's alleged injuries; Plaintiff's claims are barred by the statute of repose because Defendants' actions were made in an exercise of professional judgment and there is no evidence of ordinary negligence; and Plaintiff's SCUTPA claim–that Defendants violated the SCUTPA by failing to comply with various provisions of the Clinical Laboratory Improvement Amendments of 1988 ("CLIA")–fails as a matter of law.

III. DISCUSSION

A. Proximate Cause

Defendants first assert that the 2007 Report is not a proximate cause of Plaintiff's injuries. In a negligence action, a plaintiff must show that the (1) defendant owes a duty of care to the plaintiff, (2) defendant breached the duty by a negligent act or omission, (3) defendant's breach was the actual and proximate cause of the plaintiff's injury, and (4) plaintiff suffered an injury or damages. Steinke v. S.C. Dep't of Labor, Licensing and Regulation, 520 S.E.2d 142, 149 (S.C. 1999). Proof of proximate cause requires proof of both causation in fact and legal cause. Vinson v. Hartley, 477 S.E.2d 715, 721 (S.C. Ct. App. 1996) (citing cases). Causation in fact is proved by

establishing the injury would not have occurred "but for" the defendant's negligence. Id. (citing cases). Legal cause is proved by establishing foreseeability. Id. (citing cases). Foreseeability is determined by looking to the natural and probable consequences of the act complained of. Id. Thus, Defendants assert Plaintiff has not established a causal nexus between the 2007 Report and Decedent's treatment, regardless of whether Plaintiff alleges medical malpractice or ordinary negligence. According to Defendants:

> • Dr. Livingston, Decedent's treating physician who prescribed the medications that Plaintiff alleges caused Decedent's death, testified in his deposition that he did not see the Report or learn of its results while Decedent was alive, and, therefore, the Report did not affect his treatment of Decedent in any way. Livingston Dep. at 25:21-26:7, 27:11-17, 28:2-8, 30:18-20, 33:2-8 (Exhibit 5).
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> • The Report was not included in Dr. Livingston's file on Decedent produced in response to Defendants' subpoena, see Dr. Livingston's file (Exhibit 6), and Dr. Livingston testified that the Report was not in his records, Livingston Dep. at 25:23-24 (Exhibit 5).
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> • Athena both mailed and faxed a copy of the Report to Dr. Shoffner, the physician who ordered Decedent's SCN1A test, and those copies of the Report were included in Dr. Shoffner's file on Decedent that was produced in response to Defendants' subpoena. See copies of Report from Dr. Shoffner's file (ECF No. 127-13 and 14).
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> • Even though Athena sent the Report to Dr. Shoffner, he has no recollection of reviewing it while Decedent was alive. Shoffner Dep. at 19:15, 53:18-22, 62:17-20 (Exhibit 7).
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> • Dr. Shoffner does not know whether the Report's results were ever shared with Dr. Livingston, and he does not contest Dr. Livingston's testimony that they were not. Id. at 54:11-14, 55:24-56:5. (Exhibit 7)
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> • Dr. Kendall, Dr. Shoffner's partner who saw Decedent on two occasions after the Report was issued, testified emphatically in her deposition that she had never seen the Report or learned of its results prior to her deposition, and she has no idea what happened to the Report's results. Kendall Dep. at 34:18-22, 37:12-22, 42:20-43:5, 47:5-8 (Exhibit 8)
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> • Plaintiff insisted repeatedly in her deposition that she did not see the Report or

> learn of its results until 2014, over six years after Decedent's death. Williams Dep. at 107:15-17, 117:10-119:12, 219:10-13 ("I'm swearing I have never had knowledge of this test. . . . Prior to 2014.") (Exhibit 9).

ECF No. 167, 4.

Plaintiff contends that the proximate cause issue revolves around Defendants' failure to classify Decedent's VUS as pathogenic in 2007. To support her argument, Plaintiff argues that transmittal of Dr. Shoffner's April 29, 2007 letter most likely reached Dr. Livingston because it was Dr. Shoffner's standard practice to transmit such correspondence to the requesting physician. Id. at 8. Plaintiff further notes that, commencing in May 2007, Dr. Livingston's treatment records mirrored Dr. Shoffner's diagnosis of "probable mitochondrial encephalomyopathy" as set forth in Dr. Shoffner's April 29, 2007 letter. Plaintiff next posits that Dr. Livingston would have received the 2007 Report because of Dr. Shoffner's standard practice of forwarding test results, but that the classification of a "variant of unknown significance" would provide little to no assistance in making a specific diagnosis. Id. at 9. According to Plaintiff, literature was available to Athena in 2006 that should have caused it to label Defendant's variant as pathogenic. Thus, had the 2007 Report classified Decedent's variant as pathogenic, Dr. Livingston would have been alerted to alter Decedent's treatment. ECF No 153, 6.

Plaintiff must rely on more than conclusory allegations, mere speculation, the building of one inference upon another, or the mere existence of a scintilla of evidence. Mountain Valley Pipeline, LLC v. 0.15 Acres of Land by Hale, No. 20-1219, 2020 WL 6268085, *1 (4th Cir. Oct. 26, 2020)(quoting Dash v. Mayweather, 731 F.3d 303, 311 (4th Cir. 2013)). In this case, there is no evidence to support the notion that Dr. Livingston received the 2007 Report. The Report was not located in his file, Dr. Livingston did not recall reviewing it, and Dr. Shoffner cannot verify that the

2007 Report was reviewed by anyone in his practice or transmitted to Dr. Livingston. Further, Plaintiff's scenario requires an additional condition that, to defeat proximate cause, Decedent's variant would have been identified as pathogenic in the 2007 Report.

If the inference sought to be drawn lacks substantial probability, any attempted resolution of the question may lie within the area of surmise and conjecture. It is the duty of the court to withdraw the case from the jury when the necessary inference is so tenuous that it rests merely upon speculation and conjecture. Wratchford v. S.J. Groves & Sons Co., 405 F.2d 1061, 1066 (4th Cir. 1969). The court finds that Defendants are entitled to judgment as a matter of law as to this issue.

B. Statute of Repose

In any event, even had Dr. Livingston been in possession of the 2007 Report, the record does not support Plaintiff's premise that Decedent's VUS was wrongly classified. Further, even if the variant should have been classified as pathogenic, there is no support in the record that any error was the result of a nonmedical, administrative, or ministerial type action for which the discovery rule would apply, as urged by Plaintiff.[5]

"'Medical malpractice' means doing that which the reasonably prudent health care provider or health care institution would not do or not doing that which the reasonably prudent health care provider or health care institution would do in the same or similar circumstances." S.C. Code Ann. § 15-79-110. In medical malpractice actions, expert testimony is required to establish both the duty

[5]With respect to ordinary negligence, Defendants correctly assert that the three-year limitations period for an action for death by wrongful act accrues "upon the death of the person on account of whose death the action is brought[.]" S.C. Code Ann. § 15-3-530(6). The court will continue its analysis regarding the nature of Defendants' alleged erroneous VUS classification with respect to Plaintiff's remaining negligence causes of action. Here, Plaintiff testified in her deposition that she was unaware of any error until she received the 2007 Report and subsequent revisions in 2014 and 2015.

owed to the patient and the breach of that duty, unless the subject matter of the claim falls within a layman's common knowledge or experience. Dawkins v. Union Hosp. Dist., 758 S.E.2d 501, 504 (S.C. 2014) (citing cases). However, if the patient instead receives nonmedical, administrative, ministerial, or routine care, expert testimony establishing the standard of care is not required, and the action instead sounds in ordinary negligence. Id.

Plaintiff's expert witness, Robert Cook-Deegan, M.D., is a professor in the School for the Future of Innovation in Society and the Consortium for Science, Policy & Outcomes at Arizona State University. He previously served as a research professor in the Sanford School of Public Policy at Duke University, with secondary appointments in Internal Medicine (School of Medicine) and Biology (Trinity College of Arts & Sciences). He is a founding director of the Center for Genome Ethics, Law & Policy in Duke's Institute for Genome Sciences and Policy. ECF No. 153-3, 2. On March 2, 2020, Dr. Cook-Deegan provided an affidavit in which he made the following findings and conclusions:

> 1. Subsequent to the production of my affidavit and my December 18, 2019, deposition, Amy Williams's attorneys furnished me with a set of documents that are paginated as "DEFENDANTS_ 000857–001056." It is my understanding that the Defendants produced these documents on January 2, 2020. It is my further understanding that Quest has identified these materials as originating with the body known as the "Mutation Update Committee" identified in Dr. Izabella Karbassi's deposition on December 12, 2019. I am informed and believe that Dr. Karbassi's deposition was the first time that the existence of such a body was disclosed.
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> 2. In reviewing the documents produced on January 2, 2020, I am informed and believe that these materials consist of ongoing classification tables by Quest's Mutation Update Committee in compiling the variant database for gene SCNlA for Athena's internal use.
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> 3. According to DEFENDANTS_858–DEFENDANTS_865 (Exhibit 3), which is undated, the database reflected the Y413N variant was classified as a de novo variant of unknown significance. This information was consistent with the

"Appendix" (labeled DEFENDANTS_424–DEFENDANTS_425) (Exhibit 4 to this affidavit) which was introduced as Exhibit No. 15 in Dr. Karbassi's deposition, and dated November 30, 2006.

4. DEFENDANTS_866–DEFENDANTS_876 (Exhibit 5) is captioned "SCNlA Mutation List" and includes the notation "Last Modified by: DiVicenzo, Morneau, Towne 04/03/2009". On that document, the Y413N variant is labeled as "K", which is Quest's internal code for a pathogenic or disease-associated variant. The evidence-base cited for that classification is "Harkin 2007" and "unpublished Bionomics data."

5. This newly disclosed material has impacted my opinions substantially on the manner and degree to which QUEST/Athena erred in this case. The two evidence sources cited above and identified on the April 3, 2009 SCNlA mutation list–the evidence apparently used to reclassify the variant as disease-associated–were both demonstrably available and known to QUEST in 2007 before [redacted]'s positive test for the Y413N variant. This documentation also contradicts the deposition testimony of Dr. Sat Dev Batish that the classification was not updated until the publication of the Heron study. The newly disclosed information further undercuts the Defendants' assertion that paternity confirmation in the Canadian index case reported in the literature was necessary for a proper classification, since the change to "K" (pathogenic or disease associated) classification was made by Athena itself before publication of the Heron study.

6. Therefore, in my opinion, these newly disclosed materials show that QUEST/Athena's failure in this case was not one of genetic expertise or medical procedures, but rather a failure to update Athena's variant classifications. This is a failure of administrative oversight, not the exercise of discretion in the practice of genetics.

7. Genetics is the study of heredity and inherited characteristics. It is carried out by geneticists, who are scientists trained to study inheritance patterns, and molecules that store and transmit genetic information, particularly DNA. Medical genetics is a subset of genetics focused on inherited variations that affect human disease and health. For the purposes of this case, the geneticists employed by the Defendants were investigating the human genome. Geneticists in laboratories around the world investigate organisms ranging from plants to apes. Geneticists in the course of their employment may perform ancillary functions that contribute to their study of inherited characteristics. These ancillary functions include studying the latest literature in the field, attending conferences, corresponding with other scientists in genetics and related fields, etc. These are things that you don't need to be a geneticist to do, but because they're being performed in direct relation to the field of genetics as a field of study, they're part of being a geneticist. Keeping track of the literature

is a feature of almost any profession or scholarship, and is not distinctive to genetics. Put simply, tracking the literature and updating a database is not the practice of genetics.

8. Based on the materials furnished, I conclude that QUEST/Athena did not update its classification of the Y413N variant in SCNIA between 2004 and April 3, 2009, even though the materials that the Mutation Update Committee relied upon to change the classification were available to QUEST/Athena before it tested the decedent's samples. In fact, three of the authors of the Harkin paper that was the basis for "updating" the classification list their affiliations as Athena Diagnostics, including Dr. Sat Dev Batish, who directed the lab and signed the June 2007 report. Athena geneticists therefore had access to and knowledge of this variant's disease-associated nature. This is evidence of the lack of importance the Defendants placed on updating their variant lists: even though they had certain .knowledge identifying a variant in need of update and what the associated update should be, they still did not actually enter the updates into the database for ANTRIM until nearly three years later. This is not an error in genetics, this is a failure to administratively update databases.

9. The basis for knowing that Athena Diagnostics already had information about this mutation in hand–the information used to "update" the classification in April 2009–is the following: (1) Athena Diagnostics secured an exclusive license for US testing of the SCN1A gene from Bionomics and Australian universities in 2004. Y413N correlation with Dravet was known at that time. Presumably, the "unpublished Bionomics data" referenced in Exhibit 5 refers to material provided by Bionomics when it licensed the test to Athena. (2) A correlation between Y413N and Dravet was first published in the medical literature in 2006 (Berkovic 2006). (3) The publication by Louise Harkin and others in 2007, noted above, cited and corroborated the Berkovic 2006 correlation between Y413N and Dravet. Harkin 2007 was submitted in November 2006 and published in March 2007, three months before [redacted]'s June 2007 report was produced.

10. The mutation list curated by Athena Diagnostics in Exhibit 4 (Nov 2006) flags Y413N as "UAA?"" and lists "unpublished Bionomics data," which presumably is information transferred through the licensing agreement. In contrast, the mutation list from April 3, 2009 (and repeated in subsequent mutation lists on April 16 and May 29, 2009 and March 1, 2010), specifies the "K" (pathogenic) classification (changed from "UAA?"). The evidence cited is "Harkin 2007" and "Bionomics unpublished data." The difference is clearly having "rediscovered" Harkin 2007, the paper on which Athena has three coauthors and which was published in March 2007. Harkin 2007 thus appears to be the basis for Athena's "reclassification""to "K" status by the Mutation Update Committee in April 2009. That matters because Harkin 2007 already was published when the report about [redacted]'s mutation was prepared. This

is not lack of knowledge, but the failure to update the database according to Athena's own criteria.

11. It is my understanding that Athena has asserted the evidence it had in hand in June 2007 was reviewed and regarded as insufficient to classify the variant as "K" (disease-associated) until publication of the Heron study. Athena's own records seem to tell a different story: that when the Mutation Update Committee "rediscovered" the Harkin 2007 paper (sometime before April 3, 2009), the variant was classified as "disease-associated." Notably, confirmation of paternity in the Canadian index case (as reported in Heron 2010) was not used in Harkin. Yet the Harkin paper appears, according to the Mutation Update Committee's chronology, to be the basis for classifying the variant as disease-associated.

12. My conclusions regarding the significance of the materials produced by Quest in January 2020 are preliminary but are a reasonable interpretation of the data as it relates to the determination that Quest failed to properly categorize the mutation in its original report. Their errors did not constitute the practice of genetics but were due to administrative error or oversight. This omission constitutes a failure to properly update a database, not the exercise of clinical judgment by a geneticist.

13. This new information further supports my prior opinion that because of this failure to update the database, the physicians treating [redacted] did not receive a report that reflected the current state of knowledge about his particular SCNlA variant.

14. The revelation of materials associated with the Mutations Update Committee has had a substantial impact on my opinions of QUEST/Athena's culpability in the case. Moreover, the materials themselves contradict the testimony of the Defendants' witnesses. This suggests further lines of inquiry about facts related to the case would be helpful to confirm the Mutations Update Committee's process for collection and review of material used to reclassify the variant and update to the database itself. For example, it is my understanding from their Supplemental Requests for Discovery and Motion to Compel that the Plaintiffs are requesting the opportunity to direct inquiries to members of the Mutations Update Committee, particularly those who classified Y413N, and the process by which they did so. Additionally, it is still unclear why a year elapsed between the reclassification decision of the Mutations Update Committee and the point where the changes were finally committed to the ANTRIM database in 2010. Based on the materials produced in January 2020, the additional information requested by the Plaintiff could lead to further development of my opinions as expressed in this affidavit.

ECF No. 153-3, 5.

Dr. Cook-Deegan admitted in his deposition that the determination of whether a classification applies is something done by and within the discretion of a laboratory director. ECF No. 165-30, 7. Dr. Cook-Deegan admitted that he was unaware how Athena interpreted the category of "known disease-associated mutation" or the language Athena uses to define the term. Id. at 19. Dr. Cook-Deegan opined, however, that an English professor could make a determination whether "a paper that says this is a disease-associated variant" has been documented in the literature. Id. at 19.[6]

In the court's view, Dr. Cook-Deegan misapprehends the difference between an administrative exercise of noting the existence of scientific literature and a professional judgment as to whether the literature has been sufficiently vetted to confirm its reliability and relevance.

Izabela D. Karbassi, Ph.D., testified as a Fed. R. Civ. P. 30(b)(6) representative of Defendants. ECF No. 165-19. She reviewed the data available in 2007 and came to the same conclusion that the mutation was a VUS. Dr. Karbassi noted that there existed data in 2007 that was suggestive, but was not enough to classify the variant. Id. at 3. According to Dr. Karbassi, a mutation update committee reclassified Decedent's mutation in late spring 2009. The mutation update committee was composed of a team of Research and Development ("R&D") scientists and medical technologists working under the direct supervision of their R&D director and the laboratory director that was assigned to that gene. Id. at 5. The mutation list was created with R&D scientists and periodically updated. ECF No. 153-23, 3-4. Classifying a mutation as pathogenic required a certain level of approval that there was enough data to be called so. Id. at 4.

Dr. Karbassi testified that the mutation update committee was tasked to periodically review

[6]Plaintiff's other expert witness, Dr. Wiznitzer, acknowledged that clinical laboratory directors exercise their judgment in making a decision as to whether there is enough evidence to label a variant pathogenic. ECF No. 165-31, 3.

the literature, as well as collaborate with thought leaders and experts in the field to update the mutation list. Id. at 11. Dr. Karbassi further testified that, when a patient's chart recorded the existence of a VUS, a clinician typically would put in a request to Athena to reevaluate the variant. If the variant's classification changes, Athena will provide an updated report. Id. at 13-14.

Sat Dev Batish, Ph.D. testified that Athena used its best professional and scientific judgment in 2007 when it classified the DNA mutation in the SCN1A gene as a VUS. Dr. Batish noted that there was a question mark in the standard operating procedure where the variant was listed, indicating that the variant was reviewed in 2006 and there were some questions surrounding the variant. ECF No. 165-20, 3. Athena reported Decedent's variant as a VUS because there was only one other patient in which it had been identified and, thus, insufficient data existed to make a correlation. Id. at 4. The classification was changed upon the publication of additional literature. Id. at 9. Dr. Batish also testified that, by 2015, an additional patient carrying the SCN1A variant had been identified. Id. at 10. Dr. Batish observed that Harkin[7] and Berkovic[8] papers existed in 2006 that interpreted Decedent's SCN1A sequence, but paternity was not confirmed. Dr. Batish testified that Athena did not classify the variant as disease-causing until de novo paternity was confirmed in Heron's[9] papers in 2010. ECF No. 153-24, 3.

Defendants engaged as an expert witness Nancy B. Spinner, Ph.D., Division Chief, Division of Genomic Diagnostics; and Professor of Pathology and Laboratory Medicine, Perelman School of

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[7]Louise A. Harkin, Department of Genetic Medicine, Women's and Children's Hospital, North Adelaide, South Australia.

[8]Samuel F. Berkovic, Department of Medicine, University of Melbourne and Director of the Epilepsy Research Centre at Austin Health, Heidelberg, Victoria.

[9]Laurie Heron Seaver, M.D., Board Certified Medical Geneticist, Grand Rapids, Michigan.

Medicine, University of Pennsylvania. ECF No. 165-33. Dr. Spinner has been a Medical Geneticist since 1986 and is Board Certified as a Ph.D. Medical Geneticist and Clinical Cytogeneticist. She has run a CLIA/CAP certified diagnostic laboratory at the Children's Hospital of Philadelphia since 1992. Her laboratory performs comprehensive genomic testing of 100 genes for childhood epilepsy, including the SCN1A gene. She has worked extensively on interpretation of genomic data for clinical diagnosis. Id. at 3.

Dr. Spinner opined that Athena's classification is more detailed than that recommended by the American Board of Medical Genetics ("ACMG") guidelines and exceeds the standard of care for variant classification. Dr. Spinner noted that, despite the reporting of the prior mutation on Athena's SCN1A protocol list, "it was reasonable and within the existing standard of care to report it as of uncertain significance, and not pathogenic, since there was only a single prior patient with the mutation for whom paternity was unconfirmed. In light of all the above, it is clear that Athena followed the 2008 ACMG Guidelines' variant reporting decision tree with respect to Decedent's variant." Id. at 11. Dr. Spinner found no misclassification evidence in the 2007 Report, and that Athena's 2007 SCN1A result was correct in reporting Decedent's mutation as one of uncertain significance. Id. at 13.

Dr. Skinner further opined that Athena's decision to update Decedent's VUS upon the publication of additional evidence that the variant had been associated with SMEI is standard practice, "and quite common given the dynamic nature of genomic diagnostics due to the tremendous advancements in understanding the human genome over the past 10-15 years." Id. Dr. Skinner stated that, because of the time and financial commitment required to reclassify a variant, reclassification is done by most laboratories on a case by case, or variant by variant basis, with

initiation possibly by a referring physician contacting a laboratory with new clinical information or to seek re-evaluation of a variant.[10] Dr. Spinner observed that laboratories also may undergo systematic review of previously reported variants, instigated through the detection of a previously reported variant in a new case. Id. Further, Dr. Spinner stated that it was not inappropriate or a violation of any accepted standard of care for Athena to have issued the revised report only on the request of Plaintiff's health providers in 2014, given the transient nature of patients, the ever-changing variant database information, and the large number of samples that laboratories like Athena test and report every year. Id. at 14.

Dr. Spinner also observed:

> Finally, I have also been advised that Plaintiffs may take the position that Athena's 2007 result was reported as it was (VUS) due to a non-medical administrative error or oversight. There is nothing in my careful review of the records in this case to support such a claim. It is clear that Athena's scientists and other laboratory medicine professionals made a careful and considered judgment in this case to classify [redacted]'s variant as VUS, and I agree with that judgment as noted above. This is supported not only by the result itself, but by the testing data I have reviewed.

ECF No. 165-33, 15.

The court concludes that no reasonable jury could find Defendants erred in classifying Decedent's variant as a VUS, or that any misclassification was the result of nonmedical, administrative, ministerial, or routine care. Defendant's motion for summary judgment is granted as to this issue.

C. South Carolina Unfair Trade Practices Act

Plaintiff alleges that Defendants violated the SCUTPA by failing to comply with certain

[10]If Dr. Livingston had obtained the 2007 Report and requested a reevaluation of Decedent's VUS, the reclassification still would not have occurred until after Decedent's death in 2008.

provisions of the CLIA.

Under the SCUTPA, "[u]nfair methods of competition and unfair or deceptive acts or practices in the conduct of any trade or commerce are hereby declared unlawful." S.C. Code Ann. § 39-5-20(a) (1985).

> An unfair trade practice has been defined as a practice which is offensive to public policy or which is immoral, unethical, or oppressive. In order to be actionable under SCUTPA, the unfair or deceptive act or practice must have an impact on the public interest. [] An unfair or deceptive act or practice that affects only the parties to a trade or a commercial transaction is beyond the act's embrace.
>
> After alleging and proving facts demonstrating the potential for repetition of the defendant's actions, the plaintiff has proven an adverse effect on the public interest . . . the plaintiff need not allege or prove anything further in relation to the public interest requirement.
>
> > An impact on the public interest may be shown if the acts or practices have the potential for repetition. The potential for repetition may be shown in either of two ways: (1) by showing the same kind of actions occurred in the past, thus making it likely they will continue to occur absent deterrence; or (2) by showing the company's procedures created a potential for repetition of the unfair and deceptive acts.

Beneficial Fin. I, Inc. v. Windham, Appellate Case No. 2017-001954, Opinion No. 5753, 2020 WL 4495436 (S.C. Ct. App. Aug. 5, 2020) (internal citations and punctuation omitted) (quoting cases).

Specifically, Plaintiff contends that Defendants violated the following regulations:

> 42 C.F.R. § 493.1289 Standard: Analytic systems quality assessment.
>
> (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 analytic systems specified in §§ 493.1251 through 493.1283. . . .
>
> . . . .
>
> 42 C.F.R. § 493.1291 Standard: Test report.

. . .

(k) When errors in the reported patient test results are detected, the laboratory must do the following:

(1) Promptly notify the authorized person ordering the test and, if applicable, the individual using the test results of reporting errors.
(2) Issue corrected reports promptly to the authorized person ordering the test and, if applicable, the individual using the test results.
(3) Maintain duplicates of the original report, as well as the corrected report.

. . .

(l) Upon request by a patient (or the patient's personal representative), the laboratory may provide patients, their personal representatives, and those persons specified under 45 C.F.R. 164.524(c)(3)(ii), as applicable, with access to completed test reports that, using the laboratory's authentication process, can be identified as belonging to that patient.

Plaintiff's expert, Dr. Cook-Deegan, prepared an affidavit in which he discussed the manner in which Athena allegedly violated the CLIA regulations:

17. The revised SCNlA DNA sequencing diagnostic report issued on January 30, 2015 (Exhibit 2) by Athena Diagnostics, Inc. does not cite any new publication references used in the re-classification of this mutation (1237T>A, Y413N). This error violates CLIA regulation § 493.1241(c)(8), "any additional information relevant and necessary for a specific test to ensure accurate and timely testing and reporting of results, including interpretation."

18. In addition, the laboratory results and submitted clinical information within this revised report were signed off by Narasimhan Nagan, PhD, ABMG, Director, Genetics, and Bui Zhu, PhD, ABMG, Director, Genetics; both of whom left Athena Diagnostics, Inc. before 2009. This error violates CLIA regulation §493.1283(4) "the record system must include the records and dates of all specimen testing, including the identity of the personnel who performed the test(s)," which are necessary to assure proper identification and accurate reporting of patient test results.

19. Moreover, Athena Diagnostics, Inc. violated CLIA regulation §493.1291(1<) when it did not issue an amended report when this DNA mutation (1237T>A, Y413N) was re-classified as a "Known disease-associated mutation." "When errors in the reported patient test results are detected, the laboratory must: (k)(1) Promptly notify the authorized person ordering the test and, if applicable, the individual using

> the test results of reporting errors. (k)(2) Issue corrected reports promptly to the authorized person(s) ordering the test and, if applicable, the individual using the test results."
>
> 20. Furthermore, Athena Diagnostics, Inc. violated the patient's right to access protected health information (CLIA regulation §493.1291(1), HIPAA rule 45 CFR §164.524(c)(3)(ii)), when it refused to provide the completed test results to the patient (or the patient's personal representative) upon request in September 2014.

ECF No. 24-1, 4.

Dr. Cook-Deegan admitted in his deposition that he has never run a CLIA laboratory and has never been a laboratory director. ECF No. 165-30, 4. Dr. Cook-Deegan testified that he did not read the CLIA regulations and did not provide the language relating to the CLIA in his affidavit. Id. at 21. Dr. Cook-Deegan admitted that the regulation cited in paragraph 17 does not apply to the facts stated. Id. at 24. Regarding paragraph 18, Dr. Cook-Deegan conceded that he did not know whether Athena's records systems in 2007 or 2015 contained the information about who performed the tests, and that he may have misapprehended the language of the CLIA regulation. Id. at 26-28.

Dr. Spinner, Defendant's expert, reviewed Dr. Cook-Deegan's affidavit and determined:

> Regarding paragraph 17, CLIA Section 493.1241 applies to information to be included, by the ordering clinician, on the test requisition (request) for the testing (Standard: Test request). It has no application to information to be included on the test result by the lab.
>
> In paragraph 18, Dr. Cook-Deegan claims that inclusion of Drs. Nagan and Zhu on the revised result is a violation of the CLIA regulations. In fact, some laboratories, like Athena, issue revised results by simply reprinting the exact version of the original result with the inclusion only of a comment regarding the revision to the result. This does not violate CLIA Section 493.1283(4), which addresses "Standards: Test records." This standard requires that the records and dates of all specimen testing, including the identity of the personnel who performed the test, be kept by the laboratory. As long as Quest's record system at the time accurately recorded the then-active laboratory director and others responsible for issuing the revised result, it was in compliance with this CLIA standard. It is my understanding that Quest's system does contain this data.

> Dr. Cook-Deegan's claim that Athena violated CLIA by failing to "correct" its 2007 result is also incorrect for the reasons previously stated. CLIA Section 493.129I(k) applies to situations where the lab detects an "error" in its original report and Section 493.1289(a) requires the lab to have written procedures to address "problems identified in the analytic systems specified in §493.1251 through 493.1283." As discussed at length earlier, the 2007 report was correct and there were no "problems" with Athena's analytical systems with respect to this case.
>
> Dr. Cook-Deegan's claims that Quest violated CUA Section 493.1291(1) by "refusing" to provide the patient or her representative with requested results is also incorrect. The emails produced by Athena, and its Interrogatory responses, make it clear that it took prompt steps to comply with the patient request, while also ensuring that the result was properly updated given the classification change.

ECF No. 165-33, 15.

The court finds as an initial matter that Plaintiff has failed to establish any facts to support a cause of action under the SCUTPA. In addition, Defendants correctly assert that alleged CLIA violations cannot serve as the basis of a SCUTPA claim because CLIA does not provide for a private right of action. Accord Jewell v. Pinson, No. 255661, 2005 WL 2105417 (Mich. Ct. App. Sept. 1, 2005); Wood v. Schuen, 760 N.E.2d 651 (Ind. Ct. App. 2001); Scott v. Baylor Univ. Med. Cntr., No. Civ. A. 302CV1265-R., 2002 WL 32332551 (N.D. Tex., July 22, 2002); Whitehead v. Edmondson, No. 1:97CV29-S-D., 1998 WL 173226 (N.D. Miss. Mar. 24, 1998). This is important because the SCUTPA does not apply to "[a]ctions or transactions permitted under laws administered by any regulatory body or officer acting under statutory authority of this State or the United States or actions or transactions permitted by any other South Carolina State law." S.C. Code Ann. § 39-5-40.

The CLIA regulations set forth the conditions that all laboratories must meet to be certified to perform testing on human specimens. 42 C.F.R. § 493.1. Enforcement procedures are reserved to the Centers for Medicare and Medicaid Services and the Secretary of Health and Human Services. 42 C.F.R. §§ 1800-1850. Because Defendants are subject to this established regulatory procedure,

they are exempt from SCUTPA liability pursuant to S.C. Code Ann. § 39-5-40.

III. CONCLUSION

For the reasons stated, Defendants' motion for summary judgment (ECF No. 127) is **granted**.

IT IS SO ORDERED.

/s/ Margaret B. Seymour
Senior United States District Judge

Columbia, South Carolina

November 4, 2020.