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Perceptions of predictive testing for those at risk of developing a chronic inflammatory disease: a meta-synthesis of qualitative studies.

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Abstract

Background: The availability of tests to predict the risk of developing chronic diseases is increasing. The identification of individuals at high risk of disease can trigger early intervention to reduce the risk of disease and its severity. In order for predictive tests to be accepted and used by those at risk, there is a need to understand people's perceptions of predictive testing.

Method: A meta-synthesis of qualitative research that explored patient and public perceptions of predictive testing for chronic inflammatory diseases was conducted. Studies were coded by researchers and patient research partners, and then organised into common themes associated with the acceptability or use of predictive testing.

Results: Perceived barriers to predictive testing were identified including a concern about a lack of confidentiality around the use of risk information; a lack of motivation for change; poor communication of information; and a possible impact on emotional wellbeing. In order to reduce these barriers, the literature shows that a patient centred approach is required at each stage of the testing process. This includes the consideration of individual needs such as accessibility and building motivation for change; readily available and easy to understand pre- and post-test information; support for patients on how to deal with the implications of their results; and the development of condition specific lifestyle intervention programmes to facilitate sustainable lifestyle changes.

Conclusion: Patients and members of the public had some concerns about predictive testing, however, a number of strategies to reduce barriers and increase acceptability are available. Further research is required to inform the development of a resource that supports the individual to make an informed decision about whether

to engage in a predictive test, what test results mean, and how to access post-test support.

Key words

Chronic disease; predictive testing; perceptions; risk; qualitative research; synthesis

Background

Chronic diseases with an inflammatory aetiology, such as inflammatory bowel disease (IBD), rheumatoid arthritis (RA), cardiovascular disease (CVD) and diabetes are typically characterised by dysregulation of the immune system and long-term activation of inflammatory processes. Over time, long-term inflammation can lead to damage in affected tissues (1). For many types of chronic inflammatory disease, early intervention has substantial benefits for the disease outcome (2,3), although help seeking and referral of patients after symptom onset are often delayed (4,5).

Interest is emerging in the idea that treating people at risk of disease before symptoms appear can reduce the severity and consequences of future symptoms and may even reduce the risk of the disease developing (6,7,8). This has resulted in a drive to identify susceptibility markers in an attempt to identify individuals at risk of developing such diseases through predictive testing (i.e. genetic testing and non-genetic risk assessments). For example, diagnostic and predictive genetic tests for maturity-onset diabetes of the young (MODY) are now available for many at risk families (9). Similarly, in RA, research is increasingly focusing on methods of identifying diagnostic biomarkers and disease mechanisms which predict the transition from health to RA (4,10,11,12).

The availability of tests to predict the development of chronic inflammatory diseases is rapidly increasing, with a range of predictive tests available in various healthcare settings as well as directly available to the consumer from private companies in the form of home self-testing kits (13). Consequently the number of individuals at risk of developing a chronic inflammatory disease, who may be faced with the prospect of

predictive testing, is increasing (14). The decision whether to engage in predictive testing may be driven by personal preferences for the management of risk and risk related information, understanding of risk and the meanings associated with being labelled as an ‘individual at risk’ (15).

Previous qualitative research studies have been undertaken to explore perceptions of genetic risk in individuals at risk of developing certain illnesses, generating a broad range of themes and topics potentially important to consider before introducing new predictive testing. In particular, reviews of the qualitative literature have been undertaken within the field of cancer genetics where predictive testing is seen as important for the early detection and treatment of disease (16,17). This may, in part be driven by public perceptions about the severity of cancer and the need for early identification and intervention (18,19). Public perceptions of chronic inflammatory diseases such as RA, diabetes and CVD are more diverse (20,21,22), but often these diseases are thought to be less serious (22). Such beliefs might in turn negatively influence perceptions regarding the need for early detection and intervention for these diseases.

Understanding people’s perceptions of predictive testing is paramount if these are to be accepted and utilised by those at risk. However, no review of the qualitative literature on public perceptions of predictive testing for common chronic inflammatory diseases has been undertaken to date. The aim of the current study was therefore to identify research papers which have used qualitative methods to explore the perceptions of predictive testing held by members of the public and people at risk of developing chronic inflammatory non-malignant disease for which there are genetic

and environmental risk factors. Meta-synthesis methods were used to investigate common themes and implications for future predictive testing programmes.

Methodology

Meta-synthesis

A literature search was performed using Medline/PubMed, PsycINFO, CINAHL and Google Scholar. Only peer-reviewed papers published between 1989 and 2014 were included and the search was limited to articles published in English. The search concentrated on articles looking at predictive testing and screening for chronic inflammatory disease, including diabetes, CVD, IBD, common inflammatory rheumatic diseases (RA, ankylosing spondylitis, Sjogren's syndrome), asthma, multiple sclerosis and psoriasis. The search terms, inclusion criteria and the procedure for generation of the final sample of 11 articles presenting qualitative data relating to diabetes (n=7), CVD (n=3), and IBD (n=1) can be found in Figure 1. No relevant articles relating to any other disease were identified.

To assess the quality of the identified papers KB and RS independently carried out quality appraisals, using an existing quality appraisal framework based on a modified version of the Critical Appraisal Skills Programme (23) qualitative checklist. Each paper was rated using a three point scale (0 = Serious methodological issues; 1 = Minor methodological issues; 2 = Robust) for each of the following criteria 1) How relevant was the paper was to the present study's research question? 2) To what extent did the paper add value to answering the research question? 3) How methodologically robust was the study?

On the basis of these ratings each paper was then independently categorised as ‘key’, ‘adequate and relevant’ or ‘flawed or not relevant’. There were no disagreements between researchers and none of the 11 papers identified were classed as ‘flawed or not relevant’. As a result all 11 papers were included in the meta-synthesis.

Meta-synthesis: Data analysis and interpretation

Meta-synthesis methods were used to identify interrelated themes from the published qualitative studies (24-26). The meta-synthesis focused on two substantive areas. The first was the major themes and findings related to the perception of testing to quantify the risk of developing a chronic inflammatory disease in a) family members of patients with the disease**;** b) patients with early symptoms; and c) the general population. The second focus was on the implications and recommendations for practice that are suggested by these findings.

KB and RS independently read and analysed each of the selected articles. The data extracted from each paper were based on the original authors’ interpretation of the primary qualitative data. The themes derived from these primary data are termed first order constructs (27; Box 1). A thematic approach was then taken, grouping first order constructs from each paper into core themes. The researchers recorded which papers contributed to each theme, in terms of relevant data or contradictory or contrasting results. KB and RS then discussed and agreed on emergent themes, and their relevance to the review’s primary aims. The initial thematic framework was subsequently validated by a second round of coding of three of the 11 papers selected at random. This validation coding was conducted by three patient research

partners with a chronic inflammatory disease (two EuroTEAM patient research partners with RA and one patient with RA from the University of Manchester RA Research User Group). Their feedback and recommendations supported the initial thematic framework and no substantial changes were made.

Once the draft themes had been identified, the authors conducted a focus group with seven patient research partners (six RA patients and one patient relative recruited from the UK, Sweden, Estonia and Romania) in order to refine, prioritise and validate the themes. Two of these six individuals had also been involved in the initial validation coding of the data. During the 1.5 hour focus session the draft themes and corresponding quotes were presented and the following questions were discussed with the focus group members:

1) Do these themes and subthemes look logical?
2) Based on your experience as a patient or relative do these themes reflect reality?
3) Do you think there are any important concepts described within these subthemes?
4) Do you think that the overall themes or subthemes can be changed in anyway?
5) Do you think that our meaning/interpretation of the quotes within the papers can be changed?
6) What could be the implications of these findings for those at risk of developing a chronic illness in the future?

The answers to these questions supported the themes presented by the researchers and highlighted the need to report clear recommendations for practice in this paper. These themes are termed second-order constructs as they are interpreted from the

analysis of first order analysis of the primary data. These constructs were then interpreted and extended in the context of the wider research literature in an attempt to develop a conceptual framework (third order constructs; Box 1).

Box 1: Synthesizing Qualitative Studies (27)

First order constructs: The themes presented in papers identified by the literature search. These constructs are derived from the original authors' interpretation of the primary qualitative data.

Second order constructs: First order constructs from each paper are grouped into core themes to form second order constructs. These are presented in the results section of the present qualitative meta-synthesis.

Third order constructs: Second order constructs are interpreted and extended in the context of the wider research literature in an attempt to develop a conceptual framework and recommendations for practice.

Results

Table 1 provides an overview of the participants in each stud, the methodological strategies and the quality appraisal score for each of the articles included in the meta-synthesis. Of the studies included in this metasynthesis all were conducted in either Europe (n=8) or the USA (n=3). The synthesis of themes is presented in three sections (Box 2). The first section describes five themes which represent patient and public perceptions of predictive testing to quantify risk of disease development. The second section describes four themes that explore recommendations for practice. Relevant quotes for each theme are included with details of the person providing the

quote where available. The final section outlines the priorities of the patient partners derived from the focus group convened to discuss the themes.

Box 2: Themes
1. Perceptions of predictive testing 1.1 The perceived value of testing to establish the risk of disease development 1.2 Perceived barriers to predictive testing 1.3 The use of test information to change behaviour and modify environmental risk 1.4 Communication and understanding of information regarding risk and genetic testing 1.5 Impact of genetic testing and screening on emotional wellbeing
2. Recommendations for practice 2.1 Educating patients and their families about predictive testing 2.2 Considering the most appropriate location for and method of the test 2.3 Improving communication and the delivery of results 2.4 Supporting patients to engage in health behaviours to reduce their risk
3. The priorities of patient partners

1. Perceptions of predictive testing

The five themes related to the perceptions of predictive testing covered the perceived value of testing; the perceived barriers to testing; the use of risk information to change behaviour and modify environmental risk; the communication and understanding of information regarding risk and genetic testing; and the impact of genetic testing and screening on emotional wellbeing.

1.1 The perceived value of testing to establish the risk of disease development

Many ‘at risk’ individuals believed that predictive tests are reliable and effective at quantifying the risk of disease development (28). For diabetes, genetic predictive

testing was thought to be more certain, factual and scientific than either non-genetic risk assessment (e.g. abnormal fasting glucose or obesity) or relying on an individual's knowledge of their family history (28-30).

> *You know it instantly [whether you're at risk], by taking some blood. Do I have a predisposition, yes or no? Brief and effective; it's [DNA test] a good test.* (Female, 53, No family history of diabetes; Page 1474, Netherlands (28))

On the other hand, some participants thought that there was potential for health professionals to give false results (28), while others thought the advice on behaviour change and risk following screening was often contradictory and should therefore not be trusted (31). These participants clearly saw less value in the outcomes of predictive testing.

> *I mean there are all sorts of tosh on television telling you should eat this, you should eat that, and then, you know, in a couple of years time, I mean they were banging on about eggs being bad for you and it all comes to light that you can eat as many eggs as you like … I mean I'm 59 this year and I think all throughout my time there has been this, there has been that and you think to yourself oh God I just want it to go away you know what I mean, leave you to your own devices.* (Male, 59, high CVD risk; Page 4, UK (31))

> *If they want to prove that someone is at risk, they can manipulate the results. I don't know how, since it's quite new, quite precarious.* (Female, 69, grandmother with diabetes; Page 1478, Netherlands(28))

1.2 Perceived barriers to predictive testing

Participants identified a number of barriers to predictive testing. A common concern about testing related to the confidentiality of the information and the potential for results to be used by insurance companies and employers to discriminate against individuals (28,30,32,33). There was a fear that testing may become obligatory and affect access to life or holiday insurance (33). Participants thought that this was particularly unfair as a high risk result from a test does not guarantee that you will develop the disease in question (32). In addition to being concerned about their data being shared with employers and insurers, some patients were concerned about receiving the results of a test via the post in case someone else opened the letter (34).

> *The fact that you have predisposition for it, let's say it's in your genes, it doesn't mean it has to be there so why should you be singled-out before you actually have it…. You know, why should I be turned down beforehand before it actually manifests itself?* (IBD patient, age and gender not recorded; Page 498, USA (32))

Predictive testing was not seen as appropriate or relevant for all people. For example, personal traits such as age, heredity, lifestyle, and physical build were seen to influence the decision to take a genetic test for type 2 diabetes (33). Eastwood and colleagues (35) further reported that people from South Asian populations have a culture of using alternative therapies for common ailments and the prevention of ailments. A GP is only visited when someone is ill and not as a

preventative measure so individuals from such backgrounds may be less likely to access genetic tests. In addition, in a study of South Asians, it was highlighted that family commitments might interfere with GP visits, predictive testing and other health promoting activities.

> *There is a common saying that one should only visit a doctor if one is ill, otherwise doctors will put false doubts in one's mind. That is an understanding of people.* (South Asian female, 35, had CVD risk assessment; Page 469, UK (35))

> *... It's probably the pace of life and the stress and if they've got children as well, that plays a big factor ... so I find that people tend to neglect their own health and lifestyle ... and sometimes I find with the Indian community, I mean they might have a father-in-law or a mother-in-law, and a lot of their time's taken up with that ... so even if they did want to go to a keep-fit class or whatever, they might not be able to do it because of commitments.* (South Asian female, 45, had CVD risk assessment; Page 469, UK (35))

Some people believed that predictive testing should be focussed on certain subgroups, suggesting for example that only people with increased risk, as determined by easily accessible clinical information, or early symptoms should be tested. Certain conditions such as diabetes were not perceived to be 'severe' enough to justify genetic predictive testing (28,36).

I don't think that people will think it concerns them. People will only respond to risk information when they have physical complaints and only then they will think "Now I have to be careful". (Female, 65, mother with diabetes; Page 1475, Netherlands (28))

Genetic testing is [more than for diabetes] for serious diseases like cystic fibrosis, cancer, kidney diseases. Having a family member with one of these diseases can be a reason to have a genetic test. (Female, 69, grandmother with diabetes; Page 1474, Netherlands (28))

Negative perceptions of the test itself, including inconvenience and discomfort were also cited in some of the studies (29,33). For example, the need to fast prior to the test, having blood taken, or attending an early appointment would represent barriers, for some, to the uptake of predictive testing (33). Lengthy appointments and a long wait between tests meant that some participants in the study conducted by Adriaanse and colleagues (29) saw the process as burdensome.

I can't think there's many people, erm, you know, who have got busy lives, who are gonna give up, er, 2 or 3 hours to go and have the test unless they feel ill. (Male, 40–49, did not attend CVD screening; Page 208, UK (33))

1.3 The use of test information to change behaviour and modify environmental risk

Test results were perceived as empowering by several participants as they have the potential to motivate a person to make changes to their lifestyle to reduce their risk of

ill health (32,35,37). A high risk result was seen to add a sense of urgency to engage in healthy behaviours which exceeded the impact of receiving information about familial risk (28,30).

> *The DNA test gives the hardest "push" to live healthier. It will frighten me more than a test based on a family history, because it is the strongest evidence.* (Male, 51, father and 2 brothers with diabetes; Page 1475, Netherlands (28))

Honey and colleagues (31) reported that a high level of commitment to behaviour change was achieved when the patient was shown how their risk score was calculated on a computer screen. By presenting the information in this way, participants could see how their risk would decline if they were to improve their lifestyle.

> *That (risk score) has terrified me...and I will quit smoking. My intentions are to eat healthy, stop smoking altogether.* (Male, 60, high CVD risk; Page 5, UK (31))

When considering how a low risk result is perceived, Markowitz and her colleagues (30) reported that some patients at risk of diabetes would continue to try and reduce their risk of developing the condition regardless of whether the test results were low or high. This suggests that a predictive test might have a positive impact on health behaviour irrespective of the result.

> *Getting a 'low' genetic risk result would make me feel safer, but I would probably still do the exercise and the diet.* (Patient at high phenotypic risk of type 2 Diabetes, age and gender not recorded; Page 571, USA (30))

On the other hand, for some participants, receiving a low risk result would probably result in a reduction in the level of motivation to engage with healthy behaviours (28).

> *I can eat what I want, being fat is no problem, and being physically active, I prefer sitting behind the computer all day.* (Female, 57, no family history of diabetes; Page 1476, Netherlands (28))

An individual's pre-test motivation to lead a healthy lifestyle also influenced how they perceived their level of risk and their motivation to change their behaviour following the test result (30). For example, personal experiences with a chronic inflammatory disease, such as seeing a relative or friend suffer as a result of the condition, meant that an individual was more likely to change their lifestyle (28). Alternatively, if an individual had low motivation to engage in healthy activities prior to taking a (genetic) test, this might not necessarily change as a result of learning their test results; in some cases behavioural change would only follow the development of the relevant disease (31). Others would procrastinate, for example waiting for better weather or until after a special occasion (31).

> *I'm not motivated ... I'm not afraid. I don't think it's at that point where I've got to, you know, "rally the troops and let's get after it".* (Patient at high phenotypic risk of type 2 diabetes, age and gender not recorded; Page 570, USA (30))

Rosedale and colleagues' (34) study raised the issue of participants' choice related to knowing their risk of developing a chronic disease such as diabetes. Some people do not want to know whether they are at risk, preferring not to have to engage with the results of the predictive testing and make lifestyle changes. For example, several people did not see the test results as a prompt to lower their cardiovascular risk because they believed death from a heart attack would be preferable to dying from a protracted illness or living into extreme old age (31). This group also resisted change because they believed that a short life of indulgence was better than a long life of denial. Those participants who believed that health outcomes are already predetermined or a matter of luck, were also less motivated to change (30,31).

> *I know I am naughty because I quite like cream, you know, and things like that but I think gosh I am not going to be long on this mortal coil, I am not going to make myself miserable to the point of being really ultra miserable to maybe extend my lifespan by one or two years.* (Male, 73, high CVD risk; Page 5, UK (31))

> *You can be as careful as you want; you can eat as healthily as you want; you can do all the exercise you want and you could still get ill. It is like J's mother who lived to be 101, smoked like a trooper, never had a cigarette out of her hand and she died of something silly. But you see it's just jovial isn't it? That is why I say it is just fate. You can do all the right things and still pick things up.* (Male, 74, high CVD risk; Page 6, UK (31))

1.4 Communication and understanding of information regarding risk and genetic testing

Some participants were unclear about the roles of environmental and hereditary factors in disease development and the integration of data relating to these variables into predictive testing strategies (30). For example, Honey and colleagues (31) stated that one of their participants was sceptical about the accuracy of test results as they did not understand how the probabilities of disease were calculated.

> *I don't know whether bringing this data together into one score is a very realistic thing to do…there might be methodological problems...I don't think you can add things like that together.* (Male, 66, high CVD risk; Page 5, UK (31))

Participants invited to take part in actual screening programmes were not always aware of the reasons for their screening invitation and as a result some chose not to attend (30).

> *I don't know [why I was invited]. I thought diabetes had something to do with sugar, too, and . . . I'm not a sugar person.* (Patient at high phenotypic risk of type 2 diabetes, age and gender not recorded; Page 570, USA (30))

Understanding of the actual test results also had an impact on health behaviour. Honey and colleagues (31) found that poor communication within the consultation can be a problem, as some high risk patients did not remember receiving a detailed

explanation about their risk score or what it meant. Participants also reported confusion if they did not have any relatives with the condition, or knew little about the condition they were being tested for (29,32). Other people found positive test result difficult to conceptualise if they had no current symptoms (36). Without a real understanding of the test results, participants felt they were unable to make meaningful changes to their lifestyle.

> *I have this piece of knowledge; it makes little or no difference to physically how I am. So how helpful is it, it's not helpful is it in a way...* (Female, age not recorded, HCM gene positive (CVD); Page 90, UK (36))

1.5 Impact of genetic testing and screening on emotional wellbeing

Most participants felt that testing for whether they were at risk of disease would not have a significant impact on their emotional wellbeing (26). For example, Adriaanse and colleagues (29) found that none of their participants were alarmed by their risk of type 2 diabetes as they believed that this was a manageable condition. Similarly, Wijdenes-Pijl and colleagues (28) found that genetic testing for diabetes caused very little or no psychological harm. Emotional impact was lowest when participants knew someone that was managing the condition well. Personal experience of a condition therefore influences how individuals respond to the results of a predictive test (9;37).

> *If you've got diabetes and you, you listen to what people tell you, you can control that … I'm aware that diabetes in, in our current age, is quite treatable, hopefully, erm, so therefore, you know, even if I was diagnosed with diabetes,*

> *there's ways and means that I could continue to live a normal life.* (Male, 40–49, did not attend CVD screening; Page 207, UK (33))

> *Familial risk information will not necessarily lead to worry [about disease risk], but it can raise awareness about the risk.* (Female, 64, mother and brother with diabetes; Page 1476, Netherlands (28))

Some, participants also described how they believed that genetic predictive testing can in fact reduce anxiety (9). Participants who believed they were at a high risk due to hereditary factors describe a substantial relief and a feeling of safety when they received a low risk result (30). Many of those who perceived themselves to be at a low risk chose to take the test with the expectation of getting a 'negative' result so they did not have to worry about getting the condition. Genetic testing was therefore seen as a way to put their mind at rest (36).

> *I'd want to know whatever. You're better knowing and then you can plan, or at least get it straight in your mind, and then once it comes along you're ready for it, instead of really worrying. I mean it's always worse not knowing.* (Middle aged white male with diabetes, father of a child at risk of diabetes; Page 249, UK (9))

However, Ormondroyd and colleagues (36) have warned that health professionals need to manage the expectations of those undergoing testing, as some people in their study who had thought they were low risk were shocked by how they felt when they received a high risk result. Others reported that a positive result led to stress

among patients and family members, especially if the person was otherwise asymptomatic (32).

> *Really and truly, I was absolutely horrified when it came back and said that I had it. I mean I really was horrified... It was a dreadful shock* (Female, age not recorded, HCM gene positive (CVD); Page 90, UK (36))

Some participants were not comfortable with the uncertainty associated with predictive test results for multifactorial diseases, and the potential for unnecessary anxiety and behavioural change (35,36).

> *You see, as far as I'm concerned, it's actually become a bit of a nuisance really, unless they, until they can tighten it down to a specific thing. I didn't realise how wide, just because you have it doesn't mean you're going to get it and that's the problem isn't it?* (Female, age not recorded, HCM gene positive (CVD); Page 90, UK (36))

A stepwise approach, for example where participants visit their health professional three times to complete a genetic test for diabetes, could be effective at reducing anxiety for those undergoing predictive testing. Participants in the study conducted by Eborall and colleagues (37) perceived initial tests to be routine and rarely expected to receive a high risk result. Having three tests also allowed the participants to prepare for the results and build a rapport with the health professionals.

So I go for number one, I go for number two and then I have to go to number three. So it's a build up all the time, making me think, well OK there's a possibility you know . . . there's a strong possibility you know' in that sense [...] you've gone through the three, so your brain's adjusted anyway. (Female, 64, impaired glucose tolerance (predictive testing for diabetes); Page 3, UK (37))

When considering the impact of testing on personal relationships, a number of positive outcomes were highlighted. For example, Wijdenes-Pijl and colleagues (28) found that participants felt that a high risk result acted as a facilitator to prompt discussions within the family, encouraging the development of a support network for those who are affected. Families also benefit when a low risk result was given as this helped to reduce anxiety within a family about the participant's health. The predominant motivation for coming forward for pre-symptomatic testing was the 'need to know' (36).

I think that when you're aware of diabetes running in your family, it can help to talk about it with each other. I notice that the relationships in our family are not distorted. We talk about it [diabetes in the family] with each other. Not that we get anxious about it, but more to be supportive for other family members. It's no longer a taboo. (Female, 47, no family history of diabetes; Page 1477, Netherlands (28))

Well I suppose it's better knowing if you've got something, if it's treatable...it was a sort of joint decision really I said yes I'd go but mum and dad said

you're going anyway, so I didn't get much choice in it which is fair enough. (Male, age not recorded, HCM gene positive (CVD); Page 90, UK (36))

However, the potential for a negative impact on family relationships was also highlighted. Some participants were concerned that a positive test result may cause people to worry about the health of their children (28). Sharing results was also an issue if other family members did not want to know that they may be at risk. Finally there was the concern that someone with a high risk of developing a chronic disease may be blamed for the potential risk for other family members (28).

My relatives will get anxious [if our family history is assessed]. [...] There are some who would rather not know that. So, I think that it can cause anxiety for some people. (Female, 57, no family history of diabetes; Page 1477, Netherlands (28))

2. Recommendations for practice

Four themes were identified that related to recommendations for practice. These were: educating patients and their families about predictive testing; considering the most appropriate location for and method of the test; improving communication and delivery of results; and lastly supporting patients to engage in healthy behaviours to reduce their risk.

2.1 Educating patients and their families about predictive testing

Many members of the public lack the knowledge and understanding of (genetic) predictive testing that is necessary to be able to make an informed decision about

whether to take part in such testing and engage with test results (9,28,34,36). It is therefore important for individuals to receive intelligible pre-test information on the condition they are being tested for (34). The information should cover the risks and benefits of the test, technical aspects of testing, the implications of their results for themselves and family members, treatment options, referral information, post-test counselling and support groups (32,34). Individuals should also be made aware of the risk factors that led to their screening invitation in the first place (37). For example, when this information was provided before a genetic test for IBD (32), participants reported a reduction in fear and anxiety about testing and an increase in understanding of the test results.

> *I thought I'd be more upset about it, but I didn't seem upset. I think because we've spoken so much to everyone about it that we knew that's what we wanted to do.* (Mother of child at risk of diabetes; Page 254, UK (9))

Health professionals should also focus on motivation for testing and how the participant perceives the condition and the testing process (9). This discussion will also highlight any misconceptions about genetics and inheritance which should be addressed as well as increasing confidence in the results of genetic tests (9) and other forms of predictive testing.

2.2 Considering the most appropriate location for and method of the test

Eastwood and colleagues (35) report that certain patient groups are more likely to engage in testing if it is conducted outside of primary or secondary care. For example, South Asian populations were found to benefit from CVD screening in

religious and community settings rather than at their GP practice. These locations offered increased accessibility and the community reported a positive perception of the test if the health professionals were organised in their approach and gained the support of a respected community member (35).

> *We're screening between sixty to eighty people [in a single session] and if we're not organised it's difficult. So the first thing we did was identify someone in the temple who will take that responsibility. We will be seen as outsiders going in and imposing restrictions. So we have Dr X, who is a good organiser from the temple who is seen as a senior in the temple... along with the clinical lead from the temple for this project who* is *Dr Y.* (Clinical staff involved in CVD risk assessment, age and gender not recorded; Page 469, UK (35))

An American study by Rosedale and colleagues (34) found that diabetes screening in a dental setting encouraged patients, who would normally not have considered testing, to take part. Collecting oral blood during a routine dental examination was preferred to drawing blood from the finger as would be carried out in more traditional testing. The researchers observed that:

> *The screening approach wasn't intimidating to the patient and actually was very pleasant... according to most of the patients. Most of the patients were used to sitting in the dental chair and getting a cleaning, so the collection of oral blood wasn't a problem. Many patients stated how much they hated finger sticks because of the pain and this made potential subjects hesitant to participate.* (Page 285, USA (34))

Some participants further remarked that it was easier to talk with staff at the dental practice, rather than other medical professionals. Education about diabetic risk provided in the dental office was therefore invaluable. Participants further reported that the location was convenient, would encourage more regular testing and that they would be happy to be tested for other conditions.

> *Today, I have time and am relaxed. I go to the doctor maybe once a year for a routine check but why wait?* (Participant diabetes screening at dentist, age and gender not recorded; Page 284, USA (34))

2.3 Improving communication and the delivery of results

The manner in which information about their screening result is conveyed to participants is key to their understanding of the results and their implications (28,31). Honey and colleagues (31) found that for most participants it was acceptable to be informed about their cardiovascular risk status in person or by telephone, whereas receiving test results in a letter caused confusion for some participants as they were unable to ask questions.

> *I got a letter from the doctor's saying "as you are at a high risk of a stroke or heart attack"...well I nearly died, and I thought 'well what have my results come up as?* (Male, 66, High CVD risk; Page 3, UK (31))

Honey and colleagues (31) argue that the communication style of health professionals can also play a role in the level of motivation for behaviour change.

Participants were less motivated when the health care professional was seen to 'downplay' a high risk score by using phrases such as, 'it is only slightly higher', or make light of the risk by using humour. It is therefore important that the level of risk is accurately communicated to the patient. The person who delivers a test result also appears to have an impact on the person's understanding of these results and their level of motivation for behaviour change (31). Lewis and colleagues (32) reported that several respondents indicated that they would prefer to receive pre and post-test information from a specialist rather than their GP.

> *They [specialists] are probably more in tune to different markers, symptoms than maybe just a regular primary care physician would be.* (Unaffected first-degree relative of IBD patient, age and gender not recorded; Page 498, USA (32))

> *I think if the results are positive that they did have the disease, they are more susceptible to it, then the GI specialist, of course, should give the results.* (IBD patient, age and gender not recorded; Page 498, USA (32))

Some members of the public also believed that they should not have the responsibility of telling family members about possible risk (36). They felt that this should be done by health professionals who could also explain the risk and options for genetic testing.

> *I think it isn't perhaps something that should be left to the family to communicate because we all have our own perceptions as to whether X or Y*

is healthy or needs to have a test. How on earth do we know, when we are not qualified to make that decision. (Female, age not recorded, HCM gene positive (CVD); Page 91, UK (36))

2.4 Supporting patients to engage in health behaviours to reduce their risk

A study by Markowitz and colleagues (30) suggested that testing for type 2 diabetes should be individualised and based on each patient's risk perception and current level of motivation to engage in health behaviours to prevent diabetes. By considering current levels of motivation, the health professional can work with the patient to promote or maintain lifestyle changes when faced with a low or high risk result. It is also important for the health professional to consider how they can convey enough information about the potential consequences of the disease to justify lifestyle change, without raising anxiety to such an extent that it causes disengagement (28).To optimise motivation, the decision to undertake a predictive test should be linked with the opportunity to enrol in a lifestyle intervention programme to provide structure, education and support to facilitate sustainable lifestyle changes (30).

3. The priorities of patient partners

When considering the literature, patient partners believed that the greatest barrier to genetic testing for chronic inflammatory diseases was a current lack of knowledge about the need for testing (9,28,34). Patient partners agreed that a staged approach was required, starting before the test was administered with clear, succinct and easy to read information about the condition, and the reasons for testing (31,37). The focus group highlighted the need for health professionals to take the time to explain

the meaning of risk information, clearly stating what percentage or figure represents a ‘high risk’. A written summary of the results for the patient to take away also aids understanding and communication with family members (31).

An additional consultation to discuss possible lifestyle intervention programmes and the impact of these results on family members was also seen as good practice (30). The patient partners suggested that this session could be run by a psychologist who had skills in motivating behaviour change. This follow up care was seen as a priority in order to manage the emotional impact of the result.

Discussion

The current meta-synthesis of the qualitative literature highlights that predictive testing can be viewed by those at risk of developing chronic inflammatory disease as a reliable way to ease concerns about the risk of developing a condition, or gain valuable information to motivate behaviour change (28). However, a number of papers discuss a lack of understanding about the type of information that a predictive test can provide, what that information means in terms of level of risk and how to reduce that risk (28,31,37). These perceptions may have a negative impact on how predictive testing for chronic inflammatory disease is viewed and utilised by those at risk.

In order to reduce the barriers to the uptake to predictive testing, the literature suggests that a patient centred approach is required at each stage of the process. This can be achieved in the following ways. Firstly, there is a need for readily

available and easy to understand pre-test information (34). This should include information on genetics and inheritance, and address current concerns that the results of predictive tests are inaccurate, lack meaning or are not confidential (28,30,32,33). Further research is required to work with patients and members of the public to design an informational resource that describes technical aspects of testing, what a predictive test can tell you and how the results will be used. This will support the individual to make an informed decision about whether to engage in a predictive test (9,28,34,36). Information is also required about the condition the individual is being tested for, and the risks and benefits of a predictive test for that condition. For example, Wijdenes-Pijl et al. (28) found that people with first-hand experience of a condition are more likely to engage with predictive testing; therefore a resource such as a short film may increase understanding of the potential benefits of a predictive test and early intervention.

Perceptions of predictive testing become more positive when health professionals tailor their approach to specific groups (37). A number of variables were found to impact on an individual's decision to engage in genetic testing including age, ethnicity, current lifestyle, individual differences in the level of motivation for change and level of anxiety (33). Accessibility can be improved by taking predictive tests into the community (35), while a stepwise approach, where participants visit their health professional a number of times, has also been found to be effective at reducing anxiety (37). Further research is needed to explore the needs of different groups to understand the potential barriers to accessing predictive tests.

Once an individual has engaged with a predictive test, the health professional can work to maximise the impact of the test results by discussing what the level of risk means (31). Health professionals can also offer support to patients on how to deal with the implications of their results for themselves and their family. This may include developing a resource that provides further information on referrals, post-test counselling and support groups (32,34).

Finally, when considering the long-term impact of a predictive test, it is important to address questions about if and how risk would decline if an individual were to improve their lifestyle (31). Where appropriate the development of condition specific lifestyle intervention programmes would provide structure, education and support to facilitate sustainable lifestyle changes (30). By considering current levels of motivation, the health professional can also work with the patient to promote or maintain lifestyle changes when faced with a low or high risk result. Once again, further research is required to explore the potential barriers to implementing change to manage risk among different groups.

Comparisons with existing literature

Qualitative studies on the perceptions of predictive testing have been undertaken in the field of cancer (17). Similarities between this condition and chronic inflammatory diseases include the need for health professionals to facilitate family communication when an individual is faced with a high risk result (16,17, 28). Tensions can arise when informing family members about their practical risk, and there is little guidance on how this information should be shared (16). Heshka and colleagues (17) also found that genetic testing had little effect on behaviour, and did not change

perceived risk of cancer. This highlights the importance of the development of pre and post-test patient education strategies as recommended in the current meta-synthesis**.** Our paper also identified recommendations for practice that have not been presented in the cancer literature. These include the idea that patient groups are more likely to engage in testing if it is conducted outside of primary or secondary care, and the promotion of a lifestyle intervention programme to motivate patients to change their behaviour and reduce risk. However, there are limitations when drawing comparisons between the chronic inflammatory diseases reviewed here and the findings of cancer studies as those at risk of cancer may have greater knowledge and more complete prototypical beliefs. Furthermore perceptions of severity are usually more severe in the context of cancer (18,19,28).

Limitations

The aim of this meta-synthesis was to systematically describe the perceptions of genetic testing for chronic diseases of inflammatory origin in the general public and make recommendations for the development of future predictive tests. The review procedure only found studies from developed countries (UK, USA and the Netherlands) that explored genetic testing for diabetes, cardiovascular disease and inflammatory bowel disease, and our findings may not be generalizable to other cultures.

It is interesting to note that our search strategy failed to identify any studies that explored the views of predictive testing among patients with a range of common chronic inflammatory diseases in particular the chronic inflammatory rheumatic disease (RA, ankylosing spondylitis, SLE and Sjogren's syndrome), psoriasis and

MS, despite ongoing work to quantify disease risk in individuals from high risk populations (38). The patient research partners highlighted that RA has a number of unique features when compared to the diseases which the studies captured in this meta-synthesis focussed on. For example, deformity and chronic pain are not typically associated with diabetes and coronary heart disease. Furthermore an evidence base does not yet exist for the primary or secondary prevention of RA even if an individual is identified as being at high risk (although a number of intervention studies are ongoing in this field). Such disease specific features may influence an individual's decision as to whether or not to undergo a predictive test. Further research is needed to explore variations in perceptions of predictive testing across different disease contexts in order to develop to recommendations for practice that are specific to each condition. This meta-synthesis has also grouped different predictive tests together. Future work should compare the views of at risk subjects on different types of tests.

The authors focused on peer reviewed, published papers only. The grey literature was therefore not included. It is possible that there are other relevant studies that were not identified by our search strategy.

Competing interests

The author(s) declare that they have no competing interests.

Authors' contributions

KB contributed to the research design, completed the literature search, read and analysed each of the articles, completed the quality appraisal and drafted the paper. KR GS, MF, MH and BS contributed to the research design and the writing of the paper. RS designed and managed this meta-synthesis. She contributed to the research design, literature search, quality appraisal, analysis and writing of the paper. GS contributed to the writing of the paper. All authors read and approved the final manuscript.

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Table 1. Characteristics of papers included in the meta-synthesis

Paper (Diabetes)	**Participants**	**Country**	**Primary objectives**	**Methods and analysis**	**Type of test used**	**Score**
Adriaanse et al., 2002 (29)	20 type 2 diabetes patients (10 male, 10 female). 20 persons at increased risk of diabetes (10 male, 10 female).	NL	Explore the psychological impact of a stepwise population-screening project for Type 2 diabetes	**Methods:** semi structured interviews **Analysis:** content analysis	Symptom Risk Questionnaire Various glucose screening tests	2-1-1
Eborall et al., 2012 (33)	13 persons without a diabetes diagnosis who attended a diabetes screening 11 persons without a diabetes diagnosis who did not attend the	UK	Explore the perceptions of those invited to attend the MY-WAIST screening study for type 2 diabetes (i.e. explanations for (non) attendance; views on the specific screening	**Methods:** semi-structured interviews **Analysis:** constant comparative method	Oral glucose tolerance screening test	2-2-2

	screening (gender breakdown not provided).		strategy).			
Eborall et al., 2014 (37)	23 persons without diagnosis of diabetes attending type 2 diabetes screening process (14 male, 9 female).	UK	1. Provide insight into factors that contribute might contribute to anxiety as a result of screening 2. To explore expectations of and reactions to the screening experience of patients with positive, negative, and intermediate results.	**Methods:** prospective qualitative interview **Analysis:** Thematic analysis.	Various glucose screening tests	2-2-2
Markowitz et al., 2011 (30)	22 overweight participants at high phenotypic risk for type 2 diabetes (13 male, 9	USA	1. Explore perceptions of diabetes genetic risk testing compared to risk testing using non-genetic	**Methods:** semi-structured interviews using hypothetical	Genetic test & non-genetic risk factors	2-2-2

	female).		risk factors 2. To explore the impact of test results on motivation for behaviour change.	scenarios. **Analysis:** thematic analysis		
Rosedale et al., 2012 (34)	9 diabetes patients (gender breakdown not provided).	USA	Explore patient experiences of diabetes screening during periodontal visit (e.g. thoughts about combination with dental visit; preferences regarding FSB glucose testing compared with GCB glucose testing)	**Methods:** semi structured Interviews **Analysis:** constant comparative analysis	Blood glucose test	2-2-2
Shepherd et al., 2000 (9)	4 family members of young person at risk of	UK	To explore the perceptions genetic testing of a family	**Methods:** Open-ended	Genetic test	2-1-1

	MODY (1 male, 3 female). 6 health specialists (consultant, geneticist, genetics nurse specialist, diabetologist, paediatrician, paediatric specialist nurse. Gender breakdown not provided).		affected by MODY and the healthcare professionals involved	qualitative interviews prior and post genetic test **Analysis:** Content analysis		
Wijdenes-Pijl et al., 2011 (28)	27 persons with a family history of diabetes 18 persons without a family history (11 male, 34 female).	NL	Explore perceptions of predictive testing based on DNA test results and family history assessment.	**Methods:** Focus groups **Analysis:** thematic analysis	Genetic test and family history assessment	2-2-2
Paper (CVD)	**Participants**	**Country**	**Primary objectives**	**Methods and analysis**	**Type of test used**	**Score**

Eastwood et al., 2013 (35)	12 persons at risk of CVD (6 male, 6 female). 12 health professionals involved in risk assessments (9 male, 3 female).	UK	Explore the feasibility and potential impact of CVD risk assessment targeting South Asian groups	**Methods:** semi-structured interviews **Analysis:** thematic framework analysis	NHS Health Check (anthropometrics, blood pressure, cholesterol and glucose testing, lifestyle assessment, tailored advice)	2-2-1
Honey et al., 2014 (31)	37 patients at a high 10-year risk of a cardiovascular event (30 male, 7 female).	UK	Explore the perceptions of patients identified as being at 'high risk' of cardiovascular events, with particular reference to the potential responses to risk messages.	**Methods:** semi structured interview **Analysis:** Thematic analysis	NHS health check (calculates individual risk by physical assessment and questions about medical history, family history and lifestyle)	2-2-2
Ormondroyd	22 persons eligible for	UK	Explore perceptions of the	**Methods:** Semi	Genetic test	2-2-2

et al., 2014 (36)	pre-symptomatic genetic testing for Hypertrophic Cardiomyopathy or Long QT syndrome (9 male, 13 female).		cascade testing process, impact of pre-symptomatic genetic testing and attitudes towards direct contact as an alternative to family-mediated dissemination for inherited cardiac conditions.	structured interviews. **Analysis:** thematic analysis		
Paper (IBD)	**Participants**	**Country**	**Primary objectives**	**Methods and analysis**	**Type of test used**	**Score**
Lewis et al., 2009 (32)	30 IBD patients (14 male, 16 female). 18 unaffected first-degree family members /spouses (6 male, 12 female).	USA	Explore the perceptions, preferences, knowledge and needs surrounding genetic testing for IBD	**Methods:** focus groups **Analysis:** thematic analysis	Genetic test	2-2-2

Please note: **Score** refers to the quality appraisal of the papers carried out prior to the meta-synthesis