



## "Preimplantation Genetic Diagnosis: Practices and Attitudes of Assisted Reproductive Technology Clinics"

April, 2006

- Study Details -

### **Preimplantation Genetic Diagnosis:**

#### **Practices and Attitudes of Assisted Reproductive Technology Clinics**

Thank you for agreeing to participate in this survey of ART clinics. The Johns Hopkins University Genetics and Public Policy Center is conducting this survey with the technical assistance of Knowledge Networks and funding from The Pew Charitable Trusts. The purpose of the survey is to collect information about ART clinics' experiences with preimplantation genetic diagnosis (PGD) and ART clinic directors' perspectives about a number of issues related to PGD. We hope these data will provide an accurate snapshot of current PGD practices and attitudes. Completing this survey gives you the opportunity to weigh in on a number of important policy issues that will help us provide a complete picture of the way PGD is practiced and how it might be practiced in the future.

This study was approved by the Johns Hopkins University IRB and was reviewed and approved by the Research Committee of the Society for Assisted Reproductive Technology.

All of the information you provide in this survey will be kept in strictest confidence. Only aggregate results of the survey will be published.

During the survey, you will be given the option to authorize re-contact for a follow-up interview. No re-contact will be initiated without your authorization and unless the Johns Hopkins IRB approves a separate study protocol.

This survey should be taken **ONLY** by the medical director, laboratory director, IVF director, or his/her designee.

This survey will take approximately 20 minutes to complete

If you need to stop at any point and resume the survey at a later time, you may log in again and the survey will begin where you left off.

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Q1 Are you employed by an ART center that performs IVF?

- A. Yes
- B. No

MULTI PUNCH ANSWER

Q2 Are you the ...

- A. Medical director of an ART clinic or his/her designee
- B. Laboratory director of the ART clinic or his/her designee
- C. IVF director of the ART clinic or his/her designee
- D. None of the above

SINGLE PUNCH ANSWER

Q3 Which of the following best describes your IVF clinic?

- A. Independent/commercial/private/free-standing clinic
- B. University affiliated
- C. Part of for-profit hospital, not university-affiliated
- D. Part of non-profit hospital, not university-affiliated
- E. Other

*Q5 not used in this report*

SINGLE PUNCH ANSWER

Q6 Is your clinic a member of SART?

- A. Yes
- B. No

SINGLE PUNCH ANSWER

Q7 To the best of your recollection, approximately how many IVF cycles did you initiate in 2005?

- A. 0-100
  - B. 100-500
  - C. 500-1000
  - D. 1000-2000
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E. 2000+

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**Now we would like to ask you about different policies and procedures at your clinic. All of the information you provide in this survey will be kept in strictest confidence. Only aggregate results of the survey will be published.**

Questions about PGD

SINGLE PUNCH ANSWER

Q8 Have you provided PGD services to patients in your clinic?

- A. Yes
- B. No [go to Q70 if no or skip]

MULTI PUNCH ANSWER

Q9 Which types of PGD services has your clinic provided?

(you will have the opportunity later in the survey to describe whether you perform the genetic analysis in your clinic's own laboratory or refer it out).

- A. PGD for an autosomal single gene disorder(s), either recessive or dominant
- B. PGD for chromosomal rearrangements
- C. PGD for X-linked disease
- D. PGD for aneuploidy
- E. PGD for non-medical sex selection, for example, family balancing
- F. PGD for HLA typing

SINGLE PUNCH ANSWER

Q9b Is the PGD offered by your clinic offered:

- A. as a clinical service
- B. under a research protocol
- C. both

*Q10-14 not used in this report*

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*No questions numbered 15-17*

Q18 Approximately how many PGD cycles of each type did you perform in 2005?

	Number of PGD cycles started in 2005
PGD for single gene	
PGD for chromosomal rearrangement	
PGD for x-linked disease	
PGD for aneuploidy	
PGD for non-medical sex selection, for example, family balancing	
PGD FOR HLA TYPING	

SINGLE PUNCH ANSWER

Q19 Who performs the biopsy of the embryo for your PGD patients?

- A. In-house person performs the biopsy
- B. External person comes in to perform the biopsy

MULTI PUNCH ANSWER

Q20 What are the qualifications of the person who performs the biopsy?

Check as many as apply:

- A. Certified by the American Board of Bioanalysis (ABB)
  - B. Certified by the American Board of Medical Genetics
  - C. Certified by the American Board of Pathology
  - D. Certified by the American Board of Clinical Chemistry
  - E. Other certification
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F. Specialized training in PGD biopsy

G. Don't know

Q20d. Please describe the specialized biopsy training this person has received (fill in text box).

SINGLE PUNCH ANSWER

[FOR Q21-24, if they choose B or C but don't fill in the blank, show this prompt: "Please tell us where you send samples."]

Q21 For PGD for single gene disorders, the genetic analysis is performed

A. In my clinic's own PGD laboratory

B. In a laboratory outside my clinic (list all laboratories where sent \_\_\_\_\_)

C. Some is performed in my clinic's own PGD laboratory and some in a laboratory outside my clinic (list laboratories where sent \_\_\_\_\_)

SINGLE PUNCH ANSWER

Q22 For PGD for chromosomal rearrangements, the genetic analysis is performed:

A. In my clinic's own PGD laboratory

B. In a laboratory outside my clinic (list all laboratories where sent \_\_\_\_\_)

C. Some is performed in my clinic's own PGD laboratory and some in a laboratory outside my clinic (list laboratories where sent \_\_\_\_\_)

SINGLE PUNCH ANSWER

Q23 For PGD for x-linked disorders, the genetic analysis is performed

A. In my clinic's own PGD laboratory

B. In a laboratory outside my clinic (list all laboratories where sent \_\_\_\_\_)

C. Some is performed in my clinic's own PGD laboratory and some in a laboratory outside my clinic (list laboratories where sent \_\_\_\_\_)

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SINGLE PUNCH ANSWER

Q24 For PGD for aneuploidy, the genetic analysis is performed

- A. In my clinic's own PGD laboratory
- B. In a laboratory outside my clinic (list all laboratories where sent\_\_\_\_\_)
- C. Some is performed in my clinic's own PGD laboratory and some in a laboratory outside my clinic (list laboratories where sent\_\_\_\_\_)

*Q25 not used in this report*

MULTI PUNCH ANSWER

Q26 What technique(s) do you use in PGD? Check all that apply:

- A. Polar body analysis
- B. Single blastomere analysis
- C. Two blastomere analysis

Q27 On which day of embryonic development do you usually perform blastomere biopsy?

Day \_\_\_\_\_ (enter single digit number)

*Q28-29 not used in this report*

SINGLE PUNCH ANSWER

Q30 Has your clinic provided PGD for non-medical sex selection in the absence of another medical indication for PGD?

- A. Yes, we defer to parental preferences
- B. Only for a second or subsequent child (family balancing).
- C. No, we do not provide PGD solely for non-medical sex selection

MULTI PUNCH ANSWER

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Q31 Please indicate below the reasons that your clinic does not provide non-medical sex selection in the absence of another medical indication for PGD.

- A. We are not technically equipped to determine the sex of the embryo
- B. We have a clinic ethics policy or guidance that prohibits it
- C. We have not had any patient requests
- D. Other (fill in)

**SINGLE PUNCH ANSWER**

Q32 Which of following best describes your clinic's procedures regarding the sex of embryos.

- A. We inform parents of the sex of the unaffected embryos and comply with parental preferences to transfer only embryos of a specific sex if there are a sufficient number of embryos
- B. We inform parents of the sex of the unaffected embryos and comply with parental preferences to transfer only embryos of a specific sex if there are a sufficient number of embryos and if this is second or subsequent child.
- C. We reveal the sex and transfer embryos of the desired sex only if asked by families. We don't volunteer the option without being asked.
- D. We never reveal the sex of the embryos to the families in the absence of an X-linked disorder.
- E. Other (Please describe)

**SINGLE PUNCH ANSWER**

Q33 Have you ever provided PGD to a family seeking to select an embryo for the presence of a disability (e.g. a family wishes to have a child with a hereditary hearing loss)?

- A. Yes
- B. No

**SINGLE PUNCH ANSWER**

Q34 Have you ever provided PGD to a family seeking to avoid an adult-onset genetic disease (Huntington disease, hereditary breast cancer, Alzheimer disease)?

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- A. Yes
  - B. No

*Q35 not used in this report*

SINGLE PUNCH ANSWER

Q36 Have you ever provided PGD to a family seeking to select an HLA matched embryo for an ailing sibling in the absence of testing for a single gene mutation?

- A. Yes
- B. No

*Q37 Not used in this report*

Q38 Have you had patients request PGD for reasons you or your colleagues felt raised ethical questions or sensitivities?

- A. Yes
- B. No

Q38b Please briefly describe the circumstances and what was requested.  
(fill in) \_\_\_\_\_

Q38c. Are there other times you had requests you or your colleagues felt raised ethical questions or sensitivities?

- A. Yes [repeat for up to five circumstances]
- B. No

SINGLE PUNCH ANSWER

Q39 In your clinic, who ultimately decides whether to perform the PGD in situations that raise ethical questions or sensitivities?

- A. Individual physician who is caring for the patient in the clinic
-

- 
- B. Medical director of the clinic
  - C. Ethics committee of the clinic
  - D. Staff of clinic
  - E. Director of my clinic's PGD laboratory
  - F. Laboratory staff in my clinic's PGD laboratory
  - G. Laboratory where PGD genetic analysis is performed – external
  - H. Other \_\_\_\_\_

SINGLE PUNCH ANSWER

Q40 Does your clinic have a formal policy or policies addressing whether it will perform PGD in cases that may raise ethical questions or sensitivities?

- A. Yes
- B. No

*Q 41 not used in this report*

*Q 42 left intentionally blank*

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**QA/QC and certification/licensing/accreditation**

**Now we will ask you questions about your embryology laboratory.**

SINGLE PUNCH ANSWER

Q 43. Does the embryo biopsy take place in your IVF clinic's embryology laboratory?

- A. Yes
- B. No

Q43b. The biopsy takes place in \_\_\_\_\_ (fill in)

*Q44 left intentionally blank*

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MULTI PUNCH ANSWER

Q45 Is your IVF clinic's embryology laboratory certified/accredited/licensed by the following entities?

Please check as many as apply

- A. CLIA
- B. CAP/RLAP
- C. JCAHO
- D. COLA
- E. New York State-CLEP
- F. Other

*Q46 not used in this report*

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**Now we will ask you a few questions about your clinic's own PGD laboratory.**

Q47 For PGD genetic analysis performed in your clinic's own PGD laboratory, do you perform internal proficiency testing for the molecular or cytogenetics?

- A. Yes
- B. No

*Q48 not used in this report*

Q49 For PGD genetic analysis performed in your clinic's own PGD laboratory, do you perform positive controls?

- A. Yes
  - B. No
-

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Q50 For PGD genetic analysis performed in your clinic's own PGD laboratory, do you perform negative controls?

- A. Yes
- B. No

*Q50b, 50c not used in this report*

MULTI PUNCH ANSWER

Q51 Is your clinic's laboratory that performs PGD genetic analysis certified, accredited or licensed by the following entities? Please check as many as apply.

- A. CLIA
- B. CAP/RLAP
- C. JCAHO
- D. COLA
- E. New York State-CLEP
- F. Other \_\_\_\_\_

*Q52, 53, 54, 55, 56, 57, 58 not used in this report*

SINGLE PUNCH ANSWER

Q59 Does your clinic's PGD laboratory receive samples for genetic analysis of blastomeres or polar bodies from other IVF clinics?

- A. Yes
  - B. No
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Q59b: Previously in this survey you gave the following answers about how many PGD cycles you performed in 2005. Please clarify how many cycles were performed for your own patients and how many were performed on samples received from other centers:

For 2005:

Your previous answer		Your patients	Samples received
	Single gene		
	Chrom rearrange		
	X-linked		
	Aneuploidy		

\*Note that the numbers should automatically fill in from Q18 in the first column

*Q60, 61 not used in this report*

SINGLE PUNCH ANSWER

Q62 Among patients in your clinic, have you been aware of any inconsistencies between the PGD of embryos and later genetic testing (including analysis of additional blastomeres, amniocentesis/CVS, genetic testing of fetal tissue after miscarriage, or genetic testing of baby after birth).

- A. Yes
- B. No

Q63 Approximately how many times have you seen such inconsistencies? (fill in) \_\_\_\_\_

MULTI PUNCH ANSWER; ALLOW UP TO TWO ANSWERS

Q64 To the best of your knowledge, what is the most likely cause of the inconsistencies you observed? (choose up to two)

- A. Embryo mosaicism
  - B. Contamination
  - C. Allele drop-out / preferential amplification
-

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- D. Apparent mix-up or labeling of sample or embryo
  - E. Other human errors
  - F. Other fill in\_\_\_\_\_
- 

### **Data Collection and Follow-Up**

Q65 For your PGD patients, what do you advise regarding follow-up amnio/CVS:

- A. It is required
- B. It is recommended/suggested
- D. It is recommended against
- E. It is not mentioned

Q66 Does your clinic/laboratory keep data on PGD accuracy?

- A. Yes
- B. No

*Q67, 67b not used in this report*

*Q68-69 left intentionally blank*

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### **Questions for Non-PGD Clinics**

MULTI PUNCH ANSWER

Q70 Why do you not offer PGD?

Check as many as apply

- A. Unfamiliar with it
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- B. We “offer” it but no patient has requested it
  - C. We would like to offer it but do not have the resources (staff, money, expertise, etc) to make it available
  - D. PGD is considered experimental
  - E. We are not convinced embryo biopsy does not harm the embryo
  - F. We are concerned that the misdiagnosis rate is unacceptably high
  - G. Ethical/moral concerns about PGD’s use
  - H. Concerned about liability
  - I. Inadequate market demand
  - J. Other (fill in)\_\_\_\_\_

[If Q8 = Yes, ask Q71 and Q72 for each response list item not selected in Q9]

Response items from Q9:

- A. PGD for an autosomal single gene disorder(s), either recessive or dominant
- B. PGD for chromosomal rearrangements
- C. PGD for X-linked disease
- D. PGD for aneuploidy
- E. PGD for non-medical sex selection, for example, family balancing
- F. HLA typing

**MULTI PUNCH ANSWER**

Q71 Why do you not provide [*RESPONSE ITEM NOT SELECTED IN Q9*]?

Check as many as apply

- A. Unfamiliar with it
  - B. We “offer” it but no patient has requested it (no market demand)
  - C. We would like to offer it but do not have the resources (staff, money, expertise, etc.) to make it available
  - D. PGD is considered experimental for this indication
  - E. We are concerned that the misdiagnosis rate is unacceptably high for this indication
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- F. Ethical/moral concerns about PGD's use
  - G. We are not convinced of PGD's clinical utility for this indication
  - H. Concerned about liability
  - I. Other (fill in)\_\_\_\_\_

SINGLE PUNCH ANSWER

Q72 What is the likelihood you will offer[*RESPONSE ITEM NOT SELECTED IN Q9*] in the future?

- A. Certain we will offer – process is underway
- B. Very likely
- C. Somewhat likely
- D. Unlikely
- E. Certain we will not offer

*Q73 not used in this report*

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**In this next-to-last section, we would like to ask about your personal opinions on the topic.**

*Q74-82 not used in this report*

SINGLE PUNCH ANSWER

Please indicate your agreement or disagreement with the following statements:

- Strongly agree
  - Agree
  - Disagree
  - Strongly disagree
-

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Q83 Professional societies are best suited to create standards and guidelines relating to PGD.

Q84 There should be more professional guidelines relating to PGD.

Q85 There should be more government oversight relating to PGD.

*Q86 left intentionally blank*

*Q87 not used in this report*

*Q88-91 left intentionally blank*

**For the following questions, please answer based on your own views of how the practice of PGD will change in the next 10-15 years:**

In the future...

Strongly agree

Agree

Disagree

Strongly disagree

Q92 more major commercial laboratories will offer PGD genetic testing.

Q93 changes in technology will permit whole genome embryo screening as a routine part of IVF.

*Q94 not used in this report*

Q95 there will be restrictions on using PGD for non-medical genetic traits such as sex.

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*Q96-103 left intentionally blank*

*Q104 not used in this report*

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