

# 'A morass of considerations': exploring attitudes towards ethnicity-based haemoglobinopathy-carrier screening in primary care

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**Background.** The Netherlands does not have a national haemoglobinopathy (HbP)-carrier screening programme aimed at facilitating informed reproductive choice. HbP-carrier testing for those at risk is at best offered on the basis of anaemia. Registration of ethnicity has proved controversial and may complicate the introduction of a screening programme if based on ethnicity. However, other factors may also play a role.

**Objective.** To explore perceived barriers and attitudes among GPs and midwives regarding the registration of ethnicity and ethnicity-based HbP-carrier screening.

**Methods.** Six focus groups in Dutch primary care, with a total of 37 GPs ( $n = 9$ ) and midwives ( $n = 28$ ) were conducted, transcribed and content analysed using Atlas-ti.

**Results.** Both GPs and midwives struggled with correctly identifying ethnicities at risk for HbP. Ethical concerns regarding privacy seemed to originate from World War II experiences, when ethnic and religious registration facilitated deportation of Jewish citizens, coupled with the political climate at the time focus groups were held. Some respondents thought the ethnicity question might undermine the relationship with their clients. Software programmes prevented GPs from registering ethnicity of patients at risk. Financial implications for patients were also a concern. Despite this, respondents seemed positive about screening and were familiar with identifying ethnicity and used this for individual patient care.

**Conclusions.** Although health professionals are generally positive about screening, ethical, financial and practical issues surrounding ethnicity-based HbP-carrier screening need to be clarified before introducing such a programme. Primary care professionals can be targeted through professional organizations but they need national policy support.

**Keywords.** Barriers, carrier screening, ethnicity, haemoglobinopathies, primary care.

## Introduction

Haemoglobinopathies (HbPs), such as sickle cell disease and thalassaemia, are autosomal recessive disorders with severe anaemia, variable but lifelong morbidity and a shortened lifespan due to multi-organ ischaemic damage.<sup>1,2</sup> HbPs occur more frequently in areas such as Africa, the Mediterranean area, the Middle East and South-East Asia. Due to migration and population admixture, HbPs are now also common in other countries. Carrier prevalence amongst the general population in Northern and Western Europe

varies between 0.29% in Finland and 2.45% in France.<sup>3</sup> However, when ethnicity is taken into account, carrier prevalence may vary up to 40%,<sup>4</sup> making ethnicity an important determinant of risk. Carrier couples have a one-in-four chance in each pregnancy of giving birth to an affected child. Carrier screening would allow the identification of these couples and give them an opportunity to make informed reproductive decisions, preferably before conception. The Netherlands, as many other countries in Europe, does not have a national HbP-carrier screening programme. Although national guidelines advise HbP-carrier testing in cases

of (unresolved) anaemia or a positive family history,<sup>5,6</sup> health professionals such as GPs and midwives rarely carry this out. Being part of primary care, midwives and GPs are in a good position to provide easy access to preconception care. The vast majority of pregnant women (78%) start their maternity care in a primary care setting.<sup>7</sup> Preconception or early antenatal HbP-carrier testing would facilitate reproductive choice for (prospective) parents. Although preconception care has been implemented by midwives and is also gaining attention from GPs in the Netherlands, it is hindered by a lack of financial support.<sup>5,8</sup> Preconception care is therefore only available to those with sufficient financial means or an extensive insurance policy.

Several studies have shown that both health professionals and groups at risk support HbP-carrier screening.<sup>9-12</sup> One of the explanations for the rarity of testing may be our earlier finding in a questionnaire survey<sup>11</sup> that HbP testing is influenced by peer behaviour. As official policy is lacking and colleagues rarely offer testing,<sup>10</sup> peer influence to test for carrier state among those at risk is presumably low. As (targeted) screening has not been implemented and recessive disorders such as HbPs are often not apparent in families, carriers of HbPs remain largely unidentified.

There are several arguments for implementation of targeted screening based on ethnicity, including economic ones.<sup>13</sup> HbP-carrier status can easily and cheaply be determined by a high-performance liquid chromatography (HPLC) test. But 'before' testing is offered, couples preferably need to be accurately informed about their *a priori* risk so that health care professionals can facilitate the choice for testing. Since HbP (carrier) prevalence differs across ethnic groups, the identification of ethnicity might still be necessary. Besides, identification of ethnicity might be needed to further investigate mutations. In England, where antenatal HbP-carrier screening was introduced in 2004, self-identified ethnicity based on a family origin questionnaire is determined to assess HbP risk in both low-prevalence areas where targeted screening is practised and in high-prevalence areas where universal screening is the norm.<sup>14</sup>

Research by Dyson *et al.* showed that health care workers struggle with the ethnicity question as a screening tool and worry about the sensitivities related to ethnicity.<sup>15</sup> In the Netherlands, ethnic registration has proved to be a controversial issue rooted in the history of World War II (WWII) and it was linked to the nationalistic political climate at the time of our study (2010).<sup>16</sup> These issues came up as barriers for the introduction of HbP-carrier screening.<sup>11,17</sup> In order to offer equitable health services to all groups in society, health professionals need to be aware of ethnicity-related health needs without the fear of raising issues of discrimination or stigmatization.

### Objective

This study's objective was to gain more insight into the present attitudes towards the documentation of ethnicity in clinical records by GPs and midwives related to everyday practice, in particular to identify risk groups for preconceptional or antenatal HbP-carrier testing and to explore perceived barriers if ethnicity-based HbP-carrier screening is implemented in the future.

## Methods

### Design and setting

This qualitative study is part of a broader study examining ethnicity-based HbP screening.<sup>11,17</sup> As both 'ethnicity' and 'ancestry' are shown to be equally complex terms and ethnicity is supported by the literature as a proxy for ancestral or ethnic origin,<sup>18</sup> this term has been chosen to be used in the study. Moreover, this term is most common in areas of health care such as midwifery and obstetrics.<sup>7</sup>

A focus group study was designed as this method stimulates debate and explores ideas about the topics introduced. Moreover, we aimed to deepen the issues of interest as identified by a previous questionnaire study.<sup>11</sup>

Focus groups were homogeneous with regard to health profession in order to generate a sufficient feeling of security among participants. The semi-structured topic guide was based on the literature and the results of the aforementioned questionnaire study. The questionnaire study showed that almost half of respondents are in favour of HbP-carrier testing on the basis of ethnicity and are of the opinion that a national screening policy should be implemented. Respondents reported that they very rarely carry out HbP-carrier testing at present. Topics in the focus group discussion protocol included participants' experience with HbP-carrier testing (also related to neonatal screening for sickle cell disease, which was implemented in the Netherlands in 2007,<sup>19</sup> opinions on a possible HbP-screening programme, attitudes regarding ethnic registration in clinical records also related to HbP screening, perceived barriers and possible solutions for the potential implementation of a HbP-carrier screening programme. Prior to the start of the focus group, participants received a brief HbP fact sheet by post based on existing professional guidelines with which participants should already be familiar,<sup>6,20</sup> giving disease background and information on groups at risk. Participants were asked to complete basic background questions and provide written consent.

The focus group discussions took place at two universities and a midwifery academy; they lasted ~90 minutes and were facilitated by one of two moderators (LvT, a health scientist, or CvE, a sociologist). The primary researcher (SJ, midwife) was only involved in the GP group as note taker to prevent influencing the

discussion of her colleagues. Another assistant took notes in the midwives group. Midwives received professional register credits in return for participation, whereas GPs did not, as this was not facilitated by their professional organization. All participants received a €10 gift voucher but were not informed of this prior to the discussion.

#### *Participant recruitment and data collection*

Primary care practitioners were asked to participate by means of a letter included in the postal questionnaire (sent to all 1800 primary care midwives and a random sample of 2000 GPs) on ethnicity-based HbP screening.<sup>11</sup> To this letter, 29 GPs and 191 midwives responded. We selected a regionally based purposeful sample from among the responders, on the assumption that rural health professionals would have less experience with groups at high risk for HbP. Our sample included six focus groups of about six midwives or GPs each, with a total of 37 individuals aged 23–65 years. In the autumn of 2010, four focus groups were held with midwives ( $n = 28$ ) and two with GPs ( $n = 9$ ), after which saturation was achieved concerning the midwives. Digital audio recordings of the discussions were collected and anonymously transcribed.

Several attempts were made to recruit more GPs but these proved unsuccessful. Although most midwives were women, the mostly male GPs had more working years' experience compared with the midwives. Even so, midwives were more familiar with preconception care and looked after a larger percentage of the population at risk for HbPs (Table 1).

#### *Analysis*

The transcripts were checked once to ensure accuracy and to gain familiarity with the data and these were subsequently content analysed (SJ). Transcripts were searched for common themes but also for deviant cases and subsequently indexed as codes. The coding frame was developed by the primary researcher (SJ), in consultation with two other researchers (LH, health scientist, and CvE). SJ coded the transcripts and CvE and LH each verified one-third of the transcripts as

an inter-rater check. The codes were grouped together into key themes. Codes and emerging themes were compared for coding reliability through a process of discussion and deliberation of themes and connections (SJ, LH, and CvE). ATLAS.ti software package (version 5.2.0) was used to support the analysis of the transcripts.

## Results

General attitudes towards HbP-carrier screening were mixed. Although practitioners thought it to be good practice and that it would fulfil a health need, they also questioned the cost-effectiveness and necessity because of perceived low prevalence. Data analysis identified four themes related to ethnicity-based carrier screening: defining ethnicity; ethical dilemmas and sensitivities; ethnic awareness and good practice; and practicalities. All these themes are discussed and illustrated here by representative quotations from the discussions, translated from Dutch. The participant's group and their occupation type (GP or midwife) are given in brackets.

#### *Defining ethnicity: who is at higher risk?*

The issue that generated most discussion in all focus groups was the concern of being able to correctly identify those individuals at higher risk for HbPs. Participants wondered how ethnicities can be defined and thus how they could correctly identify those at risk: by appearance, by name, or by country of birth?

So they might look Hindustani [Surinam people of Indian origin] but there might be some black or something, (. . .) but how strong . . . wouldn't you need to test them?

Midwife (Group 6)

But no, I believe all those Mediterranean people are Caucasian too and that is clearly a different ethnicity. Because I [this GP is of Dutch ethnic origin] don't have a higher risk of HbP and they do. So yes, the way you define ethnicity puts you immediately in a morass of difficult considerations.

GP (Group 2)

Participants wondered how far back they would have to look into a person's ethnic origin to determine their risk of being a HbP carrier.

How strong is HbP in the inheritance? (. . .) How much white; lets say how much percent of something else do you need to take away the effect? This is what I don't know (. . .). Percentages will drop the more mixing there is. But it's still only a chance and you have a chance of one in two (*sic*) that it will be passed on to you and that will always remain if your parents have it.

Midwife (Group 6)

TABLE 1 Characteristics of participants in the focus groups

Characteristics	Midwives ( $n = 28$ )	GPs ( $n = 9$ )
Mean age, years (SD)	35.4 (11.3)	51.4 (11.2)
Female gender, $n$ (%)	27 (96)	2 (22)
Dutch, $n$ (%)	27 (96)	9 (100)
Mean practice experience, years (SD)	10.4 (9.2)	22.0 (11.7)
Estimated population at risk in practice, %	27	20
Urban practice, $n$ (%)	18 (64)	4 (44)
Gives preconception care, $n$ (%)	14 (50)	4 (44)

GP, general practitioner.

Both GPs and midwives also struggled with mixed backgrounds of their patients and clients, which made them feel unsure how to handle this in daily practice and uncertain whom to offer screening:

But what do you do with someone who has an Indonesian mother and an Italian father and she is married to a Dutch man, you know, these people can also have carrier (. . .) The fact that people are so intertwined, so often, well that is, we see a lot of people with mixed backgrounds and what is the limit [cut-off point], with which background?

Midwife (Group 3)

Participants seemed aware that determining ethnic origin with regard to HbP-carrier screening entails more than identifying someone's place or country of birth.

I mean, they are from Moroccan descent that's what it's all about of course, the ethnicity on the basis of genetics and not let's say . . . the ancestors, yes, so the country of birth of their parents counts, but let's say as a person I don't really have someone in front of me with a lot of Moroccan influences, her Dutch influences are much bigger.

Midwife (Group 5)

#### *Ethical dilemmas and sensitivities*

Both midwives and GPs had several, sometimes quite strong, ethical opinions concerning ethnic documentation in clinical records. Specifically, GPs felt burdened by ethical objections that stem from Dutch experiences during WWII, when ethnic and religious registration facilitated mass deportation of Jewish citizens.

But if you think a bit further, at the time of Hitler and the Jewish people et cetera; they carried out all sorts of scary experiments with different ethnic populations, that was on the basis of you know . . . Jews and Gypsies and I don't know who . . . And if we would register all of that and it would be registered somewhere that you are Caucasian; Muslim; African or whatever else: Go ahead think about it, in times of, people, well in the craziness of war strange things can happen . . .

Midwife (Group 5)

One GP said that he would not object to the documentation of ethnicity in clinical records and that it could potentially be useful, but despite this, he still felt distrust and maintains his recordkeeping to a minimum:

I register as little as possible. And well, I wouldn't object [to ethnic registration] if it was possible to do this correctly and in a way that's reproducible. I know we have a huge trauma in the Netherlands caused by World War II when Jewish people were registered with the council and as Dutch citizens we

diligently helped the Germans to deport them and we pretend to be the best country in the world. (. . .)

GP (Group 1)

Participants coupled this with the political climate (right wing and nationalist) at the time the focus groups were conducted and worried about the misuse of data that could threaten patient confidentiality. GPs especially felt very protective of their patients' privacy and had little confidence in digital record keeping:

I have no trust whatsoever in that it [electronic patient record] won't be used for anything but patient purposes. The safety and the trustworthiness of this still have to be verified and demonstrated in my opinion. I believe records can easily be hacked at the moment. Besides, I don't know what governmental authorities will do with this in the long run. At the moment we have a government which says they will only do the right thing but I am sorry the [Nationalist right wing political party] has a very large following and I am not sure what will happen in the future.

GP (Group 2)

The political climate, in which tolerance regarding ethnic and cultural differences is reduced, may also be at the root of midwives' concerns. Participants seemed keen not to be seen as supportive of this and, therefore, felt uncomfortable asking the ethnicity question. Some seemed to think that clients might interpret this as a sign of being less welcoming to them compared to a client of Dutch descent and worried that confusion over ethnicity, descent and nationality might possibly cause tension:

Are we just going to ask: 'Hey where are you from? Where are your parents from?'. Because really they are just Dutch, but that's what I find difficult; that I'm emphasizing that they are not Dutch originally. Although my impression is that they don't seem to mind themselves. It's more that it makes me feel uncomfortable.

Midwife (Group 5)

In contrast, one midwife said she did not mind at all asking her clients about their ethnicity to determine their risk; she felt the ethnicity question was a positive one, which helped her to get to know her clients:

People are proud of where they come from, so I mean, that's something; why should it [documentation of ethnicity] be an issue? And what is nice, well that is my opinion anyway, clients who come from elsewhere, they always like to, I mean they enjoy talking about their special things, their culture and traditions. It is always interesting.

Midwife (Group 4)

Midwifery participants voiced other privacy-related concerns related to the nature of genetic disorders in relation to paternity:

Well, I think it can be really tricky, it's the same with a rhesus negative status when a woman is convinced that her partner is also rhesus negative and that she doesn't want the anti-D (. . .) Some things might come out just like with this [HbP testing] he can or cannot be the father.

Midwife (Group 6)

It appears that worries such as these stem from the fact that midwives have the impression that women or couples may not be able to see the potential impact of accepting screening. Finding an appropriate way of informing women in pregnancy may thus be complicated in many ways.

#### *Ethnic awareness and good practice*

Despite the sensitivities voiced by participants, they appeared to be familiar with identifying ethnicity for (other) health purposes and seemed to believe that it is good practice to be aware of different health needs among certain ethnic groups. They use their knowledge and earlier experiences to choose the appropriate care for their patients or clients; for example, diabetes risk in Surinamese people of Indian descent. This awareness also resulted in offering certain patients or clients a test for HbP-carrier status:

We don't carry out standard [HbP carrier] screening. But we do test people with a low Hb or those who were very anaemic in a previous pregnancy and actually also those people with a Mediterranean background; we test them for sickle cell and thalassaemia when we send them for the usual Hb and Mean Corpuscular Volume (MCV) tests as our experience tells us that they are very often positive [for HbP carrier status].

Midwife (Group 5)

But again, one participant pointed out the fact that in her practice, they felt insecure about defining groups at higher risk of a positive HbP-carrier status and as a result, policy was inconsistent:

Part of our practice area is very white and another part has a lot of immigrants; probably 20–25% of our population. We screen black people for sickle cell but this is quite arbitrary because some people who are mixed [of mixed descent] are sometimes not screened, so our policy is not very consistent.

Midwife (Group 5)

Attention given to ethnic differences in clinical guidelines varies.<sup>21</sup> In this case, some participants felt supported by ethnicity-specific recommendations in their guidelines, underlining the importance of professional and national clinical guidance:

I think on the basis of anaemia, of course we currently have a beautiful guideline for this purpose which is really easy to use; you just follow it and arrive at the point where you think it has to be a haemoglobinopathy.

GP (Group 1)

#### *Practicalities in relation to (ethnicity-based) HbP-carrier screening*

Two practical barriers emerged during the focus group discussions: registration difficulties and financial issues. Despite the presented dilemmas, midwives reported that they have been registering ethnicity in the Netherlands Perinatal Register (PRN) since the 1970s, which contains national perinatal data for research purposes and care improvement. Software systems are improving the registration methods:

We register by means of our LVR [national perinatal register] but this is only a very general if not limited registration. Mediterranean, other European, the groups are very 'broad'. We have now started with [new software program for midwives] and with this program you can fill in country of birth, therefore you have the country of birth for everyone.

Midwife (Group 3)

The practical side of implementing ethnic documentation appeared to be no problem for the midwifery profession. However, this is not the case for GPs because the available GP software does not provide a simple way of doing this:

Yes, there is loads of space [in the software program] but you have to be able to locate it [the information on ethnicity]. Like, it's not a separate field [in the program] for which you can search, which you can select. (. . .) I think a computer nerd would be able to do it but not GPs in general. It has to be easy otherwise you won't do it.

GP (Group 1)

Although midwifery and GP care is exempt from financial charges to patients or clients in the Dutch health care system, they are sometimes charged for extra (laboratory) services, depending on how insurance companies apply the rules. Those midwives who already tested for HbP-carrier status expressed concern about the financial implications for their clients. Unclear financial regulations are apparently making them feel awkward about offering such tests:

That depends whether or not the insurance will pay. (. . .) And if people, I mean I don't know how much a test like that costs, but if people receive a huge bill which they have to pay themselves . . .  
Midwife (Group 4)

It appeared that midwives easily feel guilty about financially burdening their clients through the care they have proposed. Not being able to give clear explanations about financial implications may also result in complaints from clients.

## Discussion

This study took a qualitative thematic approach to provide a deeper understanding of the attitudes and it elucidates the perceived barriers of midwives and GPs with regard to ethnicity-related HbP-carrier screening. Four themes were identified, which showed that participants use ethnicity in daily practice for a tailored approach towards their patient's or client's health but find it difficult to adequately identify ethnicity whereby uneasy feelings are experienced when broaching the subject. Respondents voiced strong concerns over privacy issues of their patients and clients. GPs do not have the possibility to record ethnicity in their software programmes. Some respondents worried about the financial implications for their clients.

GPs were of an older generation compared with the midwives (none of them was old enough to remember WWII) and had more experience than the midwives, although few of them give preconception care. This limits the possibility of comparison between these two groups. The number of GP participants in our study was small. Lack of educational credits was not helpful and may have introduced some bias in recruitment. This meant that for the GPs, the intended purposeful sample became a convenience sample. Other studies have encountered similar problems in terms of response and have indicated that a high workload prevented GPs from participating in research.<sup>11,22,23</sup> Minimal interest and the GPs' popularity as research objects may also have influenced the lack of enthusiasm in participation. One GP in this study mentioned that the subject was low on her priority list considering the magnitude of (social) problems she encountered in her practice. It should further be emphasized that qualitative data are not intended to be generalized. Moreover, it is likely that participants in most qualitative studies tend to be those who have a greater-than-average interest in a subject and may therefore introduce some bias in the results.

The discussion on defining ethnicity was one of the most dominant ones in our study. Information on ethnicity can be used to determine risks for certain disorders, such as HbPs, cystic fibrosis (CF), diabetes mellitus etc., which occur more often in certain ethnic groups. However, the use of ethnicity is problematic in both social and political terms.<sup>17,24</sup> It is interesting to note that none of the participants actually thought of asking patients/clients themselves but mostly interpreted ethnicity from outward characteristics such as appearance

and/or surname. Although self-defined ethnicity is favoured by some,<sup>25,26</sup> defining ethnicity is still the subject of ongoing debate.<sup>18,27,28</sup> A generally accepted way of determining ethnic groups is by country of birth and (grand-)parents' country of birth, but this method misses third and fourth generations.<sup>28</sup> Our study results correlate with the complexity of determining ethnicity as demonstrated by others who have shown that a substantial proportion of ethnic groups give complex answers about their ethnic background (and which do not necessarily fit predetermined categories).<sup>24,27,29</sup>

As opposed to Dyson's results,<sup>29</sup> which showed no consistency in how the ethnicity question was interpreted (i.e. meaning place of birth, place of upbringing, family, ancestors or ethnic identity), the participants in this study seemed to have at least some awareness of the importance of ethnic origin with regard to HbP-carrier screening. Whether this was due to the supplied fact sheet or otherwise is unclear nor is it clear whether this awareness is also apparent in clinical practice. Our study does support Dyson's finding that midwives feel uncomfortable asking the ethnicity question. One way to reduce such feelings may be by offering combined screening for disorders, such as HbPs and CF, with different prevalences in different ethnic groups. Carrier screening for these disorders could be offered using a tool specifically designed for such purpose.<sup>12</sup>

GPs and midwives in this study are familiar with the use of ethnicity in daily practice for the purpose of tailored health care and are prepared to screen their patients for HbP-carrier status in the future. However, this also seems to create uncomfortable feelings that require further investigation. Although several instruments are available for determining ethnic HbP risk,<sup>12,30</sup> it is not known what women and their partners think about ethnicity-related health care. No feelings of stigmatization were found in case of the combined screening offer for HbP and CF mentioned above.

A previous study revealed ethnic registration to be a controversial barrier in the past, preventing the introduction of a HbP-carrier programme, which was rooted in the history of WWII and linked to the nationalistic political climate at the time the study was conducted.<sup>11,17</sup> This study shows that these issues continue to be important.

Of course, factors such as prevalence and burden of disease are determined first to assess the appropriateness of screening. However, prevalence and burden of disease vary per ethnic group; therefore, for some ethnic groups, screening is more appropriate than for others.

Considering the complexity of determining ethnicity, universal screening may be more appropriate. Once this has been decided, assessing the importance of determining ethnicity may be the next decision. Irrespective of the screening method, consideration of ethnicity is still required to adequately inform patients or clients of their HbP risk. A validated tool to support health care

professionals to determine risk should therefore be considered.<sup>12,30</sup> At the same time, support from national policy, whereby ethical and practical barriers are solved and financial issues clarified before such a programme is implemented, is crucial.

It may well be possible that for future generations, the necessity to determine ethnicity will become obsolete as ethnic admixture becomes more common in our multicultural societies and complex deoxyribonucleic acid (DNA) testing for determining a wide range of genetic disorders can be offered during preconception screening.<sup>31</sup>

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## Declaration

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