

## MEDICINE AND THE LAW

## Pre-implantation diagnosis to create 'saviour siblings': A critical discussion of the current and future legal frameworks in South Africa

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Pre-implantation genetic diagnosis (PGD) is a technology used in conjunction with *in vitro* fertilisation to screen embryos for genetic conditions prior to transfer. It was initially developed to screen mutations for severe, irreversible, genetic conditions. Currently, PGD makes it possible to select against more than 100 different genetic conditions. It has been proposed as a method for creating a tissue-matched child who can in turn serve as a compatible stem cell donor to save a sick sibling in need of a stem cell transplant. The advantage of this method is that it provides genetic information before implantation of an embryo into the womb, making it possible to ensure that only tissue-matched embryos are transferred to the uterus. A couple can therefore avoid the difficult choice of either terminating the pregnancy at a later point if the fetus is not a match, or extending their family again in the hope that their next child will be tissue compatible.

Many people have expressed disapproval of the use of PGD for this purpose, and it is associated with many conflicting interests

including religion, ethics as well as legal regulation. In order to manage these issues some jurisdictions have created legal frameworks to regulate the use of this technology. Many of these are modelled on the UK's Human Fertilisation and Embryology Authority and its guardian legislation.

This paper critiques the current and future South African legal framework to establish whether it is able to adequately regulate the use of PGD as well as guard against misuse of the technology. It concludes that changes are required to the future framework in order to ensure that it regulates the circumstances in which PGD may occur and that the Minister of Health should act expediently in finalising draft regulations which will regulate PGD in the future.

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New reproductive technology has made the impossible possible. However, this comes with a cost, as many new reproductive advances have negative and positive implications. Advances have enabled people to improve on existing biological functions. With the advent of *in vitro* fertilisation in 1978, childless couples could be assisted with procreation.<sup>1</sup> In the 1980s, scientists successfully removed early embryonic cells from human embryos for genetic screening against debilitating conditions. This can enable parents to prevent a child from inheriting serious genetic conditions. Used for other purposes, it can ensure that a child has specific characteristics such as a certain hair or eye colour, or it can even be used to create a new child who has the genetic capacity to assist a dying sibling – the so-called 'saviour sibling'.<sup>2</sup>

New reproductive technology, such as pre-implantation genetic diagnosis (PGD), poses complex and competing religious, ethical, moral and scientific interests. Accordingly, there has been a move towards its legal regulation in various jurisdictions, including South Africa. We investigate the current and future South African legal frameworks to assess whether our law allows for pre-implantation diagnosis, effectively regulates the procedure, and protects the rights of both siblings, i.e. the saviour brother or sister (if born alive) and the dying or seriously ill child.

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### Pre-implantation diagnosis – new hope for parents with dying children

PGD was developed to screen embryos for severe irreversible genetic conditions (such as sickle-cell anaemia, Tay-Sachs disease, Duchenne's muscular dystrophy and beta-thalassaemia) before *in vitro* fertilisation.<sup>3</sup> PGD can also screen for the existence of certain tissue types within the embryo, which enables a couple with a child needing a tissue transplant to 'create' a new baby who can serve as a compatible stem cell donor for its sick sibling. Parents are therefore able to use PGD together with *in vitro* fertilisation to screen their embryos and identify one or more that can provide an exact human leukocyte antigen (HLA) match for the ailing child.<sup>4</sup> Stem cells from the new baby can be used as part of the treatment for the sick sibling.<sup>5</sup> Removal of these cells at the birth of a child is painless and non-invasive.<sup>6</sup>

The main advantage of PGD is that it provides genetic information before implantation of an embryo into the womb and can ensure that only embryos that are an exact tissue match are transferred to a woman's uterus. A couple can therefore avoid the difficult decision of either terminating the pregnancy should the fetus not be a match, or extending their family again, hoping that their next child has the desired tissue type.<sup>7</sup>

### Current legal framework

South Africa has legislated on new reproductive issues through, among others, the Human Tissues Act<sup>8</sup> ('the Tissues Act'), the National Health Act<sup>9</sup> ('the Act') and the Children's Act.<sup>10</sup>

Currently, there is no reference in these laws to PGD. The Tissues Act in s18 - 23 allows the removal of blood and tissue for testing and states when and how such blood, tissues or gametes may be used. Although the Tissues Act does not refer specifically to PGD, one could argue that it is broad enough to accommodate the procedure. Furthermore, s19 enables the use of artificial fertilisation

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as a reproductive procedure in which gametes are removed for transplantation into the body of another person.<sup>11</sup>

The Tissues Act also enables individuals to remove fetal or umbilical cord tissue for possible stem cell or other future therapy. It does not prohibit the third-party use of such material. It therefore does not prohibit the harvesting of stem cells from one child for use in another, as in a saviour sibling scenario. However, ministerial consent must be obtained for the removal of the tissue, and conditions may be imposed on such an authorisation.<sup>12</sup> Accordingly, while fetal and umbilical cord tissue may be removed, the Minister may restrict its use.

The Act also does not refer to PGD but provides for the removal of gametes from a living person, which may only be used for 'medical or dental purposes'.<sup>13</sup> It prohibits the removal of such products from a person who is mentally ill, younger than 18 years old, and where the tissue is not replaceable by natural processes; or if the tissues are placental, embryonic/fetal tissue, stem cells or umbilical cord cells (excluding umbilical cord progenitor cells).<sup>14</sup> Given that umbilical cord progenitor cells are excluded, such cells may be harvested at birth and stored for later use.

The Children's Act describes rights for children as patients that protect children who participate in reproductive procedures, e.g. children who donate bone marrow to a sibling. This Act provides that children may consent independently to medical treatment from the age of 12 if they have 'sufficient maturity'.<sup>15</sup> Children may participate in decision making<sup>16</sup> and are entitled to information on their health, which must be provided in an accessible manner.<sup>17</sup> As a general principle, the Act provides that '[I]n all matters concerning the care, protection and well-being of a child the standard that the child's best interest is of paramount importance, must be applied'.<sup>18</sup>

In summary: although the current legal framework is silent on the use of PGD, we argue that the procedure is not prohibited. However, PGD is effectively unregulated, as the Tissues Act only refers in broad terms to the testing of tissues and artificial insemination without linking them. The use of stem cell tissue is regulated, as parents require ministerial consent to harvest such material. Although this acts as a form of regulation, there is no guidance on the circumstances in which ministerial permission would be granted. Finally, the rights of child participants in the PGD are protected, as the Children's Act requires them to be involved in health decisions and gives them the autonomy to decide on medical treatment (if over 12 years and sufficiently mature). However, the rights of saviour siblings, who may feel violated because they were 'created' to save an older sibling, are not protected, given the general legal principle that legal personality begins at birth.<sup>19</sup>

## Future legal framework

The future legal framework will change if five draft regulations issued in terms of the Act<sup>20</sup> are finalised and implemented, including draft regulations relating to the use of human biological material,<sup>21</sup> regulations relating to stem cell institutions or organisations,<sup>22</sup> regulations relating to tissue banks,<sup>23</sup> regulations regarding the general control of human bodies, tissue, blood or blood products and gametes,<sup>24</sup> and regulations relating to artificial fertilisation of persons.<sup>25</sup> These were published for a 3-month public comment period on 1 April 2011.

The draft **Regulations relating to the use of human biological material** are significant, as they refer expressly to PGD and will change the current framework in the following ways:

1. Creating some clarity around PGD. The regulations provide that human biological material may be removed for the purposes of DNA, RNA and chromosome-based genetic testing.<sup>26</sup> However, parents may not use genetic screening to select the sex of the

embryo for implantation, unless this is to identify and avoid 'serious sex-linked or sex-limited genetic conditions'.<sup>27</sup>

2. Ministerial consent is no longer required to remove and store umbilical cord progenitor cells. This enables the parents to harvest such cells at the birth of their child and determine how they will be used and, if needed, allows the cells to be donated to an ailing older sibling.<sup>28</sup>
3. The regulations limit the persons and institutions that can perform the procedure by providing that the removal of a female gamete or ovum can only be done by a 'competent person',<sup>29</sup> in this instance a 'gynaecologist with training in reproductive endocrinology, and in the use of ovulation-inducing agents and the hormonal control of the menstrual cycle'.<sup>30</sup> The procedure may only take place in an 'authorised' or 'prescribed institution'.<sup>31</sup>

Three draft regulations deal with the harvesting of stem cells or tissues and their storage. The **Regulations relating to stem cell institutions or organisations** provide that no person shall use or acquire stem cells unless the institution is registered to perform this function with the Department of Health<sup>32</sup> and keeps records of any stem cell donation.<sup>33</sup> The **Regulations relating to tissue banks** provide that only registered institutions that have applied to the Director-General of Health to be tissue banks may screen or test tissues.<sup>34</sup> Such institutions must store information on the results of tests (such as tissue typing tests) that are undertaken<sup>35</sup> and keep a register of all tissue donors.<sup>36</sup> The draft regulations provide an inspector of anatomy and investigating officers with powers to ensure that tissue banks maintain appropriate standards and comply with the regulations.<sup>37</sup> In the future, therefore, tissue typing as part of the PGD process may only take place at an institution that is appropriately registered as a tissue bank. These draft regulations must be read with the **Regulations regarding the general control of human bodies, tissue, blood or blood products and gametes**, which provide that tissue, blood and gametes removed or withdrawn from living persons may only be used for medical and dental purposes.<sup>38</sup> Removal must be with the written consent<sup>39</sup> of the patient or a proxy.<sup>40</sup>

Finally, the draft **Regulations relating to artificial fertilisation of persons** create a new, highly regulated environment for artificial insemination. Regulation 3(1) provides that only a 'competent person' (i.e. a health professional who is registered with the Health Professions Council of South Africa and who is '(a) a medical practitioner specialising in gynaecology with accredited training in reproductive medicine; or (b) a medical scientist, medical technologist, [or] clinical technologist, with training in reproductive biology and related laboratory procedures' may remove a gamete or cause a gamete to be removed from the body of a donor for the purpose of artificial fertilisation.<sup>41</sup> The artificial insemination may only take place at an authorised institution<sup>42</sup> and a record must be kept of the process.<sup>43</sup> All births by artificial insemination must be recorded by the institution where the delivery took place.<sup>44</sup> The mother of a child conceived by artificial insemination must ensure that the competent person who performed the procedure is informed of the birth and records the information.<sup>45</sup>

There is no change between the current and future frameworks regarding protection of the rights of the children involved in a saviour sibling process.

In summary: the new framework allows PGD, but not for sex-selection purposes. Parents could use PGD to create a saviour sibling provided they use 'competent persons' at 'authorised institutions' and comply with the requirements regarding artificial insemination. Ministerial consent will no longer be required for the harvesting of stem cells.

## Discussion

The current legal framework appears to permit PGD as a procedure, but there are no regulations dealing with when it may be used. The new framework deals expressly with PGD by providing that it is permissible, except for sex selection. However, it is a concern that both provide insufficient guidance on the circumstances in which PGD is allowable. Although prohibiting sex selection is important, it deals with one narrow aspect of PGD and does not establish general principles on other circumstances in which PGD would be inappropriate.

The current framework focuses on the harvesting of stem cells, requiring ministerial consent but provides no guidance on the circumstances in which this consent ought to be provided. The Constitutional Court has found legislation that provides unfettered discretion to public officials to be inconsistent with the Constitution. In *Dawood v Minister of Home Affairs*,<sup>46</sup> the court held that:

‘... if broad discretionary powers contain no express constraints, those who are affected by the exercise of the broad discretionary powers will not know what is relevant to the exercise of those powers or in what circumstances they are entitled to seek relief from an adverse decision.’

In the future, the draft regulations impose extensive regulation on the removal, use and donation of blood, tissues and gametes. Most of this regulation is thorough: (i) only allowing certain registered persons and institutions to perform procedures such as PGD, tissue typing and artificial insemination; and (ii) requiring the documentation of donations and procedures, e.g. at tissue banks. The Department of Health will therefore control work in this field through its registration procedures and the monitoring of databases. While this type of regulation has some value, it fails to set normative standards for the circumstances in which the procedures may be undertaken. Earlier draft regulations<sup>47</sup> addressed this as the Minister could only approve the harvesting of fetal and umbilical cord tissue on the recommendation of a sub-committee within the National Health Research Ethics Committee. This brought our framework approach closer to other jurisdictions in which powers to grant or refuse permission for certain reproductive procedures are devolved to a specialist council. Sadly, this important innovation was excluded from the current draft regulations, leaving no principles to guide the decision-making relating to PGD.

South Africa can learn from other countries in this regard, e.g. the Human Fertilisation and Embryology Authority (HFEA) in the UK established a committee to license treatments or research projects.<sup>48</sup>

The HFEA provides that PGD with HLA typing may be permitted if:

1. The affected child is suffering from a severe or life-threatening disease of a sufficient seriousness to justify the use of PGD.
2. The embryos conceived in the course of treatment should themselves be at risk from the condition by which the existing child is affected.
3. All other possibilities of treatment and sources of tissue for the affected child have been explored.
4. The techniques should not be available where the intended recipient is a parent.
5. Only cord blood should be harvested for purposes of the treatment, and not other tissues or organs.
6. Appropriate counselling around the implications of PGD should be a requirement for couples undergoing this treatment.
7. Families should be encouraged to participate in follow-up studies.
8. Embryos should not be genetically modified to provide a tissue match.<sup>49</sup>

## Conclusions and recommendations

PGD is an important medical innovation enabling families to eradicate serious genetic diseases from their family trees and, in extreme situations, to create a child that can act as a tissue donor for a sick sibling. However, it raises complex social and ethical issues that require careful consideration to ensure the affected children are protected.

Sadly, the South African legal framework is betwixt and between the new and the old. The old, in the form of the Tissues Act, caters inadequately for PGD; and new regulations under the Act have not been finalised and are in some respects inadequate. This leaves the law lagging behind new reproductive technology. Although this results in legal uncertainty, it offers an opportunity to change the framework before the draft regulations are finalised through submitting public comments. We propose the following changes to the draft April 2011 regulations:

1. Clearly establish PGD tissue typing as an acceptable therapy for use in children, rather than focusing narrowly on sex selection.
2. Establish principles to guide when PGD and the use of stem cells will be acceptable, or
3. Require the establishment of a sub-committee within the National Health Research Ethics Council to issue such guidance, as done by the HFEA in the UK.

PGD offers an unparalleled opportunity for families to save their sick children. However, it comes with the possible social cost of children who may feel that they have been ‘created’ not because they have inherent dignity and value but because their tissues are needed. Allowing the law to lag behind medical advances fails families with ill children and saviour siblings. Regulation of PGD is required to balance these conflicting interests. The Minister of Health should act urgently in revising and finalising the draft regulations to prevent the procedure from being used inappropriately and provoking a subsequent public outcry.

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