The Moral Life of Professionals in Newborn Screening in the Netherlands: A Qualitative Study

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Newborn screening (NBS) involves a complex logistical process, which depends on the close cooperation of many professionals, such as midwives, laboratory technicians, general practitioners and pediatricians. These professionals may encounter moral problems in the process, which have not been systematically studied before. This study fills this gap. We conducted interviews with 36 professionals involved in NBS in the Netherlands and made an inventory of the moral problems they encounter, as well as of the ways in which they tend to respond to them. The moral problems professionals encounter stem from interpersonal conflicts (when a professional's values conflict with those of a colleague or the NBS protocol) and intrapersonal conflicts (when one professional fulfills different roles with conflicting moral commitments, or when a person's professional role and personal moral intuitions clash). Given the complexity of the work of NBS professionals, the study suggests that the moral problems that occur on the work floor cannot easily be solved by means of offering better or more stringent policy guidelines. Rather, it should be appreciated that professionals contribute significantly to shaping the morality of NBS with the help of their daily choices, and they should be supported in carrying out this task—for example with the help of a moral training or regular multidisciplinary moral deliberation, facilitated by an ethicist.

Introduction

Newborn Screening in the Netherlands

Newborn screening (NBS) involves the collection of several drops of a child's blood on a blood spot card, in the first week of life, which are subsequently tested for a list of rare diseases. Many countries have a NBS program, but these programs differ, for example, with respect to the consent process, the technologies used and the disorders screened for (Klein, 2011; Burgard *et al.*, 2012; Loeber *et al.*, 2012). In line with the well-known Wilson and Jungner criteria (1968), the Dutch NBS program focuses on treatable diseases (Wilson and Jungner, 1968; RIVM, 2013). In its 2005 report on NBS, the Health Council of the Netherlands formulated five criteria that NBS programs have to satisfy:

- The condition should be clearly described.
- There should be a suitable method of detection.
- The condition should be treatable, and the treatment should be accessible.
- Participation of screening should be voluntary.
- Participants should be informed about the screening. (Wilson and Jungner, 1968; Health Council of the Netherlands, 2005)

In the Netherlands, the NBS program started in 1974 with phenylketonuria screening, followed in the next

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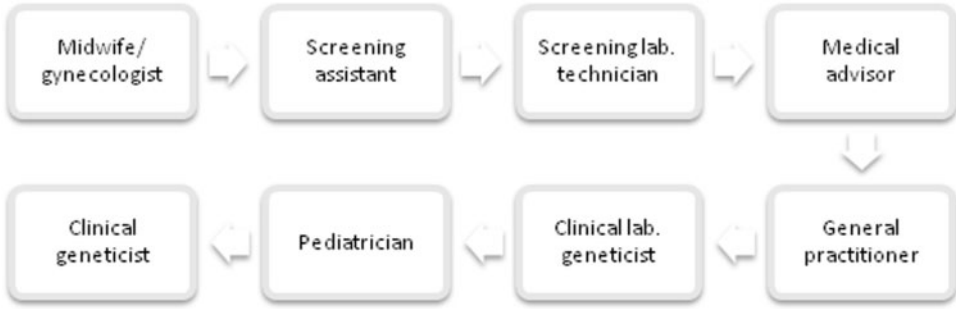

Figure 1. Chain of professionals involved in newborn screening.

decades by screening for congenital adrenal hyperplasia and congenital hypothyroidism. The most drastic change to the NBS program to date was the addition of 14 (mostly metabolic) disorders in 2007, which coincided with the introduction of tandem mass spectrometry (MS) as a screening technology in the Dutch program. In 2011, cystic fibrosis (CF) became the final disease added to the program.

Tandem MS is loaded with a test kit containing specific reagents, which determine, together with the software settings of the apparatus, which metabolites will be measured in the samples tested. Currently, the Dutch program consists of 18 disorders,[1] including metabolic disorders, hypothyroidism, congenital adrenal hypoplasia, a hemoglobinopathy and CF. These diseases have frequencies varying between 1:2500 and 1:200,000 (RIVM, 2013). Depending on the nature of the disease, treatment may consist of a specific diet (for instance limiting the intake of phenylalanine or lactose), the avoidance of fasting, management of stress or medication (for instance synthetic thyroid hormone, prophylactic antibiotics) (Health Council of the Netherlands, 2005). These therapies, when started in a presymptomatic phase, yield a significant improvement of the child's prognosis (rather than a complete cure).

As the NBS program has gone through a number of changes over time—due to the addition of new disorders or new screening technologies—the roles of professionals in NBS have also evolved. NBS involves a complex logistical process, which depends on the close cooperation of many professionals. In the Netherlands, the task of informing the parents about NBS is carried out by a midwife/gynecologist prior to the child's birth, a screening assistant takes the blood sample, laboratory technicians in screening laboratories analyze the blood spots, the medical advisor (in addition to quality control and regional program coordination) informs the general practitioner and pediatrician in case of abnormal test results, the general practitioner informs the parents about the abnormal screening test result, the hospital laboratory technician performs confirmatory diagnostic tests and the pediatrician treats the child and possibly refers the family to the clinical geneticist (see Figure 1).

Policy decisions shape the work of these professionals, since the official NBS policy forms the basis of the protocols and guidelines that NBS professionals use in their work. Yet, the ethics of NBS in a specific country are not only shaped in its policy. Professionals contribute significantly to shaping the morality of NBS on the work floor. It is therefore important to inquire into the moral problems that they encounter in their daily work, and to acquire more insight into how they deal with these problems. Moral problems occur when values and norms conflict, or when they no longer seem applicable. These conflicts may occur within a person him- or herself, for example, when different values that a person holds dear require rival actions; but there may also be disagreement between the values or norms of one person and someone else (such as a colleague), or disagreements with the laws or protocols that guide his or her activities (Kole and de Ruyter, 2007). In these situations, there is a problem that requires to be solved, for it is not possible to avoid acting (Keulartz *et al.*, 2004). The decision that is made eventually shapes the morality of the practice of NBS.

Many authors discuss the ethics of NBS, but this literature rarely focuses on the moral problems that professionals encounter. Rather, it mostly focuses on the design of NBS policy: for instance, with questions such as 'what criteria should be used to decide what to add to the program' or 'how should the informed consent process take place' (Arn, 2007; Avard *et al.*, 2007; Bodamer *et al.*, 2007; Therrell and Adams, 2007; Bailey *et al.*, 2008; Moyer *et al.*, 2008; Miller *et al.*, 2010). Until now, the moral problems that professionals encounter in NBS have not been systematically studied. In this

article, we fill this gap. We made an inventory of the moral problems that professionals encounter, and provide an overview of how they understand them and deal with them in daily practice. This study provides insight into the moral life of these professionals, and draws attention to the dependence of the success of NBS on their reflections and decisions—as well as to the need to take this more into account in the ethical debate on NBS.

Methods

Design and Recruitment

To gain in-depth knowledge of the daily practice of professionals involved in NBS in the Netherlands and the problems they encounter, we conducted semi-structured face-to-face interviews with professionals involved in NBS in the Netherlands. Qualitative interviews are useful to explore experiences, which makes this methodology suitable for our purposes. To determine the different categories of professionals involved, we used the official National Institute for Public Health and the Environment (RIVM) manual on NBS that describes these official roles and tasks (RIVM, 2013). Based on this manual, we selected our target population: midwife, gynecologist, screening laboratory professional, medical advisor, hospital laboratory professional (clinical laboratory geneticist), general practitioner, pediatrician (metabolic diseases, pulmonology, hematology, endocrinology) and clinical geneticist. The number of subjects interviewed per category was not defined in advance, but was dependent on the point of saturation: when no new information could be identified in the interviews (Pope and Mays, 2006).

Before the start of this study, contact had been established with key representatives of different professional groups. Through these representatives, we were provided with names and contact information of relevant professionals. We used purposive sampling to select the interview participants. The prospective participants were informed by email about the objective of the study and were invited to participate. Of all professionals (excluding general practitioners and midwives) approached, two declined to be interviewed (citing lack of time) and one did not respond. General practitioners and midwives, however, were less willing to participate (18 general practitioners/general practitioner's offices were approached, 3 were interviewed; 16 midwives/midwife cooperatives were approached, 44 were interviewed). Many did not respond to our invitation. Among those that did respond but declined to participate, often cited reasons were no experience with abnormal NBS results (general practitioners), lack of time and lack of interest. We think a reason for the low participation rate of these two professional groups is the fact that their involvement in NBS is modest.

Based on literature research and introductory conversations with several of the key representatives, an interview guide with open-ended questions was developed. It was pilot tested on one NBS professional and reviewed by two non-NBS researchers. With these interviews, we aimed to map ethical problems of NBS professionals as they themselves experience them. Therefore, the interview guide consisted of a general section, composed of a short introduction, followed by questions pertaining to daily practice, problems encountered in their work[2] and expectations for the future of NBS. In addition, we included questions tailored to the specific practice of the professional in question, which we thought might help to explain why a professional encounters a moral problem, or why they experience it as such.

An example of the interview guide is available from the authors upon request.

Data Collection

All interviews were held between August 2013 and February 2014 and were conducted by a trained and experienced interviewer (A.O.). Informed consent was received when making the appointment for the interview. The interviews took place in an environment of the participant's choice (usually their place of work). For practical reasons, four interviews (two general practitioners, one midwife and one screening laboratory professional) were conducted by phone. The interviews lasted between 15 and 90 min. Audio of the interviews was recorded and subsequently transcribed verbatim.

In total, we interviewed 36 professionals involved in NBS in the Netherlands (See Table 1).

Data Analysis

The interview transcripts were coded in Atlas.ti 7.1.4. The analysis was conducted using a grounded theory approach, in which the codes, themes and codebook emerge from the data (Glaser and Strauss, 1967; Lingard *et al.*, 2008). The first five individual interviews were coded by A.O. and S.v.d.B., after which discrepancies were discussed until consensus was reached. All other transcripts were coded by one researcher (A.O.). Codes that were assigned included, for example, 'physician's intuition', 'knowledge through experience',

Table 1. Participant characteristics

Type of professional	Number interviewed
Midwives	4
Gynecologist	1
Screening lab professionals	5
Medical advisors	4
Pediatricians	
Metabolic disease specialist	4 + 2[a]
Pulmonologist	3 + 1[b]
Hematologist	2
Endocrinologist	2
Clinical laboratory geneticists	4
General practitioners	3
Clinical geneticist	1

[a]Four metabolic disease pediatricians and two internal medicine specialists focused on adults with metabolic diseases.
[b]Three pediatric pulmonologists and one CF nursing specialist.

'doubts about start of treatment', 'working according to protocol' and 'diagnostic odyssey'. The interviews and the analysis were conducted in Dutch; the quotations selected for inclusion in this manuscript were translated from Dutch to English (A.O.) and subsequently checked (S.v.d.B.).

We followed the consolidated criteria for reporting qualitative research (COREQ) guideline for qualitative research in both design and analysis (Tong *et al.*, 2007).

Results

The professionals interviewed sometimes explicitly framed the problems they encounter as moral problems, but sometimes they described them as practical problems, in which we as analysts recognized a conflict of values and norms. Here, we will report about the ethical tensions that we recognized in our conversations with them.

Informing and Taking the Sample (Midwife, Screening Assistant)

In the Netherlands, midwives take care of all the uncomplicated deliveries and perform the regular health checks during pregnancy. Midwives also perform different screenings and provide the accompanying information. In the case of NBS, they provide information in the last phase of pregnancy. In some regions, midwives also take the blood sample after birth on which NBS is performed, but in most regions this is done by a screening assistant. Before taking the blood sample, the midwife/screening assistant informs parents a second time.

When midwives inform parents about NBS, they prepare them for the choices they have to make, which are threefold in the Netherlands: parents may (1) opt in or out of screening altogether, (2) accept or refuse information about the carrier status of their child (which is a by-product of screening for sickle cell disease, thalassemia and CF and (3) accept or refuse the storage of the blood spots in a biobank for 5 years after screening has been performed.

Concerning their role in NBS, midwives report experiencing a tension between two tasks: their task to inform parents about the possibility that their child is diseased and receives an abnormal screening result, and the task which they consider central to midwifery which is to reassure pregnant women and give them the self-confidence needed to handle the pregnancy and the delivery. Especially in worrisome women, midwives fear that talking at length about NBS and all its possible results may be detrimental to pregnant women's trust in themselves and their future with the child. Therefore, midwives may choose to gloss over the NBS information, without talking about its more difficult and controversial aspects.

Furthermore, midwives also stated that they often lack the time to provide all the information about NBS, and therefore they have to prioritize. As they also have to provide information about other screening programs, such as the prenatal screening for trisomy 18 and 21, midwives make a choice: they spend more time providing information about prenatal screenings, which they consider more 'controversial' because they may reveal an untreatable disorder which confronts parents with the question whether they want to terminate the pregnancy (for the relevant quotations, see Table 2).

Analyzing the Sample (Screening Laboratory Technician, Screening Laboratory Head)

Screening laboratory professionals determine whether a sample is normal or abnormal. Values that are important in their work are consistency, clarity and reliability of sample analyses. Furthermore, they closely follow national guidelines to safeguard uniformity of results between the five different screening laboratories, and to make themselves accountable for them. A trustworthy NBS program respects these values.

Table 2. Moral tensions in the information phase

Topic	Professional	Representative quotes
NBS task vs. other tasks	Midwife	*Midwives have so much to tell people, the information about the heel stick is just one very small piece. In practice, screening for Down's syndrome or trisomy 18 receives much more emphasis than all those other things. Because that's about very serious diseases with serious consequence, such as—do you want to possibly terminate the pregnancy? Those are way more difficult decisions than when you say 'okay, we're going to look if the child has a disease and if we detect it, you can prevent many very nasty consequences with a treatment', that's not something people will be against.*
	Midwife	*As a midwife in particular you are very much aimed at inspiring trust in people, like 'you can do this, you can get pregnant and deliver a child'. And especially with worrisome people you find it very difficult to discuss 'would you want to know whether your child has a disease that cannot be cured?'*
	Midwife	*The probability of them having to deal with this is negligibly small, so you'd rather just leave that story up to a professional [if it comes to that], instead of me telling everyone about it, which might cause them great distress.*

There are several situations in which these laboratory values are put under pressure. This happens most often in periods of transition; for example, when a new screening technology is introduced, or a new disease. This usually demands to adapt the established way of working. The introduction of tandem MS, for instance, marked the most radical period of change. When tandem MS was first introduced, cutoff values were predominantly based on international data, which turned out not to be as generalizable as was originally thought. The threshold to distinguish diseased from healthy children used in other NBS programs therefore had to be adapted to the genetic make-up of the Dutch population. Furthermore, there were other difficulties: such as the fact that different tandem MS machines turned out to produce slightly different results, and the timing of the blood withdrawal influenced the results. Use of tandem MS for NBS therefore necessitated adaptation, and initially led to an increased number of false positive results. This in itself produced moral problems for laboratory professionals, because they knew that false positives may lead to a decrease in the trust of parents in the NBS program, as well as a decrease in the trust of other professionals such as general practitioners. The large number of false positives had to be reduced by setting new cut-off standards. This usually involves a process in which the threshold between normal and abnormal values is at first set relatively low so that no diseased children are missed, and then it is slowly elevated. In this process the large number of false positives that is produced at first is slowly reduced with the elevation of the cut-off standard. The eventual goal is to not miss any diseased child, but to keep the number of false positives to a minimum (in other words, achieve high test sensitivity and specificity). While this process is necessary, the initially produced false positives lead to feelings of moral unease in laboratory professionals as they know that they are causing distress in parents.

In addition to the introduction of tandem MS into screening, there are also less extensive transitions that produce moral problems for laboratory professionals; such as when a new disease is added to the NBS program, or when different test kits are used to perform the screening with tandem MS. Each of these transitions may lead to false positive results, but may also produce other types of results that exceed the limitations of the program.

Laboratory professionals report, for example, that they are required to provide information just about the diseases that were included in the program at the

moment when parents consented to participate. However, sometimes—especially during periods of transition—screening laboratory professionals are confronted with results that fall outside of the scope of the official program. Acting on these results would mean going beyond the constraints of the program, and offering information that parents did not consent to obtain. This may happen, for example, during pilot tests on anonymous samples of children who were already screened, which are performed to prepare for the extension of the program. These pilot tests may occasionally reveal a possibly diseased child. Professionals indicated finding this morally troublesome, because it means that they possess knowledge that is important for the child and the parents; it could mean saving the life of that child. But passing this information on to parents *after a pilot test* means disrespecting the program's guidelines which demand sticking to the limitations that the program had at the time when the parents consented to subject their child to screening. As the disease was not part of the program when the parents gave their consent, laboratory professionals are not allowed to communicate their findings. Professionals indicated that this feels especially unjust because the disorder that is pilot tested is about to be added to the program and therefore satisfies the criteria of the NBS program.

Apart from moral problems that laboratory professionals encounter during periods of transition, they may also experience problems during regular screening, for example, when they encounter incidental findings. Incidental findings are a risk because tandem MS is able to screen for over 60 diseases, and only a subset of these diseases satisfies the criteria of the Dutch NBS program. To limit the results that tandem MS produces, a test kit is used which allows screening for this subset. However, laboratory professionals still work with 'raw' data, which means that they will sometimes see abnormal values indicative of diseases not officially included in the program. This may occasionally confront them with moral problems. Some laboratory professionals indicated, for example, that they have in the past contacted pediatricians to confer about results they thought were particularly worrying. Officially, however, they were not supposed to communicate these results for the findings do not meet the NBS criteria: the results point, for example, to a disorder with unknown prognosis, or which is untreatable. This occurred, for instance, when a screening laboratory contacted a pediatrician about a high methionine level in a sample, even though screening for this metabolite had been suspended. Although the screening laboratory technician and the pediatrician were doubtful about the benefit, in this specific situation the pediatrician felt a need to act upon the information simply because it had been disclosed. There is a tension in this situation, however, between the need to honor the informed consent of the parents and the limitations of the NBS program, and doing what is perceived to be contributing to the health of the child. Different NBS professionals have contrasting ways to respond to this problem. According to some professionals, the purposes of the NBS program can only be served if professionals follow these guidelines and do not communicate about results that go beyond the limits of the program; other professionals have difficulty adhering to the program's limitations in cases where they think parents and children can be harmed if the information is kept from them. Several professionals indicated that having to stick to the guidelines causes feelings of moral distress in these situations. While they themselves thought there were strong clinical reasons to act on results, they felt that they were forbidden to do so by the guidelines in place (for the relevant quotations, see Table 3).

Processing the Screening Results (Medical Advisor)

Medical advisors are the intermediaries between the screening laboratory and the subsequent health care. The medical advisor has a medical background and is involved in the interpretation of the results and organization of the child's referral to a specialized pediatrician. When an unclear or abnormal test result occurs in one of the screening laboratories, the medical advisor is informed and determines whether the test result warrants a visit to the pediatrician based on the national guidelines. The advisor organizes this referral visit and informs the child's general practitioner about the abnormal result, who then visits the family to inform them about the situation and assess the child's health. In addition, medical advisors run the regional program: they are concerned with program coordination, surveillance and quality control. To this end, medical advisors give (solicited and unsolicited) advice about the execution of NBS to all executive parties involved in NBS.

Essential values for the medical advisor's role are uniformity, reliability, inspiring trust, accessibility, promptness and collegiality. More than any other NBS professional, medical advisors perceive themselves as part of a chain of professionals. Several medical advisors referred to the idea of a chain being only as strong as its weakest link: to do their job in NBS well, they depend on the screening laboratories for good results and on the general practitioner and pediatrician for good follow-up care for the child and parents.

Table 3. Moral tensions in the screening phase

Topic	Professional	Representative quotes
False-positive results	Head screening laboratory	*The first year was deemed a pilot year, because only when the program actually starts running do you find out its teething problems. Of course beforehand you can validate all you want, but it will only start to become real when everything starts running. Only then, for example, did we find that the galactosemia screening was absolutely not right.*
	Head screening laboratory	*All of a sudden the method we have all been using is found to be [unavailable] and you're forced to switch to a different method and that causes a change in the entire referral trajectory, then the metabolic disease specialists are on the phone, like: 'what's happening? I have a lot of false positives'. And the entire process starts all over again. The same as in 2007: we choose the safest method, we don't want to miss anything, that is the principle of screening, so we start with the lowest cutoff value, and then you run the risk of having very many false positive referrals.*
Incidental finding pilot phase	Head screening laboratory	*And you find an MCADD, what should we do with this? You're not allowed to do anything. Parents have not given permission for screening for MCADD, that wasn't in the package. Yeah, I found that difficult.*
Incidental finding regular program	Pediatrician metabolic diseases	*But the difficult thing is, if you wouldn't have screened then you wouldn't have known. And I would rather not have known, because I can't do anything with it, but now I have to do something with it. If you wouldn't have known, you wouldn't have to tell the parents, you wouldn't have to do anything with it. When things aren't clear, you get these types of 'in between' things. So I think the clearer the guidelines are, the better.*
	Screening laboratory researcher	*Officially, we no longer screen for this disease. But you see something which makes you think 'Oh, this might actually be one' so then I contacted the metabolic disease pediatrician; asked 'We have a child with a very high methionine. What do you want us to do with this?'*

Moral problems arise for the medical advisor when others in the chain go beyond the official guidelines: for instance, when a screening laboratory reports a result pointing to a disorder that is not in the program and consults the medical advisor about how to act. Medical advisors disagree as to what is the appropriate course of action in this case. Some medical advisors do not act on this information, for it impedes on values like reliability and uniformity of the NBS program. But other advisors sometimes refer the child for further diagnosis to a pediatrician because they recognize the relevance of the finding for the health of the child and feel it is unjust to ignore it. Similarly, the interaction between medical advisors and general practitioners may also raise moral difficulties. In this interaction, medical advisors have to communicate a contradictory message: they have to explain that children need to receive a definite diagnosis urgently so that treatment can be started, but also make sure that the general practitioner does not panic and consequently overstate the urgency to the parents. While the protocol demands that medical advisors simply communicate abnormal heel stick results to the general practitioner, medical advisors often already suspect whether a result is true or false, based on the raw analysis result. Consequently, some medical advisors explain they sometimes deviate from protocol, when they

Table 4. Moral tensions in processing the screening results

Topic	Professional	Representative quotes
Interest of child vs. interest of program	Medical advisor	*In the interest of the child, everything else can disappear. A child that is sick should be told that. So yes, I will always choose the child, yes. Screw procedures. You have a different goal. If you look solely at the screening program you wouldn't report it. And we look at the patient. That is something that conflicts more often. A screening program has certain standard conditions. But we think we're there for the exceptions. I'll do things that aren't according to protocol, because I think 'How would I feel if I were the mother or father?'*
	Medical advisor	*I just have an interest in public health screening in general, the entire process. Because the entire process determines the reliability. If one link is weak then you can forget about the rest.*
	Medical advisor	[On talking to the general practitioner] *If it is a disease that occurs one in 200,000 times and it's a result right on the cutoff point you can say 'well, it's possible, but we don't know yet, we'll have to look at it further. It can still go either way, because for some diseases it very often turns out not to be anything'. And a screening is of course very straightforward and you shouldn't do that, I know that. But the GP has to go to the parents with this whole story, those parents are scared to death. So if it isn't certain that [a child is in fact sick], or maybe the therapy will be very mild, yeah, I'll say that. You can't always do that, but when it's possible, I'll do it.*
	Medical advisor	*Look, you have protocols and frameworks and those catch 80% of clinical practice, but at a certain point a professional has fingerspitzengefühl, a doctor has it, a laboratory technician has it. I always say: 'Listen, if you only follow protocols then you don't have to pay me, there's my folder, do it like this, I'll be at home. But if you want me here you should make use of me.'*

think it is very likely that the result will be a false positive or, conversely, when they are almost sure that the child will have the disease. They might indicate this to the general practitioner, to help him or her strike the right note in communication with the parents. The aim here is to prevent causing unnecessary distress in parents, and to motivate parents to act quickly when needed (for the relevant quotations, see Table 4).

Diagnosis and Treatment (General Practitioner, Clinical Laboratory Geneticist, Pediatrician, Clinical Geneticist)

General practitioners, pediatricians, clinical laboratory geneticists and clinical geneticists all have a role in setting the initial diagnosis and (if necessary) deciding about the treatment of the child. The role of the pediatrician is, however, most relevant here, for he or she encounters most moral problems. Some of these problems relate to the role of the pediatrician as a health care provider, and concern questions relating to the communication with parents, the diagnosis itself, incidental findings or the way in which they should deal with carrier status information. Furthermore, there are problems that stem from a tension between the role of the pediatrician as a health care provider and as a scientist. Values like beneficence and non-maleficence play a central role in all of these problems, but trust, honesty, an open future, care and avoidance of medicalization are also important.

Trust is the primary value at stake in the moral problems relating to the communication between pediatrician and parents. Since the relationship between pediatrician and parents is characterized by asymmetry

in knowledge (parents depend on the pediatrician to provide them with the relevant information about the disorder), the communication between them primarily serves to share knowledge and build trust in the pediatrician and trust in the future of the child with the disease. With respect to the future of the child, pediatricians indicate that values such as certainty, comfort, predictability and reassurance are of important to parents. But it is sometimes difficult for pediatricians to live up to those values. What is specifically difficult, for example, is to strike the right balance between stressing both the seriousness of the disorder (to make sure that parents adhere to the diet, medication or lifestyle rules), and at the same time reassuring parents of the favorable effect that treatment has on the quality of life of the child. These two aspects are hard to combine in a single conversation, and exactly how they need to be combined may vary between different diseases. For some diseases in the program (such as phenylketonuria (PKU)) the effects of treatment are well-known and the expected quality of life is good if treatment is consistently followed; but when the child has an extremely rare disorder (such as very-long-chain acyl-CoA dehydrogenase (VLCAD) deficiency), the pediatrician may have little or no experience with it and may be very uncertain about the effects of the advised treatment on the prognosis of the child. Since this difficulty occurs regularly (all disorders included in the NBS program are rare, with frequencies ranging from 1:2500 to 1:200,000), pediatricians frequently fear their efforts to inspire trust in parents remain lacking.

In addition to communication problems, pediatricians may also experience moral problems when they are uncertain about the diagnosis. Sometimes, when a child receives an abnormal heel stick result, the diagnostic phase does not (yet) provide enough evidence to support a clear diagnosis. In these situations, pediatricians may be uncertain about what to do: there is not enough reason to provide treatment, but it may also be unwarranted to deem the child completely healthy. In these cases, pediatricians may hesitate, and sometimes decide to monitor the child periodically, even though the child has not been diagnosed with the disorder it initially screened positively for. In these situations, pediatricians deviate from the NBS protocol, which prescribes to follow children only when they clearly have one of the diseases included in the program. Concerns for the health of the child, and concerns about the trust of parents in the health care services, may however motivate pediatricians to act otherwise.

Another example relates to the detection of carrier status. When a hemoglobinopathy carrier is detected, this is reported by letter to the child's general practitioner and to the parents.[3] The idea behind detecting and reporting hemoglobinopathy carrier status within the NBS program was to give parents the option of genetic testing to assess whether both parents are carriers and might possibly have an affected child in the future. If this is the case, they can opt to avoid the risk using different reproductive options, such as avoiding pregnancy, using donor gametes or undergo prenatal testing in future pregnancies.

The primary aim of pediatricians is to prevent pain and suffering in children and parents. Carrier status information provides this opportunity, but only if the information is adequately followed up on. According to the professionals we interviewed, this very rarely happens in reality: very few couples visit a clinical geneticist for further consultation. Several hematologists we spoke to suspected this problem lies mostly with general practitioners who do not properly understand the relevance of carrier status information, or are unable to convey its importance to the parents in question. A point of significant frustration for hematologists, as they see the potential of health benefit, but–in their view–are unable to realize this potential due to non-cooperation of a different professional. This might be due to the difference in perspective between the two professionals: the specialized physician who is very involved in the NBS program and is continuously confronted with the burden of hemoglobinopathy, and the general practitioner who is very rarely (if ever) confronted with the NBS program and is dealing with the daily grind of general practice.

Other examples relating to diagnostics include the so-called 'partial deficiencies', which are milder versions of disorders in the NBS program. These milder versions may raise moral questions regarding the decision whether to offer treatment or not. In metabolic disorders, the problem is often a deficient enzyme, causing one metabolite to build up in a child's organs and possibly damage them in the future. It is, however, often unclear how much residual enzyme activity is needed to maintain a normal, healthy life without therapy. In general, NBS programs set a certain (fairly arbitrary) threshold to determine who is labeled 'diseased' and who is labeled 'healthy'. It may therefore be that children fall on the 'diseased' side of this threshold, and receive treatment, even though their residual enzyme activity would have been enough to keep them healthy. The physicians we interviewed indicated that it would be better not to treat these children, since the burden of treatment in these cases is greater than the potential benefit. It leads to a medicalization of the children's lives.

Incidental or unsought findings may also raise moral problems for pediatricians, which relate to this problem of medicalization. An example of an unsought finding in the official program, is the detection of a disorder in the mother rather than the tested child.[4] In rare cases, a child's blood spots contain an excess amount of a certain metabolite, causing the result to be deemed abnormal and the child to be sent to a specialized pediatrician. Only after further diagnostic tests in the child and the parents, the mother is found to have the disease. Metabolic physicians who encountered this problem pointed out that these women are usually asymptomatic when they are diagnosed, even though biochemically, their values fall outside of the norms. Such a situation raises moral problems for them: should asymptomatic women be treated, should they only be monitored to make sure they stay asymptomatic, or should they be deemed healthy altogether? While the NBS protocol prescribes to treat individuals in whom the disease has been diagnosed, there is no consensus among physicians as to how to respond to asymptomatic mothers because there is lack of long-term experience with mild variants of disorders. On the basis of present knowledge, it is uncertain whether women with abnormal values at, say, age 30 will ever develop disease-related problems later in life that might have benefitted from medication or monitoring. Treating these mothers may be important to prevent disease later in life, but it may also medicalize the life of the mother who would otherwise have lived without complaints.

In all the preceding examples, pediatricians are in doubt about how they should carry out their role as health care providers, even if this sometimes conflicts with the NBS protocol. Their primary concern is to establish whether the information gained through NBS is ultimately beneficial or harmful to the patient and his or her parents, and whether acting on this information will inspire trust and empower parents to take the care for their children in their hands, or if it leads to medicalization.

Pediatricians, however, sometimes also have a role in scientific research. Part of the moral problems that they experience stem from discrepancies between what these roles require them to do. A scientific researcher aims to generate scientific evidence that is reliable, generalizable, transparent and uniformly true for large groups of subjects. But these values do not always cohere with the values of health care providers, who tailor their treatment decisions to individual patients to enhance their well-being. A conflict of values occurs, for example, in treatment of children with CF. Treatment for CF does not cure the disease, but improves a patient's quality of life and may extend his/her lifespan. In the Netherlands there are two treatment protocols, one of which is more invasive than the other. In the more invasive protocol, children undergo diagnostic interventions such as a computed tomography (CT) scan and bronchoscopy very early in life. The clinical utility of these early interventions is however a topic of debate among the pulmonologists that we interviewed. Some of them state that subjecting children to these early invasive diagnostic interventions is not in the best interest of the patient, but primarily aims at acquiring scientific data to support the clinical utility of this protocol. While obtaining this scientific knowledge may eventually benefit the CF population at large, it does not necessarily benefit the individual patient. To what extent scientific goals are mixed with clinical ones remains unclear. But it is a concern that pediatricians expressed in our interviews (for the relevant quotations, see Table 5).

Discussion

With this study, we set out to make explicit the moral commitments present in the work of NBS professionals, and how these moral commitments can lead to moral problems when they conflict. Yet, at the start of the interview, many participants expressed doubts about the very presence of a moral dimension in their work. Issues that were often brought up first were those regularly described as general moral problems in NBS literature and policy document, but they did not immediately talk about problems relevant to their own daily professional practice. Only when probing further did the participants reveal the less obvious or more subtle moral tensions in their daily work–matters they not always explicitly framed as moral problems, but often regarded as practical problems in which we, as researchers, recognized conflicting moral commitments (for example, some pediatricians regarded dilemmas surrounding the treatment of partial deficiencies as merely scientific ones, while they simultaneously described a conflict of values in their dealings with them).

Our data indicate that professionals experienced moral problems in different phases of the NBS process: from informing and sample-taking to screening, interpreting the results, setting the diagnosis and–eventually–choosing and tailoring treatment. These problems originate from different sources, and can be classified as either intrapersonal (a conflict within an individual person) or interpersonal (a conflict between different people) moral conflicts.

Table 5. Moral tensions in the diagnosis and treatment phase

Topic	Professional	Representative quotes
Balance in communication	Pediatric endocrinologist	*What you should explain to parents is that on the one hand they have a child with a serious disorder, and that medication is really necessary, you can't play that down. On the other hand those children have a high probability of normal development, provided they take their medication correctly. That balance is much more difficult for the parents than when you come in with a deathly ill child and you give them medication and they are cured. They don't actually have experience with the disease and we notice this in the course of treatment. In the first year of life the parents say: 'But do we have a sick child? Is it all correct? Is it reasonable to give the child medication?'*
Inspiring trust	Nursing specialist	*Especially in the beginning we invest a lot of time and effort in building a trust relationship with the parents that is as strong as possible. You really let them know like, we're going to do this together, it's a difficult road, but together we're a team that's behind you. Yes, you really need that [trust relationship]. Because you are both the bearer of bad news, but you are also the one people turn to.*
Partial deficiencies	Head screening laboratory	*Should you treat a partial deficiency? Well there are different opinions about that. There are many cases of galactosemia where you're like 'should we treat or not?' You don't actually want to see those. But there are exceptional cases of people with a mild manifestation in their lab values, but with clinically relevant [symptoms]. So you have to choose your cutoff values in such a way that you include some mild variants too.*
Positive screening, no diagnosis	Pediatric endocrinologist	*But of course you might consider how far you have to go to rule out diseases. A child with a positive screening, who looks healthy and whose results are negative. When are we truly and forever convinced that it is really negative? Can you really tell parents: 'We have checked everything, the child is not in danger. Your child does not have to enter the medical system'. That is my ethical dilemma.*
Different treatment protocols for CF	Pediatric pulmonologist	*Sometimes you have to wonder whether it's appropriate to start scientific research in this population so early. Yes, it's a wonderfully defined population we haven't been able to study before and it might give you starting points to improve health care. No... the research you're doing is so invasive that you have to wonder... That is something people don't know when they indicate on the heel stick form 'we want to know about this'.*

Intrapersonal conflicts arise, for example, when one professional fulfills different roles with conflicting moral commitments, such as a role within NBS, as a health care provider and as a scientist. As each role comes with its own set of distinct values, these professionals in a sense wear different 'moral hats'. Think, for instance, of the pediatrician who decides to follow-up on a child with an unclear diagnosis because she considers this to be her task as a health care provider, even though she realizes that this conflicts with the NBS protocol that explicitly asks her to focus only on treatable diseases and to which she also adheres.

Naturally, NBS professionals are also people with their own personal value system. A different form of

intrapersonal conflict occurs when there is a conflict between a person's professional role and personal moral intuitions. For example, a laboratory professional passes on to the pediatrician information that stems from a pilot test carried out prior to the introduction of medium-chain hydroxyacyl-CoA dehydrogenase deficiency (MCADD) in the screening panel, even though this disrespects the constraints of the NBS program at the time when the parents consented to participate, because she identifies with the parents and wants to prevent their child dying suddenly of this disease.

Interpersonal conflicts are a different source of moral problems. These happen, for example, when colleagues support rival norms, such as is the case when different laboratory professionals disagree as to whether an incidental finding should be communicated. Interpersonal conflicts also concern conflicts between the value basis of an individual professional and the NBS program or protocol he/she is supposed to follow. Our data reveal an abundance of examples of that, such as when medical advisors decide to follow up on findings that exceed the limitations of the program, or provide extra information which indicates whether or not the finding is suspected to be a false positive.

Generally speaking, our interview participants more easily identified interpersonal conflicts as moral issues than intrapersonal conflicts. Moral tensions stemming from intrapersonal conflicts, especially those deriving from conflicting moral commitments belonging to different professional roles were often not explicitly framed as moral problems by our participants. A possible explanation for this observation is that interpersonal conflicts are a more obvious hurdle in daily practice as they impede normal daily routines, and therefore need to be reflected on and discussed. Meanwhile, intrapersonal conflicts are more subtle, and as they do not directly obstruct the flow of the daily process, professionals give them less attention in their daily work.

Our participants had different conceptions of what constitutes good NBS practice, and what their role and responsibility within this practice should be. On one end of the spectrum, we found participants who feel very comfortable with the guidelines and protocols in place, and who generally do not question them. On the other end of the spectrum, we saw professionals who view the guidelines more as suggestions about how they should go about their job. It is these professionals who are often bothered by the guidelines and protocols, and appropriate a considerable degree of decision latitude within their NBS work.

The majority of our participants fall somewhere in the middle of this spectrum between extreme protocol adherence and extreme independent decision-making. How professionals relate to their own NBS role and the associated responsibility, influences whether and to what degree they experience moral distress. A perceived obligation to adhere to the protocol may lead to moral distress for professionals in situations where they feel they know what would be the right thing to do, while the protocol prevents them from doing it. On the other hand, professionals who feel it is their responsibility to independently decide what would be the right thing to do in a certain situation may be distressed by its heavy burden and the accompanying doubts.

It is important to have insight into how professionals experience the moral problems that they encounter, and how they respond to them, because professionals give shape to NBS policy in the daily practice of NBS. Of course, professionals such as midwives, laboratory technicians and pediatricians are also represented in NBS workgroups of the Heath Council of the Netherlands, and in committees of the National Institute for Public Health and the Environment (RIVM), which together shape the NBS policy documents. But the actual reality of NBS is given shape in the daily decisions that professionals make on the work floor. If professionals understand their relation to policy guidelines differently, then this is relevant to policy as well.

As this study reveals, there are tensions between the morality of NBS policy as it is designed on paper and the morality of NBS policy as it is practiced by professionals in daily life. While our respondents recognize that it is important to work according to policy guidelines and protocols, even the professionals who consider it their role to simply carry out protocol cannot just follow it passively, for even they will have to make decisions about individual cases that demand their professional expert judgment, for example, with respect to what information to provide to parents, how to interpret a test result and what treatment to provide or when to start providing it. Given that the identification and management of diseases included in the NBS panel are complex tasks, even those who follow protocol most strictly will be confronted with situations in which they encounter problems for which there is no cut-and-dried solution. These situations will necessitate reflection about what to do, which often also involves reflection about one's own professional role and the values that define it. Furthermore, conflicting moral commitments are and will always be a part of NBS, due to the different roles that professionals have to combine to shape it: science, public health and health care. Problems encountered in daily practice, as well as the combination of roles that professionals in NBS have to fulfill make it rather

difficult for professionals to see themselves solely as policy instruments, executers of protocols and guidelines. Rather, to do their job and realize the goals of the NBS program requires them to be independent professionals reflecting and deliberating about the best way to act in a variety of circumstances. It is precisely this professional judgment that forms the basis of good NBS practice and a strong and sound program, but it is also the source of the moral problems that professionals occasionally encounter.

Our study enriches the literature on NBS because it adds empirical content about the daily life of professionals to moral problems that have been mentioned in the literature. In the discussion about the voluntariness of the NBS program, for example, the volume of the information that needs to be provided is often called a challenge (Bailey *et al.*, 2008; Bunnik *et al.*, 2013; DeLuca *et al.*, 2013) as well as the problem of having to explain things 'without being alarmist' (Dhondt, 2007), and the difficulty of tailoring information to parents, especially when parents have different cultural backgrounds (de Montalembert *et al.*, 2005; Dhondt, 2007; Cragun *et al.*, 2015). Furthermore, the role of doctors as intermediary between screening and health care is mentioned in the literature, and consequently it is considered a problem that doctors are sometimes 'ill-prepared to talk about screening results' because of a lack of knowledge about the diseases included in the screening program (Gennaccaro *et al.*, 2005; Dhondt, 2007; Roberts *et al.*, 2014). Diagnostic difficulties—which include the detection of false positives, incidental findings, differences of interpretation of results and difficulties distinguishing mild from serious versions of disease—are also frequently described (Wilfond and Gollust, 2005; Roussey *et al.*, 2007; Dhondt, 2010; Cornel *et al.*, 2014).

What makes this study different from previous literature, however, is that until now there was little insight into how professionals experience these problems, how it raises their moral unease and how they deal with this unease. This is what this study adds: the qualitative nature of this study provides insight into the moral worlds of these professionals. More insight into this moral world also helps to make more informed suggestions as to what is needed to move forward (Guest *et al.*, 2006). In concluding this article, the question arises how to adequately deal with the problems that professionals encounter. One way of doing that would be to invest in surveillance of guideline compliance by professionals. Our study suggests, however, that this would only have limited effect. Given that NBS professionals are positioned between different forces—protocols and guidelines, private moral intuitions and professional norms and values that belong to the different roles that they play—moral problems will likely continue to turn up even if the protocol is changed. Furthermore, adding new and complex types of disorders to the NBS panel,[5] introducing new technologies (think of, for instance, whole genome sequencing; van El *et al.*, 2013; Howard *et al.*, 2015) and the growing interrelation between NBS and research will likely make the NBS program more, rather than less, complex, probably increasing the number of difficulties for professionals, including moral problems. To maintain a trustworthy and sound NBS program, it is important to anticipate these problems and acknowledge that they will continue to impose demands on professionals. Therefore, in view of current and future problems, we think a better way of helping professionals encountering moral questions would be to strengthen their own ethical reflection. This suits the role *as professional, as* they are expected to fulfill in the current NBS program, who is asked to use his or her expertise to make decisions in response to concrete situations. A sound NBS program can only be sustained by professionals who are loyal to the program as well as capable of independent decision making, which demands strong reflective skills. To help them develop these skills with respect to the moral problems they will likely encounter it may be advisable to offer ethical training or invite professionals to share these moral problems during regular interdisciplinary moral deliberation meetings, facilitated by an ethicist, as is common practice when moral problems occur in clinical settings. Such regular interdisciplinary reflection on moral problems would (a) acknowledge the fact that moral problems continue to be part of NBS practice and can never be completely avoided by improving the protocol, (b) empower professionals to develop skills to recognize and understand the field of different moral forces within which they work, orient themselves within it and provide justification for what they eventually do and (c) contribute to the shaping of a common professional practice of dealing with these problems. Ultimately, this would help professionals to optimally fulfill their role, and it would contribute to the transparency and justification of the NBS program as a whole.

Our study had several limitations. Any qualitative study carries the risk of eliciting socially desirable answers from its participants, especially when asking about moral issues. We hope to have reduced this risk by asking the participants to describe examples of problems they themselves experienced. The absolute number of participants per category in our study was relatively

small. However, when considering the labor-intensiveness of qualitative research and the fact that the field of NBS in the Netherlands consists of a relatively small number of professionals, we argue that the number of professionals included was enough to reach the point of data saturation (Guest *et al.*, 2006). Additionally, by including different types of professionals from different regions in the Netherlands, we included a breadth of perspectives, increasing the generalizability of our research. We are therefore confident our data provide a reliable basis to support our conclusions and recommendations.

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Notes

1. The screening for one of which, homocystenuria, has been suspended since October 2010 due to doubts about the quality of the test method. A recent Health Council report recommended removing homocystinuria from the program for this reason (Health Council of the Netherlands, 2015).
2. By asking explicitly about problems they encountered in their own work, our respondents generally spoke only about ethical tensions they themselves experienced or knew about. Several of the respondents fulfilled a coordinating role (medical advisor) or were part of a regional or national NBS working group, and therefore also spoke about ethical problems they encountered through this part of their work.
3. When the interviews took place, a change in policy had just been effected; where carrier status reporting used to pertain to both CF and sickle cell anemia, this changed due to several false-negative CF cases (who were incorrectly classified as carriers). From that point on, these CF 'carriers' were flagged as possibly affected children and were supposed to be sent in to the pediatrician. Therefore, currently the only carriers detected and reported in the initial phase of the program are sickle cell anemia, and—as incidental findings—some thalassemia carriers.
4. This has occurred in mothers with a thyroid disorder, and in mothers with metabolic disorders such as 3-methylcrotonyl CoA carboxylase deficiency, carnitine transporter deficiency and glutaryl-CoA dehydrogenase deficiency.
5. In fact, a recent report by the Health Council of the Netherlands recommended the addition of 14 metabolic disorders to the Dutch NBS program (Health Council of the Netherlands, 2015).

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