



## Newborn Screening, Banking, and Consent

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**Introduction.** Newborn screening (NBS) programs are implemented as government sponsored public health initiatives. They aim to identify infants affected by inborn disorders that can result in mortality or lifelong disability in the absence of immediate treatment. Once at-risk infants are detected, parents are then re-contacted to have their children undergo confirmatory diagnostic testing. Most industrialised countries screen for PKU and congenital hypothyroidism. There are also programs that target specific geographic areas, where populations are at higher risk for disorders such as thalassaemia or sickle cell disease. Some of these targeted programs are currently being integrated into universal screening programs where the population is comprised of high-risk groups for certain diseases<sup>1</sup>.

A few major trends are influencing the development of NBS programs throughout the world. At the outset, technological advances, such as the introduction of tandem mass spectrometry (MS/MS), allow bloodspot testing to identify a wider range of conditions. Indeed, at least 30 disorders, some of which are not treatable, can now be detected in a single process by using the same dried blood specimen collected in the course of routine newborn screening programmes.<sup>2</sup> The introduction of new technologies for the genetic testing of newborns, such as tandem mass spectrometry, and DNA microarray technology (DNA chip) has prompted ethical concerns about extending the range of disorders to be detected towards those which include quality of life issues and psychological benefits to the child and or parents, as well as the need for consent and to inform parents about these technological advances.

Blood spots can be used repeatedly and stored for a long time. In addition, storage practices of newborn bloodspots vary internationally. Some countries like Denmark store cards indefinitely, while others destroy their cards after completing the necessary quality check. The variability of storage practices raises concerns around newborn screening programs. For instance, what should parents be told about storage and about the subsequent use of dried blood spots? Whilst generally most routine newborn screening programs have not required parental consent for storage and future use, ethical concerns about patient privacy, confidentiality and autonomy are influencing a re-examination of this policy.

Existing policies diverge on the manner in which consent is solicited and the way information is presented to parents in the case of disorders amenable to treatment; of disorders for which no treatment is available; and of storage and of future use of stored samples. The issue of consent, refusal or choice and the practicality of written consent will vary according to circumstances.

This GenEdit critically examines how existing guidelines and policy statements have addressed: (I) consent to screening for treatable diseases, (II) consent for untreatable diseases/and a wider range of disorders, (III) consent to storage, and (IV) consent to future uses of stored samples. Finally we conclude with a few recommendations to help address the issues of informed decision-making.

## **I. Explicit or presumed consent to newborn screening for treatable disorders**

Classical newborn screening for routine/treatable disorders has been and is being carried out, in most parts of the world without explicit parental consent.(See TABLE 1) In newborn screening programs, consent is presumed and justified on the basis that when a disease is treatable, a newborn has a right to be screened and to be treated.<sup>3</sup>

What constitutes a treatable disorder? In general, newborn screening is recommended for a disorder where: 1) there is considered to be a direct benefit to the newborn from early diagnosis; 2) the benefit is reasonably balanced against financial and other costs; 3) there is a reliable test suitable for neonatal screening; and 4) there is a satisfactory system in operation to deal with diagnostic testing, counselling, treatment and follow-up of identified babies.<sup>4</sup> This is best exemplified by a disorder like PKU, where early detection by screening and treatment has been very effective in preventing neurological abnormalities.

Most newborn screening programs are part of mandated paediatric norms and are considered part of routine care, thus eliminating the requirement for a separate written consent. The rationale for this practice stems from the belief that the minimal risk of adverse medical effects associated with the collection of a few drops of blood versus the significant medical consequences of a missed case due to parental refusal can justify the absence of formal informed consent. Also consent is presumed because it is defined in term of the best interest of the child and of society. Indeed, the New York Task Force states that “that the autonomy of the parent to make health care decisions for their minor children must give way to the state’s role in protecting children from harm”<sup>5</sup>. In 1998, the World Health Organisation (WHO) declared that newborn screening should be mandatory and free of charge if early diagnosis and treatment will benefit the newborn.<sup>6</sup> Although the WHO position does not outright state that parents should not be able to refuse interventions that are beneficial to their children, some have interpreted it to mean exactly that.<sup>7</sup>

Those who support a choice approach for screening argue that obtaining consent will not compromise uptake for screening, that parents have a right not to know, and maintain the general principle of consent for any intervention.<sup>8</sup>

Though most newborn screening programs screen for treatable disorders without explicit consent, there is general agreement amongst guidelines that parents should be adequately informed about newborn screening. The International Society for Neonatal Screening (ISNS), for instance, underlines the importance of informing parents and of public education about newborn screening programmes.<sup>9</sup> It advocates that the public be kept well informed about screening programmes and that as far as possible, written information be provided to parents before screening. The Human Genetics Society of Australasia adds that written information and the opportunity for

discussion must be provided to parents before testing and that health professionals should be provided with comprehensive guidelines describing all aspects of the screening program including correct sample collection procedure.<sup>10</sup>

Both the ISNS and the HGSA make no specific mention of written consent, they do however underline the need for public education and mechanisms that appropriately address a parent's option to decline testing and that they be kept informed of the possible consequences should they chose to refuse NBS.<sup>11</sup>

The American states of Wyoming and Maryland have NBS programs, which utilize explicit written consent<sup>12</sup>, notwithstanding the Association of Public Health Laboratories' (APHL) statement to the effect that explicit parental consent is not necessary for mandated public health newborn screening<sup>13</sup>. In contrast, New York, which used to seek informed consent to test newborn blood specimens for HIV, now has added HIV to the mandatory program through legislative and regulatory amendments.<sup>14</sup>

The question arises whether the "mandatory offering" interpretation is inconsistent with the Human Genetics Society of Australasia (HGSA) position stating that participation in a newborn screening program should not be mandatory<sup>15</sup>, as well as the Institute of Medicine's recommendation promoting voluntariness of newborn screening programs<sup>16</sup>. Indeed mandatory offering provides for an opt-out but presumes consent.

The UK Newborn Screening Programme Centre is examining a different solution to written consent for screening. While recognising the importance of accurate information as essential to enable parental decision to screening, the recently proposed standards and policies reject the written consent model, opting instead for an 'informed choice' approach with recording of acceptance or decline to NBS in the mother's maternity record.<sup>17</sup>

In short, the UK<sup>18</sup>, Canada<sup>19</sup>, and the USA (See TABLE 2) have opted for the presumed consent model where parents are invited to sign admission papers and consent to NBS as part of routine paediatric procedures when they arrive at the hospital at the time of delivery.

When screening for medical conditions which when detected in the newborn period can be treated immediately, most guidelines concur that presumed consent is appropriate<sup>20</sup>. However, the policies indicate that professionals have a responsibility to inform and provide ongoing support and that explicit written consent<sup>21</sup> and/or more detailed information<sup>22</sup> is a goal to strive for.

## **II. Explicit or presumed consent to screening for a wider range of treatable and non-treatable disorders**

With the introduction of tandem mass spectrometry (MS/MS) for population based newborn screening, healthcare providers are now able to detect an increased number of disorders in a single process by using the same dried blood spot specimen collected through routine newborn screening. Unfortunately, effective treatments are not available for all diseases that can be identified by MS/MS.

NBS for new disorders, raises novel issues about consent. First a dilemma has emerged on the definition of “benefit”. Benefit has meant direct medical benefit, where early diagnosis and medical intervention improves outcomes<sup>23</sup>. Others have suggested that benefit is for the family as well as for the baby.<sup>24</sup> For example, parental knowledge has been alleged to lessen self-blame, prevent weeks or months of searching for a diagnosis and has allowed parents to take advantage of new and rapidly evolving treatment.<sup>25</sup> Early diagnosis, at birth, has also been suggested to avoid trauma and expense to the family, and allow options for family planning to be considered before other affected siblings are born.<sup>26</sup> Similarly, it has been argued that knowing an infant is a carrier, has the potential for stigmatization and alters self-perception<sup>27</sup>.

Given the absence of direct medical benefit and the deviation from the classical criteria of newborn screening, most guidelines concur that NBS programs, which elect to include new disorders, such as cystic fibrosis, that are treatable but where immediacy of detection is not an issue or conditions that are untreatable, should require explicit parental consent in the spirit of informed participation in medical procedures of limited or unproven benefit. This having been said, seeking explicit consent for the screening of untreatable disorders can have undesired outcomes. Indeed, preliminary studies from a pilot project in Scotland appear to indicate, that seeking written consent for the screening of both treatable and untreatable (Cystic Fibrosis in the case of Scotland) diseases in a single process leads to more parents refusing screening altogether.<sup>28</sup> Thus it seems logical that consent be presumed for screening of treatable disorders and explicit in the case of untreatable ones.

### **III. Explicit or presumed consent to store newborn bloodspots**

Once the newborn screening programme is completed, residual dried blood spots (DBS) are either discarded or stored in public health laboratories. Storing DBS for up to one year is indispensable for repeat-testing, normal laboratory audit and quality assurance purposes (QA). However, because bloodspots constitute valuable DNA specimens, they may also be of benefit for familial, forensic and research purposes. Indeed, stored samples could be used to conduct pre-symptomatic and susceptibility genetic testing or paternity testing, thus revealing information about children, which is not necessarily in their best interest and usually contrary to most genetic predictive testing guidelines.<sup>29</sup> The existence of these collections raises ethical concerns about access by insurers, employers, families, law enforcement agencies and others. There is currently apprehension regarding the possible misuse of stored samples as well as intrusion into privacy.

Whatever the objectives for retaining newborn blood spots, it is recommended that public health newborn screening programs thoroughly evaluate and define the rationale for storage and analysis beyond that necessary for confirmatory testing and quality control<sup>30</sup>.

A central ethical question regarding storage of newborn bloodspots is whether parental consent is needed for storage beyond the time needed for quality assurance and for subsequent research use of the samples. Based on the principles of autonomy and respect for privacy, consent is usually required when human biological materials are collected and stored for future use. In its 2003 *International Declaration on Human Genetic Data*<sup>31</sup>, UNESCO states that prior, free, informed and express consent should be obtained for the collection of human genetic data, human proteomic data or biological samples, whether through invasive or non-invasive procedures, and

for their subsequent processing, use and storage unless prescribed by domestic law consistent with the international law of human rights. It is not clear whether this declaration can be applied to the collection of newborn dried bloodspots because there is no mention of samples collected by newborn screening programs for initial diagnosis, not essentially related to the study of genetic characteristics. (See TABLE 3)

While most policy statements that address storage of newborn dried blood spots strongly agree that parents and the public in general should be informed about storage policies and practices<sup>32</sup>, most do not require written consent for storage. The American Academy of Pediatrics Newborn Screening Taskforce has recommended developing model consent forms and informational materials for parental permission for retention of DBS.<sup>33</sup> The Association française pour le dépistage et la prévention des handicaps de l'enfant (AFDPHE), for its part, has also recommended that written consent for storage be obtained when bloodspots are planned to be used for purposes other than those for which they were initially obtained.<sup>34</sup>

Another solution is the Danish approach with regard to consent to storage, where parents are informed and given the opportunity to opt-out from having their child's bloodspot stored at the time of screening.<sup>35</sup> In other words, informed refusal is generally favoured over informed consent.<sup>36</sup>

There are currently no policies governing the storage and use of residual blood samples in Canada, and a recently pilot survey demonstrates that parents are generally not informed nor are they asked to consent to storage.<sup>37</sup>

In brief, when samples are retained beyond the one-year period required for quality assurance, informing parents at the time of collection about length, purposes and methods of storage as well the confidentiality of stored samples is considered to be good practice.

#### **IV – Explicit or presumed consent to use dried bloodspot cards**

Using residual bloodspots for purposes other than those for which they were obtained can become more or less problematic depending on the use. The research use of bloodspots raises concern about confidentiality and about group harms, especially concerns about stigmatization and discrimination against those who test positive for example if the research involves genes associated with behaviour. Employers, health insurers, schools, or other institutions might want to access individual health records and dried blood samples. Seeing as a newborn's bloodspot is a valuable source of genetic information, it has been recommended that screening programs ensure that DBS and associated information are stored in a secure manner to prevent unauthorised access and secondary use of identified or identifiable samples.<sup>38</sup> While using anonymized DBS for epidemiological or public health research does not threaten patient privacy, research with coded samples create the possibility of re-contacting patients for follow-up studies, and so ethical challenges arise.

Since the primary purpose of newborn bloodspot collection is for diagnosis and confirmatory testing, most guidelines maintain that further uses of stored samples, for purposes other than screening program audit/epidemiological use, require either written permission from the

individual, the parents or guardian, a legally binding directive, or appropriate ethics committee approval for research studies.<sup>39</sup> There is also general agreement that anonymized samples are important for health surveillance studies. Consequently, they should be made available for research without parental consent and because they represent less of a threat to patient privacy than coded or identified samples.<sup>40</sup>

The prospect of storing newborn bloodspots raises questions of when and how to inform parents. Some guidelines have stated that information about additional uses of newborn samples should be conveyed to parents and that an up-front mechanism of informed consent, at the time of heel prick collection, would be a logical way of initiating the process.<sup>41</sup> Indeed, this process seems acceptable if a research project is foreseen before sample collection. But what about when consent to storage and future research use is required before the actual research protocol has been elaborated? The WHO<sup>42</sup> as well as the Danish Neonatal Screening Programme<sup>43</sup> have recommended blanket consent for all residual bloodspot use. Whilst constituting an efficient and economical method, this approach can be criticized based on the fact that it does not allow parents to fully understand the exact future purposes of stored samples and prevents them from providing a truly informed consent. It has been suggested that specific consent, through re-contact, should be obtained for each research study that requires coded or identified samples.<sup>44</sup> (See TABLE 3)

## **Conclusion and Recommendations**

Policies in favour of mandatory screening (with presumed consent) for treatable disorders, while appearing to override parental consent, are implemented in the child's best interest. The majority of policies and professional groups agree that the benefits of NBS are so great for the newborn and in the child's best interest that the screening should be universal and mandatory. While mandatory offering of newborn screening is a public health responsibility as it is the child's best interest to be screened and found, it is an area where the rights of the child and the right of the parents may appear to be in conflict. For example this conflict is reflected more and more in policies and the literature with double messages including mandatory and voluntary.

Generally, consent is presumed for treatable disorders and explicit for additional testing for new disorders and for storage. (See TABLE 4) Defining what are treatable and non treatable conditions is important because some policies have waived the need for informed parental consent stating that mandatory programs do not require obtaining parental authorization, whether explicit (written or verbal) or implicit (informed refusal).<sup>45</sup>

Where explicit consent is not a requirement, such as in the case of screening for treatable disorders, it is suggested that presumed consent should not lead to uninformed parents nor should it create confusion in the health service community as to who is responsible for providing information prior to screening. Presumed consent to routine newborn screening does not obviate the possibility of parental refusal for a specific clinical diagnosis. However once the at-risk infant is identified, it is very difficult for parents to support the authority of presumed consent because current experience demonstrates that information about parental right to refuse screening, storage and future use of coded samples is seldom provided to parents when programs are "mandated" and where written consent is not a requirement.<sup>46</sup> Nevertheless, from a child rights point of view,



presumed consent to screening is defensible because it is every child's right to be screened and found when treatments exist and are accessible.

There is no doubt that public health authorities are required to provide the best possible health services to the public and specifically to newborns where early intervention can improve the outcome. However, NBS programmes are increasingly facing new challenges which raise questions about consent and how best to inform parents. Discussing the availability and the utility of tests for treatable or non-treatable conditions with parents might prove to be worthwhile.

Providing prior written information gives parents the opportunity to learn about disorders included in the universal panel; additional disorders; disorders which are not included but for which tests do exist; storage and possible future use of stored DBS; and allows them to discuss any fears or apprehensions with their medical service provider beforehand. Some of the information may be best delivered by the obstetrician or mid-wife. With adequate training, these medical professionals will be best suited for the task as they are the ones who interact most with parents before and after birth. It is our opinion that obstetric and gynaecological societies should adopt guiding principles acknowledging the OB/GYN's privileged position and relationship with expecting parents and the importance of their role in the newborn screening process.

Indeed, as seen in the policy review, there are different ways in which information can be provided prior to newborn screening for treatable disorders, for new disorders, for storage and for the future use of stored samples. Overall there is a need for public and professional education and a better-informed public. Ideally, an informed choice model should be privileged, where information about screening and storage is conveyed to parents prior to the collection of the sample, well before the birth of the child and where presumed consent to screening is separate from explicit consent to screening for non-treatable disorders and from issues related to storage and future use of samples. This would allow the newborn to benefit from screening for treatable disorders even if parents refuse screening for conditions that are not treatable or for storage.

In all policies regarding newborn screening and DBS storage, the interest of the newborn should be paramount.

## Appendix

**Table 1. *The Ten Principles of Neonatal Screening: Consensus statement from the 1989 workshop on "Genetic Screening: From Newborns to DNA typing"***

1. Newborn genetic screening is a medical act in the context of preventive medicine.
2. Newborn genetic screening should lead to medical intervention for the benefit of the newborn.
3. Newborn genetic screening should be universally and equitably available in the population(s) to be screened.
4. Newborn genetic screening programs should inform the parent(s) and the general public of their goals and objectives, of the disorders being screened for and the tests being performed.
5. Newborn genetic screening programs should use testing procedures whose sensitivity, specificity and acceptability are known from pilot studies conducted in the population(s) to be tested.
6. Newborn genetic screening programs should inform the parent(s) of the significance of the results of screening tests and should confirm that these results are validated by the standard diagnostic tests.
7. Newborn genetic screening programs should integrate follow-up procedures into their system since it provides and validates the benefit to the newborn. Such follow-up should include referral to effective medical intervention and to other support services and resources.
8. Newborn genetic screening programs could permit the use of their blood specimens for anonymous research or surveillance provided that the following conditions are met:
  - i. Where the newborn screening programs are mandatory or operate as public health programs with an informed refusal approach:
    - a. The public should be informed that such studies are being conducted.
    - b. In surveillance for diseases where there is no effective intervention or benefit to the newborn, such surveillance should not only be anonymous but also unlinked as to avoid possible individual or population stigmatization or discrimination.
    - c. The newborn genetic screening program itself should be responsible for assuring the unlinking of the nominative data and the complete anonymity of the sample of specimens used.
    - d. Since surveillance studies benefit society, voluntary and free access to individual testing must be made available.
  - ii. When the newborn genetic screening program requires fully informed individualized consent for screening, participation in anonymous research or surveillance studies requires a specific individual authorization.
9. Newborn genetic screening programs should maintain the confidentiality of nominative information and samples unless proper authorization for release has been obtained.
10. Newborn genetic screening programs could use DNA typing as a testing procedure when genetic heterogeneity in population(s) becomes technically interpretable.

**SOURCE:** Bartha M. Knoppers & Claude M. Laberge, eds., *Genetic Screening: from newborns to DNA typing*, (Amsterdam: Excerpta Medica, 1990).





**Table 4. Consent for newborn screening: treatable vs. untreatable**

**1) When must information about newborn screening be provided to parents?**

Before the birth of a child

**2) What type of information should be provided?**

Information about treatable and untreatable disorders, opting out options, interpretation of test results, incidental findings, conditions that can be tested for but which are not included in the screening panel, etc.

**3) What type of consent is required?**

Screening for **treatable** disorders can require:

- No consent (Presumed)
- Written refusal
- Written consent

Screening for **untreatable** disorders requires:

- Written consent at all times

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<sup>1</sup> 48 states in the US have implemented universal sickle cell screening. National Newborn Screening and Genetics Resource Center, *U.S. National Screening Status Report*, Austin, Texas, July 5<sup>th</sup>, 2004. <http://genes-r-us.uthscsa.edu/nbsdisorders.pdf> (date accessed: August 3, 2004). England has started phased implementation of universal sickle cell screening. NHS Sickle Cell and Thalassaemia Screening Programme, *Policy for Newborn Screening*, July 2004. <http://www-phm.umds.ac.uk/haemscreening/Documents/NewbornScreeningPolicy.pdf> (date accessed: August 4, 2004).

<sup>2</sup> Bridget Wilcken, "Ethical issues in newborn screening and the impact of new technologies" (2003) 162 *Eur. J. Pediatr.* S62.

<sup>3</sup> Bartha M. Knoppers, "Newborn Screening and Informed Consent" in Jean-Pierre Farriaux, Jean-Louis Dhondt, eds., *New Horizons in Neonatal Screening* (Amsterdam: Excerpta Medica, 1994) 15.

<sup>4</sup> International Society for Neonatal Screening, *ISNS General Guidelines for Neonatal Screening*, 2002. <http://www.isns-neoscreening.org/FactSheets/Guidelies.htm> (date accessed: July 15th, 2004). World Health Organization (WHO), *Proposed International Guidelines on Ethical Issues in Medical Genetic Services*, Geneva, December 15-16 1997. [http://whqlibdoc.who.int/hq/1998/WHO\\_HGN\\_GL\\_ETH\\_98.1.pdf](http://whqlibdoc.who.int/hq/1998/WHO_HGN_GL_ETH_98.1.pdf)

<sup>5</sup> The New York State Task Force on Life and the Law, *Genetic Testing and Screening in the Age of Genomic Medicine*, New York, November 1, 2000. <http://www.health.state.ny.us/nysdoh/taskfcr/screening.htm> (date accessed: July 26<sup>th</sup>, 2004).

<sup>6</sup> World Health Organization (WHO), *Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetic Services*, Geneva, December 15 and 16, 1997, [http://whqlibdoc.who.int/hq/1998/WHO\\_HGN\\_GL\\_ETH\\_98.1.pdf](http://whqlibdoc.who.int/hq/1998/WHO_HGN_GL_ETH_98.1.pdf) (date accessed: July 30, 2004).

<sup>7</sup> Bridget Wilcken, "Ethical issues in newborn screening and the impact of new technologies" (2003) 162 *Eur. J. Pediatr.* S62.

<sup>8</sup> Evelyn P. Parsons & Don M. Bradley, "Newborn screening programs" in *Nature Encyclopedia of the Human Genome* (Macmillan Publishers Ltd, Nature Publishing Group, 2003) 336.

<sup>9</sup> International Society for Neonatal Screening, *ISNS General Guidelines for Neonatal Screening*, 2002. <http://www.isns-neoscreening.org/FactSheets/Guidelies.htm> (date accessed: July 15th, 2004).

<sup>10</sup> Human Genetics Society of Australasia, *Newborn Blood-Spot Screening*, Australia, March 18, 2004. <http://www.hgsa.com.au/Word/HGSApolicyStatementNewbornScreening0204-18.03.04.doc> (date accessed: July 20<sup>th</sup>, 2004).

<sup>11</sup> International Society for Neonatal Screening, *ISNS General Guidelines for Neonatal Screening*, 2002. <http://www.isns-neoscreening.org/FactSheets/Guidelies.htm> (date accessed: July 15th, 2004). Human Genetics Society of Australasia, *Newborn Blood-Spot Screening*, Australia, March 18, 2004. <http://www.hgsa.com.au/Word/HGSApolicyStatementNewbornScreening0204-18.03.04.doc> (date accessed: July 20<sup>th</sup>, 2004).

<sup>12</sup> American College of Obstetricians and Gynecologists, "Committee Opinion No. 287 on Newborn Screening" (2003) 102(4) *Obstet. Gynecol.* 887.

<sup>13</sup> Association of Public Health Laboratories (APHL), *Parental Consent in Public Health Newborn Screening Programs*, Washington, June 5, 2002, [http://www.aphl.org/Newborn\\_Screening\\_Genetics/aphlconsf1.pdf](http://www.aphl.org/Newborn_Screening_Genetics/aphlconsf1.pdf) (date accessed : October 19, 2003).

<sup>14</sup> Association of Public Health Laboratories (APHL), *Parental Consent in Public Health Newborn Screening Programs*, Washington, June 5, 2002, [http://www.aphl.org/Newborn\\_Screening\\_Genetics/aphlconsf1.pdf](http://www.aphl.org/Newborn_Screening_Genetics/aphlconsf1.pdf) (date accessed : October 19, 2003); National Newborn Screening and Genetics Resource Center, *U.S. National Screening Status Report*, Austin, Texas, July 5<sup>th</sup>, 2004. <http://genes-r-us.uthscsa.edu/nbsdisorders.pdf> (date accessed: August 3, 2004).

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- <sup>15</sup> Human Genetics Society of Australasia, *Newborn Blood-Spot Screening*, Australia, March 18, 2004. <http://www.hgsa.com.au/Word/HGSApolicyStatementNewbornScreening0204-18.03.04.doc> (date accessed: July 20<sup>th</sup>, 2004).
- <sup>16</sup> Institute of Medicine (IOM), *Assessing Genetic Risks –Implications for Health and Social Policy*, (Washington DC: National Academy Press, 1994) at 21-22.
- <sup>17</sup> UK Newborn Screening Programme Centre, *Proposed standards and policies for newborn blood spot screening – an integrated consultation*, London, June 2004. [http://www.ich.ucl.ac.uk/newborn/download/proposed\\_standards0604.pdf](http://www.ich.ucl.ac.uk/newborn/download/proposed_standards0604.pdf) (date accessed: July 26, 2004). The draft standards have not been adopted as of yet and face some obstacles particularly related to possibility of opting out from screening for one or more conditions whilst choosing the rest. It appears that if consent were offered on an individual condition basis, it could potentially increase laboratory errors, as current technology is not sophisticated enough to accommodate different work lists when screening for PKU and CHT. As stated by the UKNSPC, “there is clearly a tension between what may be an ideal process – allowing full choice for each condition – and the inability of current technology and resources to support this”.
- <sup>18</sup> For the UK, see S. Shenoy, C. Archdeacon, S. Kotecha & A.C. Elias-Jones, “Current practice for obtaining consent in UK neonatal units” (2003) 188 Bull. Med. Ethics 17.
- <sup>19</sup> Sheila Wilderman & Jocelyn Downie, “Screening Newborns for Genetic and Metabolic Disorders – The Issue of Parental Choice” (2001) 2(3) Isuma 1.
- <sup>20</sup> Association of Women’s Health, Obstetric and Neonatal Nurses (AWHONN), *National Standards for Newborn Screenings*, Washington, June 1, 2002. <http://www.awhonn.org/awhonn/?pg=875-4810-5600> (date accessed: July 15<sup>th</sup>, 2004) ; Comité d’éthique de l’Association française pour le dépistage et la prévention des handicaps de l’enfant (AFDPHE), “Les prélèvements de sang sur papier pour le dépistage néonatal. Recommandations pour leur collecte, leur traitement et leur conservation” (1995) 2 Arch. Pédiatr. 3.
- <sup>21</sup> Association of Public Health Laboratories (APHL), *Parental Consent in Public Health Newborn Screening Programs*, Washington, June 5, 2002, [http://www.aphl.org/Newborn\\_Screening\\_Genetics/aphlconsf1.pdf](http://www.aphl.org/Newborn_Screening_Genetics/aphlconsf1.pdf) (date accessed : October 19, 2003).
- <sup>22</sup> Human Genetics Society of Australasia, *Newborn Blood-Spot Screening*, Australia, March 18, 2004. <http://www.hgsa.com.au/Word/HGSApolicyStatementNewbornScreening0204-18.03.04.doc> (date accessed: July 20<sup>th</sup>, 2004). Institute of Medicine. *Assessing Genetic Risks: Implications for Health and Social Policy*. Washington, DC: National Academy Press; 1994.
- <sup>23</sup> World Health Organization (WHO), *Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetic Services*, Geneva, December 15 and 16, 1997, [http://whqlibdoc.who.int/hq/1998/WHO\\_HGN\\_GL\\_ETH\\_98.1.pdf](http://whqlibdoc.who.int/hq/1998/WHO_HGN_GL_ETH_98.1.pdf) (date accessed: May 7, 2002).
- <sup>24</sup> Human Genetics Society of Australasia, *Newborn Blood-Spot Screening*, Australia, March 18, 2004. <http://www.hgsa.com.au/Word/HGSApolicyStatementNewbornScreening0204-18.03.04.doc> (date accessed: July 20<sup>th</sup>, 2004); American College of Medical Genetics (ACMG), American Society of Human Genetics (ASHG) - Test and Technology Transfer Committee Working Group, “Tandem Mass Spectrometry in Newborn Screening”, (2001) 2:4 *Genetics in Medicine*, p. 267-269, <http://www.faseb.org/genetics/ashg/pubs/policy/pol-43.pdf> (date accessed: July 21, 2004).
- <sup>25</sup> World Health Organization, *Review of Ethical Issues in Medical Genetics*, Report of Consultants to WHO – Professors D.C. Wertz, J.C. Fletcher and K. Berg, Geneva, 2003. [http://www.who.int/genomics/publications/en/ethical\\_issuesin\\_medgenetics%20report.pdf](http://www.who.int/genomics/publications/en/ethical_issuesin_medgenetics%20report.pdf) (date accessed: July 22, 2004).
- <sup>26</sup> World Health Organization, *Review of Ethical Issues in Medical Genetics*, Report of Consultants to WHO – Professors D.C. Wertz, J.C. Fletcher and K. Berg, Geneva, 2003. [http://www.who.int/genomics/publications/en/ethical\\_issuesin\\_medgenetics%20report.pdf](http://www.who.int/genomics/publications/en/ethical_issuesin_medgenetics%20report.pdf) (date accessed: July 22, 2004).

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<sup>27</sup> Lidewij Henneman & Myriam Welkenhuysen, “Genetic Carrier Testing” in *Nature Encyclopedia of the Human Genome* (Macmillan Publishers Ltd, Nature Publishing Group, 2003) 731.

<sup>28</sup> A.J. Brown, J.M. Mackenzie, M. Fitch & A. Estell, *Impact of obtaining signed consent for newborn screening tests in Scotland*. (Scotland: NHS Greater Glasgow, 2004).

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