



*A Christian Perspective on Prenatal
Screening and Diagnosis in Australia*

Christian Medical Dental Fellowship of Australia Inc.

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CMDFA Ethics:

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BACKGROUND INFORMATION IN PRENATAL GENETIC SCREENING AND DIAGNOSIS

INTRODUCTION

Technological advances in the 21st century have given us an unprecedented and increasing ability to predict fetal outcomes. This predictive ability has brought new dilemmas in decision making. Today's families and their doctors face a range of issues that were inconceivable a generation ago. With early scans and other tests, we can now diagnose a wide range of congenital abnormalities, potentially leaving parents with decisions affecting life and death.¹ Over the course of a few decades, medical care has evolved from being primarily therapeutic and preventive to now also include prediction of risks.

In addition to rapid medical and technological progress in the last few decades, there have been powerful socio-cultural changes that alter the way we reflect on life and living. These include the increased presence of women in the workforce, with a consequent delay in the age at which they marry and start bearing children, a decrease in family size and a shift from the extended family to the nuclear family,² as well as activism and legal changes related to termination of pregnancy. Computerisation has led to greater health literacy throughout the population. Consequently, medical care, technological progress and risk prediction offer new challenges to patients and their doctors.

Modern technology is an ally in improving outcomes for the mother and the child; however, as Christians, we need to make sure that our application of science and technology is ethical. While at times Christians have feared new scientific understandings, we see that science, when ethically practised, can lead to enhanced medical practice. However, in order to determine whether a practice is ethical by biblical standards, we first need to understand the facts involved. In this paper we will give an overview of the current practices in prenatal genetic screening and diagnosis then examine the ethical issues involved and how the Christian doctor may approach them.

SCREENING IN PREGNANCY

There are many screening and diagnostic tests available to assess maternal and fetal health to ensure improved perinatal outcomes. Screening tests are easily performed and cost-effective

investigations used among large population groups to separate those who are at low risk from those who are at high risk of a disease. Screening tests provide a risk ratio, which reduces the number of people who need to progress to expensive and potentially dangerous diagnostic tests.

Most screening tests in pregnancy are well established. Maternal tests include routine tests such as blood group, rhesus status, presence of syphilis, HIV, hepatitis B, cytomegalovirus and toxoplasmosis, and prior exposure to measles, mumps and rubella, as well as monitoring of maternal full blood count, liver and kidney function and blood sugar levels. Fetal testing includes the various ultrasounds, e.g. the morphology scan at 20 weeks' gestation. Most of these investigations are motivated by a desire to improve maternal and fetal wellbeing. The focus of this paper is on issues related to prenatal genetic screening and prenatal genetic diagnosis. These more recent tests have at times been controversial and raised challenging ethical questions.

PRENATAL GENETIC SCREENING AND DIAGNOSIS

The human karyotype has 46 chromosomes: 22 pairs of autosomes and a pair of sex chromosomes, one set from the biological father and the other set from the biological mother. Genetic disorders result from either a disorder in the number of chromosomes (e.g. Down syndrome [trisomy 21] or Turner syndrome [XO]), or structure of the chromosomes (e.g. duplications/deletions/translocations/ring chromosomes). In addition, there are single gene disorders such as gene repetition disorders and uniparental disomies. Genetic or DNA testing screens for the predisposition an individual has for any of the inherited disorders and is diagnostic in many clinical contexts. It can determine a person's parentage/ancestry. Genetic tests may include biochemical testing of the markers/products of the gene or testing for the defective gene itself. A gene defect does not necessarily mean a disease state. Whether the disorder is expressed is influenced by both the nature of the disease (dominant or recessive) and the type of genetic disorder (homozygous or heterozygous). The complexity may increase due to different mutations being possible in the same gene and the actual expression of the gene disorder in an individual.

The currently available prenatal tests are:

Screening tests

- Combined first trimester screening (combining maternal blood testing and a scan of the fetus)³
- Second trimester maternal serum screening⁴
- Non-invasive prenatal testing (analysing cell-free fetal DNA from maternal blood)^{5,6}

Diagnostic tests

- Chorionic villus sampling (for chromosomes and DNA extraction)
- Amniocentesis (for chromosomes, DNA extraction and other biochemical tests)
- Microarray panels (comparative genomic hybridisation or new generation sequencing) of fetal DNA⁷

Second trimester structural anomaly scanning (morphology scan) is used for both screening and diagnosis. The number of diseases that can be screened for and diagnosed using these tests is increasing. Genome-wide prenatal testing is likely to be available soon. In the case of a negative result from screening, it is assumed that the pregnancy will continue without intervention. In the case of an indeterminate result or a positive test from screening, further assessment is recommended to establish a diagnosis. The parents have the choice to undertake diagnostic testing or to decline. Counselling is advised before the decision is made.

In the case of a positive diagnosis, all available treatment options should be offered. These may or may not be accepted by the parents. Some will choose to continue the pregnancy under careful management, usually in a tertiary care setting. Access to tertiary care is often determined by geographical location in Australia. In the case of a positive diagnosis and in the absence of any treatment or cure, outcomes include a decision by the parents to continue the pregnancy and prepare for ongoing care of the child, or a decision to terminate the pregnancy. Perinatal palliative care is an option for some parents whose child may have a severely life-limiting condition (e.g. Edward syndrome or Patau syndrome). The pace of research related to prenatal genetic conditions and their treatment is progressing rapidly;⁸ for example, it is now a possibility that Down syndrome⁹ will be treated through silencing the extra chromosome 21.¹⁰

DEFINITIONS

Modern technology and the availability of advanced life-support from 23 weeks of intrauterine life onwards have blurred perinatal timeline definitions. The following medico-legal definitions are accepted in the Western world.

Pre-viable gestation

Beginning of biological life

The potential for biological life as we know it begins at conception, and establishes itself progressively at implantation, at viability and at birth. The criteria for miscarriage have been revised to ensure that the beginnings of biological life are given sufficient margin so no errors are made in diagnosis.¹¹ A heartbeat seen by ultrasound scan is the first sign of life in an embryonic disc.

Embryonic period

The time from a missed period or positive home pregnancy test up to 12 weeks' gestation, by which time most fetal organs are formed; the fetal period dates from 12 weeks to birth.

Fetal period, weeks 12 to 22

Functionally, three fetal periods are recognised; if the fetus is born prematurely in the pre-viable period up to 22 weeks, resuscitation is normally not recommended.

Peri-viable gestation

In the peri-viable fetal period from 23 weeks to 25 weeks and 6 days, parental wishes in regard to resuscitation are taken into consideration.

Viable gestation

Beyond 25 weeks and 6 days, the fetus is considered to be viable (able to live outside of the womb with technological support and resuscitation).¹²

Birth

A birth is defined by the World Health Organization as a gestation of over 20 weeks or 500 g (though many developing countries consider this a miscarriage, with criteria for birth being

after gestation of 28 weeks or birth weight of 1000 g).¹³ It is a legal requirement to register all births after 20 weeks in Australia.

“Wrongful birth”

The concept of ‘wrongful birth’, where an unplanned child is born through medical negligence — and the parents may sue the negligent doctor to recover the costs of raising the child to maturity — was upheld by an Australian High Court by a 4:3 majority.¹⁴ Legislative changes to counter this have been proposed in the United States.¹⁵ The terms ‘wrongful birth’, ‘wrongful pregnancy’ and ‘wrongful conception’ have been variously defined, sometimes interchangeably. Strictly, what is ‘wrongful’ is the negligence, not the birth; but the label is convenient shorthand.^a This implies that parental intent is a prerequisite for rightful birth, and hence implies diminished status of the fetus where there is no parental intent.

CONTROVERSIES IN DEFINITIONS

Medicine and biology study human structure and function as well as the mechanisms in human formation. While we have elucidated the entire human genome and understand many of the mechanisms that are involved in the formation of a human being, there is still a vast amount that we do not understand. Although our understanding of the genetic and epigenetic interplay has increased in quantum leaps in the last decade, there are many more inroads still to be made before we can understand the many mysteries of cellular mechanisms. Defining human life purely in terms of biology, i.e. biochemistry, chromosomes, structure or function, is reductionist and has limitations.

Biochemical definitions

The potential for life begins with the fusion of an oocyte (n chromosomes) and a sperm (n chromosomes) to form a zygote (2n chromosomes). The exact mechanisms involved in the crossover of chromosomes (from the sperm and oocyte), spindle formation and the migration of the chromosomes to the polar bodies are not well understood. These cells rapidly multiply

^a 322 Deakin Law Review Volume 10 No 1

to separate out into the outer cell mass (trophoblastic cells/chorionic cells), which forms the placenta and membranes, and the inner cell mass, which forms the embryo/fetus. The trophoblastic cells release human chorionic gonadotropins which signal the uterus and the ovary to prepare for implantation and pregnancy. Evidence of pregnancy is biochemical, and many pregnancies are lost at this stage.

Chromosomal definitions

Once implantation occurs, some pregnancies continue without an embryonic pole. Ultrasound examination can allow the diagnosis of an “anembryonic pregnancy” to be made. Most losses in early pregnancy are likely to be from chromosomal abnormalities. The euploid human fetus is defined as 46,XX or 46,XY. Trisomy of the sex chromosomes (XXX, XXY or XYY) or of chromosome 21 is compatible with life but there are many trisomies of the autosomal chromosomes that are not compatible with life. The triploid/tetraploid pregnancy (69,XXX/92,XXXXY) that results in a partial molar pregnancy where a fetus is seen, is not compatible with life. The diploid (46,XX/46,XY) 2n chromosomes in the case of dispermy, where no chromosomes are derived from the ovum, results in a molar pregnancy where there is no recognisable fetus. It cannot be attributed any human characteristics even if the biochemistry and the karyotype are normal.

Structural definitions

The anomaly scan conducted at 18 to 20 weeks’ gestation aims to identify fetuses with a structural abnormality. It is a challenge for fetal medicine specialists to define the level of scanning required to define structural normality, and almost all reports contain a disclaimer. A fetus with anencephaly (no skull with brain tissue exposed) cannot have cortical function even though the pregnancy continues normally and the fetus could be born alive. A twin pregnancy can result in an acardiac fetus, which starts with a heart and head in early pregnancy but develops into a mass of tissue that is chromosomally normal (46,XX/XY) but structurally incompatible with life (although parents may want to name the twin). Therefore, structural characteristics from scans are inadequate to define human life. A structural abnormality may allow a child to be born alive but to only survive for a short time.

Functional definitions

In instances where the fetus has a normal head and a normal heart, the absence of kidneys or conditions that cause hypoplastic lungs are also incompatible with continuing life. Functionally, there are conditions where the fetus is structurally normal but does not move at all (fetal akinesia syndrome) and cannot breathe once it is born. It is difficult to define what functions have to be present as a minimum to define a human.

Limitations of any definition

These examples highlight the difficulty in defining a human being using any particular biological parameter. Regardless of how we define a human being or when we define the beginning of life, there are some issues that will invariably challenge our views and may not have an easy answer. We can see the difficulty of attributing human characteristics to biochemical evidence of pregnancy or to an anembryonic sac from fertilisation or to a molar pregnancy. Moreover, to be chromosomally normal does not ensure sufficient structural normality to necessarily result in a live baby (e.g. anencephaly or Potters syndrome). The falleness of the reproductive processes (as with all of creation) results in abnormalities that can exist at the biochemical, chromosomal, structural and functional level. To attribute human characteristics or to define human life in terms of karyotype/genome or in terms of structure/function would be to do it an injustice.

PROFESSIONAL GUIDELINES FOR PRENATAL SCREENING AND DIAGNOSIS

All doctors in Australia are required to comply with professional ethical standards. Several professional organisations have published guidelines on the topic of pregnant women and the disclosure of screening test availability, of which all doctors involved in antenatal care should be aware.

College guidelines

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) has a statement on prenatal screening for Down syndrome, Edward syndrome and neural tube defects with professional requirements for all obstetricians and gynaecologists on their website^b An excerpt is copied below:

^b <https://www.ranzcog.edu.au/college-statements-guidelines.html>

3. Discussion and recommendations

3.1 Prenatal tests for chromosome abnormalities

General information on prenatal screening and diagnosis

3.1.1 All pregnant women should be advised of the availability of prenatal screening and diagnosis as early as possible in pregnancy to allow time to discuss the options available and facilitate an informed choice. An informed choice is “based on relevant knowledge, consistent with the decision maker’s values and their partners, or support person if appropriate

3.1.2 Some women may make an informed decision not to proceed with any testing. Counselling should follow a shared decision-making model, where health professionals discuss information based on their expertise and respect for the woman’s values in arriving at an agreed course of action. Women electing not to have ultrasound screening in pregnancy should be aware of the other important benefits of routine scanning, including placental localisation, confirmation of gestational age, and excluding multiple pregnancy.

3.1.3 Information should be communicated using clear, simple and consistent language when discussing the tests, with confirmation that the information has been understood.

3.1.4 Information should be provided in a format that is easy to understand and accessible to pregnant women from culturally and linguistically diverse backgrounds (including Indigenous women) and women with additional needs (such as physical, sensory or learning difficulties). An interpreting service should be made available where it is required (see Appendix E).

3.1.5.7 The understanding that if an abnormality is diagnosed, women and their partners can choose whether to continue the pregnancy or have a termination. Where a genetic abnormality has been diagnosed, parents should be given sufficient information regarding the aetiology, associations, and implications of that diagnosis during pregnancy, the newborn period and beyond, in order to make an informed decision regarding pregnancy termination.

3.1.5.8 There should be an assurance that regardless of their decision, women will be offered counselling and receive ongoing care and support. In the case of continuing the pregnancy, women and their partners should be provided with appropriate antenatal care with individualised preparations for birth and neonatal management. The option of neonatal palliative care should be discussed for conditions where the prognosis is very poor. If they choose termination, they need to know that the mode of termination may be influenced by gestational age in line with local legal precedents.

Prenatal screening and diagnosis of chromosomal and genetic abnormalities in the fetus in pregnancy C-Obs 59.¹⁶

Australian Medical Council guidelines

The need for doctors to provide all available information on genetic screening to pregnant patients is supported by the Australian Medical Council's *Good Medical Practice: A code of conduct for doctors in Australia* (2009). The relevant sections are quoted below:

2.2 Good patient care

Maintaining a high level of medical competence and professional conduct is essential for good patient care. Good medical practice involves:

2.2.6 Providing treatment options based on the best available information.

2.2.12 Ensuring that your personal views do not adversely affect the care of your patient.

2.4 Decisions about access to medical care

Your decisions about patients' access to medical care need to be free from bias and discrimination. Good medical practice involves:

2.4.6 Being aware of your right to not provide or directly participate in treatments to which you conscientiously object, informing your patients and, if relevant, colleagues, of your objection, and not using your objection to impede access to treatments that are legal.

2.4.7 Not allowing your moral or religious views to deny patients access to medical care, recognizing that you are free to decline to personally provide or participate in

that care.

In the context of the guidelines above, it is imperative that all physicians involved in the care of pregnant women become conversant with prenatal screening and diagnosis, regardless of the cultural, religious and ethical views held by doctor or patient. As discussed in the Christian Medical and Dental Fellowship of Australia (CMDFA) Ethics Statement on Christian Conscience in Healthcare, we each need to decide how such requirements impact on our choices for where we practice medicine.

ETHICAL ISSUES IN PRENATAL GENETIC SCREENING AND DIAGNOSIS

INTRODUCTION

Western ethics has been traditionally based on Judeo-Christian values. In modern pluralistic Australia these values are not universally held, and the diversity of views can lead to ethical conflicts for Christian doctors.¹⁷ Furthermore, even within a Christian understanding, the right thing to do in any given situation may not be immediately apparent.

Genetic screening and diagnosis allow for improved monitoring of pregnancy and possible antenatal treatment of identified pathological conditions. A diagnosis of Down syndrome may mean that plans for a home birth need to be changed owing to the increased risk of complications at the time of the delivery. It also makes it possible for parents to prepare their home for long-term care of a child with special needs. Therefore a high regard for the moral status of the fetus does not preclude testing. One assumption underlying genetic screening and diagnosis is that genetic abnormality may lead to termination of pregnancy. The decision of whether to use the technology is complex. The numerous ethical dilemmas associated with prenatal genetic screening and diagnosis make it even more difficult.

THE INHERENT VALUE OF A FETUS

In current secular medical literature, there is no consensus regarding the moral status of the fetus. This situation is made more complicated in the fetal medicine setting, where the question of ‘fetus as patient’ arises.

The basic question here is whether we should regard the fetus as a person and thus deserving of legal protection. In secular literature it is due to the potential for maturity, or the idea of maturity increasing with development, that value may be conferred on the nascent human. This view is reflected in legislation which allows termination of pregnancy under certain circumstances. Obviously the fetus cannot exert autonomy on its own behalf. Therefore Chervenak and McCullough¹⁸ argue that, although the fetus cannot have autonomy-based protection, it could have a beneficence-based protection when the fetus is considered a patient. They further argue that the only basis upon which a pre-viable fetus can attain the status of a patient is when a pregnant woman makes a decision to confer this status onto the fetus. Under our current law, the woman is free to confer or withhold this status. Furthermore, having conferred, she is free to withdraw this status from the pre-viable fetus at any time without justification, according to her own values and beliefs.

Once a fetus has reached viability, this balance changes and some authors suggest that a direct relationship between fetus and the doctor now exists which is not dependent on maternal preference. This relationship would demand that the doctor consider the best interests of the fetus on the basis of beneficence. Chervenak and McCullough suggest that the mother should also realise that there is a beneficence-based obligation to the fetus which constrains her autonomy,¹⁸ for example, by prohibiting termination of a viable fetus. This view is obviously not universal and not supported by legislation.

INFORMED CONSENT

The requirements for informed consent are as follows: the person making the decision must be mentally competent; the choice should be voluntary (no coercion); the decision should be based on sufficient information; and the patient must have adequate understanding of what is involved.

Informed consent will be important for the pregnant woman, as whether screening is done depends entirely on her choice, although ideally this would be a family decision.

Many prenatal genetic screening tests need to be done early in a normal pregnancy, so general practitioners often order them before the mother has had her first visit with an

obstetrician. This means the doctor ordering the test may not have specialist knowledge of the conditions being tested. He or she may not be able to explain fully what is involved,¹⁹ and explanation of health risks is known to be difficult in any setting.²⁰ Often there will be time constraints, and it may be difficult to provide comprehensive genetic counselling, such as the statistical complexity of sensitivity and specificity of screening tests, in a busy general practice.²¹

General practitioners in Britain reported that it was difficult to raise possible adverse outcomes with someone who is excited at finding out she is pregnant. Although doctors offered women a choice, they knew they consciously offered genetic screening tests as routine and in a positive light due to lack of time, while being aware that a key motivation for women's positive perceptions of screening was their sense of obligation to undertake any test that would benefit their unborn baby.²² There is a need to better equip all doctors in this complex and fast-changing area.

Currently, evidence suggests that many women undergoing these tests do not understand their purpose. An Australian study²³ found that 31% did not know that miscarriage was a possible consequence of diagnostic testing subsequent to an increased risk screening result, and only 62% correctly identified that termination of pregnancy would be offered if Down syndrome were diagnosed.²³

Apart from the ethical problems involved with inadequate disclosure of all the facts, it means that women may not think through the implications of a positive diagnosis before undergoing the testing. Informed consent will also require comprehensive explanation of the results at each stage of the screening and diagnosis process, including the meaning of genetic risk, the nature of the genetic abnormality diagnosed, the concepts of variation in phenotype and genotypes, and practical information about living with disability. Currently, such information is not routinely given.²⁴ Some authors suggest it is not feasible to give so much information in a way that is meaningful to the mother. Confusingly, some authors have complained that women are already given too much information regarding prenatal genetic screening, which gives them too many choices and impairs their decision-making ability.¹⁸ This problem will only become more complex as the testing becomes more comprehensive as genetic

technology improves. Furthermore, given that our understanding of genetics is incomplete, we do not know the significance of many variations seen in genome-wide testing. This further highlights the difficulties of what women need to be told before such tests are offered.

Medical standards of patient confidentiality make it necessary for the woman to give the doctor permission before he can share such information with other parties, including the father of the fetus. Any attempts by family members to coerce the woman are also an ethical problem which the doctor should attempt to counteract.

De Jong and colleagues have suggested that informed consent may become more difficult with the introduction of non-invasive prenatal testing.²⁵ They raise the possibility that, because non-invasive testing is easy and safe and can be performed early in pregnancy, both testing and selective abortion may become ‘normalised’. It is also possible that the reduced risks involved may mean that less care will be taken with informed consent. This is particularly important, as, with widening of scope of testing and increased sensitivity of ultrasound scans, women may get unexpected results for conditions for which they did not know the fetus would be tested, and thus may not be prepared to handle. De Jong and colleagues suggest that this easy, safe, and early technique combined with testing a much broader range of abnormalities (including milder disorders or likely no disorder) will challenge the notion of prenatal screening serving reproductive autonomy. The capacity of the patient to meaningfully engage with even more diverse and complex measures of risk will further stress the doctor–patient relationship. The support of all involved by medical experts in genetic interpretation is likely to increase.

NATURE OF CONDITIONS DETECTED

Prenatal genetic screening and diagnosis was initially aimed at identifying serious conditions present at birth. With rapid broadening of the scope of genetic testing — including genome-wide molecular tests — conditions which are treatable, adult-onset or only partially penetrable (such as hereditary cancers) are now also the focus of investigations. While recommendations have been made that prenatal diagnosis not be used for minor conditions or characteristics, the question of who decides what a minor condition is is not clear.²⁶

This raises the possibility of a pregnancy being terminated for conditions which may never develop, or are treatable, or, if they do not develop until adulthood (at least 20 and up to approximately 80 years later), may be treatable by the time they occur. While many commentators suggest termination is done in the interests of the child (to avoid suffering), as the interval of anticipated disease-free life increases this is more difficult to justify.

FETAL THERAPY

In the event of a positive diagnosis, treatment of the fetus, including in-utero intervention, can be an option. Maternal–fetal surgery has significant implications and complications can occur acutely, postoperatively, for the duration of the pregnancy, and/or in subsequent pregnancies. It is a highly technical procedure with potential for significant morbidity and possibly mortality, even in the best and most experienced hands.^c Maternal consent is required for any intrauterine procedure. Treating the fetus puts the mother at risk, both in intrauterine minimally invasive procedures and open fetal surgery. Even simple transplacental therapy of the fetus has side-effects for the mother that can require intensive care admission for monitoring. Where the benefit for the fetus and the mother conflict, in our community it is generally the mother who decides which should be the goal of treatment.

When neither death nor the absence of cognitive development is certain, aggressive management of the fetal condition as consented to by the mother is the ethical standard of care in obstetrics.¹⁸ However, there are instances where the parents may not agree with the medical recommendations, and there have been no court orders to date in Australia where maternal autonomy has been challenged to enforce fetal testing or therapy.

COMPLICATIONS OF TERMINATION FOR FETAL ABNORMALITY

Prenatal testing and selective abortion to avoid the birth of a seriously impaired child is widely accepted in our community. One study²⁶ found that 79% of Americans believed that abortion should be available in this instance. Public surveys suggest that the birth of a child with a severe genetic disorder is thought to bring to a family distress, psychological harm, emotional harm and suffering, loss of opportunities, loss of freedom, isolation and financial

^c http://www.acog.org/Resources_And_Publications/Committee_Opinions/Committee_on_Obstetric_Practice/Maternal-Fetal_Surgery_for_Myelomeningocele

costs.²⁷ Concerns have also been expressed regarding the impact on the parent–child relationship and the distress of watching a wanted child die, despite the best palliative care available. However, the evidence suggests that termination of a wanted child on grounds of a genetic disorder may not lead to the best outcome for parents.

Medical termination for fetal abnormality in a wanted pregnancy is quite different from abortion of an unwanted pregnancy. It represents the loss of future hopes and entails the risk of severe and complicated grieving.²⁸ Women whose infants are diagnosed with a congenital anomaly can experience an emotional crisis²⁹ and consideration of end-of-life decisions can entail ambivalence between commitment to the pregnancy and the need to avoid the burden of disability.³⁰ A significant proportion of women undergoing termination for fetal abnormality can experience pathological levels of distress,³¹ more complicated when they were not anticipated due to inadequate information.³⁰ The long-term posttraumatic stress response and grief can last for years³¹ and is comparable to the experience of having a stillborn child.³² More research is needed to understand the long-term implications of this experience, but the assumption that early detection and termination of fetal anomaly is beneficial for women has been questioned.³³ This has important implications for the adequacy of information-giving for proper informed consent to exist.

There is concern that the easy availability of genetic screening and termination of pregnancy may reduce antenatal parental bonding. Whether an abnormal or normal result eventuates, does the idea of children needing to ‘measure up’ impair the parents’ ability to learn to love their child unconditionally? Does a false negative result make it harder for parents to accept a child born with a genetic abnormality?

In the event of a life-limiting genetic abnormality, while there is no doubt about the distress which results from perinatal death of a wanted child, many parents who have opted for neonatal palliative care have the comfort of knowing they did all they could for their child for as long as they could. Adequate counselling and support needs to be provided for those families that embrace and care for a child with disabilities. Such an experience can bring unexpected rewards as well as unanticipated challenges.

DISCRIMINATION

As already mentioned, prenatal genetic tests are usually justified in terms of maximising meaningful reproductive choice. This includes the opportunity to avoid parenting a child with a serious genetic disorder. However, there is concern that the use of prenatal genetic screening and diagnosis sends a negative message to those already living with disability. There is debate regarding whether prenatal diagnosis and selective abortion constitutes discrimination against the disabled, some suggesting that, if it is used as a tool to eradicate disabilities it does discriminate, and others suggesting that this is not a conclusive argument against the practice. De Jong and colleagues suggest that if abortion decisions remain personal and are not made instruments of societal goals to reduce the number of children with congenital disability, they are morally justified. However, some media reports have suggested that with the increased ease of testing, prenatal genetic screening and diagnosis may move from an optional extra aimed at increasing maternal freedom, to an obligation for mothers which, when rejected, is seen as negligent behaviour at risk of burdening the community with disabled offspring.

While termination of the fetus with a diagnosed genetic abnormality may be seen as an act of mercy by many people in our community, the fact that the whole genetic diagnosis process is oriented towards the autonomy of the mother raises the possibility that termination can sometimes be done to avoid anticipated suffering by the parents if they are obliged to raise a child with special needs, whether or not this suffering actually ensues. Members of the disability community have expressed dismay that others may think their lives are not worth living.³⁴

AUTONOMY OF THE CHILD

Prenatal testing has implications for the future autonomy of the child. Testing before birth removes the right of the child not to know their genetic makeup. Many people at risk of a genetic disorder (e.g. 80% of those at risk of Huntington disease) choose not to be tested to avoid the worry of impending disease, or to avoid the need to inform other parties, such as employers or insurance companies, of their genetic status. These options will no longer be possible if the testing has already been done. Solutions to this conflict between the rights of

the child and the rights of the mother have been offered in the secular literature. They include: limiting the scope of testing, avoiding the detection of risk for late-onset diseases; or allowing women to undergo these tests only if they have expressed the clear intention to abort an affected child (as the right to self-determination cannot be violated if the child does not live). As long as the right to information to allow women autonomous choice is valued so highly in our healthcare system, it is unlikely that any of these options are workable.

MEDICO-LEGAL ASPECTS

There are several factors which work together in the current medico-legal environment to create pressures for the doctor who has the responsibility of offering genetic testing to the patient.

- i. Our legal system is based on secular values and strongly supports the autonomy of the mother in genetic testing. This can be emotionally difficult for the medical practitioner who does not agree with her choices. It is also at odds with biblical directives of the responsibilities of parenting for both mothers and fathers. (Ps 103:13).
- ii. Consumerism and the threat of legal action by the parents if something is missed increase the risk of over investigation. It often allows screening tests to creep into medical practice without evidence-based benefit, rigorous debate and understanding. Obstetric specialists in Australia can be sued up to 20 years after the birth of a child for problems arising from the pregnancy and birth. As a result, medical actions can be influenced by medical defence organisations and College guidelines as they are applied to for support.
- iii. The costs involved in developing genetic tests combined with the demands of consumerism may mean that genetic tests are offered online prematurely, before they are validated. Furthermore, direct testing via the internet means that consumers are without the benefit of counselling before and after results are given, possibly resulting in unnecessary distress and confusion.

PUBLIC HEALTH PERSPECTIVES

Health screening of populations is aimed at early detection of disease to allow for prevention or early treatment. The aim of prenatal screening is to provide information for pregnant

women on fetal wellbeing. The pregnant woman uses this information to make choices whether to continue a pregnancy, seek fetal therapy if indicated, or undergo a medical termination. However, population screening also needs to be proportional. This means that which conditions should be tested for and why is not judged entirely on the balance of benefits and burdens for participants, but also for the society as a whole.

The principle of justice requires that there is equity of access to screening programs; but in a country such as Australia, where such tests are government funded, there needs to be consideration of the costs of screening, particularly in the context of financial constraints. As the number of available tests increases and the range of conditions tested for broadens, doctors and other stakeholders will need to find a balance between which disorders have sufficient impact on society to justify screening and which ones do not. This will be difficult, as there is no consensus regarding what parameters will guide such decisions. Furthermore, these decisions are likely to be revisited and revised as technology and its costs continue to change.

AN APPROACH TO READING SCRIPTURE FOR PRENATAL ETHICS

INTRODUCTION

There are many procedures involved in prenatal genetic screening and diagnosis and the subsequent management pathways that need to be considered carefully from a biblical framework. Undoubtedly, the current ethical climate in obstetric care will create dilemmas for many Christian doctors. As scripture is silent on many issues that face us in the 21st century, ethical principles need to be derived. We live in what is called the “post-modern” era. It is a time of both serious challenge to faith and a fresh opportunity to engage in ethical discussion through the Christian principles found in scripture.

Historically, Christians have made and, undoubtedly, will continue to make mistakes or misread scripture, even in the company of others. Prior to the Reformation, the Church as an institution took responsibility for making final theological decisions for the faithful. The Reformation has opened the scope of authority, with at times a loss of the moderation of the

whole catholic faith. We also issue caution. The primary message of scripture is about the love of God for the world. This love is centred in Jesus of Nazareth and his death for our salvation. Through this Christ event, we are called to a transformed way of life, to Christification. We are not advocating an “extractionist” approach to scripture outside of such a way of life.

Burridge makes these observations. “The New Testament is not a general answer book with all the answers to our ethical dilemmas. However, many of the key issues of human moral experience do appear, such as issues of violence, the state, obedience, power, war and peace; human sexuality, marriage, divorce, relationships; money, wealth and poverty, debt; life and death and the value and meaning of life.” We will therefore need to be cautious about applying biblical commands about these areas directly to our contemporary moral debates. Before we can use the biblical material, we have to deal with differences between the New Testament and Old Testament over, for example, violence (holy wars versus ‘turn the other cheek’), or whether all the laws are now binding on Christians, including things like ceremonial or purity law or circumcision. Similarly, polygamy is accepted in the patriarchal narratives, yet Jesus refers to Gen 2 on ‘one flesh’ as God’s original intention (Mark 10: 6).

In view of these concerns, Burridge makes the following proposal:

- Any attempt to apply biblical texts to ethical concerns will need to begin with the best available exegesis — to find out what this text is actually saying, taking into account historical background, genre etc.
- Equally, the reader needs to be aware of his or her own background as a filter and to make allowances for both the filter in the text and our self in the reading. To avoid negative ‘group think’, Burridge makes this suggestion. “An approach to New Testament ethics requires the interpretive community to be open and diverse; inclusive of those who might disagree with us, but who are still making their response of discipleship as we follow Christ together.”
- In a spirit of prayer, some conclusions may be provisional and thus frustrating to those who want “clear moral guidance.”

This paper seeks to provide helpful insights for Christian colleagues to approach Scripture while seeking answers or guidance for ethical concerns in health or dental care. We acknowledge a need to read Scripture with eyes respectful to past approaches and conclusions while recognising the challenges and benefits of reading scripture in our post-modern context.

It is important to recognise at this point that our reading of scripture is to be undertaken with open dependence on the Holy Spirit. It is the person of God as the Spirit who leads us into all the truth of Jesus and the love of the Father.

A CHRISTIAN APPROACH TO ETHICS — AN EXAMPLE

Allen Verhey is an ethicist who has sought to develop an integrated biblical approach to this challenge. His text, *Reading the Bible in the strange world of medicine*³⁵ develops the following considerations.

Genesis 1:26–27 states that human beings are created in the image of God. This causes Verhey to reflect ethically in a chapter ‘Mapping the Human Genome...Biblically’ and put forward an ‘orientation’ that we need to consider in the prenatal context. We should ‘orient’ ourselves not only to the values we may hope to achieve in scientific study but also and fundamentally to the values we display, i.e. our personal ethics, our transforming Kingdom lives. We should orient ourselves not only towards mastery but also toward wonder and the celebration of creation. We ‘image’ God as embodied souls. We are not separate from our bodies; and in this context, Verhey makes the comments, “We must orient ourselves to attend to whole selves. Genetic reductionism and determinism do not fit the story. We may not be reduced either to our genes or to simple transcendence over them.” We must orient ourselves to respect for the bonds of marriage. He says “We image God in marriage. This is a self-giving commitments of fidelity to care for our partners and for our children even when they don’t turn out quite like what we expected. Parenting involves the uncalculating commitment to nurture.” Verhey gets to the idea that to image God would be to sustain care for the weak and the helpless and the little ones who do not measure up. This is certainly a very important aspect of what is happening in antenatal diagnosis and treatment. His final comment is interesting: “The solution for our world is neither simply to maximize freedom

nor to eliminate it. To coerce faith or fellowship violates the freedom God gives. We must orient ourselves toward respect for freedom.”

SCRIPTURAL PRINCIPLES FOR PRENATAL ETHICS

Reading scripture for ethics in our post-modern context starts with faith and an openness inspired by the Holy Spirit. Consequently, we identify that the following approach may help us. There are cultural and literary genre issues with all biblical texts. The best information can help start the conversation around ethical concern. Ethics in scripture is always a linking of ideas, wisdom and a transformed/transforming life. For Christians, ethics starts in the community of faith where it can be grounded in worship/liturgy and the fruit of the Spirit at work among the people of God.

God’s truth providentially extends beyond the church. In humility, Christians must be subject to ‘a chastened understanding of public criticism’ if they are to participate wholeheartedly in bioethical debate (with medical/dental colleagues and others). As suggested by theologian Richard Niebuhr, “The gospel of the crucified Lord constitutes a ‘permanent revolution’ in our understanding of God, the world and ourselves”. We may need to accept a ‘provisionality’ of ethical understanding and at times revise dogmatic decisions.³⁶ There is always value in respecting the ethical understanding of the people of God who have preceded us, .e.g. papal encyclicals and international consultations.

For Christian doctors, the doctrine of man being made in the image of God will have an impact on these decisions. Genesis teaches us that human beings were made in the image of God (Gen 1:26–27), and despite the fall, we retain this likeness (Gen 9:6). However, we also know that as a result of the fall, the image is tarnished and our reasoning is flawed. Abnormalities can exist from the beginning of human development and we cannot always repair the damage. But our understanding allows us to place value on all human beings, not because of who or what they are, or what they can do, but because of the God in whose image they are made. Therefore, genetic aberrations will not change the innate value of human fetuses. As embodied souls we understand the complexity of human nature and reductionist views of human beings can be rejected.

We image God also as social and relational beings. This is not surprising given the

Trinitarian understanding of God for Christians. We are not autonomous individuals in this sense, and where possible family members beyond the mother alone should be involved in healthcare decision making. This will be influenced by our biblical view of parenthood, which sees children as a gift from God to be gratefully received, rather than a product of manufacture which can be rejected when imperfect (Ps 127:3–5). We need to remember that God has already redeemed us through the saving work of Christ Jesus, and that all creation will one day be free from suffering (Revelation 21: 4). This future hope sustains us as we seek to live according to God’s will in the transition time between Christ’s death and his return.

CONCLUSIONS

We write this at the commencement of a major change in the medical science of screening, diagnosis and management in the prenatal setting. The genetic revolution will lead to greater knowledge and, paradoxically, also often greater uncertainty. While knowledge will continually expand, the wisdom required to respond to this knowledge is mediated through the unsatisfactory prism of inherent uncertainty, or risk. This risk will be continually quantitatively improved but may not be currently clear in many cases.

Christians are inherently people who accept the imperfection of life and the associated imperfection of knowledge. We see this as an opportunity to join together with the Creator in contributing to the enhancement of what is created imperfect, each a flawed masterpiece as we inherently all are (Romans 3:23).

Others in our society may grasp at this notion of perfection which would exclude pain and suffering due to disease, death and the impact of evil in an imperfect world. This is illusory for both Christians and others. Antenatal screening and its associated science cannot protect us from the uncertainties and disappointments of life. Nevertheless, our science is understandable as people at work using God-given talents to serve others.

We seek to respectfully and humbly engage with our patients. We seek to act in accordance with the teaching and example of Jesus, the great healer who most fully understood the needs and frailties of each person he encountered. “For a Christian doctor or dentist ... the ethics of a truly transformed character will constantly challenge our clinical choices. The clinical

context is also a context of grace where the reconciling, healing, peace desiring work of God will become surprisingly apparent”.³⁷

Christians have been given a mandate to side with the oppressed, the needy and the vulnerable. This was part of Jesus’s manifesto (Luke 4:18–19). The care of orphans, widows, aliens and the poor is a recurring theme in the Old Testament (Isaiah 10:8). This mandate calls us to extend care to the fetus in its vulnerable state, and to offer prenatal diagnosis and treatment where indicated. The reality of our lives is the fallenness of our world, which disrupts all relationships, even at the level of chromosomes and genes. Parents will continue to demand prenatal genetic testing, either for knowledge or decision making regarding the continuation or discontinuation of pregnancy.

The Christian doctor could approach his or her personal role in this controversial area with the following reasoning. Given that prenatal genetic screening is legal and standard practice in Australia, the doctor who wants to continue in obstetric care needs to accept the role of working within this system. The role of conscience in medicine has been explored in an earlier CMDFA publication.³⁸ It is important to have Christian doctors in all areas of healthcare, and more so in this difficult area where there will be the opportunity to encourage parents to consider all options available to them, first in whether they want tests at all, and if so which ones. To decline prenatal genetic screening or diagnostic tests is a valid option for parents. Again, when or if an abnormality is found, the Christian doctor should encourage parents to consider all options available to them before making management decisions. Often multidisciplinary consultations give parents a balanced opinion about the anomaly seen and the management options available. There may be no simple answer to this complicated question.

Each situation and each pregnancy is different and Christian doctors need to prayerfully consider the issues surrounding prenatal genetic screening and diagnostic testing and the results from such testing. We can always offer comfort and care, hoping to restore disrupted relationships at all levels in a redemptive way. Issues in prenatal genetic screening and diagnosis challenge all doctors, and great wisdom and compassionate care for parents is required when reproductive abnormalities occur.

ADDENDUM

In this paper, we have sought to clarify the questions and uncertainties that greater yet imperfect knowledge have created in the field of prenatal screening and diagnosis. This is not positioned as a definitive checklist of mandated approaches, as this is not possible, but rather as an unravelling of the tangle of issues that face those working in the field of prenatal screening and diagnosis. We recognise that in critical areas of life there could be differences in Christian thinking and practice. The views expressed in this paper have been arrived at after discussions within the National Ethics Committee of the CMDFA and may not necessarily reflect the personal opinions of the individual members of the committee nor of individual members of CMDFA nor of the CMDFA as a whole.

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