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Does genetics matter for disease-related stigma? The impact of genetic attribution on stigma associated with rheumatic heart disease in the Western Cape, South Africa

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ABSTRACT

Introduction: A common concern in African genomic research is that such work may cause, or increase, stigma associated with particular diseases or population groups. While there is some evidence suggesting that genetic attribution of disease might impact stigma, there exists no evidence for the situation in African populations. With increasing genomic research in African populations, questions about the effect of genetic attribution on disease-related stigma are salient for stakeholders involved in implementation and regulation. To understand better the relationship between stigma and genetic attribution, we interviewed people with Rheumatic Heart Disease (RHD) in the Western Cape of South Africa.

Method: We conducted 11 focus group discussions with RHD patients of mixed-ancestry in the Western Cape, exploring the impact of genetic attribution on stigma. Participants had previously consented to participate in genomic research, attending information sessions on genetics. We explored the impact of genetic attribution by introducing both genetic and environmental causes to RHD and by specifically probing how these various causes would likely impact selected features of disease stigma.

Results: Participants reported varying experiences of stigma relating to RHD, such as labelling, social exclusion and discrimination at the workplace. They had some understanding of genetics, either in general, or in relation to their illness. Participants' understanding depicted multiple causal models to explain RHD including genetic, environmental and bacterial causation. Overall, participants did not make a connection between genetics as a cause of RHD and their experiences of stigma.

Discussion: In this study we found no support for the concern that genetic attribution of RHD, understood by participants in our study as a genetic predisposition to developing the disease, would impact stigma associated with it. Our findings provide some reassurance that genomic research may be unlikely to cause an increase in disease-related stigma in the South African context.

1. Background

1.1. Disease stigma and genetic attribution

The influence of bio-genetic explanations on disease-related stigma is an important consideration in the development and use of genomic methods (Kong et al., 2017). Genomic technologies have revolutionised the manner in which we think about health, disease and treatment (Hood and Friend, 2011), thus holding the potential to aid prevention and care significantly. There however are legitimate concerns over increasing geneticisation of diseases as a step towards eugenics (Savulescu and Kerin, 1999). Although theoretical frameworks to understand directionality have existed since the late 1990s (Lippman, 1992; Weiner, 1993), empirical studies on the impact of genetics on

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disease stigma date back only about a decade (Phelan, 2005; Phelan et al., 2002, 2006; Sankar et al., 2006).

Opposing perspectives have been proposed for understanding how genetic knowledge could impact on stigma relating to disease. The first perspective suggests that genetic knowledge could increase both public and internalised stigma. This model assumes that individuals' reactions to genetic information will reflect genetic essentialism i.e. the belief that individual health outcomes are pre-determined by genetic make-up, and in this sense that "we are our genes" (Phelan, 2005). Phelan (2005) notes three implications of this perspective for disease stigma, namely: that the individual living with a disease is essentially different; that the condition is seen as severe and unrelenting; and that there is a strong belief that family members will also develop the condition (Phelan, 2005). The second perspective argues that genetic knowledge may result in a decrease in stigma, because knowing that the cause of illness is a result of genetic variation could reduce self-blame, where individuals may be perceived as and may feel less responsible for their condition (Phelan et al., 2002). This model is rooted in attribution theory; proposed by Weiner (1985) and based on the idea that humans are interested in creating causal links between events. He asserts that when people are believed to be in control of negative outcomes (in this particular case an illness and/or associated behaviour), they are more likely to be held responsible, whereas if they are perceived to have little or no control over negative outcomes, they are more likely to be shown sympathy (Corrigan, 2000; Weiner, 1985). Empirical studies find evidence in support of both these theories.

The results of studies which investigate the impact that genetic knowledge could have on public perception reflect a complex relationship between genetic knowledge and stigma. Importantly, many empirical studies have focussed on mental illness, and all of these studies have recruit participants who do not have the specific disease being studied. Phelan and others have, through telephonic surveys of US public perceptions, tested various reactions to genetic causation vignettes. In one study, they found that reactions to presumed genetic causation were not explained by attribution theory (i.e., it had no effect on the public's perception of blame and punishment) but instead supported a genetic essentialist theory in that people were more likely to believe, if the disease was attributed to genetic causes, that mental illness was more serious and that children were also likely to develop the conditions (Phelan, 2005). In another study, Phelan and colleagues tested public perceptions of treatment recommendations and strategies in response to stimuli suggesting that depression and schizophrenia were genetic in origin. They found in cases where genetic attribution was made explicit, whilst respondents were more likely to recommend hospitalization or prescription drugs (but not more likely to prescribe seeing a mental health professional) as treatments, they also thought these were unlikely to effectively treat depression or schizophrenia (Phelan et al., 2006). A meta-analysis found contrasting evidence that while bio-genetic explanations do seem to decrease personal blame, it also heightens beliefs that people living with mental illnesses are dangerous (Kvaale et al., 2013). Additionally, a systematic review found mixed evidence, with some sources supporting essentialist claims, and others describing a negligible effect of genetic attribution (Angermeyer et al., 2011). Hamilton and Robson (2019), in their synthesis of evidence on the psychosocial impact of genomic testing in the context of cancer research, conclude that participants are unlikely to experience sustained psychological stress when receiving results about genetic mutations with high penetrance. However, they caution that such evidence is not yet available for newer technologies that may identify many variants of unknown significance. Finally, Lebowitz (2019) who explores the impact of psychiatric genomic results, notes that while genetic explanations for mental illnesses may decrease self-blame, it may increase a sense of hopeless about chances of recovering.

The results from the few studies that have focussed on investigating the impact of genetic attribution with people who actually have the disease being investigated. A US-based study, across multiple diseases, found that the effect of genetic attribution on stigma is contingent on participants' lived experience of the disease within their social and cultural contexts (Sankar et al., 2006). In a different study women with eating disorders were likely to have a decreased sense of personal responsibility with genetic attribution, and an increased sense of helplessness (Easter, 2012). Similar studies focussing on mental health have also found evidence in support of claims suggesting that bio-genetic explanations may decrease stigma (Laegsgaard et al., 2010; Meiser et al., 2005).

1.2. Stigma in African Genomics

Given the potential of genomic medicine to revolutionise healthcare, concerns have been expressed that lower- and middle income countries (LMICs), especially on the African continent, should not be excluded from its benefits, since exclusion would further contribute to global health disparities (Bustamante et al., 2011; Singer and Daar, 2001). Subsequently, there have been several genomic initiatives that have played an important role in ensuring that African scientists and populations are included in the genomic revolution (Folarin et al., 2014). A flagship initiative that has gained wide recognition is the H3Africa Consortium, which combines research in genomics and bioinformatics with work on ethical, legal and social issues. The current project was conducted under the umbrella of the H3Africa Consortium.

As in other contexts, a primary ethical concern regularly identified by stakeholders in African genomics research relates to whether, and how, genomic research could influence disease-related stigma in African research contexts. Yet there is a lack of studies that investigate the impact of genetic attribution on disease stigma in African populations. To date, there are three published studies that have investigated this relationship, with only one directly designed to investigate the impact of genetic knowledge on stigma. In this study exploring the potential for ethnic group stigma as a consequence of participation in genomic research, de Vries et al. (2012) found that genomic research would be unlikely to create stigma, although it could re-enforce existing forms of stigma, especially among groups which are already marginalised or where the research involves questions or data interpretations with particular normative implications. In a different study, Tekola et al. (2009a), working in Ethiopia with people living with podoconiosis, found that pre-existing stigma levels attached to this illness influenced decisions to be part of the study. Given the high levels of stigma attached to the condition, re-emphasising its genetic link created anxieties about increased stigma for biological relatives – called associative stigma. Participants suggested that consent needed to be sought from the entire family, not just the individuals living with the condition, as stigma could increase for the family as a whole (Tekola et al., 2009a, 2009b). In another study, Marsh et al. (2011) examined sickle cell disorder and genetics in Kenya, finding that gender often mediated experiences of stigma. Their findings show how stigma experiences in this context are not exclusively mediated by biomedical explanations but rather occur at the complex intersection of genetics, culture, and structural economic and gendered inequalities.

To shed light on the relationship between genetic attribution and stigma relating to disease in the African context, we set out to conduct research with participants of ongoing H3Africa genomic studies taking place in the Western Cape, South Africa. These included two studies: one involving patients with a diagnosis of schizophrenia and the other patients with a diagnosis of rheumatic heart disease. These two studies provided the opportunity to undertake interviews with the research participants, to investigate both internalised stigma and associative stigma. In this study, we employ a broad definition of internalised stigma to mean both the experience of being aware of stigma towards oneself as a result of having a specific disease (often referred as felt stigma), or accepting negative perceptions or consequences as a result of having a particular disease (often referred to as self-stigma) (Livingston and Boyd, 2010). Associative stigma is the stigma that is

experienced by others as a result of their biological or other association with patients living with a stigmatised condition (Bos et al., 2013). In this paper, we report on our data regarding patients with Rheumatic Heart Disease. Rheumatic Heart Disease (RHD) is a chronic acquired heart condition that is caused by untreated infection with Streptococcus pharyngitis that can led to permanent damage to heart valve tissue (Carapetis et al., 2005). RHD largely affects only the global poor, with the condition virtually having been eradicated in high-income countries (Carapetis et al., 2005; Robertson and Mayosi, 2008). The enduring high prevalence of this illness in LMICs is especially problematic given that preventative measures are both effective and inexpensive (Irlam et al., 2013). As in other countries, South African incidences of RHD are correlated with socio-economic metrics of inequity, such as poverty, over-crowded housing, poor nutrition and low levels of education (Barth et al., 2015). Living with RHD has high and enduring social and economic costs for both patients and families of patients. Some of these costs are educational limitations due to increased school drop-out rates among children due to parental absenteeism, financial limitations due to loss of employment, loss of income, dependence on others, as well as lifestyle limitations (Robertson and Mayosi, 2008).

Given that there is mixed evidence for the impact of genetic attribution on disease stigma, and the dearth of evidence in LMICs, this study investigates the impact of genetic attribution on disease stigma among patients with RHD in the Western Cape of South Africa.

2. Methods

2.1. Participants and recruitment

We conducted 11 focus group discussions (FGDs) with patients (n = 52) living with Rheumatic Heart Disease (RHD) between March and November 2017. For the initial recruitment we used the contact details in the study database for the parent genomic study called the Genetics of Rheumatic Heart Disease project (RHDGen). The RHDGen project aimed to investigate genetic factors that contribute to the development of Rheumatic Heart Disease. All FGDs were conducted at a public hospital in Cape Town.

The participants enrolled in our study had previously consented to participate in the RHDGen genomic study. Many of these participants would have been involved in RHD patient awareness days where talks and information sessions on RHD and genetics were offered (Zühlke et al., 2018). However, we also had to recruit from other databases of RHD patients, given that many of the patients in the RHDGen contact database had either changed their contact details and not informed us; could not make any of the scheduled FGDs due to poor health; work commitments; or were deceased. In total, about half (n = 30) of the participants we recruited had previously enrolled in RHDGen. The other participants (n = 22) had previously enrolled in other RHD-related research but not in genomic research specifically. Although the participants enrolled in the RHDGen study were exposed to previous explanations of genomics during the consent process of those studies, we did not observe any differences in how they responded to questions seeking to probe genetic literacy in the FGDs as compared to participants who had not previously participated in genomics research. In other words, participants generally had a basic understanding of genetics (as will be illustrated in the next section), regardless of whether they had been enrolled in the RHDGen study.

All of the participants were of mixed-ancestry descent and regularly attended the public hospital where we conducted our research. In South Africa, the mixed-ancestry population makes up 8% of the national population and 49% of the population in the Western Cape (Africa, 2012). This racial minority is the second poorest in the country, an economic trend that reflects South Africa's history of racialised inequality.

In terms of gender, 90% of our sample were women. This gender proportion is reflective of the broader RHD patient community. For example, in the RHDGen project, 71% of the participants were female. One possible explanation for this gender disparity is that women may be more likely to use health services consistently and actively seek and adhere to treatment (Noone and Stephens, 2008; Oksuzyan et al., 2008). Another is that RHD is more likely to be diagnosed in pregnancy, meaning that men may go undiagnosed. The average age of participants was 59 years old, with the youngest participant being 35 and the oldest 78 years old. In terms of education, 4% of participants completed post-secondary school (professional or university) education. Additionally, 80% of participants received a government pension or disability grant as their primary form of income, which provides a monthly income of R1500 (121 USD).

2.2. The use of focus group discussions

We opted to use focus group discussions for this research. Our reasons were partly related to the paucity of African data on the relation between genetic attribution and stigma and the absence of any kind of evidence of disease stigma in the mixed ancestry population of South Africa. Against that background, we felt FGDs were a suitable method of initial exploration to help elucidate the relationship between genetic attribution and disease stigma.

2.3. Discussion guides

Discussion guides were developed as part of the larger Stigma in African Genomics project of which this project was part. The guides used vignettes to explore various questions relating to genetic attribution and RHD.

The vignettes had three primary stages. The first stage explored general questions relating to genetics, during which we probed knowledge of genetics. In this stage we also explored general ideas and beliefs around RHD, for example we asked people what RHD is and what causes it. The second stage of the vignettes explored experiences of living with RHD with specific reference to internalised forms of stigma (i.e. both as felt and self-stigma) as it relates to genetics factors – here we probed the impact of having RHD on participants' lives, the impact on finding employment, and whether having RHD would influence decisions on getting married and having children. We did this by exploring how knowing that the disease was genetic would impact these domains by asking questions like, “would knowing that the disease is caused by genetics change the way family/friends relate to someone with RHD?”, “would knowing that the disease is genetic affect one's desire to be friends/marry a person with RHD?” and “how would knowing the cause of disease impact the chances of getting employment?”. In the third stage of the vignette, we focussed on exploring associative stigma. In this stage, we asked if participants would be willing to be associated with or married to someone who has RHD, again with a focus on exploring how genetic attribution would change this. We therefore asked questions like, “If you knew that the disease was genetic, how would you feel about marrying someone with RHD?” and “How would you feel about being friends with a person who has RHD?”

2.4. Analysis

FGDs were conducted in English and Afrikaans. Recordings were transcribed verbatim and the Afrikaans FGDs were translated to English by a professional translator. Transcripts were analysed using NVivo11 software (“NVivo 11,”). Thematic analysis was used to analyse the data, with all codes being derived from the data (Braun and Clarke, 2006). Two researchers independently developed an initial coding scheme using the same three transcripts. After meeting to compare the resulting open -coding schemes, one researcher developed a hierarchal coding scheme that was then applied to a separate fourth transcript. Following discussion with the research team, we made final changes to the

hierarchical coding scheme with one researcher coding the transcript and another researcher checking the coding scheme used it to code all transcripts. At each stage, two researchers met to discuss each step of the analysis and the interpretation of study results.

2.5. Ethics approval

Ethics approval for the study was received from the University of Cape Town's Ethics Committee (FHS204-2015). All participants signed a consent form agreeing to be part of focus groups discussions and for the discussion to be audio recorded.

3. Results

3.1. Experiences of stigma

While participants generally reported that living with RHD did not significantly impact their ability to live a "normal life", some participants reported that they struggled to sleep as a result of the unfamiliar sound of the mechanical valve inserted to replace the damaged heart valve. Participants often reflected on how having RHD limited their ability to perform ordinary tasks. For instance, many women reported not being able to do as many household chores as they previously could. While some of these experiences were not reported as stigma, many participants also described experiences of stigma related to living with RHD. In some instances, having RHD compromised participants' ability to engage in physical activities, which often involved being part of a community or social group. This often resulted in being excluded from peer-group activities that were meaningful to participants. One participant noted that:

> It was a problem because I loved athletics, I loved ballet … and I couldn't be part of it anymore because of my getting tired, and so I just had to take a step back and realize, ok, I can't …

While for some participants, having RHD meant they were not able to take part in personally meaningful activities resulting in a degree of labelling and social exclusion. One participant, whose son also developed RHD, reflected on his son's experience of being labelled as 'sick' resulting in social exclusion:

> [My] son didn't have friends, he stayed indoors because he couldn't play the way he wanted to … They [his friends] didn't want to play with him, they teased him, they were scared, they were scared to play with him because he was sick …

Participants also described experiences of stigma relating to employment. Some participants reported being excluded from employment as a result of having to manage their heart condition. Specifically, barriers to employment related to missing work because of regular hospital appointments, ill health, or not being able to do specific kinds of work (i.e. manual labour, which for individuals with a low level of education is the most likely form of work in South Africa). For example, one participant noted, "When I was working, my boss said, if I stay out of work every time to go to hospital then I might as well stay at home." In South Africa, most people are forced to rely on under-resourced public healthcare systems, which results in having to spend a large part of the day waiting to see a doctor and receive medication. In addition, the patients enrolled in our study were managed in a district hospital close to Cape Town's centre. Yet these patients live on the outskirts of the city, meaning they have to travel by public transport for an hour or two. These combined factors mean that patients frequently needed to take off an entire day, which had repercussions on their ability to seek or maintain employment.

On the other hand, participants also described situations where their employers were generous and gave them time off to attend hospital and, in some instances, also provided transport to the hospital. However, many of these examples were explained as a result of "proving" to their employer that they were not "lazy" or "taking advantage".

3.2. Understanding genetics

In the FGDs, before exploring experiences of stigma, we established what participants understood about genetics. Many of the participants recruited in this study showed a degree of familiarity with genetics. When participants were asked, "What do you know about genetics?", they described genetics as "something passed down in the blood" and similar explanations. Overall, participants had a general understanding of genetics as something inherited across generations. Some even articulated that a disease passed down may skip a generation. Participants often used metaphors like that of a seed or tree growing:

> Genetics is something that comes down in the family, it grows … like a cell, it develops forward, it's been planted in the family as it goes, the family tree, it carries on from the one to the other one, sometimes it skips the first … and then the second one gets it, so it runs in the family …

Although participants had difficulty describing exactly how genes are passed on, we found that overall, participants had a clear understanding of genetics as being something that was passed on through family lineage or 'in the blood'. Importantly, these insights into genetics were shared across all the focus groups, regardless of whether participants had previously participated in a genomic study.

3.3. RHD, genetic attribution and stigma

One way we explored the impact of genetic attribution was to investigate how participants understood the cause of RHD. In the vignettes offered to participants as part of the FGD, we introduced both genetic (heritable) and environmental causes of the illness. In the ensuing discussions, we probed whether and how these various causes would impact people's relationships, decisions to get married and have children, and employment. In our study, participants largely seemed to understand 'genetic attribution' in terms of a greater genetic predisposition to developing RHD.

When we asked participants what caused them to develop RHD, some reported that having RHD was a result of their family's genetics. In these instances, participants usually directly traced a history of heart disease in their family. For example, one participant could trace back a history of heart disease to both her maternal and paternal ancestors:

> …my mother has chest problems, this asthma, my father's side are heart people, my father's brothers, many of them are died because of heart attacks, and one of the doctors said it's in our genes.

In other instances when participants used genetic explanations, they did not think about genetics as determinative but rather described it in non-absolute terms, e.g. that it may not necessarily be passed on to every generation or may not be an influencing factor at all. Another participant who demonstrated knowledge that a genetic disease may skip a generation said:

> For me, it's not like your children will inherit it, because they told me I have rheumatic fever, my son didn't inherit it, and I have two sons, one is 54 and the other 34, and they both didn't inherit it, so their children's children might inherit it, but they didn't get it, perhaps it skip two generations or so, then it will come up again, but it might come out somewhere in the generations.

Participants displayed some understanding of the relationship between genetics and disease, and they also reported experiences of stigma as a result of living with RHD. However, few participants seemed to consider directly that genetic attribution – or speaking about their illness as being caused by a genetic predisposition – would be likely to impact experiences of disease stigma. In fact, only one

participant equated genetics with stigma, namely:

> Genetics, people will think it's a disorder, some disorder that you have, in your genetic system, your combination of human being, so there's something wrong.

However, for the majority of participants, genetics was not associated with any form of social stigma or judgement.

3.4. Minimizing the impact of genetic attribution

In the previous two sections we established that participants reported stigmatising experiences. We also showed that participants had some understanding of genetic causation with regards to disease. Yet when we probed whether and how genetic attribution of RHD would shift experiences of stigma for these patients, we found very little evidence that this would be the case. When exploring our data to understand why that could be, we found that one explanation is that participants attributed their condition to a range of causes, and genetics was only one of three models participants employed to explain their disease aetiology.

Besides genetics as a cause of RHD, participants also reported that RHD was caused by bacteria. RHD is caused by an auto-immune response to a streptococcal infection, often first manifested as a sore throat, and participants accurately described the cause of RHD as a "germ", "bug" or "bacteria" and generally linked it to having a sore throat as a child. One participant related the following:

> I suffered a lot with tonsillitis … the doctor always explained to me, the germs that's here, closest organ is your heart so that's where your germs go settle …

However, most importantly our participants drew on issues of poverty and racialised inequality to explain their heart condition. One participant reflected on the fact that it was after her family was forced to move to the Cape Flats, where houses did not have proper insulation, that she started to get throat infections:

> …we moved to Lavender Hill, we were the first people that moved … the walls and stuff, it was very cold … from there onwards, I was sick …

Lavender Hill was one of the areas to which people of colour were forced to move during Apartheid. These areas usually have small houses, are built very close together, and often must accommodate large, and sometimes multiple families. Some participants linked the development of RHD to living in small spaces or reflected on poverty and restricted access to proper nutrition. These participants described inherited social vulnerability as the cause of RHD.

While participants did not explicitly express such conditions as being stigmatised, effects of structural inequality in the South African context, which exists across entire generations, translate into life experiences that are stigmatising of their own accord. For example, living in poverty, not being able to provide for your family, having been subjected to sexual or physical violence, or living in communities ridden by constant gang violence translates into conditions or experiences that are stigmatising.

4. Discussion

Our study aimed to explore the impact of genetic attribution on disease stigma in the mixed ancestry population living with RHD in the Western Cape, South Africa. We found no support for the concern that genetic attribution of this condition, understood by participants in our study as a greater genetic predisposition to developing RHD, would likely cause or increase stigma associated with this condition. We suspect that this may be because the possible impact of genetic attribution on disease stigma is displaced by stigmatising features of other causal beliefs and of the environment in which many of our participants live.

Participants employed multiple casual models to explain their condition, including genetic explanations, where participants shared how other members of their family also suffered heart disease; infectious disease explanations, where participants attributed their illness to germs or bacteria; and experiences of living in poverty. For participants in this study, genetics was one of many ways to make sense of their condition.

In addition to multiple explanatory models, we also found that participants were managing more immediate challenges and conditions, some of which may already be stigmatising. These relate to the effects of transgenerational poverty and living in areas with high incidences of gang violence and rape. Coping with these realities and associated stigma appeared to mitigate the relative importance of any stigma associated with RHD.

The findings on use of multiple disease explanatory models are consistent with other research investigating the relationship between genetic attribution and disease stigma. For example a study by Condit (1999) shows the complexity and variation of aetiological frameworks used by laypeople, with genetics being only one factor in how laypeople understand disease causation. With regards to how bio-genetic explanations can be made sense of in relation to other stigmatising conditions, Kong et al. (2017) notes that one weakness of relying exclusively on bio-genetic explanations to reduce stigma in the context of mental health is that such a focus does not consider how stigma relates to structural factors such as economic or gender. Furthermore, our observation that context matters in understanding the relation between genetic attribution and illness strongly resonates with the conclusion reached by Parens and Appelbaum (2019), in the introduction of a recently published special issue of the *Hastings Centre Report*. In that, they call attention to the importance of context in understanding the psychosocial impact of genetic testing, cautioning against generalising findings from one context to others.

Overall, our findings suggest that multiple causal models, of which genetics is one, and navigating more immediate stigmatising conditions, displaces the potential impact of genetic attribution on stigma in this context. This is a novel finding given that this is the first study of the impact of genetic attribution on stigma in South Africa, and the first purposely designed study to investigate this relation on the continent. It is also novel in that it does not support either of the two primary theoretical models, genetic essentialism or genetic attribution, even though supporting evidence has been found for both theories in Western contexts. Although this does not preclude the impact of genetic attribution on disease stigma, it suggests that the impact of genetic knowledge on stigma in contexts like South Africa is more complex and that mitigating factors must be considered.

What such a finding also implies is that understanding disease stigma, as opposed to other forms of stigma, is complicated not in the least because it is difficult to isolate one source of stigma from another. For example, in our study when participants report on discrimination in the workplace, this could be a result other forms of discrimination related to class or the lack of skills and not just result from having a chronic heart condition. In a research context where illness, poverty and inequality collide, it is important to be cognisant of the compounded nature of the stigma that participants experience.

There are three important limitations to our study. The first relates to the challenge of isolating the potential impact of genetic attribution from other causal models when mediating discussions with research participants with only moderate prior knowledge about genetics. We sought to address this challenge by specifically aiming to recruit participants who had previously consented to participate in a genomic study and who were likely to have received some information about genetics. We also presented specific genetic causation models in the vignettes. However, we recognise that our analysis is only an initial attempt at elucidating the possible relationship between genetic knowledge and disease stigma in the African research context. Furthermore, we recognise that focus groups are limited in their ability

to highlight causality and caution that such findings must be interpreted alongside other studies in similar contexts, when they become available, employing a variety of other methods which together can perhaps provide more definitive conclusions. A second limitation is our focus only on the mixed-ancestry population in the Western Cape of South Africa. Participants enrolled were a relatively socio-economically homogenous group, although religiously and linguistically diverse. Research is needed with a more diverse sample in terms of race, ethnicity, geographic location and socio-economic characteristics. This could potentially provide greater insight into mechanisms of stigma related to RHD (or other cardiovascular diseases). Another limitation of this study was that the findings may offer limited insight into the relationship between genetic attribution and stigma insofar as RHD is not a severely stigmatised disease. Whilst our findings discredit the concern that genetic attribution could cause stigma where there is none – a concern habitually identified in the context of African genomics research – more research focussing on this relationship in non-Western contexts is needed for diseases that are more explicitly stigmatised. Studies which focus on genetic diseases in more affluent contexts in South Africa would be especially instructive given that the impact of genetic attribution may be clearer in the absence of impoverished material circumstances that compound the effect of disease stigma.

Overall, our findings suggest that when genomics research is conducted in areas where participants are juggling the compounded stigmatising effects of structural inequalities, enduring discrimination, and poverty, there is little evidence to suggest that genetic attribution alone could significantly impact the stigma experienced by patients for the condition under study. What is rather more likely is that any effect that genetic explanations of illness could have on disease-related stigma would be displaced by the stigma associated with those other conditions.

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Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.socscimed.2019.112619.

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