THE CENTRE FOR BIOETHICS
Improving health care through Bioethics

The Centre for Bioethics

Bioethics Bulletin - **Volume 1, 2018**

Editorial

The draft policy for the prevention and control of hemoglobinopathies speaks of a much needed intention to address this great burden on the people of our country. It is at times like these when documents are written down, that we as responsible citizens of our country have the opportunity to reflect on certain issues of great significance that we might otherwise overlook.

Is it acceptable in our country today that a family that struggles to get a single wholesome meal a day should suffer the further indignity of selling all their possessions for a blood transfusion or a hospitalisation for a critical illness that one of their children requires from Sickle Cell Disease or Thalassemia? Is it acceptable that children with hemoglobinopathies should die of easily preventable infections simply because their families could not afford the crucial vaccines that were probably never even offered to them? The policy addresses many of these issues admirably, aiming to strategically channel resources to effectively deliver optimal care for people with hemoglobinopathies in India.

When it comes to prevention, a whole new set of questions arise. Is it acceptable to deny life to a person in the womb because they are less than "perfect" and high maintenance? Is it acceptable to create pathways that have the potential to pressurise pregnant mothers to terminate their children in case found less than "perfect"? Is it acceptable to pave a path that is likely to make people with disabilities feel unwanted, burdensome, alienated and stripped of the dignity that is due them because they are human? The policy's strategies for prevention, albeit well intentioned, certainly deserve a closer look.

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"Policy for Prevention and Control of Hemoglobinopathies — Thalassemia, Sickle Cell Disease and variant Hemoglobin's in India"

The hemoglobinopathies can be divided into two general types: the thalassemias (which are disorders of decreased globin chain production) and the hemoglobin structural variants (eg, hemoglobin S, hemoglobin C); a combination of the two is also possible. The thalassemia and structural hemoglobin variants are the commonest monogenic disorders globally. India has a huge burden with an estimated 100,000 patients with a β thalassemia syndrome and around 150,000 patients with sickle cell disease, but few among them are optimally managed. The β thalassemia and sickle cell disorders pose a significant health burden in India. The average prevalence of β thalassemia carriers is 3–4% which translates to 35 to 45 million carriers in our multi-ethnic and culturally and linguistically diverse population of 1.21 billion people which also includes around 8% of tribal groups according to the Census of India 2011. India with its diversity has several ethnic groups have a much higher prevalence (4–17%).[1,2]

Hemoglobinopathies represent a unique set of genetic disorders. The March of Dimes Global Report on Birth Defects has estimated that the prevalence of pathological hemoglobinopathies in India is 1.2 per 1000 live births. It has been suggested that there would be 32,400 babies with a serious hemoglobin disorder born each year based on 27 million births per year in India.[4,5]Of the 10,000 to 12,000 thalassemic children born annually in India, very few are optimally managed mainly in urban regions although the Government of India has included the care and management of patients of thalassemia and sickle cell disease in the 12th Five Year Plan. It has been estimated that 2 million units of packed red cells would be needed for transfusion of thalassemia patients in the country.[6,7] Better management for β thalassemia major patients mainly in urban regions in India with regular and safe blood transfusions and adequate iron chelation allows them to have a better quality of life. However, as they grow older, multi-disciplinary care is required. Although blood is now provided free of cost for patients with hemoglobinopathies and gradually iron chelators are also provided in many states, there are still other expenses for testing, processing and leucodepletion. Thus, majority of the patients do not receive optimum care.

Formerly, many affected individuals did not survive to childbearing age. Affected women now commonly reach childbearing age and desire pregnancy. Successful pregnancy is possible in many cases with carefully coordinated obstetric and medical management. Genetic screening and prenatal diagnosis is an important aspect of prenatal care in these disorders. DNA mutation analysis offers rapid and accurate fetal diagnosis.

Pregnancy also offers a unique situation in that cord blood has become a valuable source of stem cells for transplant. This allows the potential role of the unaffected fetus as a donor for affected siblings. Women with sickle cell disease who are pregnant also are at increased risk of

spontaneous abortion, stillbirth, and preterm labor. Cesarean delivery is not recommended in women with sickle cell disease, and it should be performed only for obstetric indications. Epidural analgesia is usually well tolerated as long as hypotension and hypoxemia are avoided. Ideally, however, pregnant women with sickle cell disease should receive care at institutions that can manage complications of the disease and high-risk pregnancies.

Women with sickle cell disease who are pregnant are at increased risk of morbidity and mortality because of the combination of underlying hemolytic anemia and multiorgan dysfunction associated with this disorder. These patients need increased prenatal folic acid supplementation. However, the standard of 1 mg of folate in prenatal vitamins is not adequate for patients with hemoglobinopathies; instead, 4 mg per day of folic acid is recommended.

Early diagnosis can facilitate implementation of proper preventive health measures, education of the parents regarding their carrier status, and provide the child with ongoing comprehensive care. Prenatal diagnosis is ideally done in the first trimester of pregnancy (10–12 weeks gestation) by chorionic villus sampling and DNA analysis which requires well trained obstetricians and sonologists for fetal tissue sampling as well as a competent laboratory for molecular diagnosis. Amniotic fluid can also be used for DNA-based prenatal diagnosis.

In India, many couples at-risk are identified late and fetal blood sampling by cordocentesis at 18–19 weeks gestation and HPLC based analysis of fetal blood also continues to be done. However, the results have to be interpreted with caution as certain mutations like the poly A (T > C) can lead to relatively high HbA levels and a misdiagnosis.[9,10]

There are very limited centers in the country where training is given to obstetricians and sonologists for fetal tissue and fetal blood sampling. The issue of safety of the procedures used for PND is worth mentioning first. Though they are found to be relatively safe, there is still a chance of a miscarriage following chorionic villus sampling and amniocentesis (worse with CVS and usually multi-factorial) .[4,5]

Abortion of the affected fetus is regarded as a consequence component of PND in most cases. In the case of the fetus having the SS genotype, the ethical question arises whether to have an abortion or to keep the pregnancy. The decision whether to terminate a pregnancy based on a positive result is usually a difficult one that involves religious, psychosocial and cultural considerations.[1] The purpose is to allow parents to make reproductive choices based on this information and, in the case of alpha-thalassemia major, to monitor the pregnancy for nonimmune hydrops fetalis and potentially intervene. The clinical sequelae of other hemoglobinopathies manifest later in life and have no adverse effects on the fetus, mother, or neonate.

Allogeneic stem cell transplant remains the only option for a cure for β thalassemia major patients. The chance of a successful transplant is >90% in patients with good risk features while the outcome is still challenging for high risk patients.[6] The high cost is also not affordable by majority of the families with a thalassemia major child. Thus, prevention of the birth of an affected child is a feasible and realist option. In addition the fetus may be able to act as a donor of stem cells for an affected mother. Despite current screening recommendations, as many couples are not aware that they are carriers, it is common for a child to be born with an unexpected, serious hemoglobinopathy. For this reason, newborn screening programs have been suggested to be introduced in most high-risk areas.

Since the last 30–40 years different Institutions in India as well as social organizations like the Rotary Clubs, Lions Clubs and various NGOs and Thalassemia Parents-Patients societies have been conducting education and awareness programs. Yet, awareness about β thalassemia among pregnant women was very limited. Another study about β thalassemia among a high- risk group, the Aroras in north India from a rural area in Rohtak district in Haryana and an urban area in New Delhi showed that a large number of individuals from the rural setting had not heard about thalassemia and the etiology of the disease was believed to depend on orthodox feelings like the sins committed by the parents.[14] Further, >50% of individuals from the urban area were not willing for pre-marital screening for β thalassemia.[11] This emphasizes the need for more intensive education and awareness programs in different parts of the country.

A feasible option for control is to promote education and awareness programs and develop adequate facilities for genetic counseling and prenatal diagnosis in public sector Institutions. The process involves pre-test counseling as well as post-test counseling to enable the individuals to face the situation and take appropriate decisions with the right frame of mind.

Major ethical principles which govern the attitudes and actions of counselors include: respect for patient autonomy, non-maleficence, beneficence, or taking action to help benefit others and prevent harm, both physical and mental, and justice, which requires that services be distributed fairly to those in need. Other moral issues include veracity, the duty to disclose information or to be truthful, and respect for patient confidentiality. Nondirective counseling, a hallmark of this profession, is in accordance with the principle of individual autonomy.

A critical mass of trained genetic counselors who have understanding of the ethical issues and its appropriate handling with the required sensitivity is needed in India. Moral problems arise constantly in social life with the need to resolve conflicts between moral rules and principles to help, regulate and modify desires.[12]

When genetic risks are high, the desire to have a healthy child and to avoid danger to oneself, family and society are frequently in conflict. Although more than 90 per cent of the counseling sessions end well with no or very little chances for the occurrence of the disease, the remaining people in the high risk category are left with options like (i) prenatal diagnosis and abortion if required, (ii) artificial insemination, and (iii) gene therapy. Genetic counseling and allogeneic stem cell transplant is unaffordable for the majority of families. Government and non-government organizations have been working towards this goal for the last 3 to 4 decades but community control in a vast and diverse country is challenging and a national program reaching all rural regions where almost 70% of the population resides is yet to begin. The practical method of calculating risk figures, intended for information regarding the unborn, and we ought to use it in an efficient manner but in a direction, which our ethics and morality point to. The decision taken by the parents after the counseling session must leave them satisfied instead of placing them in a state of dilemma.[13]

It is important that genetic counselors continue being patient advocates while ethical guidelines for their use with appropriate counseling procedures are developed after ensuring adequate human resource in this area. Policy makers and the professionals have equal responsibility in this regard.[12] Public awareness is the need of the hour. The crux of genetic counseling is to make the family aware of the genetic disorder, its clinical presentation and severity along with the risk of recurrence and mortality associated with the disease. Communication should be in simple language taking

care of the family history, psychological issues, ethical issues and cultural and religious challenges.[7] The family should be given all the available options including prenatal diagnosis along with a reassurance of having an unaffected child after prenatal intervention. Couples for whom termination of pregnancy is unacceptable due to cultural or religious reasons can be given an option of pre -implantation diagnosis or artificial insemination with a donor who is not a carrier of β thalassemia or another hemoglobinopathy or even adoption of a child. However, there are very few trained counselors in the country and this aspect needs strengthening.[8] Hence, before implementing it as a policy all these aspects need to be put in scrutiny before making it into a blanket rule which might only end up in increased burden on the people where proper government back up is not available to get either the test or the back up for the results of the tests.

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Reflection on Non-Invasive Prenatal Testing

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A. Background

The recent draft policy on For Prevention and Control of Hemoglobinopathies – Thalassemia, Sickle Cell Disease and variant Hemoglobin's in India is an excellent and comprehensive one. This policy on hemoglobinopathies encompasses the public health goals of prevention to reduce their prevalence, empower prospective parents to exercise their right not to have a child with a serious genetic disorder but have a normal child, and protects the rights of an affected child to have access to optimal care. The policy also talks about screening of pregnant women, preferably in the first trimester, for carrier status, and for those who test positive, screen their husbands and enable the at-risk couples to avail services for prenatal diagnosis to prevent the birth of an affected child. (1)

Though the policy talks about prenatal diagnosis, it is silent about what next, if the child is tested positive. In one sense the silence is good, since as a nation we do not have a framework to guide, control or limit the outcomes of pre-natal testing other than the MTP act. But it does leave one with a disturbing issue for the future. If as a nation pre-natal testing becomes a policy, would it lead to an increase in numbers of termination of pregnancy like what has happened in other nations?

A review of international research studies done on Downs syndrome (DS) has found that NIPT (Non-invasive prenatal testing) identifies 97% of fetuses with Down's Syndrome. In the UK, statistical predictions have calculated that NIPT would result in around 3,368 fewer invasive tests and therefore an estimated 17 fewer miscarriages per year. However, the same predictions also calculate that 195 more fetuses with DS would be detected each year. Since the proportion of women having a termination after a diagnosis of DS ranges from 89–95%, this would result in around 180 babies with DS being aborted each year than is currently the case. (2)

If this is the case for a small nation like UK, what would be its equivalent on the societal attitudes in a nation like India, where NIPT has the potential to throw up large numbers of diseases diagnosed early in pregnancy thus leading to potential increases in medical termination of pregnancy?

Few days back, 29 weeks gestation Primi gravida, was brought with fetal hydrops, with very poor chances of survival for the baby, to our labor room. The family wanted the pregnancy to be terminated. But the questions that challenged our team was, though the fetus has very low chances of survival, can we terminate the pregnancy? Since there was no threat to mother's health and the pregnancy had crossed the limits of legal termination of 20 weeks, we had to counsel the family to allow the pregnancy to continue till an intra uterine death happens or carry till full term, which the family was unwilling to.

Two clinical situations where pre-natal testing leaves one with confusing questions: One - if early diagnosis of non-life-threatening illnesses is made, would it lead to potential increase in terminations and change in societal attitudes? Two - At the same time for life threatening illnesses of the unborn child, what are the options available if we need to provide a support system for families?

B. What is Prenatal screening and NIPT or Non-invasive prenatal testing

Prenatal screening encompasses the entire array of medical tests that all pregnant women – depending on institutional and financial accessibility – qualify for during pregnancy. This is distinct from tests that may be indicated for some individual pregnant women in the context of looking for a specific abnormality that has already affected a child in their family. The latter cases constitute an individual diagnostic process and not population-based screening.

In recent years, new screening tests have been developed to identify chromosomal and genetic abnormalities of embryos and foetuses. These tests are non-invasive (NIPT) making use of DNA from the embryo or foetus circulating in the mother's blood.

Screening for chromosomal abnormalities is at present a stepped procedure: the first step is called a 'combined' or 'first-trimester test', which is performed at a pregnancy term of eleven to fourteen weeks. It combines ultrasound with biochemical examinations from maternal blood. So they are non-invasive as well, though typically the notion NIPT is only used for the new genetic tests for circulating DNA.

Pregnant women, in whom the combined test indicates an elevated risk of a child with an abnormality, are offered follow-up testing to determine whether the foetus has indeed a chromosomal abnormality. This is done using amniocentesis (at fifteen to eighteen weeks) or chorionic villus sampling (at eleven to fourteen weeks).

Currently, in several countries NIPT is offered as a second screening test to pregnant women diagnosed with an elevated risk due to a positive combined test. The advantage is that pregnant women receive a rapid, reliable and almost always reassuring (negative) test result. An invasive test would only follow in the event of an unfavorable (positive) NIPT result, because a final diagnosis is needed if the pregnant woman is to consider terminating the pregnancy. (3)

C.What are some of the prevailing frame works and goals for NIPT?

When prenatal diagnosis was introduced in countries in the late 1960s and early 1970s, there were three main motives for it:

1) the woman's health, quality of life and self-determination (or autonomy),
2) the child's health and quality of life and
3) economic considerations.

From the prevailing common understanding of NIPT, the goals of prenatal diagnosis can be summarized in the following points:

- To enable assessment of fetal health - to enable parents to make informed choices about how, when and what children they want (family planning)
- To enable the pregnant woman to make an informed choice about whether to carry the pregnancy to term
- To prevent individual suffering by treating a discovered impairment or disease in the fetus
- To prevent individual suffering by preventing the birth of a child with profound disabilities
- To improve public health by reducing the number of people suffering from severe diseases or disabilities

- To improve public health by reducing the number of carriers of severe hereditary diseases
- To save society money by reducing the cost of caring for people with severe disabilities and diseases. (4)

D. What are some of the challenges these goals throw up?

- **Autonomy of the mother and reproductive choices /rights and freedom of the mother**

The question of autonomy is a confusing one in the context of NIPT. Whose autonomy are we talking about? Is it autonomy of the mother, the family and or the unborn child? If unborn child, it throws up the issue of understanding the rights of the unborn, which is addressed below.

Reproductive choices about genetics has been an issue of ongoing controversy. While some people claim that parents have the right to make far reaching choices concerning their offspring, others hold that they should refrain from genetic testing of their future children if it is not necessary for their health. Some people underline the responsibility of parents to strive for the best possible health of their children even by intervening in their genes, whereas others stress the right of every human being to have an unmanipulated genetic make-up, so that nobody is brought into existence due to choices of other human beings with respect to their biological starting configuration, if it poses no risk of developing a disease of severity. (4)

- **Moral status and the rights on the unborn child**

There are diverging views in society about what moral status should be accorded to a fertilised egg. Three different positions can be distinguished: (4)

Human life begins at conception and the fertilised egg has full human dignity, i.e. a right to protection and an unconditional right to life.

The genesis of human life is a process in which the fertilised egg is a life in the making and has a certain moral status. This moral status increases gradually as the foetus develops. At the point in time when the foetus can survive outside its mother's body, its moral status becomes human dignity.

The fertilised egg has development potential but has no moral status.

NIPT and its outcomes are influenced by the understanding of these rights in a culture and nation. As a nation, we do not have a commonly accepted moral standard. Though by law, life in India is regarded to have started at the time of conception of child itself (debatable since the law is not clear on this), there are loop holes in existing acts for an unborn fetus to be terminated.

- **Protection of the rights on the unborn**

Many legal instruments exist to protect the welfare of those individuals with inability to fight for their own rights, legal representatives being one of them. However, all these instruments cannot protect the unborn child from being genetically tested in a comprehensive way, considering that the unborn child has a different moral and legal status in different countries.

- **The responsibility to future generations**

The responsibility to future generations is important because it respects the rights of those coming

into life later on. It is also important for our social relationships, for a society in solidarity and for justice between all peoples to keep in mind that the respect for the dignity of every human being entails the duty to refrain from making her or him a mere instrument for the fulfilment of the wishes and preferences of others. (3)

- **NIPT as routinization and institutionalization of the choice of not giving birth to an ill or disabled child**

The introduction of noninvasive prenatal diagnosis is being increasingly implemented as a routine measure during early stages of pregnancy, especially in countries with an established system of technique-based pregnancy care. This could have a major impact not only on reproductive freedom, but also on the perception of disability and on societal solidarity with disabled people and women who give birth to them.

The disadvantage of a simple, safe test may be that participation is considered self-evident and presented as such by care providers, especially when financed by health insurance. This may lead to pregnant women (and their partners) not fully realising that the test results may leave them with a major and possibly extremely difficult decision. Ironically, the introduction of a test that may bring informed choice to more pregnant women may undermine this goal in practice, if NIPT is used without thinking enough about its impact.

Furthermore, there is the risk that pregnant women with a positive result will not await the validation of the result through invasive diagnostics, but immediately choose to abort the embryo or foetus, without adequate counselling about the relevance of the detected abnormality. Also, women may feel pressured to submit to such screening. They might be stigmatized if they refuse to take the test. (3)

- **Erroneous or misinterpreted results and relevance of the unknown**

Pre-natal diagnosis is a developing field. We do not have adequate evidence for many newer tests and methodologies. A widespread use of NIPT to analyze more and more genetic features up to the entire genome would mean that the complexity of data would lead to a significant increase of false-positives, requiring a confirmation by invasive tests or of abnormalities whose relevance is not known at all, but this unknown might lead the parents not to take any risk.

- **The consequence of detecting a genetic abnormality therapeutic vs non-therapeutic**

Though newer NIPTs are currently promoted by the market forces as therapeutic in nature, there are very few clinical situations where a pre-natal therapeutic intervention is possible other than termination of pregnancy.

- **Perception of disability and on societal solidarity with disabled people and women who give birth to them**

A widespread use of NIPT, namely as general screening to detect abnormalities, followed by an abortion, is perceived by some people as an evidence of the will to avoid permanent pain in a lifetime, by others as a sign of a situation of exclusion that society gives to people affected by this illness, meaning indirectly, that certain lives are worth living, and others less. (3)

- **Population based testing and the potential impacts of the same**

Population screening is defined as the offering of medical investigations to people who have no

symptoms or other reasons to seek medical care for the conditions that are the target of the investigation. Screening is only justified if the usefulness of the intervention has been proven, and the advantages for the participants clearly outweigh the disadvantages. For most forms of screening, this means that health gains may be achieved through timely treatment or prevention. This also applies to prenatal screening programs for infectious diseases and blood group antigens.

The potential long-term outcome of screening of hemoglobinopathies in India, with no prenatal treatment or prevention interventions available, is an issue that needs much more careful consideration.

- **Public health justice and economy**

In a privatized and insurance-based economy, public health decisions are linked to cost of care. Some nations have already gone the way of calculating costs for caring of a disabled child and advocating for early termination as a potential intervention for cost effective health care access. Would that be the way for our nation in future if prenatal testing becomes routine and insurance systems come up?

E. What are some principles that should undergird our responses?

There are many more such challenges, but with these few in our purview, it is important to reflect on some principles to guide our responses as a Judeo-Christian value-based community.

a. What is our understanding of the moral rights of the unborn and when life begins.

If we believe that human life begins at conception and the fertilised egg has full human dignity, i.e. a right to protection and an unconditional right to life, then our approach to NIPT and its outcomes would be clearly guided by this overarching principle.

b. Our understanding of Autonomy

Christians are to seek autonomy that is not only limited by God's sovereignty, but also by concerns for the community and its common good. God has not only made us independent individuals but also placed us in families where 'mutual burdensomeness' is part of the created order. None of us can act in isolation and affect no one else. All of our decisions should take account of the needs of these shared relationships, not just our own wants and desires. (2) This understanding should guide our discussions on reproductive rights and choices.

c. Is prenatal diagnosis morally licit?

The Catholic church's stand is quite clear and succinct –

"If prenatal diagnosis respects the life and integrity of the embryo and the human foetus and is directed towards its safeguarding or healing as an individual, then the answer is affirmative.

But this diagnosis is gravely opposed to the moral law when it is done with the thought of possibly inducing an abortion depending upon the results: a diagnosis which shows the existence of a malformation or a hereditary illness must not be the equivalent of a death-sentence.

Thus a woman would be committing a gravely illicit act if she were to request such a diagnosis with the deliberate intention of having an abortion should the results confirm the existence of a

malformation or abnormality. The spouse or relatives or anyone else would similarly be acting in a manner contrary to the moral law if they were to counsel or impose such a diagnostic procedure on the expectant mother with the same intention of possibly proceeding to an abortion. So too the specialist would be guilty of illicit collaboration if, in conducting the diagnosis and in communicating its results, he were deliberately to contribute to establishing or favoring a link between prenatal diagnosis and abortion.

In conclusion, any directive or programme of the civil and health authorities or of scientific organizations which in any way were to favour a link between prenatal diagnosis and abortion, or which were to go as far as directly to induce expectant mothers to submit to prenatal diagnosis planned for the purpose of eliminating foetuses which are affected by malformations or which are carriers of hereditary illness, is to be condemned as a violation of the unborn child's right to life and as an abuse of the prior rights and duties of the spouses."

This understanding goes a long way in the way we would approach the matter personally and as a professional who must advise his or her patients and their families.

d. Family and its effects of having a disabled child

The belief that parents and families will be damaged by having a disabled child, and that this damage can be limited through abortion, is almost unquestioned today. However, psychological morbidity for mothers following abortion for disability is considerable and associated with long-lasting consequences for a substantial number of women. Research has found that women who terminate pregnancies for fetal anomalies experience grief as intense as those who experience spontaneous perinatal loss. (2)

e. Care for the weak

Throughout the Bible we see God's concern for the weak and, as stewards of his creation, we are called to emulate this; 'bearing one another's burdens' lies at the very heart of Christian morality. We must 'defend the weak' and 'help the weak'. This mandate involves compassionate caring, rather than seeking our own human means to obliterate weakness (and the weak) from the world. (2)

f. Providing a support for the weak and challenged

The Christian answer is clear that bearing one another's burdens is at the very heart of the gospel. We walk in the steps of the all-powerful Creator who laid aside everything and entered this world at great personal cost to rescue, care and serve. There is no doubt that providing life-time support for people with genetic disabilities can be costly in emotional and economic terms. This is where the Church is needed, to provide these most vulnerable of human beings with the honor, respect, love and protection that they deserve and to be places of support to their parents and families. (2)

This leaves us Christian health care professionals and the church at large, with the following questions and challenges.

- How can we develop a comprehensive understanding of NIPT and its potential outcomes for the individual, the family and society at large based on the values we hold on to?
- How can we advocate, influence the culture, the health care profession and policies at large by communicating the values we hold on to – life, when does it begin, the challenged and the

unborn and their rights, the value for the weak, and understanding of autonomy?

- How can we bear one another's burdens even the unborn and facilitate a community that is willing to bear one another's burden?
- How can we provide unbiased information to all stakeholders, foundation being our values, but at the same time being compassionate and caring, understanding the challenges the mother and family is going through?
- How can we facilitate the best medical care for all members of our society, especially the most vulnerable?
- How can we proactively support their families struggling with these issues and the child born with challenges in the longer term?

A comprehensive value-based understanding of the issues involved is important if we are to be the salt and light in the context of our nation, that does not seem to have an undergirding moral frame work to guide it policies.

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Should they be tracked down?

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The hemoglobinopathies comprise inherited disorders of the structure or synthesis of hemoglobin. They are the commonest single gene disorders in the world. It is estimated that about 4,50,000 infants with hemoglobinopathies are born each year in the world. 85.9% of these hemoglobinopathies are sickle cell disease (SCD). 1 Every year about 10,000 to 15,000 children are born in India with Thalassemia Major.

Considering the significance of this genetic disorder, Ministry of Health and Family Welfare, GOI, with the help of experts has developed a comprehensive, "Guidelines of Hemoglobinopathies in India". The purpose of the guidelines is to provide not only a better future for all patients affected by Thalassemia or Sickle Cell Disease, but also to **prevent the birth of children with such diseases…2**

The draft policy for prevention and control of hemoglobinopathies in India is a well thought out, comprehensive document. It has excellent ideas for early detection and prompt treatment of those suffering from hemoglobinopathies. However, **in the areas of prevention & control of hemoglobinopathies, there are deep ethical concerns.**

Adolescent screening:

In targeted carrier detection, positive results could burden young minds, hamper growth, prevent pursuing lives' goals, instill fear & mental stress. Adolescents who become aware that they are carriers, could become depressed and could attempt suicide.

Extended family screening:

Screening of all known and detected carriers and patients could lead to identifying a population of carriers, who may not be preferred for marriage, job opportunities etc. thus creating a sub set of communities.

Stigma & Discrimination:

Mapping of carrier status could create stigma and discrimination against those who are carriers. This could have huge repercussions in the society.

Violation of reproductive rights:

Counselling couples who are carriers not to have babies, will be intrusion into their private lives and violation of their reproductive rights.

Mandatory Screening:

Making it compulsory for ante natal mothers to undergo carrier detection, it will hinder the autonomy of the individual / couple to make informed decisions.

Maintaining confidentiality:

Screening adolescents, extended family members and couples for carrier status could make it almost impossible to keep the status confidential.

Subjecting vulnerable pregnant women to risks:

Amniocentesis, is an invasive test, by which 30ml of amniotic fluid in which the baby is floating is removed at 14-16 weeks. It is a risky procedure. It could lead to leaking amniotic fluid, miscarriage (6%), Needle injury to baby, RH sensitization, Uterine Infection and Transmission of Hep C, HIV etc. from mother to child. It is unethical to subject pregnant women to such risks.

Coercion to abort:

If the child within is found to be positive, the pregnant woman/couple could be forced to abort, thereby making them childless.

Right to be born:

Each human being has intrinsic value and so each human life has the right to be born irrespective of its potential abilities or limitations. With all the technological advances in medicine, every effort should be made to meet the specific needs of the population with hemoglobinopathies.

Negative Eugenics:

Eliminating the pre-born babies with hemoglobinopathies, is to say that only perfect babies should be allowed to be born. This negative eugenics could be a dangerous position to hold on to, as this could be extended to a number of other genetically transmitted diseases.

Dr. Jameela George MIRB
Executive Director, TCB
Manager, Research & Bioethics EHA

Draft policy on prevention and control of hemoglobinopathies

Jubin Verghese

How can one respond to the draft policy on prevention and control of hemoglobinopathies with sensitivity? It is possible to be logical and firm about one's opinions and yet, what we are talking about are lives and stories of parents and children. How can I possibly empathise with families who are living daily with the reality of the diagnosis and treatment of their children? And yet, I worry about the doors that will open with the screening of carriers and prenatal diagnosis of hemoglobinopathies.

Most tests used in medicine are used to treat disorders or at least to alleviate the suffering. It is often considered unethical to screen apparently health people if no treatment is available. Even in HIV counselling, the person is given the choice as to whether they would like to get tested or not, as part of the pre-test counselling. Our experience with the HIV counselling and testing has shown us that confidentiality does not really work in our country. If apparently healthy young people begin to be screened and labelled, who will take responsibility for the distress and discrimination they will face because of the test result?

The other concern is that the only option we are able to offer parents following a prenatal diagnosis is abortion. When did abortion become a treatment option? How long will it be before we move from offering abortion as an option(out of compassion) to expecting mothers to abort their affected fetus as a 'duty' to society to prevent the economic drain that their affected child will be? What of mothers who decline testing or decide to have the child despite the testing? Will they be considered 'socially irresponsible' for having had the child? Also, while counselling may be offered soon before the prenatal testing, what services will be offered to couples after the abortion to deal with the psychological consequences of it? Do we have the capacity to set up such systems?

Prenatal testing for hemoglobinopathies also opens the door wide for other disorders like spina bifida, Down's syndrome, muscular dystrophy etc., some of which is already being practiced. So where will we draw the line? As different genes get isolated for more and more disorders, will we do the same for all these as well, till only the fittest may survive?

Interestingly in these conversations the voices of those with these disorders is missing. Would they have preferred not be born at all than to have lived with this condition? Under the Rights for persons with disabilities act, 2016, thalassemia, sickle cell anaemia, haemophilia have been termed as a disability. Where are the voices of the disability activists in this issue? With regard to prenatal diagnosis, here is what Martha Saxton, one activist had to say," The message at the heart of prenatal diagnosis is the greatest insult: some of us are too flawed in our very DNA to exist. We are unworthy of being born…Fighting for this issue, for our right and worthiness to be born, is the fundamental challenge to disability oppression; it underpins our most basic claim to justice and equality – we are indeed worthy to be born, worth the help and expense and we know it !"

Finally, in the words of a bioethicist," It is claimed that antenatal screening is just another example of preventative medicine. But this is a novel redefinition of the word 'prevention'. To be strictly accurate, prenatal screening and termination do not prevent disability; they eliminate disabled individuals '. Is this the road we want to be on?

Jubin Varghese
Deputy Director, Community Health & Development Programme

Aims & Objectives of TCB

1. To be a Christian voice on ethical issues based on Biblical values

2. To analyze, interpret and engage with the existing and emerging bioethical issues pertaining to health care and research

3. To facilitate upholding the sanctity of life and dignity of humans in medical practice and research

4. To promote ethical medical practice

5. To build leadership in the field of Bioethics, in the areas of Medical education, Medical practice and Medical research

Prayer support:

TCB needs your prayer support to make a significant contribution to improve healthcare in India, through Bioethics.

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