UK National Screening Committee

Consultation on Screening for cfDNA in pregnancy

Submission on behalf of the Christian Medical Fellowship

The Christian Medical Fellowship (CMF) is an interdenominational Christian organisation with about 4,500 British doctors as members, practising in all branches of the profession. Through the International Christian Medical and Dental Association we are linked with like-minded colleagues in over 100 other countries.

CMF regularly makes submissions on ethical and professional matters to Government committees and official bodies. One of CMF's aims is 'to promote Christian values, especially in bioethics and healthcare among doctors and medical students, in the church and in society'. Many of our members are directly involved 'on the front line' in diagnosing, treating and caring for pregnant women, as well as people with disabilities.

As a Christian organisation, we encourage our members to be advocates for those who are weak, sick, marginalised and disabled and to seek to love and care for them to the utmost of their abilities.

Reflections on UK NSC recommendation

CMF is supportive of reducing the unintended adverse consequences of screening procedures but we do not welcome the proposal to add cfDNA analysis as a contingent test for those in whom initial screening has indicated a >1:150 risk, for the reasons outlined in this submission.

This development purports to reduce the overall number of referrals for invasive diagnostic procedures (amniocentesis or chorionic villous sampling) and thereby reduce the number of miscarriages that arise as the unintended result of those procedures, of babies that are almost always healthy.

However a concomitant effect would be an increase in the number of babies with Down Syndrome (DS) which would be lost.

The Convention of the Rights of Persons with Disabilities (CRPD)

The Convention of the Rights of Persons with Disabilities (CRPD) is relevant here. The United Nations (UN) General Assembly adopted the CRPD[1], together with its Optional Protocol[2], by consensus on December 13, 2006[3]. The Convention provides, in the form of a legally binding core human rights convention, a disability-specific framework for the civil, political, economic, social and cultural rights of persons with disabilities. The CRPD is a legally binding core human rights convention, which has been signed by the United Kingdom in 2007 and ratified in 2009.

The general principles in the CRPD that apply to health policies include, but are not limited to:

- Respect for inherent dignity;

- Non-discrimination;
- Full and effective participation and inclusion in society;
- Respect for difference and acceptance of persons with disabilities as part of human diversity and humanity;

Policies that potentially offend one or more of the general principles of human rights law must be treated with extreme caution and heightened scrutiny and should be subject to immediate review.

Major discrepancies in figures

There is also a major discrepancy in the figures between the two projections from the Systematic Review and the RAPID study which seriously undermines confidence in the data on which the UK NSC recommendation is based.

According to the Systematic Review's projected figures, for DS there would be 20 more cases detected per year, if the cfDNA test was to be offered on a contingent basis. Assuming a 90% abortion rate among these additional cases[4], it would in theory imply 18 additional abortions for DS per year. Their figures also suggest that there would be a 'saving' of 43 normal pregnancies per year, as a result of many fewer invasive procedures being necessary, and therefore proportionately fewer miscarriages. On these projections, the result would be an overall *decrease* of 25 per year in the number of fetal lives lost.

However, the RAPID study suggests that the overall number of abortions for babies with trisomy is actually likely to be much higher as a consequence of introducing the new test. The RAPID study projections suggest that there would be 102 more T21 (Down Syndrome) pregnancies detected per year, with the 1:150 threshold protocol in place. Assuming the same 90% subsequent abortion rate, this would translate into 92 'additional' abortions per year. Their figures also predict a reduction in the rate of miscarriage of 25 per year. An overall *increase* in fetal loss, therefore, of 66 per year.

Further concerns

However, we also have concerns of a more fundamental nature, that are to do with the assumptions that lie behind current screening policy, and the lack of opportunity for reflection and means of support for those women (and their families) learning that their fetus is affected by trisomy. We explain and amplify these reservations below and make a number of recommendations in conclusion.

Other reflections on the NSC documents:

- The greater accuracy of the cfDNA test (over the existing combined test) will surely lead to public demand that it be made available as part of routine initial antenatal screening, particularly given the trend towards increasing average maternal age, when risks are higher.
- The NSC summary estimates that using the new test as part of *primary* screening would detect 289 more trisomy babies annually. This figure appears at odds with the more general comment (page 4, para 2) suggesting that at lower thresholds the number of detections does not increase greatly. The impression that the threshold has been set so as to be cost-neutral is difficult to avoid.

- As awareness of the test increases, and its cost comes down, then many pregnant women will access the test privately if it is not routinely available on the NHS. They will receive results outlining all manner of variable predictive risks faced by their babies, but will not have the context in which to discuss the relevance of those results. This will increase anxiety and make abortion a more likely choice, sometimes without evidence of trisomy. What measures are in place to educate women on not making a decision to abort straight after cfDNA testing? [5]
- The same technology that allows cfDNA testing to detect trisomy also detects gender[6] and will in time permit the detection of a wide range of genetic 'conditions' and predispositions. A widespread use of cfDNA testing to analyse more and more genetic features up to the entire genome would mean the complexity of data would lead to a significant increase of false-positives, requiring a confirmation by invasive tests of abnormalities whose relevance is not known at all. This unknown might lead the parents not to take any risk, with the resulting paradox: the number of invasive diagnostics would rise because of the use of the new test that should precisely be diminishing the use of invasive diagnostics. With that increase in the number of invasive tests would come an increase in the number of unintended miscarriages of often normal futuses. Further, what safeguards will be in place to prevent abortion on the grounds of gender where specific sex-linked diseases are NOT implicated? Where are lines to be drawn? Which conditions will be included in screening protocols? What data protection policies will be in place to prevent 'genetic disadvantage' leading to discrimination in the job market, insurance provision and the like? Until the ethical implications of the more widespread uses of the technique have been thoroughly considered, we consider its introduction to be irresponsible and unethical.
- On what logical grounds can permission to abort up to birth, for 'serious' genetic disability, be prevented from extension to infanticide for the same conditions? If it is legal and ethical to end such a life up to birth, why is it not so a day later? If it is not legal and ethical after birth, why is it so a day earlier?
- The language of 'screening' suggests that tests are intended to uncover conditions for which appropriate therapy will then be made available. In the case of Down Syndrome, the only 'therapy' on offer is to terminate the life of the fetus. The notion of 'screening in order to eliminate' has sinister undertones. (This is explored further in the next section.)

Concerns about assumptions

The World Health Organisation screening guidelines, often referred to as Wilson's Criteria, were published in 1968, but are still applicable today[7].

In the case of prenatal screening for trisomies, the second of Wilson's ten criteria, namely that 'there should be a treatment for the condition', does not apply. Trisomy screening is carried out not to identify individuals with special needs in order that they may be more effectively treated, but rather with the express purpose of eliminating them from the population. This type of screening offers no benefit to the fetus being screened and also results in collateral damage in that many unaffected fetuses also die in the process.

We would submit that this is contrary to the Hippocratic Oath, the Declaration of Geneva and to the general strategy of medicine. But we would also take issue with the underlying assumption that such screening benefits the health of pregnant women and their families (see below).

It is assumed that a woman who discovers that she is carrying a fetus affected by Down Syndrome will want to terminate her pregnancy. The UK Abortion Law, under Ground E, permits an abortion to take place up to birth if 'there is a substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously handicapped'. The terms 'substantial' and 'serious handicap' are not defined and have been applied in practice to cover many conditions that are compatible with life outside the womb. Down Syndrome is one such condition, and those with it may live for 50 or 60 years, depending on co-morbidities, finding fulfilment and contributing greatly to family and community life. Research published in the American Journal of Medical Genetics[8] found that nearly 99% of people with Down Syndrome are happy with their lives, more than three-quarters of parents of a child with Down Syndrome had a more positive outlook on life and almost 90% of siblings said they considered themselves better people because of their family member with Down Syndrome.

To assume Ground E provision should automatically apply to Down Syndrome is to stretch the law to the point of completely misshaping it. In our opinion, DS should not be classed as a 'serious handicap' and screening for it should only be offered to mothers in order to prepare them and their families better to be joined by a child with special needs. Further, to give DS Ground E status implies that the life of a person with Down Syndrome would have been better terminated before birth – a life not worth living. Ultimately, it fosters in society the notion that only the (genetically) perfect are acceptable and that it is socially desirable to prevent people with disabilities from being born[9] – an insidious and discriminatory eugenics practised for its perceived social or economic benefit to individuals other than those directly affected. It attempts to control human reproduction in order to 'improve' the genetic characteristics of the next generation.

These concerns are echoed by the International Bioethics Committee of UNESCO that comments: 'The widespread use of genetic screening and in particular of NIPT may foster a culture of 'perfectionism' or 'zero defect' and even renew some 'eugenic trends', with the consequence that it could become more and more difficult to accept imperfection and disability as a part of normal human life and a component of the diversity we are all called on to acknowledge and respect. The anxiogenic effect is also to be considered. The right of an individual to make autonomous choices is to be made consistent with the right not to be subjected to discrimination or stigmatization based on genetic characteristics and the duty to respect every human being in her or his uniqueness[10].

Without question, caring for a child with Down Syndrome brings added pressures to parents and siblings but also particular and rewarding joys. The possibility that raising a child with Down Syndrome could be a positive, life-affirming experience is nowhere mentioned in the NSC review. We would like to see, as a statutory requirement, provision for informed 'reflection before decision' for every woman receiving the news that she is carrying a fetus affected by Down Syndrome (and other trisomies). This should include the opportunity wherever possible to talk to someone with that diagnosis or a similar condition, a family who has a child with that diagnosis or a similar condition[11]. At the very least, printed information written by those who have the same disabilities, and their families, should be made available. Health professionals should signpost families receiving a

diagnosis of disability to information leaflets covering all their options, to telephone and online helplines manned by trained professional counsellors, and to local and national support groups for those with specific conditions. Following her decision, and regardless of what choice the woman and her family may make, ongoing support must be part of that provision.

Current practices leave little room for such reflection. The assumptions behind current practice mean that opportunity for such reflection and support is bypassed in the rush to terminate. Subtle or direct pressure may be placed on parents who decide not to abort[12]. Parents can be made to feel that to bring into the world a child with known disability is somehow irresponsible and blameworthy[13]. **We should not underestimate the coercive power present in a system where a conveyor belt of expectation moves in the direction of choosing not to give birth to children with special needs who are regarded either as a burden or as, in some sense, not fully human[14].**

Another assumption is the belief that parents and families will be damaged by having a disabled child, and that this damage can be limited through abortion. This is not supported by research; indeed, the opposite may be the case. Psychological trauma following abortion appears to be least when the pregnancy is early, when there is no maternal ambiguity about the decision and when the child was most definitely not wanted.

Abortions for congenital abnormality usually occur in pregnancies that are both late and wanted. It is not surprising therefore that psychological morbidity is considerable. Such morbidity following termination of pregnancy for fetal disability has been shown to be both prevalent and persistent[15] and associated with long-lasting consequences for a substantial number of women[16]. Rather than leading to psychological well-being, termination of pregnancy for fetal disability can be an emotionally traumatic major life event which leads to severe post-traumatic stress response and intense grief reactions that are still detectable some years later[17].

People do not easily 'get over it' although proper support during the loss can lessen psychological morbidity[18]. In fact women who terminate pregnancies for fetal anomalies experience grief as intense as those who experience spontaneous perinatal loss with approximately a fifth developing major depression and/or requiring psychiatric intervention[19]. Their families are also not immune with even very young children and those sheltered from knowledge of the event showing reactions to their parents' distress and maternal absence[20].

Some may experience an acute grief reaction or be plagued by guilt and fear that can precipitate marital breakdown. Additionally, there is a risk that through striving to eradicate congenital disability, a community risks promoting a culture of perfectionism that may have discriminatory effects on disabled people[21].

By contrast, current data on children and families affected by disabilities indicate that disability does not preclude a satisfying life. Many problems attributed to the existence of a disability actually stem from inadequate social arrangements that public health professionals should work to change[22]. This, along with the psychological morbidity often accompanying abortion for fetal disability has led many to conclude that abortion for even severe fetal disability, as well as taking the life of a disabled person, is also worse for the parents and families concerned.

Janet Goodall, a paediatrician with a lifetime's experience in caring for severely disabled children, describes the 'pearl effect'.

'In a culture that views success and failure in materialistic terms, many perceive disabled children as an extra burden. But paradoxically, divorce rates and unhappiness are no more common in the families of disabled children than in those with healthy children. Like the grit in the oyster that causes a pearl to form, caring for a child with special needs often strengthens relational bonds and can act as a catalyst for maturity and stability.'[23]

If people with disabilities were fully integrated into society, there would be less impetus for testing and termination because those with disabilities would be seen as full, valuable and equal members of the community. The Christian ethic, which calls the strong to make sacrifices for the weak, leads to a strengthening of family and society, by combating discrimination and strengthening human virtues of patience, perseverance and altruism.

In summary, we recommend:

- Diagnostic and prognostic information, including information on the risks of abortion for fetal disability, must be conveyed in a way that is genuinely neutral, balanced, compassionate and well-informed.
- Advice and counselling should be provided by qualified and trained counsellors.
- Parents should be offered the option to meet others who have first-hand experience of the condition or disability in question. This includes affected patients and their families, disability specific support groups, healthcare professionals caring for babies, children and adults with the relevant condition. Reading testimonies of women who have chosen to continue with their pregnancies, such as those collected by Amy Kuebelbeck[24], may also be helpful.
- Bringing up a child with special needs often involves substantial emotional and financial cost. Practical support for the longer term must be in place for families, and access routes to financial and emotional support as well as treatment need to be clearly signposted. These should include routes for exploring adoption for those families who feel personally ill-equipped but who wish to offer their child 'the gift of life'.
- More statutory funding should be provided for information, care and support groups and organisations for those with disabilities.
- Perinatal palliative care is an appropriate option for patients whose babies are diagnosed antenatally with a severe or terminal disability. In one British study, when parents were offered perinatal hospice as an option, 40% chose to continue with their pregnancies[25].

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[1] Convention on the Rights of Persons with Disabilities, *opened for signature* Mar. 30, 2007, 46 I.L.M. 433 [hereinafter Convention or CRPD]. The CRPD text, along with its drafting history, resolutions, and updated list of signatories and States Parties is posted on the United Nations Enable website at http://www.un.org/esa/socdev/enable/rights/convtexte.htm.

[2] Optional Protocol to the Convention on the Rights of Persons with Disabilities, *opened for signature* Mar. 30, 2007, 46 I.L.M. 433 [hereinafter Optional Protocol].
[3] *See* CRPD, G.A. Res. 61/106 (2007); Optional Protocol, G.A. Res. 61/106 (2007).
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