Human
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AUTHORITY

Outcome of the Public Consultation on Preimplantation Genetic Diagnosis

Foreword

In November 1999 the Human Fertilisation and Embryology Authority and the then Advisory Committee on Genetic Testing issued a consultation document on preimplantation genetic diagnosis (PGD). The Human Genetics Commission took over responsibility for this subject when it was formed in December 1999.

PGD is a technique that combines genetic testing and IVF in order to offer those who are at significant risk of passing on a serious genetic condition the choice of selecting embryos that are unaffected before a pregnancy is begun. However, the wider implications of the technique are of concern to a great many people as the responses received and the forthright views expressed showed. We are grateful to all those who responded for their valuable contribution to this important debate.

In order to consider these responses a Joint HFEA / HGC Working Party was established in late 2000. It worked quickly in a spirit of collaboration that has characterised this consultation exercise throughout. We would like to thank the Joint Working Party for their work in reviewing the responses and preparing their conclusions. A summary of their discussions and recommendations is presented here.

The Joint Working Party concluded that in general most respondents agreed with the current approach of licensing centres to test for a limited number of specific and serious conditions. The existing (interim) guidance was felt to have worked well in this respect. However, the JWP also provided valuable recommendations aimed at strengthening the expertise available to HFEA in reviewing applications for PGD licences and increasing the support and counselling available to those seeking treatment.

The HFEA, the body responsible for licensing and regulating PGD in the United Kingdom, will consider these recommendations in preparing new licensing guidance for centres wishing to conduct PGD. The HGC has also identified the wider issue of genetic testing and reproductive issues as an important topic for further work during 2002 and will want to continue its work with HFEA and others to promote informed public debate about the long term ethical and social issues that must be confronted in the light of developments in human genetics. We consider that the completion of this phase of public consultation on PGD marks an important step in the debate.

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Further copies of this document are available from:
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It is also available on the HFEA and HGC websites (www.hfea.gov.uk and www.hgc.gov.uk/business_publications.htm respectively)

The Human Fertilisation and Embryology Authority/Advisory Committee on Genetic Testing Consultation Document on Preimplantation Genetic Diagnosis is downloadable in portable document format (pdf) (http://www.hfea.gov.uk/pgd/pgdpaper.pdf).

The Joint Working Group met 4 times between December 2000 and May 2001, and the minutes of its meetings have been published on the HGC website (www.hgc.gov.uk/business_groups.htm#jwp).

Outcome of the Public Consultation on Preimplantation Genetic Diagnosis

Contains the Summary of Discussions and Recommendations of the HFEA/HGC Joint Working Party and the Analysis of Responses carried out by the HFEA

November 2001

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HUMAN FERTILISATION AND EMBRYOLOGY AUTHORITY/HUMAN GENETICS COMMISSION JOINT WORKING PARTY ON PREIMPLANTATION GENETIC DIAGNOSIS

Summary of Discussions and Recommendations

Introduction

1. Responding to concerns raised by potential uses of the technique of preimplantation genetic diagnosis (PGD), the Human Fertilisation and Embryology Authority (HFEA) and the then Advisory Committee on Genetic Testing (now subsumed into the Human Genetics Commission (HGC)) published a joint public consultation document in November 1999. Responses were solicited until March 2000 and these were analysed by the HFEA. (The analysis of responses follows.) In August 2000 Ruth Deech, chairman of the HFEA, wrote to Helena Kennedy, chair of the HGC, suggesting the establishment of a joint working party to take forward the consultation exercise.

2. The objectives and terms of reference of the HFEA/HGC Joint Working Party on PGD (JWP) were as follows:

   - to make recommendations concerning the HFEA's licensing of PGD;
   - to make recommendations concerning the nature of guidance as to when PGD should be offered in clinical treatment in the light of ethical issues raised by the technique; these recommendations to include recommendations relating to preimplantation genetic screening (PGS), for example, aneuploidy screening;
   - to report the Working Party's discussions to the Human Fertilisation and Embryology Authority and the Human Genetics Commission;
   - to make recommendations concerning a joint public response based on the outcome of the PGD consultation exercise.

3. (A list of members of the Joint Working Party is given in the Annex).

Licensing of PGD

4. Under the HFEA's interim PGD licensing guidance centres wishing to offer a PGD service are required to apply to the HFEA for a variation to their licence. Before licensing a particular test

for use at a centre the HFEA will require evidence about the safety and efficacy of the tests and the centre's competence in the techniques used, as well as notification of the information given to patients and the means by which the decision to use PGD has been reached. In the case of ethically difficult decisions centres may seek advice from a treatment ethics committee and this may be taken into account by a licence committee when considering an application. All PGD applications are sent out to a minimum of two peer reviewers and decisions are taken by HFEA licence committees in consideration of the scientific and clinical information currently available, and in the light of prevailing moral attitudes towards the proposed treatment. (This procedure was broadly endorsed by the majority of responses to the consultation: paras 28, 58 (pt 1) and 60.)

5. The JWP agreed that in general the interim licensing guidance had worked effectively but wished to recommend developments in a number of areas, many of which were highlighted in public responses to the consultation.

6. The first of these areas was increasing the input of those with specific genetic expertise to the consideration of licence applications. Since under the Human Fertilisation and Embryology Act 1990 (HFE Act) HFEA licence committees must be composed exclusively of members of the Authority, the JWP considered the possibility of establishing a dedicated PGD advisory group to provide additional counsel. This suggestion was not favoured however, not least because of the delay that involving such a group would be likely to impose on the provision of treatment. After much discussion the JWP considered that the most effective way of achieving this objective would be through the peer review mechanism, and agreed to recommend that peer reviewers with expertise in all disciplines connected with genetic illness and disability should be sought.

7. **<u>Recommendation 1</u> The list of peer reviewers should be expanded to include clinical geneticists, molecular geneticists, cytogeneticists, and genetic counsellors.**

8. In order to make best use of this expertise the JWP agreed that the input of peer reviewers should be structured in a manner that focussed attention on the issues peculiar to PGD and the conditions with which it was concerned. Accordingly, two further recommendations were made, relating to information to be provided by applicants and peer reviewers.

9. **<u>Recommendation 2</u> A specialised peer review form for PGD applications should be developed.**

10. **<u>Recommendation 3</u> Peer reviewers should also be asked to comment on the seriousness of the disorder for which the centre is applying to use PGD in the light of guidance given in the Notes for Centres.**

11. Additionally the JWP considered that it would be useful for centres to provide further information of a non-clinical nature in their applications for the benefit of lay members.

12. Owing to the difficulty of obtaining this information in the case of rare genetic disorders the JWP agreed that two types of information could be considered, namely (1) information about the full range of the experiences of affected individuals and their families with an indication of where this information could be found, and (2) information provided by specialist clinicians with experience of the disorder in question.

13. **<u>Recommendation 4</u> Centres should include in their application a paragraph describing in lay terms the condition for which they are applying to use PGD, incorporating descriptions of the full range of the experiences of affected individuals and their families where appropriate.**

Multidisciplinary team

14. Many responses to the consultation highlighted the fact that the interim licensing of PGD had functioned effectively, but would benefit from being broadened to include more specific guidance relating to the other relevant disciplines including medical ethicists. The guidance should also include the importance of the clinical team working together, in the provision of a comprehensive PGD service.

15. **<u>Recommendation 5</u> A multidisciplinary team including reproductive specialists, embryologists, clinical geneticists, genetic counsellors, cytogeneticists and molecular biologists should be involved in the PGD service at all licensed centres. This team should maintain close contact with the primary care physician or referring clinician and treatment should also encompass continued support of patients following PGD.**

Genetic Consultation

16. The role of genetic counsellors in the management of those requesting PGD was discussed at length by the JWP. Note was taken of the fact that that the route into treatment for those seeking PGD was typically very different from that of IVF patients, since they would ordinarily be seeking treatment as a result of a preexisting indication of a genetic disorder rather than infertility. The JWP felt that the role of genetic consultation in the PGD service would benefit from greater emphasis than was given in the interim guidance. However since this aspect of the service could not be inspected directly by the HFEA, it was agreed that reference should be made in the guidance to some formal system of accreditation. Whilst it was felt that ideally those seeking PGD treatment should be referred by a Regional Genetics Centre it was acknowledged that this might discriminate against some of those seeking treatment privately, particularly those from abroad.

17. **<u>Recommendation 6</u> The HFEA/HGC Joint Working Party made the following recommendations regarding genetic counselling:**

- **that this aspect of the PGD service should be referred to as 'genetic consultation,' as those seeking treatment should have access to both clinical geneticists and genetic counsellors;**
- **that, ideally, people seeking treatment should be referred to the centre by a Regional Genetics Centre; however, all those seeking treatment should at least be known to an accredited clinical geneticist;**
- **that centres should work closely with the local genetics team of the people seeking treatment.**

Accreditation and inspection of genetics laboratories

18. Similarly the regulatory jurisdiction of the HFEA does not extend to genetics laboratories. Consequently the JWP considered that emphasis on a standard of professional accreditation was important. Whilst it was recognised that genetics laboratories could only be visited at the invitation of the laboratory concerned, it was felt that the opportunity to review the relevant facilities was highly desirable.

19. **<u>Recommendation 7</u> The HFEA inspection should include all aspects of the PGD service, including genetics laboratories.**

20. **<u>Recommendation 8</u> All genetics laboratories used for PGD should be CPA accredited (or equivalent) or at least be working towards Clinical Pathology Accreditation (or equivalent), with accreditation to be completed within five years.**

Decision-making regarding the use of preimplantation genetic diagnosis

21. Concern was expressed, both by respondents to the consultation and by the Joint Working Party, about the recommendation in the interim guidance to make use of the Royal College of Obstetricians and Gynaecologists (RCOG) document *Termination of Pregnancy for Fetal Abnormality in England Wales and Scotland* (RCOG, 1996). Reservations centred around two areas: first, the crucial importance of the views and experiences of those seeking treatment in decision-making, and second, the dependency of the RCOG guidance on the World Health Organization (WHO) definition of 'serious disability' which was felt to be based on a view that emphasised a discriminatory conception of disability.

22. It was noted that the WHO's revised classification, *ICIDH-2—International Classification of Functioning, Disability and Health* (WHO, 2000), was currently in prefinal draft but the JWP felt that that this did not provide a standard by which to judge serious disability that was appropriate to the very personal decision to seek PGD treatment. Accordingly it was felt that reference to the RCOG guidance should be replaced by dedicated PGD guidance and the JWP made a number of recommendations concerning how this should be developed. This reflected the view that PGD guidance should support difficult parental choices rather than appearing to discriminate against individuals with certain conditions.

23. In line with the overwhelming weight of responses to the PGD consultation (para.35, pt 1) the JWP agreed that the guidance given on the use of PGD should not comprise a prescriptive list of 'serious conditions' for which the use of the technique was thought to be appropriate. The 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.

24. **<u>Recommendation 9</u> Guidance on the appropriate use of PGD should concern factors which should be taken into account and the process by which the decision to use PGD is reached rather than comprising a list of 'serious conditions' for which PGD is considered acceptable.**

Restrictions on the use of PGD

25. Respondents to the consultation (paras 30, 35 (pt 2) & 52) indicated strongly that restrictions should be placed on the use of PGD to prevent it being used for frivolous or 'social' reasons, or for eugenic purposes. Whilst the JWP appreciated that significant differences existed between the techniques of PGD and post-implantation prenatal diagnosis (PND) it agreed as a precautionary principle that the use of PGD should be consistent with the use of PND.

26. **<u>Recommendation 10</u> The guidance should state that indications for the use of PGD should be consistent with current practice in the use of PND.**

27. The JWP agreed that in its recommendations, whilst references to the risk of the occurrence and the seriousness of the condition should be retained, these should be coupled with further recommendations concerning how judgements about these should be reached in individual cases.

28. **<u>Recommendation 11</u> 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.**

29. It was noted that a recommendation of this nature should relate to individual tests thereby implicitly ruling out Human Leukocyte Antigen (HLA) typing (where an embryo is selected to provide a tissue match for transplant to an existing family member). The JWP discussed this extension of PGD, which had not been mentioned specifically in the 1999 consultation document, and agreed that the there were sufficient ethical difficulties with this approach that it should be subject to further discussion before its use was considered.

30. Reference to the 'embryo' in this recommendation was made in the awareness that this would be consistent with any future decision to license the detection of aneuploidy (numerical chromosomal abnormalities) in embryos. Members of the JWP agreed that the value of

screening had yet to be demonstrated and as a result they strongly suggested that if it were to be licensed in future this should be within the context of a clinical study.

31. **<u>Recommendation 12</u> Screening for aneuploidy to improve IVF outcomes should be further considered by the HFEA as its safety and efficacy has not yet been sufficiently established.**

Significance of risk of occurrence and seriousness of the condition to be diagnosed

32. The JWP discussed at length the nature of the decision to pursue treatment involving PGD. Consistent with the majority of responses to the consultation (paras 47 (pt 2) & 52) it agreed that the personal nature of this decision 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.

33. **<u>Recommendation 13</u> 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.**

34. In particular the JWP was concerned about the nature and extent of information provided to those seeking treatment to help them reach in reaching an informed judgement on the seriousness of the condition for which PGD was being considered. It was felt that information should be balanced and include that provided by disabled people and their families about their experiences of living with disability.

35. **<u>Recommendation 14</u> The guidance should indicate that the seriousness of a condition should be a matter for discussion between the people seeking treatment and the clinical team. Information provided to those seeking treatment to be taken into account 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.**

Other factors relevant to decision-making

36. In line with responses to the consultation (paras 36, 38, 45, & 47 (pt 1)) 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.

37. **<u>Recommendation 15</u> 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 impairment;**
- **the extent of social support available; and**
- **the family circumstances of the people seeking treatment.**

38. Embarking on preimplantation testing presents patients and clinicians with a further set of choices to be made after the outcome of the test is known. Difficult choices might arise if the test is inconclusive, if only affected embryos are available for transfer, or if collateral information about the genetic status of other family members was discovered.

39. In view of this the JWP agreed that in the case of chromosomal rearrangements or autosomal recessive conditions, if it is possible to exclude affected embryos without discovering the carrier status of others and without compromising the accuracy of the test, then this is to be preferred.

40. **<u>Recommendation 16</u> The possible outcomes of genetic testing and their implications should have been fully explored with the people seeking treatment prior to PGD being undertaken.**

41. In formulating its recommendations the JWP is cognizant of the fact that the technology associated with preimplantation genetic diagnosis is continually developing and it recognises that there will be a need to keep the framework for licensing treatments under review in order to ensure that this is able to accommodate novel techniques and applications.

JUNE 18, 2001

Annex

Membership of the HFEA/HGC Joint Working Party on Preimplantation Genetic Diagnosis

Professor Allan Templeton	Chairman
Dr William Albert	Member of the HGC
Dr Frances Flinter	Co-optee on HGC Genetic Testing Sub-group
Professor Christine Gosden	Member of the HFEA
Dr Hilary Harris	Member of the HGC
Professor Henry Leese	Member of the HFEA
Professor Stuart Lewis	Member of the HFEA
Mr Phillip Webb	Member of the HGC

Analysis of the Responses to the Joint HFEA/ACGT Consultation Paper on PGD

Introduction

1. The HFEA/ACGT joint consultation on preimplantation genetic diagnosis (PGD) ended in March 2000. A total of 171 responses were received; 124 individual responses and 47 responses from organisations.

2. This paper gives:

- the data for all responses received;
- a breakdown of data for the responses from individuals; and
- a breakdown of the data for the responses from organisations.

3. Many respondents answered only some of the questions. Further, 32 of the 124 individuals who replied did not answer the specific questions asked. Instead they expressed a general opinion about PGD. Their responses, therefore, have only been included in the data given in tables 1 and 2 below which relate to a general statement of opinion. A few responses were too ambiguous to be recorded.

4. In addition, many respondents qualified their answers. A selection of the most common and/or significant qualifications has been included with respect to each question. A section giving other general comments can be found at the end of the paper.

Categorisation of Responses

Individuals

5. Those individuals who answered one or more of the specific questions posed in the consultation document were grouped into three categories: 'disability'; 'clinical'; and 'other'. These categories include the following:

- 'Disability' - those individuals who indicated some experience of disability, including carers, families as well as disabled individuals themselves;
- 'Clinical' - those with some experience in the wider field of obstetrics and gynaecology and fetal and maternal medicine as well as those more immediately involved in fertility treatment. This category also includes individuals who identified themselves as counsellors, nurses or others working in this field; and
- 'Other' – any individuals who did not fall into the above categories. Most did not indicate an allegiance to, or experience in, any particular field.

Organisations

6. The organisations that responded to the consultation (given in the Annex) were subdivided into the following five categories: 'clinical/scientific'; 'bioethical/social science'; 'consumer groups'; 'disability'; and 'religious or pro-life'. Two organisations formally

acknowledged the consultation but chose not to comment (NGDT, Wellcome) and were therefore not included in the data.

Statements in favour/against PGD in general

7. Of the total 171 responses received, 68% (117 responses) expressed a clear opinion in favour of PGD in general, unrelated to the question of regulation or the specific questions posed in the consultation document. 31% (53 responses) were against PGD on the same terms.

8. Of the individual responses, 70% were in favour of PGD in general and 30% against as shown subdivided below:

Table 1

Category	Total	For	Against
Disability	20	12	8
Clinical	42	42	0
Other	30	23	7
Non-specific resp.	32	10	22

9. Of the responses received from organisations, 62% were in favour of PGD in general and 34% were against as shown subdivided below:

Table 2

Category	Total	In favour	Against
Clinical/Scientific	9	8	0
Bioethical/Soc. sc.	6	4	2
Consumer groups	8	8	0
Disability groups	10	7	2
Religious or Pro-life	14	2	12

Para 28: Do you agree with the proposal that, subject to appropriate clinical considerations, the current practice of licensing clinics to perform PGD for a limited number of specific serious inherited conditions, including sex linked disorders and chromosome abnormalities should continue?

a) total respondents

10. Of the 119 respondents that addressed this specific question, 74% (88) agreed with the proposal. In many cases, however, this agreement was subject to caveats such, for example, that legislation should not be over-restrictive, that patients' choices should be respected, or that balanced information about life as a disabled person should be provided to potential parents. These caveats were more fully elaborated in answer to the other questions posed in the consultation document.

11. Of those that did not agree with the proposal, 20% (24) argued that the licensing system should place greater restrictions on PGD than at present. Some of these responses maintained that PGD was a process in which living beings would be discarded. Concerns were also raised

that PGD contributed to a society in which the disabled were devalued and that it was inherently eugenic.

12. 8% (9) of responses argued in favour of a lower level of regulation of PGD. Some of these respondents argued that, as long as a medical indication exists, it should be legitimate to perform PGD for less serious disorders. Concern was also expressed that the proposed system of regulation was in itself eugenic because of the role of a statutory authority in the definition of normality and 'acceptable disabilities'. This argument asserted that the danger of eugenics increased with a greater level of regulation.

b) individual respondents

13. 77% (60) of the individual respondents agreed with the proposal. 12% (11) argued that greater restriction was needed while 8% (7) argued for fewer restrictions as shown below:

Table 3

Category	**Yes**	**No – greater restriction needed**	**No – less restriction needed**
Disability (20)	10	5	0
Clinical (42)	30	0	6
Other (30)	20	6	1

c) organisation respondents

14. 65% (28) of these respondents agreed with the proposal and 30% (13) argued in favour of greater restriction. Arguments in favour of less regulation were noted in a couple of responses.

Table 4

Category	**Yes**	**No – greater regulation**
Clinical/Scientific (9)	7	0
Bioethical/Soc. sc. (6)	3	2
Consumer groups (8)	7	0
Disability groups (10)	8	2
Religious or Pro-life (14)	3	9

Para 30: In due course should there be restrictions on who might have access to PGD?

a) total respondents

15. Of the 99 responses to this question, 81% (80) agreed that there should be restrictions as to who might have access to PGD. In responding to this question, some people confused the issue of regulatory restrictions with the issue of funding. The latter responses have not been included in this data.

16. Of these 81%, 85% (68) agreed that PGD should be restricted to cases where there is a known family history of a serious genetic disorder or to cases of aneuploidy. The majority of these responses felt that PGD should only ever be intended for such cases. However, the opinion that PGD should be more widely available once its safety was more firmly established was also expressed with the implication that, in the future, PGD might also be available for less serious conditions. A few respondents who objected to the more general use envisaged in the consultation document still felt that PGD could be beneficial in cases of aneuploidy, where the technique would be geared towards ensuring that an embryo was carried to term.

17. 8% (8) argued that all access to PGD should be denied, and 11% (11) argued that there should be unrestricted access to be PGD or that it should be offered routinely in IVF. These responses stressed the primacy of reproductive choice or the fact that the intention in all cases would be to have a 'healthy child'. Other stressed that parents would only be likely to consider PGD if there was a prior indication of a genetic disorder.

b) individual respondents

18. Of the individual respondents, 86% (57) argued that access should be restricted and 73% (48) thought that PGD should be restricted to cases where there is a known family history of a serious genetic disorder or to cases of aneuploidy as shown by subdivision below:

Table 5

Category	**Yes – unspecified**	**Yes – known family history of serious genetic disorder or aneuploidy**	**All access denied**	**No – unrestricted use or offered routinely in IVF**
Disabled (20)	1	8	1	0
Clinical (42)	1	27	0	4
Other (30)	7	13	2	2

c) organisation responses

19. Of the responses to this question received from organisations, 70% (23) argued that access should be restricted and 61% (20) thought that PGD should be restricted to cases where there is a known family history of a serious genetic disorder or to cases of aneuploidy.

Table 6

Category	**Yes – unspecified**	**Yes – known family history of serious genetic disorder or aneuploidy**	**All access denied**	**No – unrestricted use or offered routinely in IVF**
Clinical/Scientific (9)	0	4	0	1
Bioethical/Soc. Sc.(6)	0	3	1	2
Consumer groups (8)	1	3	0	1
Disability groups (10)	2	5	0	1
Religious or Pro-life (14)	0	5	4	0

Para 35: Part 1 – Should the seriousness of a genetic condition be a matter of clinical judgement based on general guidance?

a) total respondents

20. Of the 81 responses to this question, 80% (65) agreed with the proposal that the seriousness of a condition should be a matter of clinical judgement based on general guidance. 20% (16) argued that the seriousness of a genetic condition should be determined by a specified list. These responses tended to see a danger in allowing individual preference to be a determining factor in the use of PGD.

b) individual responses

21. Of the 58 individual responses to this question, 79% (46) agreed with this proposal. Over 25% (12) of these responses stressed the importance of patient input in this decision making process. 21% (12) argued in favour of a specified list.

Table 7

Category	Yes	No – specified list
Disability (20)	6	3
Clinical (42)	25	3
Other (30)	15	6

c) organisation responses

22. Of the 23 responses received from organisations, 83% (19) agreed with the proposal.

Table 8

Category	Yes	No – specified list
Clinical/Scientific (9)	5	0
Bioethical/Soc. Sc. (6)	3	1
Consumer groups (8)	4	0
Disability groups (10)	5	2
Religious or Pro-life (14)	2	1

Part 2 – If so, what aspects might such general guidance cover?

23. Of the 40 responses received to the second element of this question, 30% (12) argued that general guidance should be based on the RCOG criteria for termination of pregnancy. The case for this was most strongly argued by the 'clinical' category of respondents and often emphasised that it would be illogical to restrict PGD to a greater extent than the more ethically difficult PND. Some responses also argued that PND carries a greater risk than PGD.

24. 63% (25) argued that the guidance should be based on one or more of the following factors:

- disabled people's experience of the severity of a disorder, including sociological or psychological studies of this nature. Several responses stressed that disabled individuals should be involved in the provision of "accurate" information to patients about the reality of living with a disability. This has been represented as 'multidisciplinary' in the tables below;
- the clinical and bioethical respondents who are represented under 'other' in the following tables, recommended that guidelines should take into account the following factors: degree of suffering; available medical therapy or effective treatment; individual circumstances of the family or woman; speed of degeneration; extent of mental impairment; and sensitivity and specificity of the test;
- the patient and consumer groups represented under 'other' stressed the importance of the involvement of non-medical groups such as ethics committees in the decision making process and the need for patient information to be clear and accessible.
- three respondents also suggested the WHO guidelines as a basis for general guidance.

b) individual respondents

25. Of the 26 individual responses, 35% (9) argued that general guidance should be based on the RCOG guidelines for the termination of pregnancy. 54% (14) argued that the general guidance should be based on a 'multidisciplinary approach' or on 'other' factors.

Table 9

Category	**Based on PND (RCOG TOP)**	**Multidisciplinary approach**	**Other**
Disability (20)	0	3	2
Clinical (42)	8	1	2
Other (30)	1	1	5

c) organisation respondents

26. Of the 14 responses from organisations, 79% (11) argued that general guidance should be based on a 'multidisciplinary approach' or on 'other' factors.

Table 10

Category	**Based on PND (RCOG TOP)**	**Multidisciplinary approach**	**Other**
Clinical/Scientific (9)	1	0	3
Bioethical/Soc. Sc.(6)	0	0	1
Consumer groups (8)	1	1	3
Disability groups (10)	0	3	0
Religious or Pro-life (14)	1	0	0

Para 36: Have you any comments on the general issue of replacing carrier embryos?

a) total respondents

27. There were 89 responses to this question. 38% (34) of responses felt that carrier embryos should be replaced. It was, however, often unclear whether those responding felt that carrier embryos should be replaced as a matter of principle (that the aim to eradicate a gene from the gene pool has eugenic implications) or whether carrier embryos merely could be replaced, for example, in the event that they were the only good embryos available. Some responses argued that it would be improper to test for carrier status and that this should be avoided where possible. 51% (45) of responses argued that the choice of whether to replace carrier embryos should remain with the patients. 11% (10) of the responses argued that carrier embryos should not be replaced. The argument often presented in support of this was that the aim of PGD should be to eradicate disorders entirely.

b) individual respondents

28. Of the 63 individual responses to this question, 33% (21) argued that carrier embryos should be replaced. 57% (36) argued that the choice should remain with the patients. In connection with this question, two of the individual clinical respondents drew a distinction between autosomal recessive and x-linked conditions with respect to this question.

Table 11

Category	**Should be replaced**	**Should not be replaced**	**Patients choice**
Disability (20)	5	0	5
Clinical (42)	3	5	22
Other (30)	13	1	9

c) organisation respondents

29. Of the 26 responses to this question, 50% (13) argued that carrier embryos should be replaced and 35% (9) argued that the choice should remain with the patients.

Table 12

Category	**Should be replaced**	**Should not be replaced**	**Patients choice**
Clinical/Scientific (9)	2	1	1
Bioethical/Soc. Sc.(6)	1	0	2
Consumer groups (8)	1	1	3
Disability groups (10)	3	2	2
Religious or Pro-life (14)	6	0	1

Para 38: Can the principle of the Welfare of the Child ever be compatible with the decision to begin a pregnancy knowing that a child will be born with a genetic disorder?

a) total respondents

30. Of the 98 who responded to this question, 47% (46) argued that the principle of the welfare of the child could be compatible with the decision to begin a pregnancy knowing that a child will be born with a genetic disorder. The majority of these responses qualified their answers by stating that this might only be appropriate in certain circumstances, depending on severity of the condition, family situation and other similar considerations. In addition, most of the 'yes' responses concerned only the question of affected embryos where there were no unaffected ones for transfer, but rejected PGD as a method of choosing an affected embryo for social or other reasons.

31. 31% (30) of responses felt that this was an issue that should be left to the choice of the patients. 22% (22) of respondents argued that starting such a pregnancy could never be compatible with the welfare of the child. It should also be noted that many respondents were unfamiliar with the terminology of 'welfare of the child' and some respondents questioned its meaning, range or relevance.

b) individual respondents

32. Of 66 individual responses, 45% (30) argued that such a decision could be compatible with the welfare of the child. 27% (18) argued that the decision should be left with the patients concerned. A further 27% (18) argued that the decision to begin a pregnancy could never be compatible with the welfare of the child principle. The majority of the respondents who argued that affected embryos should not be replaced were part of the 'clinical' group of respondents.

Table 13

Category	Yes	No	Patient's choice
Disability (20)	6	0	3
Clinical (42)	13	13	7
Other (30)	11	5	8

c) organisation respondents

33. Of the 32 responses from organisations, 50% (16) argued that replacing affected embryos could be compatible with the welfare of the child. 38% (12) argued that the decision should be left to patients.

Table 14

Category	Yes	No	Patient's choice
Clinical/Scientific (9)	1	0	3
Bioethical/Soc. Sc. (6)	1	0	3
Consumer groups (8)	4	2	3
Disability groups (10)	4	2	2
Religious or Pro-life (14)	6	0	1

Para 45: It is suggested that if a disorder is of late onset, this should be one of a number of factors, but not an overriding factor, in determining whether PGD should be offered. Do respondents consider this to be the correct approach?

a) total respondents

34. Of 88 responses to this question, 59% (52) agreed with the proposition in this paragraph. Many of these responses drew a distinction between highly predictive late onset disorders and other types. 24% (21) disagreed with the proposition. These responses included both those who felt that the fact that a disorder is late onset should not effect an evaluation of its severity, and those who argued that PGD should not be carried out for late onset disorders at all. 17% (15) argued that this question should be a patient's decision.

b) individual respondents

35. Of 63 responses from individuals, 62% (39) agreed with the proposition, 22% (14) disagreed and 16% (10) said that this should be a decision for the patients.

Table 15

Category	Yes	No	Patient choice
Disability (20)	4	5	3
Clinical (42)	25	1	5
Other (30)	10	8	2

c) organisation respondents

36. Of 25 responses from organisations, 52% (13) agreed with the proposition and 28% (7) disagreed. Those that disagreed with the proposition all stated that PGD should not be performed if the disorder is late onset. 20% (5) said that this should be for the patients to decide. The consumer groups in particular stressed the importance of counselling in allowing patients to make a decision in cases of late onset disorders.

Table 16

Category	Yes	No	Patient's choice
Clinical/Scientific (9)	3	0	1
Bioethical/Soc. Sc. (6)	3	0	0
Consumer groups (8)	3	1	2
Disability groups (10)	4	1	1
Religious or Pro-life (14)	0	5	1

Para 47: Part 1 – Should guidance distinguish between PGD for genes that are highly predictive of a serious disorder and those where the genetic component is more complex?

a) total respondents

37. Of 66 responses to this question, 94% (62) agreed with this proposition. Their position was based both on ethical considerations surrounding such use of PGD and on concerns that the science is not sufficiently developed for accurate prediction in cases of multifactorial and polygenic disorders.

b) individual respondents

38. Of 45 individual responses to this question, 96% (43) agreed with this proposition.

Table 17

Category	Yes	No
Disability (20)	6	1
Clinical (42)	23	1
Other (30)	14	0

c) organisation respondents

39. Of 21 responses from organisations to this question, 90% (19) agreed with this proposition.

Table 18

Category	Yes	No
Clinical/Scientific (9)	3	0
Bioethical/Soc. Sc. (6)	3	1
Consumer groups (8)	5	0
Disability groups (10)	4	1
Religious or Pro-life (14)	4	0

Part 2 – Should the use of PGD for any indication be the subject of clinical judgement, and as such left to practitioners and individual patients to decide?

a) total respondents

40. Of 41 responses to this question, 66% (27) agreed with the proposition in this question. Most of these appeared to be qualified in that the decision should be taken by the clinician and individual involved only as long as it was within certain established boundaries. 34% (14) argued that the seriousness of a condition should be established in a central list.

b) individual respondents

41. Of 30 responses received from individuals, 70% (21) agreed with the proposition in this question.

Table 19

Category	Yes	No (specified list)
Disability (20)	2	1
Clinical (42)	11	2
Other (30)	8	6

c) organisation respondents

42. Of 11 responses from organisations, 55% (6) agreed with the proposition.

Table 20

Category	Yes	No (specified list)
Clinical/Scientific (9)	1	1
Bioethical/Soc. Sc. (6)	1	1
Consumer groups (8)	2	0
Disability groups (10)	2	1
Religious or Pro-life (14)	0	2

Para 52: In the context of PGD, and given the current practical limitations, should there be any restrictions on the number and range of tests to be carried out in the absence of a clear genetic or medical indication?

a) total respondents

43. Of the 88 responses received to this question, 88% (77) agreed with the proposition in this question that there should be restrictions on the number and range of tests to be carried out in the absence of a clear genetic or medical indication. The clinical and consumer groups stressed the central role of patient and doctor decision making in deciding the number and range of tests to be carried out. Responses from the clinical group also raised the issue of aneuploidy screening as an instance for which testing should be available.

b) individual respondents

44. Of the 65 responses from individuals to this question, 88% (57) agreed with the proposition.

Table 21

Category	Yes	No
Disability (20)	9	0
Clinical (42)	25	6
Other (30)	23	2

c) organisation respondents

45. Of the 23 responses from organisations, 87% (20) agreed with the proposition.

Table 22

Category	Yes	No
Clinical/Scientific (9)	4	0
Bioethical/Soc. Sc. (6)	2	1
Consumer groups (8)	4	2
Disability groups (10)	7	0
Religious or Pro-life (14)	3	0

Para 58: Part 1 – Should centres be licensed for PGD in general or in relation to each specific test and condition?

a) total respondents

46. Of the 78 responses to this question, 37% (29) argued that centres should be licensed for PGD in general. One organisation (BFS) suggested that the HFEA should regulate laboratory conditions and techniques but not the circumstances in which PGD may be carried out. 63% (49) argued that centres should be licensed in relation to each specific test and condition. Some of these responses stressed the importance of regularly reviewing any list of specific tests.

b) individual respondents

47. Of the 57 individual responses, 42% (24) argued that centres should be licensed in general and 58% (33) argued that centres should be licensed in relation to each specific test and condition.

Table 23

Category	In general	Specific condition
Disability (20)	1	5
Clinical (42)	9	20
Other (30)	14	8

c) organisation respondents

48. Of the 21 responses received from organisations, 24% (5) argued that centres should be licensed for PGD in general and 76% (16) argued that centres should be licensed in relation to each specific test and condition.

Table 24

Category	In general	Specific condition
Clinical/Scientific (9)	1	3
Bioethical/Soc. Sc. (6)	0	3
Consumer groups (8)	4	3
Disability groups (10)	0	3
Religious or Pro-life (14)	0	4

Part 2 – Should the HFEA record each new condition, mutation or test carried out by individual centres?

a) total respondents

49. Of 54 responses to this question, 100% (54) of responses agreed that each new condition, mutation or test should be recorded by the HFEA.

b) individual respondents

50. There were 40 individual responses to this question.

Table 25

Category	Yes	No
Disability (20)	5	0
Clinical (42)	13	0
Other (30)	22	0

c) organisation respondents

51. There were 14 responses from organisations to this question.

Table 26

Category	Yes	No
Clinical/Scientific (9)	3	0
Bioethical/Soc. Sc. (6)	3	0
Consumer groups (8)	3	0
Disability groups (10)	2	0
Religious or Pro-life (14)	3	0

Para 60: Do respondents think that the general approaches proposed for the regulation of PGD are appropriate?

a) total respondents

52. Of 75 responses to this question, 75% (56) agreed that the general approaches proposed in the consultation document were adequate. 25% (19) did not agree.

b) individual respondents

53. Of 56 individual responses to this question, 75% (42) agreed with the proposition in the question.

Table 26

Category	Yes	No
Disability (20)	4	7
Clinical (42)	25	2
Other (30)	13	5

c) organisation respondents

54. Of the 19 responses received from organisations, 74% (14) agreed with the proposition.

Table 27

Category	Yes	No
Clinical/Scientific (9)	3	0
Bioethical/Soc. Sc. (6)	3	0
Consumer groups (8)	3	0
Disability groups (10)	3	3
Religious or Pro-life (14)	2	2

General Comments

55. Many of the responses also contained comments which were not directly related to any particular question. The following is a summary of some of the relevant and more prevalent points made.

- the issue of funding for PGD was widely mentioned both with regard to natural justice and with respect to the concerns that a privileged super-class with access to this technique may emerge. A number of NHS trusts also requested that the HGC and HFEA should propose a process for assisting the NHS in deciding on the cost effectivess of PGD and when it should no longer be considered an experimental procedure;

- many respondents addressed the issue of the status of disabled people within society and stressed that the most disabling factor for people with serious impairments or conditions is the attitude of, or barriers within, society. Many of these respondents were concerned that PGD would reinforce social prejudice against the disabled and that the proponents of PGD saw disability in an entirely negative light. Others argued that the desire to alleviate disability does not imply disrespect for disabled individuals;

- a few respondents mentioned that the real needs of families affected by genetic disorders, which may partly be met by PGD, should not be lost sight of due to an undue fear of a hypothetical scenario about the 'abuse' of the technology.

- the regulation of PGD must place greater emphasis on counselling and the provision of information to patients. Patient groups also stressed that choosing not to undergo PGD and to risk having a child with a disorder should not be seen as irresponsible. The decision to have PGD must be the patients' own;

- some responses suggested that the success rates for PGD should be published;

- many of the 'clinical' responses stressed the importance of follow up studies as a means of directing the future regulation of PGD. Consumer groups also stressed the need for follow-up studies on the social and psychological welfare of people undergoing PGD and of the families where it had been successful;

- one clinical response suggested that the use of PGD could be widened to allow identification of sex or diagnosis of y-linked defects in male infertility cases treated by ICSI (to prevent transmission of genetic defects affecting infertility);

- it was suggested that aneuploidy screening should be licensed separately from PGD for other reasons;

- concern was expressed about pressure exerted on healthcare professionals to provide a service with which they are not comfortable;

- some responses criticised the consultation document itself. A few responses argued that it was too much concerned with procedures rather than ethical principles and that the consultation period had come too late as PGD was already licensed by the HFEA. Some of these responses called for a moratorium to enable a more effective discussion to take place concerning the ethics of PGD. Others, however, expressed regret that the consultation document did not cover the practical aspects of the licensing procedures including the training and assessment of the biopsy practitioner and the proposed accreditation mechanisms for genetic laboratories.

- a few responses asked that research be undertaken to develop alternative procedures which do not involve the destruction of the embryo, i.e. diagnostic techniques involving the ova and spermatozoa;

- a number of responses expressed the concern that PGD would draw funding away from the development of treatments for diseases and care for people with disorders.

Summary

56. The following conclusions have been drawn from the data extracted from this consultation:

a) All respondents

i) 70% of all respondents, including those who did not address specific questions, were in favour of PGD in principle.

b) Respondents who addressed specific questions

ii) 74% were in favour both of PGD in principle and of its continued regulation by the HFEA. 20% argued that the licensing system should place greater restrictions than at present and 8% argued for a lower level of regulation.

iii) 81% indicated that the use of PGD should be restricted to cases in which there is a known family history of serious genetic disorder or for the detection of aneuploidy. 8% agreed that all access to PGD should be denied and 11% argued that there should be unrestricted access.

iv) 80% agreed that the seriousness of a genetic condition should be a matter of clinical judgement based on general guidance. The majority of these suggested that such guidance should be based either on the RCOG Termination of Pregnancy criteria or on a range of additional considerations outlined in the analysis. 20% argued that the seriousness of a genetic condition should be determined by a specified list.

v) 51% agreed that patients should judge whether carrier embryos should be replaced. 38% agreed that carrier embryos should be replaced and 11% argued that they should not be replaced.

vi) 47% acknowledged that there might be situations in which the replacement of an affected embryo would be compatible with the principle of the welfare of the child. 31% argued that this should be a decision left to the parents. 22% argued that starting such a pregnancy could never be compatible with this principle.

vii) 59% agreed with the approach suggested in the consultation that the fact that a disorder is late onset should be one of a number of factors, but not an overriding factor, in determining whether PGD should be offered. 24% disagreed and 17% argued that this should be the patient's decision.

viii) 94% felt that guidance should distinguish between PGD for genes that are highly predictive and those disorders where the genetic component is more complex.

ix) 88% agreed that there should be restrictions on the number and range of tests to be carried out in the absence of a clear genetic or medical indication.

x) 63% felt that centres should be licensed for each specific test or condition, rather than for PGD in general. 37% argued that centres should be licensed for PGD in general. It was unanimously agreed that the HFEA should record each new condition, mutation or test.

xi) 75% agreed that the general approaches to regulation outlined in the document were appropriate.

Annex

Organisations that responded to the consultation on PGD

Clinical/Scientific

British Fertility Society
British Infertility Counselling Association
British Medical Association
ESHRE PGD Consortium
Medical Research Council
Public Health Genetics Network
Royal College of Nursing
Royal College of Obstetricians and Gynaecologists
The Royal Society of Edinburgh

Bioethical/Social Science

Centre for Family Research
European Bioethical Research
Nuffield Council on Bioethics
Progress Educational Trust
Public Health Genetics Unit
The Social Council on Human Bioethics

Consumer Groups

Antenatal Results and Choices
Child
Children in Scotland
Directorate of Women & Children's Health
Issue
National Council of Women of Great Britain
PROGAR
Townswomen's Guilds

Disability

ADAPT
Association of Camphill Communities
Cystic Fibrosis Trust
Disability Awareness in Action
Disabled Peoples' International Europe
Genetic Interest Group
John Grooms
Thalidomide Society
The Association of Cystic Fibrosis Adults
The Campaign Against Human Genetic Engineering

Religious or Pro-Life

Catholic Child Welfare Council
Catholic Children's Rescue Society
Centre for Bioethics and Public Policy
Christian Action Research and Education
Comment on Reproductive Ethics
Image
Labour Life Group
LIFE
Public Morals Committee of the Reformed Presbyterian Church
Society for the Protection of Unborn Children
The Church of England's Board for Social Responsibility
The Fellowship of Independent Evangelical Churches
Upminster Christian Concerns