ARCHDIOCESE OF MALTA

EMBRYO PROTECTION ACT (AMENDMENT) BILL NO.5

A POSITION PAPER

JUNE 2022

church.mt

Contents

Executive Summary

This position paper, prepared by a multidisciplinary group of experts, seeks to respond to the amendments that the Government is planning on introducing to the Embryo Protection Act. We believe that the 'Objects and Reasons' stated at the end of the Bill do not reveal the real objective of one of the most morally sensitive interventions that would be permitted under the proposed amendments to the Act.

The actual focus of the Bill appears to be pre-implantation genetic diagnosis (PGD) of monogenic disorders. How this fits into the objectives of the Bill is not clear.

The objective of the Embryo Protection Act is the protection of the human embryo and the legitimate interests of the future child. Introducing in the principal Act a provision designed directly to prevent a human embryo from being born, because the embryo has a genetic disorder, makes a mockery of what the principal Act intended originally to do.

The proposed amendment provides the option that embryos with a genetic condition may be offered for adoption. This is not a realistic offer but a 'mute option' from the outset and an 'on-paper only' exercise. Even worse, it is a ploy to assuage the moral concerns related to the discarding of such embryos. Among other concerns we list the following:

First, the Bill states that a Protocol will be drawn up by the Embryo Protection Authority, listing the monogenic conditions that will be subject to this kind of selection. So far, the list of conditions that will be included in the Protocol is unknown and therefore legislators cannot deliberate on the consequences of the Bill until there is full disclosure.

Second, irrespective of the conditions that will be listed by the Protocol, no embryo diagnosed with any condition should be subject to perpetual cryopreservation. To choose between embryos on the basis of their genetic make-up is discriminatory and constitutes a eugenic practice.

Third, the possibility of adoption of these embryos is highly unlikely. The reason is that so far, none of the 'surplus' embryos that have been cryopreserved since 2018 have been adopted by unrelated couples. It is obvious, therefore, that these embryos will be preserved for perpetuity.

Fourth, the Bill states that it is the Embryo Protection Authority which determines what constitutes and what does not constitute a eugenic practice and what is the maximum number of fertilized egg cells to be allowed in each treatment cycle. This shifts the decision-making authority from Parliament to the Embryo Protection Authority. The Bill removes the regulation of fundamental and controversial decisions from scrutiny by the House of Representatives, indispensable to a well-functioning democracy, and confers this function on unaccountable and political persons of trust.

Therefore, rather than protecting the embryo, the proposed amendments violate its dignity, by freezing embryos with certain genetic defects forever. Moreover, the selection of embryos to be cryopreserved, as the Protocol may determine at some future date, amounts to a eugenic practice and undermines the dignity of persons already living with such conditions.

Position Paper

Introduction

1. This position paper, which has been prepared by a group of experts in clinical medicine, basic sciences, embryology, health sciences, law, psychology, well-being, social policy, family studies, disability studies, philosophy and theology, is not meant to discuss all the ethical aspects of assisted reproductive technology (ART), including *in vitro* fertilization (IVF), but to respond to the amendments that the Government is planning to introduce to the *Embryo Protection Act*. Generally speaking, the section on the 'Objects and Reasons' of the Bill should provide a helpful context for interpreting the amendments that are proposed. In this case, however, the 'Objects and Reasons' that are listed do not reveal the objective of one of the most morally sensitive interventions that would be permitted under the proposed amendments to the *Embryo Protection Act.*

2. The 'Objects and Reasons' are: to provide the necessary legal framework to facilitate oocyte and sperm distribution, to increase the opportunity for more individuals to have a successful IVF outcome and to provide the necessary legal framework to facilitate embryo adoption for the prospective parent or parents. How pre-implantation genetic diagnosis (PGD) for monogenic disorders[1] fits within this set of reasons and objectives is difficult to understand, unless it is implied in what it is being described as "a successful IVF outcome". If that is the case, the genetic testing of human embryos, conceived *in vitro* to detect hereditary genetic disorders prior to implantation, is a game-changer to the existing legislation.

The Embryo Protection Act

3. The very title of the *Embryo Protection Act* encapsulates what should be its basic concern and central ethical principle. Its objective should clearly be the protection of the human embryo and the legitimate interests of the future child. Introducing in the principal Act a provision designed directly to prevent a human embryo from being born, because the embryo has a genetic disorder, makes a mockery of what the principal Act originally intended to do.

4. In the existing law, the central focus is supposed to be the dignity of the human embryo - a dignity that calls for protection. The freezing of human embryos introduced in the 2018 legislation has already gravely weakened the protection of the human embryo. Currently, there are in Malta more than 300 human embryos who are frozen waiting to be adopted. So far no request for adoption of these embryos in suspended frozen animation has been submitted, even though they have not been tested for genetic disorders.

5. Yet the proposed amendment provides the option that embryos which will now have a confirmed genetic disorder may be offered for adoption. This is not a

[1] The Bill does not itself make reference to monogenic disorders. These have been reported by the Minister of Health during the press conference held on 26th May 2022.

realistic offer at all; this is a 'mute option' from the outset and no more than an 'on-paper only' exercise. Even worse, it is a ploy to assuage the moral concerns related to the discarding of such embryos.

Ethical Aspects

6. The proposed amendments may give one the impression that chromosomal and monogenetic disorders have contributed to the relatively low success rates of IVF procedures which have recently come under scrutiny in the media.[2] It is pertinent to ask whether formal local studies have revealed the statistical contribution of chromosomal and monogenic disorders to the IVF success rate in Malta. The proposed amendments will clearly shift the *locus standi* of the embryo from central to distant peripheral. It is but a matter of time between the *freezing* of a 'defective embryo' for an indefinite period of time to its *elimination*, as a 'defective foetus', once detected in early pregnancy, a few months later.

7. Indeed, the status of the human embryo has been downgraded from a crucial player with independent rights and deserving of dignified respect, to little more than a commodity. The amendments proposed in Bill No.5 continue to dismantle rather than consolidate the legal protection of human embryos since they broaden the options of assisted procreation precisely at the expense of the full protection of the human embryo, the most vulnerable human being in society.

8. Act XXI of 2012 specifically prohibits eugenics, that is, the intention of creating a society based on 'good genes'. In its present format, the Bill No.5 proposes that "in paragraph (e) thereof the words "eugenic purposes;" shall be substituted by the words "eugenic purposes:" and immediately thereafter there shall be added the following new proviso: 'Provided that the Protocol may specify that certain exceptional circumstances shall not constitute selection of embryos for eugenic purposes.'" Though the Bill maintains the clause "forbidding eugenics", its import is immediately nullified by allowing *exceptions* to be included in the, as yet, unrevealed protocol that will build upon the proposed amendments. In effect, embryo selection for eugenic purposes will be allowed after all!

9. During a press conference, held on 26th May 2022, the Minister of Health, the Hon. Dr Chris Fearne, stated that discarded embryos which have genetic defects will be frozen for eventual adoption. What the Minister failed to mention during the press conference was that according to the Annual Report of the Embryo Protection Authority that he himself presented to Parliament on the 16th May 2022, "since the introduction of the amendment law in 2018, a total of seven hundred and fifty-two (752) embryos have been cryopreserved for future use, thus bringing the total to seven hundred and fifty-nine (759) cryopreserved embryos. Noteworthy is the fact that four hundred and eighteen of which were cryopreserved during the calendar year of 2021. ….. Whilst two hundred and twenty-nine (229) embryos were thawed to be used in embryo transfer procedures carried out in 2021. All embryos thawed survived the thawing

[2] "[T]he maximum percentage success rate, or 'Take Home Baby' rate for calendar year 2021 is 21.13%", Embryo Protection Authority, *Annual Work Report: Trends and Figures for Fertility Treatments in Malta for 2021*:190.

process. Thus, the total number of embryos cryopreserved as at the end of 2021 stood at three hundred and eighty-eight (388), nearly double the number of embryos stored as recorded at the end of 2020 which stood at 197 embryos."[3]

10. Over the past two years, the number of frozen embryos has risen from 180 to 388, yet not a single embryo from this substantial stockpile has been adopted by a third party. It is therefore highly unrealistic to expect that any human embryo known to have a significant genetic defect and already discarded by its own parents, would be considered for adoption by unrelated couples. By definition, these embryos will be classed as 'rejects' and they would be eventually discarded. A eugenic practice will be introduced 'by stealth', under the umbrella of embryo freezing and the unrealistic assumption of eventual adoption. The Bill in article 5 states that the Embryo Protection Authority is "obliged to provide the prospective adoptive parents with medical and, or diagnostic information concerning the embryo, as specified in the Protocol."

11. The Bill attempts to make the proposed changes more palatable to the moral conscience of the general public. It is also an attempt to politically hedge the issue of eugenics that is being proposed in the Bill. The fact remains that some embryos will be selected for elimination through perpetual freezing. Our moral and political conscience cannot be appeased by giving the impression that Malta's stand on abortion is not being changed, when it is evident that with the introduction of the proposed amendments those embryos which are unwanted because of some genetic defect will be frozen for ever.

12. The Bill proposes that a Protocol, drawn up by the Embryo Protection Authority, should list the genetic conditions that permit the freezing of the embryo. To date, this protocol has not been published, while the Bill is being rushed through Parliament. How can the legislators deliberate on the consequences of the proposed Bill without having full disclosure? It would be interesting to know the list of conditions which the Government and medical personnel deem to be unacceptable in our society.

13. The argument that an embryo with a genetic condition, if it is frozen, can still benefit from what medicine might discover in future to deal with genetic defects and may be eventually adopted, is based on a narrow model of thinking about disability. In fact, it runs counter to what has been achieved in many countries, including ours, as a result of a social model of thinking about disability. According to this model, society has a particular responsibility to provide persons with disabilities with those facilities and resources that enable them to meet their own special needs in order to flourish.

14. When a human embryo is created *in vitro*, both its parents and the medical experts are more compelled to think that, because they invest so much in the process – emotionally, financially and in terms of technical expertise – they have a 'duty' to ensure that the finished 'product' meets everyone's expectations by subjecting the human embryo to quality control. The reasoning behind pre-

[3] Embryo Protection Authority, *Annual Work Report: Trends and Figures for Fertility Treatments in Malta for 2021*, 142.

implantation genetic diagnosis reflects this technical and consumerist mindset. While apparently seeking the best outcome for all concerned, it transforms the human embryo into a commodity that can be manipulated to achieve a predetermined 'ideal' or desired standard. PGD is not a guarantee of a newborn without genetic abnormalities. PGD itself has many limitations such as in detecting microdeletions and microduplictions, *de novo* variants, imprinting disorders and the detection of mosaicism during PGD aneuploidy.

15. Diagnosis before implantation is usually immediately followed by the elimination of an embryo suspected/found to have genetic or chromosomal defects or having other qualities that are not wanted. Cases are becoming more prevalent in which couples who have no fertility problems are using artificial means of procreation in order to engage in genetic selection of their offspring. Children have the right to be welcomed unconditionally from the start of their life. Human dignity belongs equally to every single human being, irrespective of one's parents' desire, quality of life and level of physical or mental development. All deliberate discarding or destruction of human embryos on the basis of disability or undesirable traits is ethically unacceptable. Society has a moral duty to protect and safeguard the vulnerable human embryo from any form of injustice and discrimination.

16. Genetic screening of pre-implanted embryos is fundamentally discriminatory - targetting some embryos as not being fit to be born. By choosing to discard embryos with genetic impairments, it is being assumed that the life of someone with an impairment is not worth living. Who is to tell, or indeed predict, what one's life will be like once one is born? Who is going to decide which conditions are acceptable and which are not? What are the criteria that this Authority will adopt to make its decisions? The emphasis here is only on the 'burden' or the 'suffering' of a person who has an impairment. Indeed, there is much more to the life of a person who has a disability. Instead of discarding 'imperfect' embryos, we should be focusing on providing more support services, an environment which is more accessible to persons with disabilities, and a more inclusive society that is free from such assumptions and myths. People with disabilities can contribute much to society if provided with the right support and opportunities - but they have to be given a chance to live first!

Medical Considerations

17. The Bill does not specify which technique of pre-implantation genetic testing (PGT) will be introduced in Malta. PGT consists of either pre-implantation genetic diagnosis (PGD) or pre-implantation genetic screening (PGS). PGD is offered to a fertile couple who are known to be carriers of a particular monogenic disorder, in order to prevent the couple from having an offspring with that particular disorder. PGD does not increase the success rate of IVF, because the couple do not actually have a problem of infertility. It is PGS which may increase success rates in infertile couples who have recurrent IVF failures or unexplained recurrent miscarriages. However, PGS is never recommended for routine use, as there is no evidence that it actually improves the take-home baby rate of IVF.

18. The principal scope behind PGD is to ensure that offspring conceived by IVF do not carry chromosomal or monogenic disorders. The process involves the study of the cell's chromosomes for chromosome gains/losses, such as those brought about by translocations, inversions, deletions, and insertions, related to parental structural chromosomal abnormalities. PGD can be carried out at the (1) pre-fertilization phase by testing the polar body associated with the maternal ovum, or (2) post-fertilization by testing cells obtained by biopsy from the periphery of the blastocyst or trophoectoderm before embryo transfer and implantation.[4]

19. The former, i.e. polar body testing, has the advantage that the test is done prior to fertilization and it would, therefore, not involve unwanted embryos that are then discarded or frozen in perpetuity. Its disadvantage is that it provides information only about the maternal contribution to the embryo and gives no information about the paternal contribution. Unlike post-fertilization testing methods, polar body biopsy does not raise any concerns about the potential risks of embryo biopsy, and extended embryo culture. Post-fertilization biopsy removes cells that are destined to form the placenta, increasing the risk of pathological placentation, and has been associated with the development of pre-eclampsia and reduced foetal growth – conditions associated with significant maternal and foetal morbidity and mortality.

20. Blastocyst biopsy has the advantage of giving a definite whole genetic picture of the produced embryo checking for both the maternal and paternal genetic contributions. Its disadvantage is that it requires the process of fertilization and therefore embryos found with a genetic variant are then discarded or in the case of the proposed Maltese legislation cryopreserved in perpetuity. This is nothing more than selective eugenics where identified genetically defective embryos are not given the opportunity to develop as other embryos.

21. Currently, PGD focuses on three domains: identification of (1) single gene defects; (2) chromosomal abnormalities and (3) triploidy. In the light of the above discussion, it is anticipated, that PGD will be introduced by degrees, starting with a very small number of rare conditions with single gene defects with extremely poor clinical outcomes and for which there is no cure (e.g. GM1 gangliosidosis that results in approximately 1 case every 2-5 years in Malta). Indeed, selecting out these embryos and not opting to use them for implantation in an IVF programme will be seen as an act of kindness to many. The introduction of this practice will, however, open the path for the introduction of PGD for other diseases due to single genetic mutations where treatment options are plentiful. Cystic fibrosis (CF) is one such example, with an incidence of 1 in 5-10,000 and where great efforts as well as finances are directed toward their care per annum.

22. Indeed, the Government has only recently approved a new, extremely expensive drug for use in a subgroup of CF patients, with hugely encouraging results. Any proposal that allows the introduction of a very small number of rare, single gene diseases, yet with the option to allow the Embryo Protection Authority to decide on the addition of other diseases will, yet again, widen the scope for selective

[4] E Greco, K Litwicka, MG Minasi, E Cursio, PF Greco, P Barillari. "Preimplantation Genetic Testing: Where We Are Today?" *Int. J. Mol. Sci.,* 21, no. 12(2020): 4381; https://doi.org/10.3390/ijms21124381

eugenics. PGD for chromosomal problems such as the trisomies (e.g. Trisomy 21, Down Syndrome, with an incidence of 1 in 600 live births, being the most common), is likely to be met with opposition from interested parties. PGD for triploidy, a very rare condition that usually results in miscarriage or very early death, will not have a significant impact.

23. One may argue that it is in the best interest of human embryos detected with a debilitating disease to be frozen until a therapy has been developed for these conditions. There is no guarantee that gene-editing techniques would be fast enough to offer these embryos such a therapy in the near future. Gene therapy (despite of the advances made with the use of CRISPR-Cas9 technology) is still inherently difficult and unlikely to be offered in the near future. Moreover, clinical application of gene therapy is still fraught with many uncertainties, knowledge-gaps and safety/risk assessment issues. In fact, there are no scientific publications which show the application of gene therapy on embryos produced by IVF leading to a livebirth. This is because these embryos can only be used for experimentation. Thanks to advances in medicine, an ever-increasing number of genetic diseases can be treated after birth. Thus this PGD would have prevented the child with a condition where already existing and developing therapies can treat this child. However there are other conditions which may be more difficult to treat adequately and where this perpetual freezing following PGD will simply discard some individuals as unfit to live - converting the curative part of a doctor's role to the individual to that of a gatekeeper of the Eugenic Health of a society.

Legal Considerations

24. The Bill, by allowing the Embryo Protection Authority (a) to adopt a Protocol to determine which exceptional circumstances do not constitute selection of embryos for eugenic purposes; and (b) what maximum amount of fertilized egg cells is to be allowed in each treatment cycle, is shifting the decision making authority from Parliament to the Embryo Protection Authority. Democratically speaking, this is a move in the wrong direction as the authority of Parliament is being bypassed and Parliament’s role in the Embryo Protection Act as it exists today and as it has so existed since the enactment of this law, is now being assigned to a government-appointed authority. The Bill removes the regulation of fundamental and controversial decisions from the scrutiny by the House of Representatives, indispensable to a well-functioning democracy, to confer this function on unaccountable and political persons of trust.

    Whilst parliamentary sessions are held in public which can follow the debate even on Parliament’s television channel with the possibility for consultation and discussion with civil society organisations and organised interests, this popular consultation, discussion, and participation will be entirely removed when the 2022 amendments come into force as it is the Embryo Protection Authority that will reach its decisions on these fundamental matters without the public’s involvement and behind closed doors.

25. From a rule of law perspective, the democratic element is cheapened and the Parliament’s authority is being bypassed in favour of a government-appointed authority. This proposed abdication of responsibility by Parliament will diminish

its control role over the Government and will not allow it, any longer, to exercise its supervisory function over Government to ensure good governance. This is indeed not a measure that leads to enriched good governance and augmented transparency.

Moreover, there is no requirement that before its approval the said Protocol will be published in draft form, tabled in the House of Representatives and subjected to a consultation process. On the contrary, both Parliament and the public will be left out of the whole approval procedure and faced with a *fait accompli*. It is only discussed in the Parliamentary Committee for Health *after* its publication and the said Committee has no power to change it or amend it.

26. Furthermore, there is no time-limit established within the date for the publication of the protocol. If at all, it should be published in *The Malta Government Gazette*, in two daily newspapers (one in English and one in Maltese) within a week from its adoption by the Embryo Protection Authority and on the Embryo Protection Authority's website. The minister responsible for health should also lay it on the table of the House of Representatives following its publication in the *Government Gazette* within a week if Parliament is in session; if not, within one week after resumption of sessions.

    Finally, as the protocol will not be published as a Legal Notice, it cannot be challenged in the House of Representatives through the negative resolution procedure. It is not even clear whether it can be challenged in court or, should this be possible, whether its effects are to be suspended until the courts would have decided any litigation brought in relation to the Protocol.

Concluding Reflections

27. We recognise and share the pain and anguish of those prospective parents who are unable to have children of their own or of couples burdened with the knowledge that they might be carriers of a genetic variant. We therefore reiterate our heartfelt appeal to the authorities not to continue using a rhetoric which only increases the desire for a baby without any 'defects' at all costs which, notwithstanding everything, remains elusive. Instead, we call for more support services for couples going through fertility treatment, education to end the stigma of infertility, smoothening of complex bureaucratic adoption procedures, better genetic counselling services and a culture that is more welcoming to persons with disabilities.

28. Polar body biopsy is a viable alternative to preimplantation genetic diagnosis (PGD). Polar bodies are by-products of the meiotic cell cycle which have no influence on further embryo development. The biopsy of polar bodies can be accomplished either by zona drilling or laser drilling within a very short time period. However, the paternal contribution to the genetic constitution of the developing embryo cannot be diagnosed by polar body biopsy. The major application of polar body biopsy is the detection of maternally derived chromosomal aneuploidies and translocations in oocytes. For these indications, polar biopsy may offer a viable alternative to blastomere biopsy as the embryo's integrity remains unaffected, in contrast to preimplantation genetic diagnosis

(PGD) by blastomere biopsy. Compared to a blastocyst biopsy, a polar body biopsy can potentially be of lower costs, cause less harmful side-effects, and can be more sensitive in detecting abnormalities.[5] Polar body biopsy has been proven to be sufficiently effective for the diagnosis of structural and numeric chromosome aberrations in human oocytes with the use of FISH and array-CGH.[6]

29. In situations where the paternal rather than the maternal genetic makeup is abnormal, there exist alternative artificial reproductive technology (ART) options, such as gamete or embryo adoption, to help the couple achieve a healthy viable pregnancy. The latter would help mitigate the increasing problem of frozen embryos destined to suspended animation for perpetuity. One may find gamete donation morally objectionable but why should the law allow a genetically defective embryo be frozen forever when the problem can be legally solved by gamete donation?

30. One can anticipate that the amendment are likely to allow for the introduction of PGD for a select number of extremely rare and single gene disorders, but with a covert provision to expand the list of named disorders by devolving responsibility to a named committee. The door will be opened to a myriad of other, single gene disorders and, eventually, other chromosomal defects, and pre-implantation genetic screening. The resulting defective embryos will be rejected by the biological parents and, in all probability, by any third party. Although the current amendment allows for these embryos to be offered for adoption, this is hopelessly unrealistic. An ever-increasing number of such unwanted embryos will be frozen, until such time that further amendments to the Bill will ensure that they can be disposed of legally. The Bill, in its current format, is facilitating selective eugenics and paving the way for the destruction of human life.

[5] A Kuliev, S Rechitsky. "Polar body-based preimplantation genetic diagnosis for Mendelian disorders," *Molecular Human Reproduction*,17, no.5 (2011):275-285, https://doi.org/10.1093/molehr/gar012; RTScott, NR Treff, J Stevens, EJ Forman, KH Hong, MG Katz-Jaffe, WB Schoolcraft, "Delivery of a chromosomally normal child from an oocyte with reciprocal aneuploid polar bodies." *J Assist Reprod Genet*, 29, no.6 (2012):533–537, https://link.springer.com/article/10.1007/s10815-012-9746-6

[6] M Montag, M Köster, T Strowitzki, B Toth, "Polar Body Biopsy," *Fertility & Sterility*, 10093: (2013): 603-607, https://www.fertstert.org/article/S0015-0282(13)00694-8/fulltext.

Signatories

Mgr Professor Emmanuel Agius, Professor, Department of Moral Theology
Professor Kevin Aquilina, Professor, Department of Communications and Technology Law
Dr Michael Asciak, Senior Lecturer, Institute of Applied Sciences
Dr Josephine Attard, Senior Lecturer, Department of Midwifery
Rev. Dr Stefan Attard, Senior Lecturer, Department of Sacred Scripture (Dean, Faculty of Theology)
Professor Simon Attard Montalto, Professor, Department of Paediatrics
Professor Andrew Azzopardi, Professor of Disability Studies and Community Development (Dean, Faculty for Social Wellbeing)
Dr Daniel Bianchi, Lecturer, Department of Media, Communications and Technology Law
Judge Emeritus Giovanni Bonello
Professor Tonio Borg, Professor, Department of Public Law
Dr Roderick Bugeja, Lecturer, Department of Nursing
Professor George G. Buttigieg, Consultant in Obstetrics and Gynaecology/Medico-Legal Studies
Rev. Dr Carlo Calleja, Lecturer, Department of Moral Theology
Professor Jean Calleja-Agius, Professor, Department of Anatomy
Professor Ann Marie Callus, Associate Professor, Department of Disability Studies
Ms Astrid Camilleri, Visiting Senior Lecturer, Department of Obstetrics and Gynaecology
Dr Maria Cassar, Senior Lecturer, Department of Nursing
Professor Maureen Cole, Associate Professor, Department of Social Policy and Social Work
Professor Nadia Delicata, Professor, Department of Moral Theology
Dr Bridget Ellul, ELSI Adviser to Biobank, Centre for Molecular Medicine and Biobanking
Dr Ruth Falzon, Senior Lecturer, Department of Counselling
Dr Rosienne Farrugia, Senior Lecturer, Department of Applied Biomedical Science
Professor Alex Felice, Professor, Faculty of Medicine and Surgery
Mr Ivan Galea, Consultant in Obstetrics and Gynaecology
Dr Vickie Gauci, Lecturer, Department of Disability Studies
Mgr Professor George Grima, Professor, Department of Moral Theology
Mr John Mamo, Consultant in Obstetrics and Gynaecology
Dr Nikolai P. Pace, Senior Lecturer, Department of Anatomy
Dr Roberta Sammut, Senior Lecturer, Department of Nursing
Dr Mario Thomas Vassallo, Senior Lecturer, Department of Public Policy
Professor Charles Savona Ventura, Professor, Department of Obstetrics and Gynaecology
Mr Albert Paul Scerri, Consultant in Obstetrics and Gynaecology
Professor Pierre Schembri Wismayer, Professor, Department of Anatomy
Rev. Professor Mark Sultana, Professor at the Department of Philosophy
Dr Josef Trapani, Senior Lecturer, Department of Nursing (Deputy Dean, Faculty of Health Sciences)
Rev. Dr Ray Zammit, Senior Lecturer, Department of Moral Theology

Addendum

Observations on the draft Protocol tabled in Parliament on Wednesday 8th June 2022

1. With reference to the draft Protocol including the list of conditions approved by the Embryo Protection Authority for pre-implantation genetic testing for monogenic diseases (PGTM), one notes that 8 out of the 9 listed conditions are *autosomal recessive*.

2. These *autosomal recessive* conditions are: Finnish Nephrotic Syndrome; Gangliosidosis; Joubert Syndrome; Maple Syrup Urine Syndrome; Nemaline Myopathy; Spinal Muscular Atrophy; Tay-Sachs Disease and Walker-Warburg Syndrome.

3. These conditions can be tested using genetic testing on the parents, followed, if need be, by polar body biopsy of the oocyte.

    - If the mother is not a carrier, then she does not need to be tested and the embryo will not be affected by the condition. In fact, if only the father is the carrier, the embryo will have a 50% chance of being completely normal and not a carrier, and a 50% chance of being a carrier, like the father, without ever expressing the disease.

    - If the mother is indeed a carrier of an autosomal recessive gene, then polar body biopsy will help determine which oocytes do not have the autosomal recessive gene, and these are the ones chosen for fertilisation to create the embryo. In this case, the embryo will have a 100% chance of being completely normal and not a carrier. In case that the father is also a carrier, then the embryo has a 50% chance of being normal and 50% chance of being a carrier like the father, without ever expressing the disease.

4. There is absolutely no added advantage to offer PGTM on the embryo, over polar body biopsy on the oocyte, because if the oocyte is not affected by the recessive gene, then the embryo will also not be affected and will not be a carrier either.

5. Out of this list, Huntington Disease is the only disease which is *autosomal dominant*, meaning that even one copy from either the affected mother or affected father will lead to the disease in the affected parent, and even in the embryo. However Huntington's is a particular disease which will show up later in life (typically after the age of 40 years). The parents will be tested to check whether they are carrying that gene, and if one of them does, it is clear that there is a 50% chance that their offspring will develop the disease later in life (above the age of 40 years). This means that this condtion is not immediately

life threatening, and there is medication which can help to alter the course of the disease (eg. Xenazine (tetrabenazine).

6. Just in the case of cystic fibrosis (which we gladly note is not included in the list of proposed diseases), medical advances are continously being made to treat these conditions, where effective treatment like Kaftrio are now available and are even being provided for free by the Maltese government to affectd patents (https://timesofmalta.com/articles/view/life-saving-cystic-fibrosis-drugs-to-be-added-to-government-formulary.859111). This is what needs to be encouraged – elimination of the illness and not of the ill persons.

ARCHDIOCESE
OF MALTA