

# THE APPROPRIATE EXTENT OF PRE-IMPLANTATION GENETIC DIAGNOSIS: HEALTH PROFESSIONALS' AND SCIENTISTS' VIEWS ON THE REQUIREMENT FOR A 'SIGNIFICANT RISK OF A SERIOUS GENETIC CONDITION'

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## I. INTRODUCTION

If a couple knows or fears that they are at risk of having a child with a genetic condition or disease, they may seek *in vitro* fertilisation (IVF) treatment and pre-implantation genetic diagnosis (PGD). Although success cannot be guaranteed, the purpose of PGD is to enable parents to have a child without a genetic impairment and, in so doing, to avoid moral or other difficulties in the termination of an already-begun pregnancy. PGD can be performed when there is a 'significant risk of a serious genetic condition being present in the embryo', the criteria established by the Human Fertilisation and Embryology Authority (HFEA) and Human Genetics Commission (HGC).<sup>1</sup> The technique involves genetic testing of a cell extracted from a six-to-ten-cell (day 3) embryo fertilised and grown *in vitro*. When a couple approaches a PGD clinic, they may find that the condition of concern to them is one for which the HFEA has already granted a licence

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<sup>1</sup> HFEA and HGC, *Outcome of the Public Consultation on Preimplantation Genetic Diagnosis*, 18 June 2001, Rec. 11.

(through a licence committee). Or it may be that, as a result of discussions with a couple, the clinic applies for a licence on their behalf for their condition for the first time. Either way, patients will discuss their concerns and reasons for seeking treatment with clinicians and other professionals who provide PGD. Moreover, these health professionals and scientists will have views about the criteria indicating a 'significant risk of a serious genetic condition' and the operation of these in practice, particularly the extent to which a given condition can be regarded as 'serious'.

This paper explores these views by considering the experience and thoughts of health professionals and scientists working at one UK site that provides PGD. It also explores these professionals' views with reference to the factors that the HFEA and HGC recommend clinics to consider in deciding whether to offer PGD for a given condition, such as 'the view of those seeking treatment of the condition' and 'the likely degree of suffering associated with the condition'. We consider the views of these health professionals and scientists as revealed in transcripts both of interviews with individual staff members and of ethics discussion groups (EDGs). These groups consisted of four to six members of staff and were facilitated by a philosopher. We also relate staff's views and experiences to some key arguments about the appropriate scope of PGD and prenatal diagnosis (PND). In particular, we consider the question of whether, and if so how, prospective parents' interests should be taken into account in decision-making.

## II. THE RECOMMENDATIONS ON RISK AND SERIOUSNESS

Health professionals and scientists involved in PGD (and HFEA licence committees) operate under the criteria established jointly by the HFEA and HGC. Following public response to a Consultation Document, the two issued a report (the 'Outcome Document')<sup>2</sup> with a number of recommendations on the provision of PGD, including the key criteria on risk and seriousness. Those recommendations were subsequently embodied within the HFEA's Sixth Code of Practice.<sup>3</sup> The criteria for PGD, the views of those who sit on the HFEA's licence committees and of health professionals and scientists who provide PGD, will each have a bearing on the extent to which a couple can choose to try and avoid the birth of a child with a certain condition and so upon the extent of their reproductive autonomy. The appropriate scope of such autonomy is controversial, particularly in this context when we are

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<sup>2</sup> *Supra* n. 1.

<sup>3</sup> HFEA, *Code of Practice*, 6th Edn., (2003).

considering to what extent a couple should be able to try to avoid the birth of a particular child, for instance because of a certain genetic condition, in favour of another child without that condition.

Before we turn to explore the way health professionals and scientists perceive and experience the criteria governing PGD and the factors to take into account in deciding whether to offer any given test, it is worth considering how the HFEA and HGC decided on these criteria and how they envisaged they would operate, especially the way in which access to treatment would be discussed and negotiated. For instance, how did the HFEA and HGC foresee the relationship between health professionals and prospective parents?

Prior to the consultation exercise, PGD had been licensed in line with the criteria of the disability ground of the Abortion Act 1967 (as amended by the Human Fertilisation and Embryology Act 1990). That ground requires that two clinicians consider that there is a 'substantial risk' of a child being born 'seriously handicapped'.<sup>4</sup> Following the consultation, the HFEA and HGC decided to continue the 'precautionary principle' of broadly aligning the criteria for PGD with those for selective abortion.<sup>5</sup> Recommendation 11 contains the chosen criteria for PGD: 'The guidance should indicate that PGD should only be available where there is a significant risk of a serious genetic condition being present in the embryo'.

We can immediately see that there are differences between the wording in the Abortion Act and that in Recommendation 11. Most obviously, perhaps, the level of risk is not the same. For abortion, this must be 'substantial'; for PGD, it must be 'significant'. The first term implies a certain quantitative level of risk; it is not clear what the second implies. It could refer to a quantitative element, including a degree of risk that is less than substantial, but still of some weight. In this sense, the concern would be the statistical risk of the condition (depending on whether it is dominant, recessive, random but high, or sporadic but low). Or the term 'significant risk' could imply a degree of risk with a certain meaning to the couple seeking treatment: *i.e.* the perception, or experience, of the couple of the impact of the disorder. Or the term could be intended to mean both of these. Indeed, if we explore more deeply the recommendations governing PGD, we see

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<sup>4</sup> The relevant part of s.1(1)(1) of the Act reads: 'Subject to the provisions of this section, a person shall not be guilty of an offence under the law relating to abortion when a pregnancy is terminated by a registered medical practitioner if two registered medical practitioners are of the opinion, formed in good faith... (d) that there is a substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously handicapped'.

<sup>5</sup> *Supra* n. 1. The reference to the 'precautionary principle' is in para. 25.

that while the HFEA and HGC profess broadly to align the criteria for PGD with those for selective abortion, the discussions and recommendations within the Outcome Document explicitly mention and give weight to the views of prospective parents on the two criteria. This is in contrast to the relevant section of the Abortion Act, which makes no mention of the views of prospective parents and explicitly requires that the two clinicians are of the view, formed in good faith, that the criteria of 'substantial risk' and 'serious handicap' are satisfied.

For instance, one section of the Outcome Document on PGD notes that the Joint Working Party (JWP) of the HFEA and HGC agreed that 'the nature of the decision to pursue treatment involving PGD... meant that a central role in the judgement about the significance of the risk and the seriousness of the condition should be given to the people seeking treatment'.<sup>6</sup> Another stresses the 'crucial importance of the views and experiences of those seeking treatment in decision-making'.<sup>7</sup> Here the personal nature of the issues in question is apparently being emphasised. This may begin to explain the difference in the nature of the risk specified in the Abortion Act on the one hand and the criteria for PGD on the other, although it is not clear that the degree of risk (or views on the seriousness of a condition) in the abortion arena is any less personal. The appropriate degree of risk in PGD compared with PND was discussed by the JWP<sup>8</sup>:

Some members pointed out that the risk should be substantial, and that people seeking treatment should not be allowed to pursue treatment where there was only a minimal risk (as they would not in PND), although it was acknowledged that this could potentially invite a conflict between people seeking treatment involving a particular test and clinicians who do not consider that the risk warrants it.

The passage seems to show two concerns: first, that parents might want to test too freely; second, that to require a substantial degree of risk could be problematic in relations between those seeking treatment and clinicians. So what is a 'significant' risk?

The JWP recommended that 'the significance of the risk to people seeking treatment, not the level of risk itself, should be judged by

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<sup>6</sup> *Supra* n. 1, para. 32 and Rec. 13, our emphases. Para. 34 addresses the issue of information provision, especially the need for balanced information that includes 'that provided by disabled people and their families', an issue to which we return later.

<sup>7</sup> *Ibid.* para. 21. This paragraph is concerned with the relevance of RGOG guidance on termination of pregnancy, to which we return in due course.

<sup>8</sup> HFEA/HGC, *Minutes of Joint Working Party on Preimplantation Genetic Diagnosis*, 30 March 2001, para. 5.4.

agreement between the people seeking treatment and the clinical team'.<sup>9</sup> This passage seems to acknowledge that the way a couple perceives a risk is not governed solely by how great that risk is in percentage terms. The passage also suggests that the couple and the clinical team should discuss and agree upon the significance of the risk. There is a further recommendation of relevance here, as Recommendation 13 states: 'The guidance should indicate that the perception of the level of risk by the people seeking treatment is an important factor in the decision-making process'. If we look at minutes of a JWP meeting on this point, we find 'that what [is] intended [is] the subjective evaluation of the level of risk in the minds of the people seeking treatment'.<sup>10</sup> Ultimately then, prospective parents' perception of the risk is described as 'an important factor'; at the same time, 'agreement' is required as to the significance of the risk. In this way, the term 'significant risk' in Recommendation 11 should perhaps be understood as one that acknowledges the weight of prospective parents' views, but also requires that they discuss their views with clinicians, who may or may not agree that a risk is indeed 'significant'. The implication is that if they do not, the criteria for PGD are not satisfied. In fact, it is highly unlikely that any of the health professionals or scientists at a clinic will be or has been in the same position as those seeking treatment. So there may sometimes be difficulties in agreeing upon the significance of the risk.

When the HFEA and HGC turned to consider the criterion of seriousness, there was a concern, which seems to reflect that in relation to the degree of risk, 'that the decision should not be made solely by the parents as some had very wide definitions of what counts as serious, which many others would not agree with'.<sup>11</sup> The final recommendation implicitly recognises that prospective parents will have views on this matter, but also requires that those views are discussed with clinicians: Recommendation 14 states that 'the seriousness of a condition should be a matter for discussion between the people seeking treatment and the clinical team'.<sup>12</sup> We do not find quite the same attention here (as we

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<sup>9</sup> *Ibid.* para. 5.11.

<sup>10</sup> HFEA/HGC, *Minutes of Joint Working Party on Preimplantation Genetic Diagnosis*, 11 May 2001, para. 5.15, our emphasis.

<sup>11</sup> HGC, *Minutes of Genetic Testing Sub-group*, 12 Jan. 2001, para. 5.4, per Dr Flinter, our emphases.

<sup>12</sup> *Supra* n. 1. The recommendation goes on to state that 'information provided to those seeking treatment . . . should include genetic and clinical information about the specific condition; its likely impact on those affected and their families; information about treatment and social support available; and the testimony of families and individuals about the full range of experiences of living with the condition'. Curiously, a somewhat puzzling variation of the point in Rec. 14 can be found in the Summary of the Responses to

did in relation to the degree of risk) to the views of parents and there is no overt suggestion that seriousness is a subjective matter. Yet, since the recommendation explicitly requires that seriousness should be a matter of discussion, there appears to be a realisation that there may be a range of views on the matter. This is in contrast to the disability ground of the Abortion Act, which makes no reference to the views of prospective parents or to discussions between them and health professionals. (It should be remembered here that abortion itself is a crime, to which the opinion of two doctors that one of the grounds for abortion is made out provides a defence.<sup>13</sup>) It may well be that increasing awareness of the difficulty of defining 'serious' underlies this recommendation: notably, the HGC observed: 'It has proved impossible to define what "serious" should mean in this context'.<sup>14</sup>

On the whole, the HFEA and HGC seem to have tried both to recognise, to some extent at least, the personal nature of the issues at stake in PGD, but also to observe limits to the acceptability of the views of prospective parents and so, ultimately, to reproductive autonomy. This is clearly expressed in the statement that

[t]he JWP agreed the importance of placing greater emphasis on the role of those seeking treatment in reaching the decision about when treatment was appropriate, whilst at the same time maintaining that this should not imply that this treatment should be available on demand.<sup>15</sup>

The idea, it seems, is that this delicate balance will be achieved by means of the stated criteria ('significant risk of a serious genetic condition') as discussed, interpreted and agreed between prospective parents and health professionals on the one hand and clinics and licence committees on the other.

What is it like for PGD health professionals and scientists to operate under these criteria? How do they feel about trying to achieve the balance between respect for personal views on the one hand and the acceptable limits to reproductive autonomy on the other that seems to be sought by the HFEA and HGC? How do health professionals and scientists perceive the risks and the seriousness of the genetic conditions

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the Consultation Document, which at one point observes that '80% agreed that the seriousness of a genetic condition should be a matter of *clinical judgment* based on general guidance'. HFEA/HGC, *Analysis of the Responses to the Joint HFEA/AGCT Consultation on PGD*, para. 56(iv).

<sup>13</sup> Offences against the Person Act, 1861, ss. 58, 59.

<sup>14</sup> HGC, *Response to the Human Fertilisation and Embryology Authority on the Consultation on Preimplantation Genetic Diagnosis*, para. 6.

<sup>15</sup> *Supra* n. 1, para. 23.

in question? What do they think about prospective parents' views about these matters? Do they see themselves as being some kind of a 'check' on the potentially excessively wide views of prospective parents?

### III. HEALTH PROFESSIONALS' AND SCIENTISTS' EXPERIENCE AND VIEWS

This central section of our paper considers health professionals' and scientists' experience and views of the twin criteria of 'significant risk' and 'serious genetic condition'. In due course, the discussion is developed with explicit reference to the factors that the HFEA and HGC recommended should be considered in the decision to offer PGD, such as 'the view of those seeking treatment of the condition' and 'the likely degree of suffering associated with the condition'. In effect, these factors help elucidate the meaning of the criteria for PGD.

#### A. '*Significant Risk*'

The question of the degree of risk facing a couple was touched on in several of the EDGs. Some members of staff appear to see the issue of risk in quantitative terms. For instance, Scientist 19 refers to the idea of a 'significantly increased risk above population risk'.<sup>16</sup> This same scientist also states that there is no need for additional tests that will compromise accuracy, when people are not 'at risk'.<sup>17</sup> Another member of staff (Nurse 13) comments that if the risk is greater, this helps you to 'feel easier maybe about what you're doing'.<sup>18</sup> The relationship between the criteria is also touched on in discussions. For instance, Scientist 2 comments that although there may only be a risk rather than a certainty of a condition occurring in a child, the risk could relate to a very serious condition.<sup>19</sup>

Some of the health professionals and scientists also work in the area of prenatal screening, diagnosis and termination, and the recognition that risk may have a subjective component is clearly revealed in the account of a woman who was already pregnant and undergoing prenatal screening. The account is given by Counsellor 28.<sup>20</sup> The woman in question had previously given birth to a child with a 'very nasty disease' who

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<sup>16</sup> Ethics Discussion Group (EDG) 1, Scientist 19, 6.

<sup>17</sup> EDG 1, Scientist 19, 11.

<sup>18</sup> EDG 3, Nurse 13, 8. For a detailed discussion of the staff's attitudes to embryos, see also K. Ehrich, C. Williams, B. Farsides, J. Sandall and R. Scott, 'Choosing Embryos: Ethical Complexity in Staff accounts of Preimplantation Genetic Diagnosis', (2007) *Sociology of Health and Illness* (forthcoming).

<sup>19</sup> EDG 5, Scientist 2, 19.

<sup>20</sup> EDG 2, Counsellor 28, 13–14.

'died very horribly'. She now had a new partner, so that her risk of having another affected child was 'really very small'. 'But... she was at the stage of her life that she was so anxious about what might happen and the pregnancy brought back all the kinds of memories and feelings that losing her baby obviously reminded her of'. A test for the condition that affected her child was negative, but a scan revealed a relatively mild and unconnected physical malformation that could be corrected once the child had been born.

And that was the only abnormality with this baby, but the girl became so convinced that this was like a sign, a premonition, you know, a sort of marker, that this was all going to go wrong again. And as a result of that, she became... very mentally unstable. And we really felt that if she had been forced to continue with that pregnancy, she may have ended her life.

This case highlights the potentially subjective nature of risk. Clearly, it also raises questions about seriousness. Counsellor 28 notes that the relatively minor physical malformation would not have satisfied the requirement of seriousness under the disability ground of the Abortion Act. However, in light of the woman's fears about something else being wrong with the foetus, the health professionals were in fact concerned about a risk to her mental health. Although the account does not record on what grounds an abortion was sanctioned, it is likely that this would have been under section 1(1)(a). This permits abortion where 'the pregnancy has not exceeded twenty-four weeks and... the continuance of the pregnancy would involve risk, greater than if the pregnancy were terminated, of injury to the physical or mental health of the pregnant woman or any existing children of her family'. A set of Royal College of Obstetrician and Gynaecologists' guidance implicitly notes the potential use of this ground for foetal disability in the following terms<sup>21</sup>:

Women vary in their reaction to being told that their fetus is, or may be, abnormal. Occasionally a woman feels strongly that she is unable to accept a probability of risk or a degree of handicap that her medical practitioners consider less than substantial or

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<sup>21</sup> RCOG, *Termination of Pregnancy for Fetal Abnormality in England, Wales and Scotland* (RCOG Press, Jan. 1996) at para. 3.4. The guidance continues: 'After 24 weeks the grounds for abortion for mental health are more stringent; the continuation of the pregnancy must result in grave permanent damage to mental health. Such damage to mental health is unlikely to result from a woman's concern about a fetal abnormality that her doctors do not consider serious enough to satisfy the law. In effect this means that after 24 weeks the abortion decision must be based only on the anticipated risk that the child would be seriously handicapped'.



serious. Under such circumstances, and only when the gestation is less than 24 weeks, the practitioners may decide that abortion has become necessary to protect her mental health.

In such circumstances, a woman's views about the degree of risk or the seriousness of the disability are in effect being taken into account by use of another ground of the Abortion Act. As noted previously, the actual disability ground of the Act makes no mention of a woman's (or her partner's) views about disability. Curiously, perhaps, the Outcome Document on PGD makes no reference to the use of section 1(1)(a) of the Abortion Act in the context of disability.

It is interesting to speculate what would have happened if, instead of becoming pregnant with her new partner, the woman had instead sought PGD, for instance, for the condition which had affected her first child. As noted above, her risk (with her new partner) of having another child with the same condition was now 'very small'. Understandably, however, she was very fearful of something going wrong with a subsequent pregnancy. To what extent could these fears have been relevant to a decision to provide PGD of some kind? Two of the factors that the HFEA and HGC recommend should be considered when deciding the appropriateness of PGD (discussed further below) could have been particularly relevant: the view of those seeking treatment of the condition and their previous reproductive experience. However, although the woman may have been very fearful, where the risk is in fact 'very small', it is not clear that her subjective experience can gain her access to PGD. Indeed, as we saw above, while the HFEA and HGC intended the term 'significant risk' to mean 'the subjective evaluation of the level of risk in the minds of the people seeking treatment' and this is 'an important factor', agreement is still required as to the significance of the risk. It seems unlikely that health professionals and scientists would have agreed that the risk was 'significant'. Undoubtedly, resource considerations will also be relevant here in the NHS context. Further, since the woman in this revised scenario is not already pregnant, there would not be the immediate concern with her mental health—and the need, in some sense, to 'rescue' her—that there was in the original case.

### *B. 'Serious Genetic Condition'*

We have discussed a case from the context of prenatal screening and selective abortion that highlighted the potentially subjective nature of risk and speculated on the degree to which this can be relevant in the context of PGD. The case also raised issues about seriousness, a question about which health professionals and scientists had a great deal to say in the interviews and discussion groups. By way of introduction

to this issue, it may be useful to note that recollection of the case above led Counsellor 28 to observe<sup>22</sup>:

I've always thought, perhaps it's one of the reasons it hasn't been done in law, is that you can't kind of... write a list of things or conditions that you consider serious... And I think one of the reasons it's not been done in law is that it – the perception and the reality of seriousness isn't just about the condition itself.

Having recounted the above case, Counsellor 28 continued: '[I]f you did have a list, I think you would be less of a clinician, because the notion of clinical judgement and care of your patient or family would have to go out the window'.<sup>23</sup> The suggestion here seems to be that the perceptions of the prospective parents and the situation of the wider family may be relevant to an assessment of seriousness. As we shall see, this suggestion is given further weight by the views of other staff.

Before turning to their views, we might note—not surprisingly in light of Counsellor 28's comments about the difficulties of drawing up a list—that the HFEA and HGC rejected the idea of a 'prescriptive list of "serious conditions" for which PGD was thought to be appropriate'.<sup>24</sup> (Parliament likewise rejected the idea of such a list in relation to abortion for foetal disability on the basis that this would interfere with clinical discretion. This is particularly important because conditions may vary in their severity.<sup>25</sup>) Instead of producing a list of serious conditions, '[i]n line with responses to the consultation... the JWP considered that it would be appropriate to specify factors that should be taken into account in reaching a decision to provide PGD treatment'.<sup>26</sup> This list is contained in Recommendation 15<sup>27</sup>:

The guidance should indicate that in any particular situation the following factors should be considered when deciding *the appropriateness of PGD*: the view of those seeking treatment of the condition; their previous reproductive experience; the likely degree of suffering associated with the condition; the availability of effective therapy or management now and in the future; the speed of degeneration in progressive disorders; the extent of any intellectual

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<sup>22</sup> EDG 2, Counsellor 28, 13.

<sup>23</sup> *Ibid.* 14.

<sup>24</sup> *Supra* n. 1, para. 23.

<sup>25</sup> On the approval of medical discretion generally in the application of the Act, see *Hansard*, 21 June 1990, vol. 174, col. 1156, per Mrs Maria Fyfe: 'If the law sets parameters that do not allow medical judgments to be freely exercised, it must be defective'.

<sup>26</sup> *Supra* n. 1, para. 37.

<sup>27</sup> *Ibid.*, our emphasis. See also HFEA, *supra* n. 3, para. 14.23.

impairment; the extent of social support available; and the family circumstances of the people seeking treatment.

All of these factors could be helpful in our exploration of health professionals' and scientists' views on seriousness, some particularly so. We do not have the scope here to give them equal weight in our discussion, and we do not necessarily look at them in the order listed above.

At the outset, it is worth noting how decisions about seriousness are made in this clinic. Given that different professionals will hold a range of views about the seriousness of conditions and the appropriateness of testing for them, the clinic discusses decisions on a case-by-case basis. Where it wishes to start testing for a new condition, the clinic holds multidisciplinary meetings approximately every six weeks prior to applying for a licence from the HFEA. Present at these are reproductive clinicians, geneticists, molecular biologists, counsellors, clinical geneticists, embryologists, cytogeneticists, specialist clinicians in haematology and dermatology and administrators involved in planning cases.

### 1. The View of Those Seeking Treatment of the Condition

This factor explicitly refers to the need to take into account prospective parents' views. As we have seen, the HGC has stated that it is 'impossible' to define seriousness. What we might call the 'subjectivity' of seriousness was a much-discussed issue in the groups. For instance, Doctor 24 says '[w]hat I feel is serious, minor, is a very subjective issue especially to parents'.<sup>28</sup> He/she expands:<sup>29</sup>

Maybe for a parent having a child with six fingers, is minor. That's the only thing I can think of myself. Anything beyond that, any handicap in a child, I would not consider *not* serious. So, we have to – it's a very personal opinion as to – I mean I can't off hand think of any condition which should not be tested because it's not serious.

Similarly, Counsellor 18 says of the notion of seriousness that '[w]e all have a different idea of what it is'.<sup>30</sup> In light of the developments in PGD technology ('it is just going to develop and progress over the centuries') Counsellor 18 continues<sup>31</sup>:

[T]here will always be people pushing the boundaries, and it's a serious condition this week, but like we've said, nobody can

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<sup>28</sup> EDG 1, Doctor 24, 7.

<sup>29</sup> Our emphasis.

<sup>30</sup> EDG 3, Counsellor 18, 15.

<sup>31</sup> EDG 3, Counsellor 18, 23.

really define seriousness. . . and what is serious for you won't be for me. And I think it's this, and you think it's that. And in ten years' time, they'll think. . .

Differences of view about seriousness are also noted by Embryologist 33<sup>32</sup>: 'I think how one person copes with it and how another person copes with 'serious' is going to be different as well. . . What one person thinks is a serious condition, the next person might not'.

Importantly, however, within the groups there was also a sense that although there is a subjective element to seriousness, at the same time there must be a limit to what could in any sense be regarded as a 'serious genetic condition': it cannot be that 'anything goes'. For instance, in response to the last comment above, Scientist 2 observes<sup>33</sup>: 'No, but, when it's serious, their life is completely debilitated by it. . . it's medically serious'. Later, he/she adds: 'If we said the possibility is it's going to be mild, we wouldn't do PGD for it. But if the possibility is there that it's going to be fairly serious, that's why it's done'. Further, as we saw above, Doctor 24 suggested that having an extra finger could not in any sense be 'serious', thus seeming to place extra digits at the non-serious end of the spectrum of 'abnormality'. As for other conditions which may not be serious, Nurse 13 suggests that a condition such as cleft palate is not serious, although 'people are clamouring for' the relevant testing.<sup>34</sup> Not surprisingly, these observations show that health professionals and scientists have views—although not necessarily homogeneous ones—about which conditions will satisfy the criterion of 'a serious genetic condition'. In any case, as we know, the HFEA and HGC require (in Recommendation 14) 'that the seriousness of a condition should be a matter for discussion between the people seeking treatment and the clinical team'.

So far, we have seen that although these health professionals and scientists consider that 'serious' has a subjective component, at the same time several members of staff think it is not a 'free-for-all'. It may now be helpful to think more closely about the possible meanings of 'serious' by considering whose interests may be at stake in PGD. In this regard, a very important question was posed by Doctor 11: 'Is seriousness. . . [about] the person that's going to be affected by that condition? Is it serious at their level, not so much our level?'<sup>35</sup>

This question has been considered in the related context of PND and selective abortion. Notably, Sally Sheldon and Stephen Wilkinson have

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<sup>32</sup> EDG 5, Embryologist 33, 19.

<sup>33</sup> EDG 5, Scientist 2, 19.

<sup>34</sup> EDG 3, Nurse 13, 24.

<sup>35</sup> EDG 4, Doctor 11, 27.

helpfully clarified this issue in relation to the interpretation of the disability ground of the Abortion Act. Sheldon and Wilkinson argue that when the foetus would be born as a child with what many would regard as an extremely serious condition (perhaps Tay–Sachs),<sup>36</sup> termination may well be in the foetus's interests, a position in accordance with other philosophical analyses.<sup>37</sup> Since it *may* be that if born such a child would say her life was not worth living (see further below), it may also be that, if not terminated *in utero*, the subsequently born child could claim, ethically speaking, to have a wrongful life.<sup>38</sup> Yet, there are remarkably few conditions that might be potentially serious in this way. Recognition of this requires that we think of other possible justifications for PND and termination and indeed for PGD.

Where a child would be born with a condition that would mean she still thought she had a life worth living, a more satisfactory rationale for use of the disability ground of the Abortion Act might be found in the notion of 'parental interests', as Sheldon and Wilkinson have argued.<sup>39</sup> This type of argument could also be applied in the context

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<sup>36</sup> 'The classical form of Tay–Sachs disease (TSD) is a fatal genetic disorder in children that causes progressive destruction of the central nervous system... By about two years of age, most children experience recurrent seizures and diminishing mental function. The infant gradually regresses, losing skills one by one, and is eventually unable to crawl, turn over, sit, or reach out. Other symptoms include increasing loss of coordination, progressive inability to swallow and breathing difficulties. Eventually, the child becomes blind, mentally retarded, paralyzed, and non-responsive to his or her environment. To date, there is no cure or effective treatment for TSD'. National Tay–Sachs and Allied Diseases Association, Inc. <http://www.ntsad.org/pages/t-sachs.html>.

<sup>37</sup> S. Sheldon and S. Wilkinson, 'Termination of Pregnancy for Reason of Foetal Disability: Are there Grounds for a Special Exception in Law?', (2001) 9 *Med. L. Rev.* 85, 88ff. A second rationale for the disability ground of the Abortion Act discussed and ultimately rejected by Sheldon and Wilkinson is the so-called "replacement argument". *Ibid.* 93ff. For a compatible analysis, see, e.g. A. Buchanan, D. Brock, N. Daniels and D. Wikler, *From Chance to Choice: Genetics and Justice* (Cambridge University Press, 2000) at 235.

<sup>38</sup> See e.g. A. Buchanan *et al.*, *supra* n. 37, *ibid.* As Sheldon and Wilkinson note, however, such a claim has not been recognised in English law. Sheldon and Wilkinson, *supra* n. 37, 89, and referring to *McKay v. Essex A.H.A.* [1982] 2 *W.L.R.* 890.

<sup>39</sup> S. Sheldon and S. Wilkinson, *supra* n. 37, 99ff. The legal objection to this argument, originally explored by Derek Morgan (D. Morgan, 'Abortion: the Unexamined Ground' [1990] *Crim. L.R.* 687, 692) is that a 'parental interests' interpretation of s.1(1)(d) of the Act would essentially repeat s.1(1)(a) of the Act, making s.1(1)(d) redundant. Sheldon and Wilkinson observe that, from an ethical viewpoint, it may simply be that there is more than one justification for this section. From the viewpoint of statutory interpretation, however, they suggest that 'there are some grounds for questioning' whether Parliament can have intended that s.1(1)(d) should protect parents' interests. S.1(1)(d) can never be entirely redundant in that, unlike s.1(1)(a), it allows terminations up until birth. Apart from this point and more fundamentally, it may be that as a society

of PGD, the rationale for which is of course our main consideration here. The thought is that PND or PGD for conditions that are not as serious as (for instance) Tay–Sachs, coupled with the option of termination or embryo discard, may potentially respond to parents' concerns and interests.

At the first mention of this kind of possible parental interest, it should immediately be noted that, although this paper discusses staff's views about parents' concerns and relates these to ethical arguments about whether it can be appropriate to take account of parents' possible interests in the evaluation of 'seriousness', nothing of what follows should be taken as implying that raising a child with a disability is necessarily more demanding than raising a non-disabled child. Further, we make no judgment as to the balance of rewards and demands in raising a disabled, as compared with a non-disabled child. This is a judgment for parents to make. Experiences will vary widely and some or many people may say that raising a disabled child is particularly rewarding.

Before leaving the PND context, if parents are to have the legal option of termination in any given case, two clinicians must judge in good faith that the condition is 'serious'. (It is likely that parents would only have been offered screening or testing aimed at 'serious' conditions of some kind, although a less serious feature might happen to be found on a scan.) In effect, then, where a foetus would be born as a child with a life she thought worth living, but clinicians still judge a given condition to amount to a 'serious handicap' for the purposes of the disability section of the Abortion Act, they are effectively determining that although the condition would leave a child with a life worth living, it has an element of seriousness which implicates its parents' interests in child-raising and, moreover, to an extent that justifies termination. Since there is a dearth of case-law on the Abortion Act, there is as yet no overt recognition of this point, although the law has recognised that parents can sue for the lost opportunity to abort a disabled child and that to some extent this is because of their interests in child-raising.<sup>40</sup>

Returning directly to PGD, here too we face the point that very few genetic conditions might be so serious as to give rise to a life that a child thought was not worth living. (We consider health professionals'

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we have not necessarily been honest about whose interests the disability section of the Act is capable of protecting: it is to some degree taboo to say that parents have an interest in choosing to accept or avoid the birth of an impaired child and those debating the Act in Parliament may well have sensed this.

<sup>40</sup> For discussion of relevant wrongful birth case law (and interpretation of the disability ground of the Abortion Act generally), see R. Scott, 'Interpreting the Disability Ground of the Abortion Act', (2005) 64/1 *C.L.J.* 388.

and scientists' thoughts about some specific contenders for the description 'very serious' below.) This means that it may be appropriate to recognise that at least some forms of PGD are conducted for the prospective parents. Scientist 21 makes this point very clearly<sup>41</sup>:

I was at a conference. . . Parent Project UK, which is a charity which is aimed at . . . therapy for Duchenne muscular dystrophy people, and they were all parents. So one presented a talk actually which I found very interesting, and they looked at the quality of life for families with boys with Duchenne muscular dystrophy, which is a severe disease. The average lifespan now is about 19. And the quality of life of – the perception of the quality of life of the affected boy was rated differently by parents, by the clinicians looking after them and by the boys themselves. *And the boys themselves. . . gave their rating of quality of life the same as any healthy controlled sample.* And the parents gave them the lowest quality and the clinicians gave them somewhere in between the two, which was interesting, I thought. . . . *So that implies we're doing this for the parents and not for the child in some respects.*

Earlier, Scientist 21 observed<sup>42</sup>: 'Obviously people want to have children and when they have children with disability or handicap, to some extent that makes their life a bit more miserable compared to what they're hoping for'. The potential impact of a child's disability on parents was observed by a number of participants. Scientist 8 refers to the 'huge burden' that parents may experience.<sup>43</sup> He/she also alludes to the idea of undoubtedly serious conditions on the one hand and conditions about which parents may disagree on the other<sup>44</sup>:

I mean I think there are conditions which are under all circumstances, horrendous. And can be put very firmly on that list. But I completely agree with [participant], I mean I think there are lots of conditions which aren't clear-cut and which for some families might be considered serious and others not.

Further, Administrator 31 observes: 'I think you need to look at people's – you know seriousness, to a certain extent, has to be based on people's perception of their ability to cope'. Ethically speaking, we could relate the idea of the 'ability to cope' to the question of the extensive positive moral duties that parents must undertake in child-raising and to the idea that people might reasonably disagree about how much is

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<sup>41</sup> EDG 2, Scientist 21, 3–4, our emphases.

<sup>42</sup> EDG 2, Scientist 21, 2.

<sup>43</sup> EDG 2, Scientist 8, 5.

<sup>44</sup> *Ibid.* 14.

reasonable to have to 'take on' (assuming there is a choice at the stage of selection by PGD or PND). So, each of these members of staff seems to recognise and accept that parents' interests are significantly implicated in the possible wish to avoid a child with a serious genetic condition.

Turning now to the interaction between health professionals and prospective parents, in which they may (explicitly or otherwise) discuss the notion of seriousness, it appears that meeting the couple and learning something about their experience can considerably affect the views of staff. For instance, Doctor 11 observes<sup>45</sup>: '[K]nowing the couple can have an amazing effect on how you feel about them'. Counsellor 17 adds<sup>46</sup>: "I guess if you do get the opportunity to see, hear a little bit more about the whole story behind... the couple, you've got more of an idea of the issues then, for them too". The philosopher facilitating the discussion suggested to one of the groups<sup>47</sup>: 'So you think if someone has personally experienced something, they're in a position to tell you, in a real sense, how serious it is to them, in a way that might not seem obvious from the outside?', to which Doctor 11 responds '[t]hat's where it sits with me'. This perspective is echoed by Embryologist 15<sup>48</sup>: '[I]n general, if I was pushed about it, I would probably still lean towards the side of the parents for opting to go through it because they obviously have either seen their child go through that or someone in their family'.

Generally, there did not seem to be any strong disagreement with the idea, expressed by Scientist 2, that 'you have to be able to see patients' perspectives'.<sup>49</sup> Referring to the way the clinic works as a team, he/she continues 'and then [you] open it to everyone who works in the unit, and then their perspectives as well'. On the one hand, then, there are discussions between people seeking treatment and one or more members of staff; on the other, the staff will then discuss a given request for PGD among themselves (as noted earlier). Since the staff are actively involved in all the processes in PGD, they need to feel as comfortable as they can with the acceptability of PGD for a given condition. The ethical dimensions of staff involvement in PGD was a much-discussed issue in the groups, but not one on which we can focus here.<sup>50</sup>

In this section we have reviewed these health professionals' and scientists' thoughts about the 'subjective' notion of seriousness, but at the

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<sup>45</sup> EDG 4, Doctor 11, 24.

<sup>46</sup> EDG 4, Counsellor 17, 24.

<sup>47</sup> B. Farsides, EDG 4, 28.

<sup>48</sup> Interview, Embryologist 15, 2.

<sup>49</sup> EDG 5, Scientist 2, 27.

<sup>50</sup> *E.g.* in EDG 1, 2 and 5. The issue was also referred to by various members of staff in interview, *e.g.* Scientist 2, Doctor 6 and Embryologist 15.



same time noted that they do not necessarily think that *any* condition can reasonably be seen as serious. We have also noted, in response to Doctor 11's question ('Is it serious at their level, not so much our level?'), that in reflecting on seriousness, it is important to think about 'serious for whom?'. As others have recognised, there is a distinction between conditions that are so serious that it may, conceivably, not be in the embryo's or foetus's interests to be born on the one hand and conditions that are less serious and that may have a significant impact on a child's life but, more particularly, on its parents on the other.<sup>51</sup> We then considered the recognition, by staff, of the importance of the views of prospective parents about the seriousness of a condition.

It should be noted in passing that an alternative way of reflecting on selection practices would be to adopt a non-person-affecting perspective. On this approach, we would be thinking of the lives of different possible people and recognising that, other things being equal, it may be preferable if people are born who at least start life without impairments or disorders that may impede their capacity for flourishing and welfare. Not surprisingly perhaps, no distinction is made between a person- and a non-person-affecting approach in the 1990 Act, nor in the HFEA's Sixth Code of Practice. More particularly, we shall not discuss non-person-affecting principles here, because these were not identified as relevant by the professionals at the clinic (with one possible exception).

We now need to look at something of the spectrum of possible conditions in order to further disentangle the embryo's or foetus's interests from those of its parents or the wider family, where these may diverge. We do this with reference to another factor that the HFEA and HGC recommend should be considered in relation to the decision to offer PGD, namely 'the likely degree of suffering associated with the condition'.

## 2. The Likely Degree of Suffering Associated with the Condition

### 2.1 *Very serious conditions*

Clearly, suffering could be of a mental or physical form. Either way, it may impact on someone's quality of life. The difficulty with this important factor is that we have to estimate the degree of suffering in conditions we have not experienced. Arguably, anyone with a given condition will be the best informed about it and, in the PGD context,

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<sup>51</sup> The reference to interests is used loosely, since it may be argued that the embryo has none until it acquires them at sentience. On the link between interests and sentience, see B. Steinbock, *Life Before Birth* (New York: Oxford University Press, 1992) at 24, n. 24. On the notion of interests, see J. Feinberg, *Harm to Others* (New York: Oxford University Press, 1984) at 34: 'In general, a person has a stake in X... when he stands to gain or lose depending on the nature or condition of X'.

sometimes prospective parents will themselves be in a position to offer insights. We start with some of the conditions staff mentioned that are often regarded as undoubtedly serious. Recall, for instance, Scientist 2's reference to a life being 'completely debilitated' by a 'medically serious condition'.

'I think I would consider a serious condition, for instance, a genetic condition called Tay–Sachs disease, where the children die in childhood... I would consider that a serious condition because a child being so sick in early life, and going through all the things...', observes Doctor 32.<sup>52</sup> Tay–Sachs is often cited as a classic case of a very serious condition. Since a child with Tay–Sachs lives such a short time and in a state of rapid and marked physical and mental deterioration, it may not be feasible to obtain the views of such a child on his or her suffering. Looking from the outside, however, it may be plausible to say that the suffering would be very great.

It might be useful at this point to think about the significance of the potential losses—of experiences, activities or opportunities—involved in disease or disability. In this respect a key idea may be that of flourishing. Glover writes about disability in this way. He suggests that disability is a 'functional limitation, which (either on its own or – more usually – in combination with social disadvantage) impairs the capacity for human flourishing'.<sup>53</sup> When we have not experienced a given condition, he asks whether, and if so how, we can make the judgment that it amounts to a disability. Glover suggests that strong weight should be given to the accounts and views of those with disabilities. This implies that we should take the views, noted earlier, of the boys with Duchenne muscular dystrophy very seriously. However, because people's identities are to some degree inherent in their disabilities, Glover suggests that these views 'need to be interpreted with alertness to possible biases'.<sup>54</sup> He also notes that, in reflecting on whether a condition amounts to a disability, we have other sources upon which we can draw: on the one, ideas about the components of human flourishing; on the other, the knowledge that people who can see and hear, for instance, can give about these senses and their experience of them.<sup>55</sup>

In this light, and returning to the example of Duchenne muscular dystrophy, it may be that although these boys rated their quality of life as high as that of healthy boys, it could be argued that the condition brings with it the possibility of suffering and loss of the opportunity to

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<sup>52</sup> EDG 3, Doctor 32, 18.

<sup>53</sup> J. Glover, *Choosing Children: Genes, Disability and Design* (Oxford: Oxford University Press, 2006) at 9.

<sup>54</sup> *Ibid.* 22.

<sup>55</sup> *Ibid.* 22–23.

flourish. In this way, Duchenne muscular dystrophy may reasonably be viewed, as Scientist 21 suggests, as a 'severe' disease and perhaps a 'very severe' one. However, since judgments by third parties about the quality of life of possible others are obviously difficult and sensitive, as Glover has suggested, rather than attempting to make clear-cut judgments about conditions that we have not experienced, it may be better to restrict ourselves to the question of whether there is a *serious risk* that a child would have a life that was not worth living.<sup>56</sup> Glover thinks this might be true of a condition such as Lesch–Nyhan disease.<sup>57</sup> It also seems likely that we can say there is such a risk in the case of Tay–Sachs. At the same time, it is *not* clear whether we can say this in relation to Duchenne muscular dystrophy. With regard to the boys who have Duchenne muscular dystrophy, Scientist 8 observed<sup>58</sup>: 'I think what's not predictable is that they would not see themselves as having any different quality of life than normal boys. I think it's quite predictable that they would say that they're glad they're alive'. However, in all these cases, the impact on parents could be considerable. On this point, Doctor 32 added in relation to Tay–Sachs that the condition would be serious 'for the child, for the parents, just as bad. So that I would consider serious on those two accounts'.<sup>59</sup>

Glover's warning about making judgments that there is a risk that someone will have a very low quality of life is salutary. In one of the discussion groups, Scientist 8 observed that one clinician had given a talk in which he/she had said that the only condition in relation to which he/she could ever recall that people said they would prefer not to be born

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<sup>56</sup> *Ibid.* 60.

<sup>57</sup> Lesch–Nyhan syndrome (LNS) is a rare, inherited disorder caused by a deficiency of the enzyme *hypoxanthine-guanine phosphoribosyltransferase* (HPRT). LNS is an X-linked recessive disease – the gene is carried by the mother and passed on to her son. LNS is present at birth in baby boys. The lack of HPRT causes a build-up of uric acid in all body fluids, and leads to symptoms such as severe gout, poor muscle control, and moderate retardation, which appear in the first year of life. A striking feature of LNS is self-mutilating behaviors – characterized by lip and finger biting – that begin in the second year of life. Abnormally high uric acid levels can cause sodium urate crystals to form in the joints, kidneys, central nervous system, and other tissues of the body, leading to gout-like swelling in the joints and severe kidney problems. Neurological symptoms include facial grimacing, involuntary writhing, and repetitive movements of the arms and legs similar to those seen in Huntington's disease. Because a lack of HPRT causes the body to poorly utilize vitamin B12, some boys may develop a rare disorder called megaloblastic anemia.... The prognosis for individuals with LNS is poor. Death is usually due to renal failure in the first or second decade of life'. National Institute of Neurological Disorders and Stroke, [http://www.ninds.nih.gov/disorders/lesch\\_nyhan/lesch\\_nyhan.html](http://www.ninds.nih.gov/disorders/lesch_nyhan/lesch_nyhan.html).

<sup>58</sup> EDG 2, Scientist 8, 48.

<sup>59</sup> EDG 3, Doctor 32, 19.

was epidermolysis bullosa (EB).<sup>60</sup> This suggests that EB would be a strong contender for being at the top of the list of very serious conditions, although we have noted that it may be very hard to obtain the views of a child with Tay–Sachs, who will be subject to rapid physical and mental deterioration and will die very young.

## 2.2 *Less serious conditions*

We now move away from conditions that some people might reasonably regard as very serious (even though a person with a given very serious condition may not say that his or her life is not worth living). Conditions that might be seen as less serious include Cystic Fibrosis and Down syndrome, amongst others. What do PGD health professionals and scientists think about testing for these conditions?

**2.2.1 Cystic Fibrosis.** A number of members of staff discussed Cystic Fibrosis. Since someone with this (or any genetic) condition could only ever exist with Cystic Fibrosis, we need to think about how severe the condition is and whether it should be avoided for the ‘person’s sake’, as it were. What did members of staff think about Cystic Fibrosis?

Scientist 21 observes: ‘I would say our role was to enable somebody to have a healthy baby when, in the first instance they’ve got a fairly substantial risk of having an unhealthy baby. And so I guess... the dividing line then is, what you classify as healthy and unhealthy and how severe...’.<sup>61</sup> He/she later refers to Cystic Fibrosis as ‘major’. Scientist 19 describes Cystic Fibrosis as ‘quite serious’.<sup>62</sup> Doctor 24 thinks it is ‘fairly serious’.<sup>63</sup> ‘Major’, ‘quite serious’ and ‘fairly serious’ all seem to indicate a degree of uncertainty about the severity of Cystic Fibrosis. The issue was touched on in one of the discussion groups. The philosopher observed<sup>64</sup>:

BF: [O]ne of the groups that might... have serious questions to ask of the work you do, are people who are born with and living with disability, because what they might say is, ‘Actually we live

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<sup>60</sup> EDG 2, Scientist 8, 4. ‘Epidermolysis bullosa (EB) is the name given to a group of genetic disorders that lead to fragile skin. This fragility leads to blistering and shearing of the skin as a response to friction and everyday knocks and bumps... There are three major types of EB, but the general symptoms in all forms of EB are skin fragility and blistering. In some types the internal linings of the body can be affected, as well as the cornea of the eye. Healing with scarring seen in some forms of EB can also lead to worsening disability. Within each type there are sub-groups with a huge variety of symptoms and prognosis’. N.H.S. Direct Health Encyclopaedia, ‘Epidermolysis bullosa’, <http://www.nhsdirect.nhs.uk/articles/article.aspx?articleId=560&PrintPage=1>.

<sup>61</sup> Interview, Scientist 21, 13.

<sup>62</sup> Interview, Scientist 19, 4.

<sup>63</sup> Interview, Doctor 24, 5.

<sup>64</sup> B. Farsides, EDG 4, 32.

perfectly good lives, we enjoy a good quality of life. Yet if you match up the lives we're living against the lives you're selecting out, they don't look much different. . .'

In response, Doctor 11 observed: 'You could use that argument quite easily for Cystic Fibrosis, couldn't you?' This suggests we need to reflect again on whose interests are being protected, or most protected, in PGD, this time in relation to Cystic Fibrosis testing. In this regard, Doctor 20 recalls an interesting couple<sup>65</sup>:

I saw a couple last week who came for Cystic Fibrosis, a fertile, intelligent couple, who have a Cystic Fibrosis child. And I said, 'What are you doing this for? Why don't you just have another pregnancy?' And they couldn't consider terminating a Cystic child because, firstly they said, 'we do not want to have another child that we have to watch die or be very ill. But on the other hand, if we kind of [terminate the] pregnancy it's like terminating [our existing child]. . . And we feel we can't do that. And we want some other way of approaching this.'

In this case, the testing seems very much in the interests of the parents: although PGD is not 100% accurate, these parents clearly saw the possibility of a pregnancy achieved through PGD as a way of avoiding the potentially very painful issues they might face if a foetus tested positive for Cystic Fibrosis. The question of trying to avoid the dilemmas around termination is extremely important in PGD and is often referred to by members of staff both in interviews and in the discussion groups.<sup>66</sup> The issue may be particularly acute because, as with the parents in Doctor 20's example, the parents have already had another child with the relevant condition; or they may have terminated one or more pregnancies; or they may have miscarried earlier pregnancies and want to avoid the risk of miscarriage inherent in diagnostic testing in pregnancy.

Overall, then, it may be defensible to describe Cystic Fibrosis (which can vary in its severity) as 'serious', provided that we recognise that it does not seem to be so serious as to give rise to a risk of a life of very low quality, one that may not be worth living. Further, since someone with Cystic Fibrosis could only ever exist as that person, the seriousness of the condition may really relate more to their parents' interests. Recall again the comment of Scientist 21 (in relation to another condition) that: 'So that implies we're doing this for the parents and not for the child in some respects'. What we have seen so far is that although this could be true in relation to what might be seen as very serious

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<sup>65</sup> Interview, Doctor 20, 20.

<sup>66</sup> E.g. EDG 5, Scientist 2; and in interview Scientist 8, 5, 17.

conditions (such as Tay–Sachs), it is particularly the case in relation to less serious ones. We discuss the moral acceptability of PGD that may ultimately be predominantly in the parents' interests below when we turn to consider Down syndrome.

When prospective parents discuss the seriousness of a given condition with members of staff, they need to know about what that condition involves. The question of information provision is in fact part of Recommendation 14 in the Outcome Document, the recommendation (to recap) that states 'that the seriousness of a condition should be a matter for discussion between the people seeking treatment and the clinical team'. The recommendation in fact goes on to state<sup>67</sup>:

[I]nformation provided to those seeking treatment... should include genetic and clinical information about the specific condition; its likely impact on those affected and their families; information about treatment and social support available; and the testimony of families and individuals about the full range of experiences of living with the condition.

In turn, this is embodied in the HFEA's latest Code of Practice.<sup>68</sup>

Now, an important aspect of PGD for a condition such as Cystic Fibrosis is that the people who seek PGD for this condition will have a reason to think that they are at risk. This may be because they have had a child with Cystic Fibrosis, or someone in their wider family may have the condition or one of the couple has the condition. (In contrast, in dominant conditions—such as Huntington's chorea, achondroplasia and myotonic dystrophy—one of the parents is *always* affected (or will be in due course if it is late-onset) and will therefore have a particularly personal view of the situation.) Indeed, it will likely be for one of these reasons that they come to the clinic seeking PGD. The couple will therefore have some knowledge about what living with Cystic Fibrosis is like. This, in turn, would mean (for instance, if PGD for Cystic Fibrosis had not yet been licensed and, therefore, a decision had to be made about whether to offer such testing) that their views as to its seriousness would carry some considerable weight. Generally, it may mean that these couples are likely to have less need for information about the condition than other people may have. In contrast, if a couple presented for consultation having had a child who was very mildly affected by Cystic Fibrosis, they may well need to know more about the condition. The general point here is that people seeking PGD for Cystic Fibrosis

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<sup>67</sup> *Supra* n. 1, para. 32 and Rec. 13. Para. 34 addresses the issue of information provision, especially the need for balanced information, which includes 'that provided by disabled people and their families', an issue to which we return later.

<sup>68</sup> *Supra* n. 3.

will typically have a good idea of what it involves, even if they may benefit from further information about (say) the spectrum of severity; this may help them gauge the severity of what they have 'experienced' so far.

An additional point to note here is that when people approach a clinic about the possibility of PGD for something they have experienced in some way, they must think that it is important enough to try to 'do something about it'. As one member of staff put this, 'of course it must be serious for them to come here'.<sup>69</sup> This does not tell us 'for whom' the condition is serious, but it does tell us that people have experienced something as a problem and have decided to go out of their way to try to avoid it. In relation to experience of a condition and subsequent knowledge of it, the situation may be very different in relation to the second condition we now discuss, Down syndrome.

*2.2.2 Down syndrome.* At the time of the interviews and discussion groups, one of the issues the clinic was considering was whether to offer PGD for Down syndrome to patients coming through for PGD for other conditions. New developments in PGD technology are behind this move, which would mean that identification for trisomy 21 could be 'added in' to other PGD testing. We now explore some of the issues at stake in this idea, as revealed through the interviews and discussion groups.

To offer women testing for Down syndrome under these circumstances would be to offer them testing for a condition which was not the primary focus of their PGD attempt. It would typically mean that staff took the initiative of offering this test in the course of discussions with a couple. For older women, the issue of Down syndrome may well be in their minds before any overt offer of a test. For many younger women, the issue may not be so prominent although it is likely that, if they were to become pregnant, they would face the issue later given the current national screening programme for Down syndrome. At the PGD stage, however, since the offer of testing for Down syndrome would be in addition to testing for another condition, and since the primary focus of couples in coming to the clinic would have been on that other condition, testing for Down syndrome would be different in various ways from testing for conditions such as Duchenne muscular dystrophy or Cystic Fibrosis. We would like to explore some of these differences with reference to staff's views about Down syndrome and PGD for this condition.

A preliminary point to make is that the accuracy of testing for Down syndrome is an important issue. This is noted by Scientist 21, who seemed to be in favour of PGD for Down syndrome, subject to concerns

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<sup>69</sup> Interview, Doctor 6, 13.

about accuracy in the test itself and the possibility of then having to exclude what were in fact normal embryos.<sup>70</sup> We cannot discuss the scientific aspects here. Our discussion can only proceed on the basis that testing for Down syndrome is, or could become, sufficiently accurate to justify its inclusion at the PGD stage. To some degree, this is a matter of judgment that needs to be related to the significance of the risk and the perceived seriousness of the condition. As we have seen, these are not purely objective matters.

What are some of the attitudes of staff to Down syndrome? In one of the discussion groups, Embryologist 33 says that ‘people [with Down syndrome] can still have fully functional lives...’.<sup>71</sup> Scientist 2 later observes: ‘...and they feel they have a fulfilling life, and that’s what counts, how they feel. If they feel it’s fulfilling, it doesn’t matter what anyone else says, if they feel they have had a worthwhile life and what they’ve achieved...’.<sup>72</sup> In another discussion group, Scientist 8 observes<sup>73</sup>: ‘You mentioned earlier about Down’s, which used to be the end of the world, and now it’s a much more accepted condition. And as time moves on and people’s ideas change, you can’t have hard and fast rules’. None of these comments suggests that these members of staff thought that Down syndrome should be avoided for the sake of the person with the condition. Those groups that did touch upon the issue of Down syndrome then had to face the question of why there might be screening for this condition.

To begin, there was some discussion of screening for Down syndrome in the PND context. For instance, in response to Scientist 8’s comment above about ideas changing, Counsellor 28 observes<sup>74</sup>: ‘Right, but it’s still the most common condition for which we’ve screened’. In another group, Doctor 24 observes ‘[t]here’s such a lot going on about Down’s syndrome. There’s so much work going on, there’s so much apprehension going on...’.<sup>75</sup> Who is this work for? Although the severity of Down syndrome varies considerably, the idea that it is better not to be born than to have Down syndrome is highly implausible. We must face the recognition then, that Down syndrome screening is really aimed at prospective parents. Is this morally acceptable? To offer any thoughts on this, we need to think both about prospective parents and about the embryo or foetus.

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<sup>70</sup> Interview, Scientist 21, 23–24.

<sup>71</sup> EDG 5, Embryologist 33, 33–34.

<sup>72</sup> *Ibid.* Scientist 2.

<sup>73</sup> EDG 2, Scientist 8, 14–15.

<sup>74</sup> EDG 2, Counsellor 28, 14–15, part cited above.

<sup>75</sup> EDG 1, Doctor 24, 13.



Turning to the parents, it could be argued that they should have some choice in reproduction about whether to have a child with a disability, including one that is not so serious as to lead to a very low quality of life for a child, such as Cystic Fibrosis or Down syndrome. The idea that reproductive autonomy should include, to some extent, the ability to avoid congenital disability in future children is supported by a number of different writers, some from a broadly liberal perspective.<sup>76</sup> Perhaps most strikingly, there is some support for this idea from the disability critique of prenatal screening and selective abortion.<sup>77</sup> For instance, Shakespeare observes generally<sup>78</sup>:

[T]here are reasons to want to prevent the birth of a child affected by impairment which do not reflect discrimination against disabled people: for example, the desire to avoid the early death or suffering of a loved child, or a feeling that a family will be unable to cope with the strain of looking after a very impaired member.

(He also emphasises the need 'for better provision of welfare services and financial benefits to parents of disabled children, in order to make it easier for parents to choose to... continue such a pregnancy'.<sup>79</sup>)

Turning now to the embryo or foetus, the acceptability of selection will depend, in part, on the question of the moral status of each. The argument that parents are entitled to have some choice about whether to have a child with a disability entails the view that the moral status of the embryo (at the time of PGD) or the foetus (for instance, at the time of a nuchal fold scan for Down syndrome and chorionic villus sampling) is not so great as to outweigh the prospective parents' reproductive autonomy interest in avoiding disability in a future child. In contrast, if we were to give greater moral importance to the embryo or foetus or both, we might say that it is only morally acceptable to test (with possible discard or termination) where there is a serious risk that a child would have a life of very low quality. (For some, of course, even this would not be acceptable.) As we have seen, however, several members of staff at this PGD clinic seem to think that it is

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<sup>76</sup> E.g. A. Buchanan *et al.*, *supra* n. 37; J. Robertson, 'Genetic Selection of Offspring Characteristics', (1996) 76 *Boston University Law Review* 421; B. Steinbock, 'Disability, Prenatal Testing and Selective Abortion', in E. Parens and A. Asch (eds), *Prenatal Testing and Disability Rights* (Georgetown University Press, 2000) 108; J. Botkin, 'Line Drawing: Developing Professional Standards for Prenatal Diagnostic Services', in E. Parens and A. Asch, *ibid.* 288.

<sup>77</sup> See e.g. T. Shakespeare, 'Choices and Rights: Eugenics, Genetics and Disability Equality', (1998) 13/5 *Disability and Society* 665, 672.

<sup>78</sup> T. Shakespeare, "'Losing the Plot?' Medical and Activist Discourses of Contemporary Genetics and Disability', (1999) 21/5 *Sociology of Health and Illness* 669, 681.

<sup>79</sup> T. Shakespeare, *supra* n. 77, 672.

appropriate to take account of parental interests in PGD and were prepared to say so quite explicitly in relation to very serious conditions as well as in relation to Cystic Fibrosis. This suggests that they consider that in such cases the parental interests outweigh the moral claims of the embryo.

More specifically, why might parents be interested in the option of avoiding the birth of a child with Down syndrome and how might this compare with their interests in relation to Cystic Fibrosis? In the latter case, it may be that prospective parents are likely to think primarily about the avoidance of suffering. Arguably, they can think of Cystic Fibrosis testing as 'testing which avoids suffering'. This is so even though to be born with this condition often appears compatible with a reasonable quality of life and a life worth living. In the case of Down syndrome, there may be physical suffering in severe cases and prospective parents may worry, in particular, about the risk of cardiac defects, particularly inoperable ones. However, it may be that prospective parents are more likely to think about the question of mental impairment and to worry about how severe this may be. Although mental impairment may give rise to a loss of opportunity in the life of someone with Down syndrome, it will not necessarily mean that that person is unhappy or feels unfulfilled themselves, although they may suffer from social stigma. Despite this, in light of the potential health problems and a degree of mental impairment, it seems unlikely that prospective parents would *hope* for a child with Down syndrome. At the same time, it may be hard for prospective parents to face—let alone admit to—the possibility that Down syndrome testing may really be about their own potential interests and concerns. Interestingly, this difficulty appears to be echoed among the staff at the clinic (many of whom are likely to be parents) in their discussions of Down syndrome, since it seemed hard for anyone to state explicitly that testing for Down syndrome might really concern parental interests although Scientist 8 (below) touches on this. Further, out of the 26 interviews conducted at this site, only 4 brought up the example of Down syndrome in their one-to-one interviews, although the interviewer occasionally raised it.<sup>80</sup> (Of course, in part, this is not surprising in that PGD for Down

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<sup>80</sup> Scientist 9 raises the issue of PGD for Down syndrome in addition to testing for other conditions. Scientist 3 mentions Down syndrome, but this is simply in connection with analysing amniocentesis results. Administrator 12 comments on whether people necessarily abort a foetus with Down syndrome. Counsellor 17 raises the issue of adding in PGD for Down syndrome in relation to concerns about decreasing the accuracy of test results. Scientist 21 discusses Down syndrome and PGD for Down syndrome, although the interviewer was the first to mention the issue. Doctor 6 responded to a question about Down syndrome by the interviewer; Doctor 24

syndrome had only just become part of the agenda for discussion at the clinic.) Rather, typically the issue was brought up by the philosopher in the discussion groups.

The important point about whose interests may be at stake (and why) in testing for Down syndrome was put to one of the groups by the philosopher<sup>81</sup>:

[I]n the past people were very ready to make quite sort of broad sweeping statements about the quality of life of somebody with Down's syndrome. But people now no longer feel happy making [those statements]. The shift of emphasis is on, 'Well what does it mean to the family to have a child that will remain dependent for much longer in their life, that might have... health problems etc? What does it mean to the other siblings?' And some people are less comfortable with that sort of more global calculation than they would be with the calculation in the case where you actually look at the condition and...

Here, she suggests some plausible reasons as to why parents may be concerned to be able to choose whether or not to have a child with Down syndrome and, at the same time, acknowledges that recognition of these reasons may be uncomfortable or unacceptable for some. Of course, if we do accept that parents are entitled to avoid the birth of children even when they would have conditions, such as Cystic Fibrosis, that might well enable them to have a reasonable quality of life, then we have no reason to say that it is not acceptable for parents to be concerned with the similarly less serious (though very different) condition of Down syndrome. However, it could be argued that it may be harder to acknowledge this openly in the case of Down syndrome because of the element of mental impairment: in particular, we speculate that it may be easier for parents to say that they want to avoid suffering (for instance, as in the case of Cystic Fibrosis) than to say that they want to avoid mental impairment in their child (in the case of Down syndrome). What did staff think about the philosopher's suggestion about what may be the real purpose of Down syndrome testing?

At this point, Doctor 22 commented that 'those children are very happy children often aren't they?'. Scientist 8 responded<sup>82</sup>:

But that's exactly the point. In that situation we're talking about existing individuals and their quality of life. When we're looking

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responded to two such questions; Nurse 25 responds to a question about Down syndrome in connection with PGD.

<sup>81</sup> B. Farsides, EDG 2, 5.

<sup>82</sup> EDG 2, Scientist 8, 5.

at a PGD family, we're not saying, we're not making any judgement about an existing individual by saying that on average these people do not have the opportunities and quality of life that normal people have, the parents have a huge burden. So we're not, there's no living person that we're saying we're going to take away their life, this is not about that. We're not going to deprive some individual, of life, by doing the test. I mean it is different – a non-existing person does not have the same weight in the balance as the existing person. Isn't that right?

Scientist 8 seems to envisage here the idea of PGD for Down syndrome, although he/she may also be alluding to PGD more generally. There are a number of important ideas embedded in what he/she says and it is worth trying to spell these out: first, that when people reflect that children with Down syndrome often seem 'very happy', they are thinking of people who actually exist and their quality of life; and, therefore, secondly, deliberations in PGD do not entail judgments about the quality of lives of existing people; rather, thirdly, they entail general reflection on the opportunities and quality of life 'on average'; fourthly, there is the suggestion (which may or may not include Down syndrome) that parents 'have a huge burden'; fifthly, the point is made that no-one's life is being taken away by doing 'the test', coupled with the thought that 'non-existing' people have less weight in any moral equation than existing ones. Arguably, if we were to take the view that the foetus has greater moral status than the embryo (and various members of staff seemed to think this was the case, while others did not),<sup>83</sup> this last statement would apply more strongly in the PGD than the PND context. However, since on many moral views also the 12-week foetus is not a person,<sup>84</sup> the statement may also apply in the context of PND. Each of these statements seems concerned to justify the idea of PGD, including PGD for Down syndrome, and among them we find the notion of a 'burden' for parents. Interestingly, we find the statement about parents' interests mixed in with a number of important statements about the justifiability of testing. This may again highlight the difficulty that people may seem to feel about pointing too overtly to parents' potential interests in relation to Down syndrome testing.

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<sup>83</sup> Examples of staff in favour of a gradualist approach to the embryo's and foetus's moral status were Scientist 8, 4, and Embryologist 15, 16; an example of a member of staff who attributed a high moral status to the embryo was Scientist 2, 15–16. We do not attempt here to represent the proportion of staff with given views.

<sup>84</sup> In contrast, even the embryo has the moral status of a born child on a potentiality account. For an adherent, see J. Finnis, 'The Rights and Wrongs of Abortion: A Reply to Judith Thomson', (1973) 2 *Philosophy and Public Affairs* 117.

Since Down syndrome is a condition about which people may be fearful, they may also be misinformed. In particular, people may overestimate the impact on them of the birth of a child with Down syndrome. Being well or fully informed about the life of someone with Down syndrome will most likely not be true of the majority of the couples coming through for PGD for another condition. One reason for this is that the syndrome arises from a chromosomal trisomy and is generally not inherited. Even if a couple has met or known a child or an adult with Down syndrome, they will not necessarily know about the range of severity of effects, both physical and mental. Information provision in relation to a condition such as Down syndrome would therefore be very important if PGD for Down syndrome were offered at the clinic. Shakespeare has suggested that

Decisions about screening should be based on good information: rather than evaluating screening programmes in terms of those who undergo tests and terminations, programmes should be evaluated in terms of the proportion of people who were empowered to make an informed choice.<sup>85</sup>

The possibly unbalanced nature of information about Down syndrome has been explored in the context of PND.<sup>86</sup> It would, therefore, be crucial that this issue is adequately addressed in the context of PGD.

In one of the groups, staff showed an awareness of the importance of information provision. With reference to Down syndrome being the most common condition for which there is prenatal screening, Scientist 8 observed<sup>87</sup>: 'But that's presumably because the women who accept the screening and prenatal testing, are ones who know about the condition enough to know that they will not necessarily want to have a Down's baby'. However, Counsellor 28 responded: 'Except I think they don't...'. The concern that people may take up the offer of Down syndrome screening and testing without adequate reflection (either in the PND or PGD context) is well caught by Counsellor 10<sup>88</sup>: 'As long as the medical profession is vigilant in providing unbiased, accurate information, and not just putting these couples or women on a conveyor belt, you know, like what sort of has happened with the Down's syndrome screening, then I think you're empowering people to make an individual choice'.

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<sup>85</sup> T. Shakespeare, *Supra* n. 78, 685.

<sup>86</sup> See C. Williams, P. Alderson and B. Farsides, 'What Constitutes 'Balanced Information in the Practitioners' Portrayals of Down's Syndrome?', (2002) 18 *Midwifery* 230.

<sup>87</sup> EDG 2, Scientist 8, 14–15.

<sup>88</sup> EDG 1, Counsellor 10, 29.

We have suggested why parents may want to be informed about Down syndrome and why they may or may not want to accept the offer of testing for it. We have also suggested that there are arguments as to why testing for Down syndrome may legitimately be seen as part of reproductive autonomy (at least at the stage of selection by PGD and earlier rather than later in a pregnancy) just as testing for other conditions that leave someone with a life they think worth living, such as Cystic Fibrosis, may be.

We now turn very briefly to note the remaining factors that the HFEA recommends should be considered in relation to the decision to undertake particular PGD tests and to draw out some relevant comments of the clinic's staff. There is only scope to give a flavour of the views of health professionals and scientists in relation to these factors.

### **3. The Extent of Any Intellectual Impairment**

The possible distinction between a child's and its parents' interests can also be noted in relation to 'the extent of any intellectual impairment'. A child with moderate intellectual impairment may well make considerable educational progress and subsequently lead a life of some independence.<sup>89</sup> This may be the case with more mild expressions of Down syndrome. Yet, if the people seeking treatment have some interest in which children are born, then their views and interests will have some weight. Apparently in support of this, Administrator 12 observed: 'I don't say that a child with learning difficulties or anything like that isn't going to be able to live a good life, good quality of life. But I think the couples should be able to make that decision themselves'.

### **4. The Extent of Social Support Available**

Clearly, good social support will be in the interests of both a child and its parents. The issue was touched on in one of the discussion groups, as Doctor 32 observed: 'But if you talk about it basically, you know, why can't a child with a disability, with Down syndrome or something else, not be perfectly okay if it's adequately supported?'<sup>90</sup> We can relate this observation to the distinction between 'medical' and 'social' models of disability. The medical model has tended to dominate understandings of what it is to be disabled.<sup>91</sup> We may have seen an example of this above when Scientist 2 observed: 'No, but, when it's serious, their life is completely debilitated by it... it's *medically* serious'.<sup>92</sup> The medical

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<sup>89</sup> On Down syndrome, see the Down's Syndrome Association, at <http://www.downs-syndrome.org.uk>.

<sup>90</sup> EDG 3, Doctor 32, 26.

<sup>91</sup> See e.g. J. Harris, 'Is There a Coherent Social Conception of Disability?', (2000) 26 *J.M.E.* 95.

<sup>92</sup> Our emphasis.

model has been subject to a critique in the form of the 'social' model.<sup>93</sup> The former locates disability within the individual, the latter in the social environment the individual inhabits, so that the individual is 'impaired', not 'disabled'. Over time, some proponents of either model have accepted to some degree the limitations of their respective approaches.<sup>94</sup> And it is now clear that some combination of the models is best placed to account for the significance of impairment.<sup>95</sup> In turn, it can reasonably be acknowledged, as Shakespeare has, that despite good social support there may still be considerable difficulties in raising an 'affected' child.<sup>96</sup> Therefore, if prospective parents' interests are relevant to selection decisions, there may still be reason to test for a given condition, even when good social support would be available (which it will not always be).

### 5. The Family Circumstances of the People Seeking Treatment

There are various elements of possible relevance here. Prospective parents may already have other affected children. Or this factor might allow for concerns as to the possible effects, one way or the other, of the birth of a seriously impaired child in a family generally. The welfare-of-the-child requirement of the Human Fertilisation and Embryology Act allows for the interests of other children to be considered.<sup>97</sup> Whether 'family circumstances' is confined to the nuclear family or whether a couple must already have children in order for this factor to be relevant is unclear. Arguably, some flexibility here is desirable given the variation in families' experiences. The relevance of family circumstances to all the conditions discussed here was noted implicitly or explicitly in earlier discussion, for instance, in relation to Duchenne muscular dystrophy, Cystic Fibrosis and Down syndrome. Further, there were numerous observations in the discussion groups and interviews that revealed great empathy with couples in light of their family experience to date. For instance, in relation to cancer in the family, Nurse 4 observes<sup>98</sup>:

So if someone felt so passionate about it, I don't feel that I can judge or say what they - because I don't know, unless you're in someone else's shoes, I don't feel that we can make such a judgment really.

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<sup>93</sup> See e.g. T. Shakespeare, *supra* n. 77.

<sup>94</sup> E.g. J. Harris and T. Shakespeare, *supra* n. 91 and 77.

<sup>95</sup> J. Glover, *supra* n. 53, pp. 7-8.

<sup>96</sup> See e.g. T. Shakespeare, *supra* n. 77.

<sup>97</sup> The HFE Act, s.13(5) states: 'A woman shall not be provided with treatment services unless account has been taken of the welfare of any child who may be born as a result of the treatment (including the need of that child for a father), and of any other child who may be affected by the birth'.

<sup>98</sup> Interview, Nurse 4, 8.

So if someone has got a family history, a very strong family history of cancer, then ... who am I to say?

Doctor 6 also alludes generally to the experience with which a couple presents at the clinic<sup>99</sup>:

There are very few grey areas that I find in my mind. I think that anybody... that comes here seeking PGD has been on such a roller coaster ride and been through so much, that one has to appreciate just the sheer emotional turmoil they've been through.

Doctor 6 also notes the difficulty of making judgments about seriousness unless one has experience of a condition:

...I think you can only make decisions on such subjects if you have experience in that. I don't. I couldn't tell you whether... was serious enough, but I bet if we had a roomful of parents and families with children affected with it, I'm sure they would voice their opinion - in their situation it is a hugely important, devastating thing.

Further, Counsellor 17 observes that 'at the end of the day, it should be - we should be led by what parents want to do, they're in the best position to know what's right for them and their family'.<sup>100</sup>

## **6. The Availability of Effective Therapy or Management Now and in the Future**

The availability of therapy or management now is potentially relevant to many conditions. The possibility of future treatment is of particular significance in relation to testing for late onset or lower penetrance disorders, especially certain breast cancers. (It is beyond our scope to consider the HFEA's recent consultation and decision in relation to these.<sup>101</sup>) For instance, in one of the discussion groups Scientist 34 observed<sup>102</sup>: '[W]ith breast cancer you can have all this, you know, preventative treatment and treatment is a lot better'. Later, the discussion proceeded as follows:

Scientist 2: But it's treatable to the point where you will have a normal lifespan. . .

Embryologist 33: Not in everybody.

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<sup>99</sup> Interview, Doctor 6, 4.

<sup>100</sup> Interview, Counsellor 17, 18-19.

<sup>101</sup> See HFEA, *Choices and Boundaries* (Nov. 2005); HFEA, 'Authority Decision on the Use of PGD for Lower Penetrance, Later Onset Inherited Conditions' (10 May 2006).

<sup>102</sup> EDG 5, Scientist 34, 25.



Scientist 2: No, but it can be now because you can have a mastectomy and that's it, you're cured for life.

There is not scope here to discuss other aspects of treatment by mastectomy and the suitability of PGD for breast cancer. It would be interesting to know whether there would ever be a situation in which the clinical staff thought that the relevant treatment was sufficient, but prospective parents did not. Given the empathy with a couple's family experience that we saw in relation to the previous factor, it is possible that most members of staff would defer to patients on the question of the acceptability of a given treatment. Inevitably, however, there remains the issue of uncertainty as to future treatments.

### **7. The Speed of Degeneration in Progressive Disorders**

Once again, both the child's and the prospective parents' interests could be relevant. In the extreme case of Tay-Sachs, where a child will live at most four to five years and will suffer greatly during this time, birth is unlikely to be either in its or its parents' interests and so PGD for Tay-Sachs is arguably strongly supportable. The clinic's staff were highly sensitive to the impact of a child's death on its parents. For instance, from one of the discussion groups<sup>103</sup>:

Doctor 32: For the parents, the death of a child is very serious... I can only, of course, judge from the outside experience, you know, because I've been an oncologist for a long time. And I've seen children die and what that did to the parents throughout the illness and the death and everything. So that's where I'm coming from...

It is beyond our scope to discuss the quality of life of someone who dies, say, in the teenage or early to mid-adult years. However, when someone has lived that much longer, perhaps into early to mid-adulthood, then it may be that prospective parents' interests may have less weight in selection decisions, although this will be complicated by the severity and certainty of the relevant condition. Testing for Huntington's Chorea is taking place and, generally, the staff were very open to the views of people who had experience, in one way or another, of Huntington's. For instance, in interview Nurse 4 observed: 'when you've got something like Huntington's... I mean the parents are so aware of how devastating it is'.<sup>104</sup>

### **8. Their Previous Reproductive Experience**

Lastly, great sensitivity to a couple's reproductive past was shown in very many of the interviews and discussion groups. There was discussion

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<sup>103</sup> EDG 3, Doctor 32, 19, partly noted earlier.

<sup>104</sup> Interview, Nurse 4, 7-8.

of the experience of couples with another affected child (or children), of couples who had frequently miscarried and of couples who had felt the need to terminate previous pregnancies.

In relation to the advantages of PGD over PND, Scientist 8 observes: 'I would look at it very much from the woman's point of view, that it's rescuing women from having to make those appalling decisions'.<sup>105</sup> Similarly, Embryologist 15 observes<sup>106</sup>:

I think I've got a leaning towards PGD more from a patient-orientated view, inasmuch as what the patients have actually been through to actually be at that stage, and offering a treatment that doesn't involve termination of a fetus when it's actually growing inside them, I think it's a really viable way of bypassing a lot of problems with terminations of pregnancy for what are often the most atrocious diseases, you know, really they've been through a lot of trauma by the time they actually come to see us.

The avoidance of further difficulties or suffering was also relevant in relation to patients who had suffered from miscarriages due to chromosomal translocations. Doctor 14 observes<sup>107</sup>:

And, to me, it is legitimate to avoid them going through this heartbreaking experience and the anticipation and the torture of not knowing a) that it will happen, b) that if it happens and they became pregnant and that pregnancy would be doomed to another miscarriage – so, in my mind, that is a sensible indication to give them the benefit of treatment and the technique that will increase the probability of them having a continuous pregnancy as opposed to going through that experience again.

Here, there is the highly compassionate suggestion that a couple should not be expected to suffer just because the risk of miscarriage in relation to a given translocation is not certain.

#### IV. CONCLUSIONS

At the start of this paper, we reviewed the HFEA and HGC recommendations and discussions relating to the PGD criteria of 'a significant risk of a serious genetic condition'. We saw that the recommendations sought to recognise, at least to some extent, the personal nature of the issues at stake in PGD, but also to observe limits to the legitimacy of prospective parents' views. In summary, this was to be achieved by

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<sup>105</sup> Interview, Scientist 8, 2.

<sup>106</sup> Interview, Embryologist 15, 2.

<sup>107</sup> Interview, Doctor 14, 8.

requiring discussion between prospective parents and health professionals about the degree of risk and the seriousness of any given condition. We suggested that the HFEA and HGC had, in effect, put these professionals in the position of being something of a 'check' on what these bodies saw as the potentially excessively wide views of those seeking treatment.<sup>108</sup> Have the views of staff that we have explored here indeed shown these health professionals and scientists to be in this gatekeeper role?

In relation to the degree of risk, we saw that health professionals and scientists tended to view risk in quantitative terms. (Of course, the Abortion Act relies on a quantitative notion of 'substantial risk'.) This was interesting, given that a possibly more subjective notion of risk was intended by the JWP.<sup>109</sup> One exception to this is Scientist 21, who commented on how his/her own experience of pregnancy changed his/her perception of risk<sup>110</sup>:

So, what I'm saying, actually putting myself in that position of the patient, it completely changed my perspective... subsequently, when I've been giving risk figures out to people, because I can imagine what it's like getting that figure, that result myself now.

As we have seen, the HFEA and HGC also envisaged that the degree of risk should be a matter for discussion and agreement.

Most of our discussion concentrated on the thorny issue of seriousness. We saw that health professionals and scientists saw some conditions as being very definitely serious, such as Tay-Sachs, but that many saw less serious conditions, such as Cystic Fibrosis (and potentially Down syndrome) as sufficiently serious to meet the criteria for PGD.<sup>111</sup> Given that these conditions were less serious, however, the question then arose 'serious for whom?'. Here we saw the view, at least on the part of some staff, that sometimes seriousness concerns possible parental interests. In relation to Down syndrome, this tended to be implied rather than stated. In line with the recognition generally of the legitimacy of parental interests, great weight was given to the views of prospective parents by staff. They were mindful of couples' previous reproductive experience—particularly the traumas of abortion and miscarriage—and also of the experience that those seeking treatment might have of a given

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<sup>108</sup> HFEA licence committees are of course a further possible check. However, it is likely that if a clinic supports an application for PGD, the licence committee will usually agree.

<sup>109</sup> *ibid* and *Supra* n. 10.

<sup>110</sup> Interview, Scientist 21, 27.

<sup>111</sup> The possibility of testing for Down syndrome, as an addition, was the subject of discussion at the clinic.

condition, for instance through the previous birth of an affected child. In this process staff were, in effect, attending to many of the factors that the HFEA and HGC recommended should be considered in the decision to offer PGD.

However, although staff were sensitive to prospective parents' views and experiences, this did not mean that they necessarily thought that 'anything goes'. While they were often prepared to defer to prospective patients in relation to 'grey areas', this was largely because they thought it would be inappropriate to judge the seriousness of a condition of which they had no experience. Further, by definition of course, a 'grey area' is not one that concerns trivial features or conditions. Indeed, there was a recognition that people would only be motivated to seek PGD and, in particular, go through with the attendant IVF, where they truly experienced something as a problem—as 'serious'. In these cases, it is likely (though not certain) that staff (and perhaps HFEA licence committees when the case is made out by staff) will agree that a condition is sufficiently serious to justify PGD. This is not to deny that there may always be conditions that may provoke debate and disagreement, perhaps between staff and perhaps also, though not necessarily, between staff and prospective parents. The clinic's regular case-by-case and multi-disciplinary meetings (to discuss new conditions) were noted above.

In summary, it appears that if these health professionals and scientists were truly to doubt the seriousness of a given condition for which a couple is seeking PGD, they would act as a check on their views, and would be likely to deny the provision of PGD. In any event, in response to the idea that relatively mild conditions might be the subject of testing, Scientist 8 reminds us of the 'limitations of PGD in terms of the number of embryos you have available'.<sup>112</sup> The implication of this limitation is that at some point a couple's desire to have a child at all could come into conflict with their desire to have a child without a very much less serious condition. In other words, they would have to reflect carefully on their true goal.

Overall, these health professionals and scientists showed great empathy with the experiences of prospective parents and were likely to defer to their views. In response to the interviewer's question, '[s]hould we always give patients what they want?', Doctor 14 replies: 'No, no. It's not in absolute terms. They want it for a legitimate reason'.<sup>113</sup> Of another staff member, the interviewer asks: '[i]f they wanted to do something, anything, would it be acceptable because it

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<sup>112</sup> EDG 2, Scientist 8, 19.

<sup>113</sup> Interview, Doctor 14, 2. The interviewer was K. Ehrlich.

is what they wish?’ Scientist 9 responds<sup>114</sup>: ‘I think it depends on, it’s within reason, isn’t it?’ This notion of reason is crucial. The scientist continues: ‘But what is reason, whose reason, whose reason is it?’ More generally, this is a central question in medical ethics and law. The best answer that we can give in this context is that as long as a condition lies in that ‘grey area’, its seriousness can be a matter of reasonable disagreement. For this reason, it could be appropriate to offer PGD.

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<sup>114</sup> Interview, Scientist 9, 7. The interviewer was K. Ehrich.