When Does an Illness Begin: Genetic Discrimination and Disease Manifestation

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I. Introduction

Congress passed the Genetic Information Nondiscrimination Act of 2008 (GINA) in order to remove a perceived barrier to clinical genetic testing. By banning health insurance companies and employers from discriminating against an individual based on his or her genetic information, legislators hoped that patients would be encouraged to seek genetic testing that could improve health outcomes and provide opportunities for preventive measures. Their explicit legislative goal was "to fully protect the public from discrimination and allay their concerns about the potential for discrimination, thereby allowing individuals to take advantage of genetic testing, technologies, research, and new therapies."[1]

However, GINA left a number of issues unresolved, most notably failing to define the concept of disease manifestation. GINA was structured such that it only provides protection against misuse of genetic information up until the point when an individual's disease has manifested.[2] It protects an individual with a genetic predisposition for a disease, but not an individual actively suffering from that disease.[3] This would not be problematic if the exact moment of disease manifestation was always obvious, but diseases often develop in a predictably non-predictable manner, and doctors often find it difficult to pinpoint the root cause of symptoms at the beginning stages of a disease or condition.

This is particularly worrisome given the fact that GINA was premised on the genetic science of the 1990s and early 2000s. Early in the field of genetic medicine, except for a handful of well-studied variants that had been clearly linked to specific diseases (e.g., BRCA and breast cancer), our understanding of the relationship between genetic variation and human disease was relatively thin. In this scientific environment, there was a distinct conceptual divide between genetic tests and the eventual manifestation of actual disease. Scientific advances over the past decade or so have given us the tools to much more clearly understand the pathways through which genotype and phenotype are related. We are now beginning to comprehend the cellular pathways and biological mechanisms that explain the specific ways in which genetic variants can impact human health.[4] As we continue to elucidate these bio-

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logical processes, and particularly as we develop the technical ability to measure the subtle chemical and physiological changes associated with the development of genetically-based human disease, the notion of a genetic test as being distinct from actual disease will dissipate, making it that much harder to clearly establish when a disease has manifested.

The extent of legal protection under GINA rests on how manifestation is defined and subsequently interpreted. If the concept of disease manifestation is not carefully drawn, it could undermine the very protections GINA was designed to provide. When Congress passed GINA, the concept of manifestation appeared four times in the legislation, but the bill did not define the term.[5] Early critiques of GINA noted that a clarification of what legislators meant by manifestation was needed, especially because the vast majority of those who will interpret the law will not have medical backgrounds.[6] Eventually, regulators settled on a definition, but it remains to be seen whether it is the one that best advances the protective goals of GINA. This paper examines the range of possible legal definitions of disease manifestation and explores the historical struggle that courts have faced when trying to apply these different definitions.

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Section II provides essential background about the law and discusses GINA in the context of subsequent health care reform. Although health care reform has drastically changed the health insurance landscape, we make the case that analysis of GINA implementation remains relevant and essential. The importance of appropriately implementing GINA is particularly true given the existing gap between when the protections of GINA end and when the protections of the ADA begin. Section III examines how courts in the past have interpreted the concept of manifestation. There are three common interpretations of manifestation of a disease: (1) manifestation as apparent symptoms, (2) manifestation as patient action, and (3) manifestation as physician action. The final section analyzes the definition of manifestation adopted in GINA. We argue that the definition of manifestation adopted by GINA is, on its face, comprehensive, but that courts may have difficulty adequately applying the definition during litigation involving very early stages of a disease. This could lead to a potential for extensive battles between experts about whether the disease in question has manifested. Due to this potential for unwieldy litigation and gaps in protections in the law, we argue that courts should view information about the manifestation of a disease in the light most favorable to the plaintiff in order to fulfill the protective goals of GINA.

II. Background

GINA is something of a legislative experiment, whose success or failure could influence future efforts to regulate genetic discrimination. GINA was heralded as the "first civil rights bill of the 21st Century,"[7] but GINA is very different from other existing civil rights bills. First, while prior civil rights bills were enacted to address growing problems of actual discrimination, GINA was passed largely to address fear of potential discrimination and to prevent discrimination from occurring in the future.[8] Second, GINA is more limited in scope than many previous civil rights bills because it only applies to employers and health insurance companies. Finally, unlike other civil rights bills, GINA goes beyond simply preventing employers and health insurance companies from discriminating against an individual due to his or her genetic information. Instead, GINA seeks to prophylactically prevent discrimination by prohibiting employers and insurance companies from even obtaining genetic information in the first place.

GINA was passed shortly before the Patient Protection and Affordable Care Act, which seemingly undercut the need for genetic-specific legislation. Most notably, one of the major provisions in the health care reform bill prohibits insurance companies from discriminating on the basis of pre-existing conditions.[9] There are a number of reasons, however, why analysis of GINA implementation remains important and relevant. First, the protections for adults with pre-existing conditions will not go into effect until 2014. In the next few years, many individuals will rely on GINA's protections, which will take on increasing relevance as genetic testing technology continues to advance. Second, especially given the contentious political environment surrounding health reform, it is unclear whether the health reform legislation will be overturned or

altered. Third, GINA applies to both employment and health insurance. Therefore, this discussion remains very salient for the employment context since the health reform bill regulates only health insurers in the context of pre-existing conditions. Fourth, and most importantly, the definition of pre-existing condition in health care reform is currently being worked out in the relevant regulations. It is vital to fully understand both the definition of manifestation in GINA and the definition of pre-existing condition in health care reform to ensure that there is no gap between the definitions and to fully understand the interplay between them.

Furthermore, to the extent that GINA serves as a model for future legislation on genetic discrimination in long-term care, disability, or life insurance, settling the question of disease manifestation will be extremely important. Many states are continuing to introduce legislation to fill the gaps that GINA does not cover.[10] These legislative efforts will inevitably turn to GINA's regulations for guidance. For example, SB 559, a civil rights bill recently introduced and passed in California, protects genetic discrimination in housing, employment, life, long-term care, and disability insurances, and other businesses.[11] This bill borrows directly from GINA, adopting a similar definition of genetic information that includes the manifestation of disease in family members.[12]

Finally, under any definition or interpretation of manifestation, there will be a gap between when an asymptomatic individual will be protected by GINA and when their symptoms will rise to the definition of disability protected under the Americans with Disabilities Act (ADA).[13] Under the ADA, an individual meets the definition of disability if their symptoms substantially limit a major life activity. There is arguably no protection for individuals who have manifested some symptoms, but whose symptoms have not risen to the level of substantial limitations. Prior to GINA, in 1995, the EEOC issued guidance stating that discrimination based on genetic information could constitute disability discrimination under the "regarded as" prong of the ADA.[14] However, since that guidance, scholars have questioned whether courts would rule this way in practice.[15]

In order to fully protect individuals in employment across the continuum from genetic predisposition to manifested symptoms that substantially limit, new legislation would be needed.[16] For example, health care reform helped to bridge the gap between GINA's health insurance provisions and previous insurance laws by banning insurance companies from denying coverage for preexisting conditions. However, until legislation in the employment context comes to fruition, a gap between GINA and ADA will exist, making it even more important to appropriately define the concept of manifestation in a way that minimizes that gap. For this reason, we argue that courts should interpret GINA's definition of manifestation broadly in order to protect as many individuals, avoiding the potential doughnut hole between the protections afforded by GINA and the ADA.

Settling on an appropriate legal definition for disease manifestation is more than just an academic exercise. Proper implementation of GINA is important for a number of reasons. The first, and most obvious, involves justice and equity. GINA creates the promise of protecting individuals from genetic discrimination and therefore encourages individuals to take genetic testing and participate in research. Second, it is essential to identify unintended, and undesirable, consequences of the regulations in order to ensure that the law achieves its anticipated effects in both the health insurance and employment sections of the law.

III. Disease Manifestation Jurisprudence

Courts have long struggled with the issue of when a disease first manifests. This paper's analysis includes research from cases that interpreted the Wartime Disability Compensation Act (WDCA) or the National Childhood Vaccine Act (NCVA)[17] and cases that discussed the initial onset of a disease in other contexts. In addition to interpretations of the NCVA, the analysis of manifestation commonly arises in two contexts. First, statute of limitations cases raise questions about whether a plaintiff manifested a disease within sufficient time to have a cause of action. Second, in insurance questions, a court must often determine whether a plaintiff has a pre-existing condition or whether his disease manifested after his health insurance became effective.

The strategies of interpreting and defining manifestation in these different areas have varied widely among courts. This paper introduces an analytic framework that divides the courts' approaches into three categories: (1) manifestation as apparent symptoms, (2) manifestation as patient action, and (3) manifestation as physician action. The next section highlights paradigmatic cases, and discusses the benefits, drawbacks, and unanswered questions presented by each of the possible frameworks.

Manifestation as Apparent Symptoms

CASES

Under a "manifestation as apparent symptoms" analysis, a disease manifests when the symptoms of the disease begin, not when a diagnosis occurs. For example, in *Cardamone v. Allstate Insurance Company*,[18] the

plaintiff visited her doctor due to stomach pains, one day prior to the effective date of the her health insurance policy. The doctor mentioned six possible causes of the plaintiff's pain, including gallstones, but noted that further testing was needed to determine the true source of the pain. The following day, the plaintiff's health insurance took effect. Subsequently, the plaintiff had X-rays taken, and the doctor confirmed the diagnosis of gallstones. The court held that even though the patient's diagnosis was not definitive, her gallstones had manifested at the time of her first appointment because the symptoms were "clear" and "unmistakable."[19] If X-rays had been taken earlier, the plaintiff's disease could have been diagnosed immediately. Therefore, because the symptoms of the plaintiff's disease were present before the effective date of her insurance policy, the court found that the disease had manifested. As a result, she was disqualified from compensation for gallbladder surgery due to the pre-existing condition clause of the insurance policy.[20]

Other cases have followed similar manifestation as apparent symptoms analysis. For example, in *Life General Security Insurance Company v. Cook*, the Fourth District Court of Appeals of Florida held that despite an earlier misdiagnosis, the insured patient had a pre-existing condition due to the presence of symptoms during the exclusionary period of her health insurance.[21] During this exclusionary period, the patient saw her doctor for abdominal pain and diarrhea and was incorrectly diagnosed with gastritis. After the exclusionary period ended, the insured saw a specialist who correctly diagnosed the symptoms as Crohn's disease. The court reversed the lower court's finding in favor of the insured patient because the symptoms were present before the policy began.[22]

ANALYSIS OF "MANIFESTATION AS APPARENT SYMPTOM" CASES

Courts arguably choose the definition of manifestation as apparent symptoms in cases involving questions of pre-existing conditions because it provides an objective and consistent framework for analysis. It does not matter in these situations what an individual's condition is or turns out to be. In these contexts, what matters is whether the insured had any pre-existing condition at all. Accordingly, courts have focused on general symptoms to determine a pre-existing condition, not specific diagnoses.

This approach is attractive because it does not rely on human action. In the other two approaches, discussed below, the outcome of each analysis depends on either the patient's or the doctor's actions. The same type of disease or symptoms in those contexts could result in a different outcome depending on what actions the players take. Additionally, this approach is ostensibly more objective because it looks only at the factual symptoms and disease itself, rather than the more subjective beliefs of individuals.

Although it is helpful for a court to look only at the symptoms of a disease and not the action of a patient or doctor, this approach has potential drawbacks, most notably associated with the risks of over-inclusiveness and inequity. For example, it is easy to imagine a situation where an individual has slight, recurring stomach pain and does not go to see a doctor for the pain because the problem is not unduly burdensome. However, once the individual has health insurance, she visits a doctor and discovers that the mild symptoms indicate a serious condition. Under the manifestation as apparent symptoms analysis, a court could find that these slight stomach pains constituted a pre-existing condition; however, it can be argued that this result is not fair if the symptoms were not serious and if the individual did not take any action or know about a more serious complication.

This definition of manifestation is unsatisfactory due in part because the definition is simply a direct rewording of the legal question and does not provide any additional explanation or clarification. Under this definition, a disease manifests when its symptoms begin. But it does not resolve the problem of determining the exact time when a nuanced disease manifests. Specifically, do very slight symptoms constitute the manifestation of a disease? The court in *Cardamone* recognizes this potential problem: "While we are not certain when her symptoms first appeared, it is clear that by this date they were sufficiently pronounced for her to seek medical attention."[23] Although often it is clear when symptoms of a disease have passed the threshold of development, this definition of manifestation is insufficient because it relies on establishing when symptoms first begin, a much more difficult proposition.

Manifestation as Patient Action

CASES

A second approach measures manifestation from the point when a patient takes action based on symptoms. Although the *Cardamone* court does not explicitly analyze how the patient's actions relate to the manifestation of a disease, it hints that the disease had manifested partly because the patient sought medical attention.[24]

Other courts have more explicitly linked manifestation with patient intent. In *Doroshow v. Hartford Life and Accident Insurance Company*, the Third Circuit Court of Appeals found that, based on the insurance contract, a disease had manifested when the patient

sought advice with a specific concern in mind.[25] The plaintiff sued his insurance company after he was denied benefits due to a pre-existing condition. The plaintiff was suffering from symptoms that indicated a motor neuron disease, and he was eventually diagnosed with amyotrophic lateral sclerosis (ALS) after the exclusionary period of his insurance ended. The plaintiff had visited a doctor during the exclusionary period and was misdiagnosed. In the doctor's notes, there were even comments that he did not feel that the symptoms were caused by ALS.[26]

Despite the doctor's early findings, the court found that the insurance company could deny benefits for the subsequent diagnosis and treatment of ALS based on the pre-existing condition exclusion. The court noted that there is a difference between seeking care or advice for a particular condition versus seeking treatment for a "nebulous or unspecified medical problem."[27] In this case, because the plaintiff sought advice specifically for a motor neuron disease, not vague symptoms, the court found that the ALS had manifested despite the misdiagnosis. Thus, it was the plaintiff's intent, not the diagnosis, which indicated that the disease had manifested.

Other courts have defined manifestation in a similar manner by examining patient action rather than patient intent. In these circumstances, the courts found that a disease manifests when the individual "became aware of the disease, or should have become aware of it in the exercise of reasonable diligence.[28] For example, in *Tillman v. Lykes Brothers Steamship Company*, the Southern District of Texas found that the plaintiff did not have a cause of action because his disease manifested after the statute of limitations had run. The plaintiff argued that his asbestosis had manifested prior to the relevant date because he experienced symptoms such as shortness of breath.[29] The court found, however, that because the plaintiff neither was aware nor should have been aware that the symptoms were caused by asbestosis prior to relevant date, his disease had not manifested before the statute of limitations.

This interpretation is slightly different from *Doroshow* because in *Tillman*, the court found that the disease did not manifest until it was clear that it was asbestosis, not in the years prior when the plaintiff suffered from breathing problems.[30] However, the court's inclusion of the exercise of a reasonable diligence standard demonstrates that, in order for a disease to manifest, the patient must have, or should have, acted on symptoms.

Analysis of "Manifestation as Patient Action" Cases

In this line of cases, manifestation of a disease occurs when symptoms become so clear that the patient acts upon their symptoms. The court in *Doroshow* followed the framework of patient action because, similar to the other pre-existing condition contexts discussed above, the insurance companies' policies of excluding pre-existing conditions is not dependent on the final diagnosis.[31] However, the *Doroshow* court seemed to depart from simply viewing manifestation as symptoms because of the challenges of determining when slight or vague symptoms manifest. By focusing on patient intent rather than symptoms, the *Doroshow* court could more clearly define this threshold. Symptoms are clear or strong enough to constitute a manifest disease when a patient intends to seek advice for those symptoms and their possible causes.

The *Tillman* court had a different motivation for examining manifestation as patient action. *Tillman* was a worker's compensation determination brought under the Longshoreman and Harbor Worker's Compensation Act (LHWCA).[32] As the court noted, "The LHWCA is not concerned with pathology, but with industrial disability; and a disease is no disease until it manifests itself."[33] In the context of worker's compensation, a patient action framework is appropriate. An individual may be experiencing symptoms of a disease, but unless these symptoms are severe enough to cause her to seek medical attention, she probably does not have a condition sufficient to impair an ability to work and thus constitute an industrial disability.

The manifestation as patient action framework is beneficial because it answers some of the lingering questions left by the apparent symptoms framework. The patient action framework takes into account that the manifestation of a disease is a cumulative process, as discussed under the Wartime Disability Compensation Act.[34] Therefore, these courts have decided to interpret the patient action as the catalyst, rather than the more difficult standard to determine: when the symptoms first begin. Additionally, this framework is reasonable because it generally is the individual who has the disease who is bringing the court action. Thus, it is appropriate to examine the plaintiff's intent or actions to determine when his or her disease manifested.

The downside of this framework is that each patient has different levels of risk aversion, medical knowledge, and access to health care. By focusing on patient action, rather than only on the symptoms of the disease, this framework adds a degree of variability to the analysis, which makes it a difficult standard for courts to interpret. For example, in *Doroshow*, the court

noted that seeking advice for a nebulous or unspecified condition does not constitute the manifestation of a disease.[35] It did not, however, clarify what makes a condition nebulous or unspecified. Additionally, by focusing on patient action, the courts run the risk of potentially punishing those individuals who are proactive about their health care and who pay attention to and seek advice for changes in their bodies.

By relying on lay individuals the patient action framework is also problematic because it leaves unanswered questions about what happens when a patient has incorrect information about his or her condition. It is easy to imagine a situation where a patient believes that they may have a specific health condition, but it is ultimately determined that they do not have that disease. This is especially a concern in the context of genetics because an individual with a family history of a disease may be more likely to think that he or she has a specific, familial health condition.

Similarly, under a *Tillman* analysis, one can imagine a patient who thinks that he or she has a disease, but it is actually a misdiagnosis. Was the incorrect disease manifest at these times? In this way the patient action framework is problematic due to the uncertainties that are common when patients and doctors are trying to make an initial diagnosis.

Manifestation as Physician Action

CASES

The most common framework that courts have utilized when determining when a disease manifests is physician action. Under this framework, a disease is considered manifest at the point when a doctor could diagnose the disease based on present symptoms. For example, in *Dowdall v. Commercial Travelers Mutual Accident Association of America*, the Supreme Judicial Court of Massachusetts held that a disease is manifest when "there is a distinct symptom or condition from which one learned in medicine can diagnose the disease."[36] In *Dowdall*, the insurance company refused to pay for treatment for the plaintiff's multiple sclerosis (MS) treatment because it was a pre-existing condition. The plaintiff was not definitively diagnosed with MS until after his health insurance policy became effective; however, the court held that knowledge of the existence of the disease on the part of the plaintiff was not necessary for manifestation. The doctor's testimony revealed that, although he did not inform the plaintiff at that time, he had reasonable cause to believe that the plaintiff had MS prior to the effective date of the health insurance policy based on the symptoms. The court therefore held that the plaintiff's MS had manifested prior to the policy and affirmed the lower courts motion for directed verdict in favor of the defendant.[37]

Under this framework, a disease can be manifest, not only when the individual has no knowledge of the disease, but also when the doctor has no knowledge of the disease. In this analysis, a disease manifests when a doctor *could have* diagnosed the disease. In *Dirgo v. Associated Hospitals Service, Incorporated*, the plaintiff suffered from fatigue and lower abdominal discomfort.[38] Both he and his doctor were unsure what was causing these symptoms until two weeks after the effective date of the plaintiff's health insurance policy. However, the doctor testified that he could have diagnosed the condition, prior to the effective date, based on the symptoms if he had taken an X-ray. The Supreme Court of Iowa therefore held that the plaintiff's disease had manifest because the doctor could have diagnosed the symptoms.[39]

ANALYSIS OF "MANIFESTATION AS PHYSICIAN ACTION" CASES

The courts have adopted the manifestation as physician action analysis because of a recognition that diseases develop slowly. In *Dowdall*, the court noted "the mere presence of latent germs or seeds of illness in the body prior to the issuance of such a policy would not preclude recovery. 'Few adults are not diseased, if by that one means only that the seeds of future troubles are not already planted.'"[40] Because the Dowdall court recognized that many individuals have latent diseases, they used the physician action framework to define manifestation.

This framework is beneficial because, in the context of pre-existing condition litigation, it creates a balance between protecting insurance companies from applicants fraudulently obtaining coverage for a disease of which they possess current knowledge and protecting those insurance applicants who are unaware of a latent disease they have when they apply for health insurance.[41] By relying on the physician as an ostensibly neutral third party, this framework creates balance by insulating the decision of when a disease manifests to a player that is neither the insurance company nor the patient. Like the patient action framework, it also recognizes the cumulative and possible latent nature of diseases.

One drawback of the physician action framework is that it does not require a definitive diagnosis by a doctor. The initial diagnosis of a condition is not always an easy task. Different physicians would give tests at different times, or try to rule out different diseases before narrowing the diagnosis. During the initial stages of a disease, a physician is not likely to jump to the conclusion that the patient has a rare genetic

condition. She will likely test common explanations first. However, it is possible that the physician could have diagnosed the rare condition at the time. As we discuss in more detail later, this creates the potential for litigants to find expert witnesses that would argue that if they were the plaintiff's physician, they would have diagnosed the disease earlier. This might lead to a battle between experts in complicated cases. Competing experts could argue whether the doctors could have diagnosed a disease given a patient's condition at the time in question. Individual plaintiffs will generally be at a disadvantage in this situation because insurance companies will have superior resources to find and hire relevant experts.

IV. GINA's Definition of Manifestation

Congress passed GINA in order to protect individuals from discrimination based on a family history of, or genetic predisposition to, a disease. The law is based on the premise that it would be unfair for an employer or health insurance company to make a decision about an individual based on a condition that may or may not actually develop in the future. Thus, GINA bans discrimination based on information about the probability of getting a condition. For this reason, GINA only covers genetic information and not symptomatic genetic conditions. Because the manifestation of a disease establishes a threshold of protection for individuals under GINA, the definition of manifestation is crucial to the scope of the bill's protections.[42]

The GINA legislation includes the concept of disease manifestation in four different contexts. First, GINA includes many rules of construction sections that clarify that the law does not prohibit insurance companies from altering premiums based on an individual's manifested disease.[43] Second, the definition of genetic information includes "the manifestation of a disease or disorder in family members of such individual."[44] Third, one exception to the definition of a genetic test is "an analysis of proteins or metabolites that is directly related to a manifested disease, disorder, or pathological condition that could reasonably be detected by a health care professional with appropriate training and expertise in the field of medicine involved."[45] Finally, under the employment section of GINA, an employer is not be considered to be in violation of GINA if it acquires or uses medical information that is not genetic information about a manifested disease, disorder, or pathological condition.[46] Despite the inclusion of manifested disease in the legislation, Congress did not provide a definition of manifestation in the bill.

The final regulations by the Equal Employment Opportunity Commission (EEOC) states that a disease is manifested when:

> an individual has been or could reasonably be diagnosed with the disease, disorder, or pathological condition by a health care professional with appropriate training and expertise in the field of medicine involved. For purposes of this part, a disease, disorder or pathological condition is not manifested if the diagnosis is based principally on genetic information.[47]

Like the majority of the previous court cases that have analyzed the manifestation of a disease, the GINA definition follows the manifestation as physician action framework.

Compared to the other two frameworks, we agree that physician action is the most appropriate for the GINA context. The "manifestation as apparent symptoms" framework would be difficult to apply given the gradual onset of many genetic diseases. Many genetic conditions begin with subclinical symptoms that a patient may not be aware of, but that a diagnostic test could in some situations detect, like hemochromatosis.[48] Thus, it is difficult to ascertain in these contexts when a disease crosses the threshold from presymptomatic or subclinical symptoms to a manifested disease.

"Manifestation as patient action" is also problematic in the realm of genetics because genetic diseases are often associated with family history. An individual with a family history of a disease is more likely to go to the doctor with the intent to seek advice or treatment for a particular genetic disease. However, the definition of manifestation in GINA makes it clear that a disease is not manifest if the diagnosis is based principally on genetic information. GINA defines genetic information broadly to include genetic test results, the use of genetic services, and family medical history. Accordingly, the patient intent framework does not conform with the goals of protecting individuals against genetic discrimination since under this framework, an individual with a family history of a genetic disease would be more likely to be found to have a manifested disease than one without a family history.

Therefore, the physician action framework appears to be the best framework for the definition of manifestation, given the slow development of many genetic conditions and the likelihood that individuals with family histories of a genetic condition will go to doctors more than others. It is also the best framework because the onset of genetic diseases and conditions are often complicated. It is beneficial to have the defi-

nition rest upon the judgment of a doctor with medical training, rather than the patient or symptoms. Patient reactions can vary, but by having a physician standard that includes the "could have" portion of the definition, it ensures that the general standards of the medical field will be taken into consideration, not solely individual reactions.

Potentially Insufficient Protection

Despite the strengths of the physician action framework, however, there are still questions about the adequacy of the way that manifestation is specifically defined under GINA. First, there is a key unanswered question in the definition as it stands: how should courts interpret the "has been or could reasonably be diagnosed" clause?[49] Genetic diseases can be extraordinarily rare and complicated. What might clearly appear to be a genetic disease in hindsight might not be so clear during a patient's diagnostic odyssey. If, like in *Cardamone*, a physician surmises that her patient has one of a number of possible diseases, including a genetic one, has the disease manifested? If the initial physician did not diagnose the genetic disease, how much evidence is sufficient for a defendant to argue that a definitive diagnosis could have been made?

Second, the definition has a narrower view of who counts as a physician than previous court analyses. GINA requires diagnosis by a health care professional with appropriate training and expertise *in the field of medicine involved*. Previous case law has required that *one learned in medicine* make the diagnosis. Narrowing the definition to those with training in the field of medicine involved is intended to ensure that the individual making the manifestation determination has sufficient knowledge. This portion of the definition, however, could be problematic for plaintiffs. Many patients are treated by their general doctor, who will often not have detailed knowledge of genetic conditions. A defendant could argue that this general practitioner does not have a specific enough field of knowledge to diagnosis the disease. Thus, the defendant could argue that if the patient had seen a specialist with expertise in genetics (or with expertise in the genetics of that disease), then it would have been possible to have diagnosed the disease. Many patients, however, do not have ready access to specialists. Additionally, if the patients are relying on their general doctor for referrals, they may not know to ask to see a specialist or demand a second opinion from a specialist.

Further complicating matters, the definition of manifestation also includes a stipulation that a disease cannot be considered manifest if the diagnosis is based primarily on genetic information or tests. This is included in order to protect those with genetic conditions from discrimination, but may not completely fulfill its goal because genetic information can suggest the need for non-genetic testing, which can in turn lead to a diagnosis. To illustrate this concern, imagine an individual, James, who has a family history of hemochromatosis. Worried about developing this condition, James takes a genetic test to determine whether or not he has inherited his family's predisposition to the disease. The genetic test comes back positive. Although James is pre-symptomatic at the time, because of the genetic test results his doctor suggests diagnostic testing. The diagnostic test reveals elevated iron levels in his bloodstream, the earliest indicator of the disease. By catching the disease before James became symptomatic, his doctors can treat him through a process similar to blood letting, avoiding potential organ failure.[50]

Under GINA's definition of manifestation, James' hemochromatosis could be considered as manifested since a physician could have been able to diagnosis the disease with a simple blood test. However, the only reason that James and the doctor knew to take the blood test was due to the genetic test and James' family history, not due to any current medical symptoms. If James had gotten insurance coverage between receiving a positive genetic test and taking the blood test, there is a possibility that the insurance company could drop him from coverage because he had a pre-existing condition that a physician should have been able to diagnose.

Suggestions for Future Interpretation of GINA's Definition of Disease Manifestation

GINA's regulations use the physician action framework to define manifestation. Although we argue that this is the best framework to utilize in this law generally, the concerns raised above suggest potential gaps in GINA's protection. Given the underlying purpose of GINA — to encourage patients to take genetic testing and participate in genetic research — we argue that the definition of manifestation of a disease should be read in the light most favorable to the plaintiff. We suggest the following recommendations to mitigate these potential problems with GINA's definition of manifestation.

First, courts should only use the subjective "could reasonably be diagnosed" portion of the definition in those cases where a diagnostic test should have been administered, given symptoms. It should not be invoked at those early stages of disease development when individuals seem asymptomatic until genetic information suggests that further diagnostics should be taken. In order for GINA's goals to be reached, the

disease should not be considered manifest if the doctor could have diagnosed the disease based only on family history information alone.

Second, courts can encourage an appropriate application of GINA by placing the burden on the insurance companies and employers when arguing that a physician could have been able to diagnose the disease. GINA's manifestation definition subjective prong states that a disease is manifest when "an individual has been or *could reasonably be* diagnosed with the disease."[51] The inclusion of a reasonable standard in the definition shows that the regulations do not envision a broad use of the subjective portion of the definition.

Therefore, in order to show that an individual could have *reasonably* been diagnosed by a physician, the courts should place the burden on the defendants to prove both that the physician would have been able to diagnosis the disease with a test *and* that other physicians in the same position would have administered the diagnostic test without knowing the genetic information of the individual. For example, in James' situation, if an insurance company were trying to argue that the hemochromatosis was a pre-existing condition, it would have to prove two things. First, it would have to show that a physician would have been able to diagnose the hemochromatosis through a blood sample before the insurance plan went into effect. Second, it would have to provide evidence to show that other physicians in the same position would have given James the blood test even if they had not known James' family history or genetic test results.

This second step of the proof is essential to help protect individuals from genetic discrimination. The goals of GINA are undermined if an insurance company or employer can argue that a disease is manifest because a doctor could have diagnosed a condition at a time when the individual did not have any outward signs or symptoms. If the only way that a physician would know to give the diagnostic test was because of a family history or genetic test, then this is ultimately genetic discrimination. A patient's disease should not be considered manifest if there are so few symptoms that a doctor would not perform diagnostic measures without knowing about genetic information. By requiring the defendants in GINA cases to bear the burden of proving that a physician "could reasonably" diagnose a patient, without reliance on solely genetic information, courts will help to uphold the underlying purpose of GINA.

Conclusion

In March 2011, the EEOC began seeking public comment on a plan to retroactively review major regulations.[52] This new plan gives a unique opportunity to reexamine parts of regulations, such as GINA, that may not be completely clear. Our hope is that during this process, careful thought is given to how manifestation should best be defined under GINA. Ultimately, the physician action definition used in GINA is most appropriate for this difficult concept. Clarification is needed, however, as courts begin to grapple with the concept of manifestation. They should not allow GINA's definition of manifestation to provide defendants with an avenue for arguments that circumvent the protections GINA was designed to provide. Instead, in order to further the important goals of GINA, the definition should be viewed in the light most favorable to the plaintiff and the burden should be placed on the defendants to prove that a physician could reasonably have diagnosed a patient if they are utilizing the subjective prong of the manifestation definition.

In order to further the important goals of GINA, the definition should be viewed in the light most favorable to the plaintiff and the burden should be placed on the defendants to prove that a physician could reasonably have diagnosed a patient if they are utilizing the subjective prong of the manifestation definition.

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References

1. *Genetic Information NonDiscrimination Act*, § 2(5) (2008).
2. 29 U.S.C.A. § 1182(a)(3)(B) (2008); 42 U.S.C.A. § 2000ff-9 (2008).
3. Note: An individual's genetic information is always protected under GINA. However any information about an individual's manifested condition or current symptoms is not protected. For example, if an individual has a family history of heart dis-

ease, this fact would be protected, but the individual's current high blood pressure is not.

4. E. D. Green, M. S. Guyer, and the National Human Genome Research Institute, "Charting a Course for Genomic Medicine from Base Pairs to Bedside," *Nature* 470, no. 7333 (2011): 204-213.
5. *Genetic Information Nondiscrimination Act (GINA) of 2008*.
6. M. Rothstein, "GINA, the ADA, and Genetic Discrimination in Employment," *Journal of Law, Medicine & Ethics* 36, no. 4 (2008): 837-840.
7. G. J. Annas, P. Roche, and R. C. Green, "GINA, Genism, and Civil Rights," *Bioethics* 22, no. 7 (November 7, 2008): ii-iv, at iii.
8. *Id.*
9. *Patient Protection and Affordable Care Act*, Public Law 111–148 (2010).
10. D. Vorhaus, "Is the Genetic Rights Movement Picking Up Steam?" *Genomic Law Report*, March 16, 2011, *available at* <http://www.genomicslawreport.com/index.php/2011/03/16/is-the-genetic-rights-movement-picking-up-steam/> (last visited September 6, 2012).
11. California Senate Bill No. 559, Introduced by Senator Padilla, February 17, 2011, *available at* <http://www.leginfo.ca.gov/pub/11-12/bill/sen/sb_0551-0600/sb_559_bill_20110217_introduced.pdf> (last visited September 6, 2012).
12. Id.
13. See Rothstein, *supra* note 6.
14. EEOC Guidance, Section 902 Definition of the Term Disability. (Note: While the ADAAA regulations altered much of the Section 902 definition guidance, those regulations did not address genetic information).
15. W. J. McDevitt, "I Dream of GINA: Understanding the Employment Provisions of the Genetic Information Nondiscrimination Act of 2008," *Villanova Law Review* 54, no. 91 (2009): 91-116, at 104.
16. See Rothstein, *supra* note 6.
17. There are two federal statutes that include the concept of manifestation, the *Wartime Disability Compensation Act (WDCA)*, 38 U. S. C. § 1110 (1998), and the *National Childhood Vaccine Act (NCVA)*, 42 U.S.C.A. § 300aa-2, et. seq. (2003). The WDCA introduces a complicated concept of manifestation without ever defining what events or symptoms make a disease manifest. Under the WDCA, a disease is considered "incurred in or aggravated by" service in the military if it becomes "manifest to a degree of 10 percent" within a certain time period after the service. The statute does not include a discussion of what it means to have a disease manifest to a certain percentage and therefore lacks guidance in GINA analysis. Although this was researched, no cases interpreting the WDCA were found. The NCVA also introduces the concept of manifestation without providing a definition within the statute. The NCVA allows individuals to recover compensation for vaccine related injury or death. Section §300aa-11(c)(1)(C)(i) permits individuals to receive compensation for illnesses, disabilities, injuries, or conditions that manifested within the time period set forth in an accompanying Vaccine Injury Table.
18. *Cardamone v. Allstate Ins. Co.*, 364 N.E.2d 460, 462 (Ill. App. 1977).
19. *Id.*
20. *Id.*, at 463.
21. 648 So.2d 237 (1994).
22. *Life General Sec. Ins. Co. v. Cook*, 648 So.2d 237 (1994).
23. *Cardamone v. Allstate Ins. Co.*, 364 N.E.2d 460, 462 (Ill. App. 1977).
24. *Id.*
25. 574 F.3d 230, 236 (3rd Cir. 2009).
26. *Id.*, at 232.
27. *Id.*, at 235.
28. *Tillman v. Lykes Brothers Steamship Company*, 732 F.Supp. 1402, 1405 (S.D. Texas 1990).
29. *Id.*
30. *Id.*
31. 574 F.3d 230, 236 (3rd Cir 2009).
32. 732 F.Supp. 1402, 1404.
33. *Id.*, at 1405.
34. *Wartime Disability Compensation Act (WDCA)*, 38 U. S.C. § 1110; Supra note 10.
35. *Doroshow v. Hartford Life and Accident Insurance Company*, 574 F.3d 230, 235 (3rd Cir. 2009).
36. 344 Mass. 71, 74 (Mass. 1962).
37. *Id.*
38. 210 N.W.2d 647, 648 (Supreme Court of Iowa, 1973).
39. *Id.*, at 651.
40. 344 Mass. 71, 74 (Mass. 1962), quoting *Grain Handling Co., Inc. v. Sweeney*, 102 F.2d 464, 466 (2nd Cir.).
41. *Mutual Hospital Ins., Inc. v. Klapper*, 153 Ind. App. 555, 560 (Ind. App. 1972).
42. See Rothstein, *supra* note 6.
43. E.g., 29 U.S.C.A. § 1182(a)(3)(B) (2008).
44. 29 U.S.C.A. § 1191b(d)(6)(A)(iii) (2008).
45. 29 U.S.C.A. § 1191b(d)(7)(B)(ii) (2008).
46. 42 U.S.C.A. § 2000ff-9
47. 29 C.F.R. § 1635.3(g) (2011); The most recent health insurance regulations have a similar definition of manifestation. 26 C.F.R. §54.9802-3T(a)(6)(i) (2009).
48. Hemochromatosis is a genetic disease that causes iron levels to build up in the body; without treatment, this can lead to organ damage or failure.
49. 29 CFR § 1635.3(g); 26 CFR 54.9802-3T(a)(6)(i).
50. S. Baruch, "Your Genes Aren't Covered for That: One Year Later, Gaps in Genetic Discrimination Legislation Reveal the Challenges Ahead," *Science Progress* (2009), *available at* <http://www.scienceprogress.org/2009/06/gina-challenges/> (last visited September 6, 2012).
51. 29 CFR § 1635.3(g) (emphasis added).
52. U.S. Equal Employment Opportunity Commission, "EEOC's Final Plan for Retrospective Review of Significant Regulations," *available at* <http://www.eeoc.gov/laws/regulations/comment_retrospective.cfm> (last visited September 11, 2012).