

Establishing the demographics and rationale for use of Preimplantation Genetic Screening and Diagnosis in Arizona

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INTRODUCTION

- Over the last 30 years, the growth of In-vitro Fertilization (IVF) pregnancies and techniques has led to the era of preimplantation genetic diagnosis (PGD)
- With PGD came the ability to anticipate implantation and prenatal diagnosis, identifying the genetic predispositions of fertilized eggs prior to transfer
- PGD continues to expand, ensuring the genetic health of the implanted embryo, with the number of successful births at now more than 10,000 (Simpson, 2010), and still rising
- With the growth, questions have been raised over what utilizing preimplantation genetic testing might mean both to the parents, the subsequent fetus, as well as implications on the families and society as a whole (Iwarsson 2011, Zeiler 2007, Knoppers 2006)

DEFINITIONS

American Society for Reproductive Medicine (ASRM) 2008 definitions:

- Preimplantation Genetic Diagnosis (PGD) applies when one or both genetic parents carry a gene mutation or balanced chromosomal rearrangement and testing is performed to determine whether that specific mutation or an unbalanced chromosomal complement has been transmitted to oocyte or embryo
- Preimplantation Genetic Screening (PGS) applies when the genetic parents are known or presumed to be chromosomally normal and their embryos are screened for aneuploidy

STUDY QUESTION

What is the current utilization of Preimplantation Genetic Diagnosis (PGD) and Preimplantation Genetic Screening (PGS) in Arizona Infertility clinics, including the demographics of the patient population and their reasons for pursuing PGD and PGS?

STUDY SIGNIFICANCE

- No report exists on the relationship of PGD implementation to usage, demographics or financial considerations, as previous studies have focused on practices and procedures
- This current study integrates the qualitative surveying of clinicians into the more quantitative structure of a survey tool, allowing for statistical comparison
- Arizona study group:
 - Well-defined set of IVF clinics, reportable as statistically exhaustive
 - Patient population with larger minority percentages than national average, provides insight of their contribution to PGD usage

METHODS

- Survey tool designed with the help of statistician, focusing on
 - Assessment of practice policies
 - Patient population
 - Provider experience
 - Individual IVF clinic's opinion on political and social questions related to use of PGD
- Capture-Recapture method to identify all Clinics providing IVF in Arizona (n=11)
- All clinics contacted by phone to ensure use of their use of PGD and willingness to participate in study
- Clinics responding to survey remained anonymous
- Completed surveys were analyzed, numerical data compared, significance determined

RESULTS

- As a result of the capture recapture method used in this study all IVF clinics currently in operation in the state of Arizona were found, 11 in total
- The overall adjusted response rate, was 82%, 9 distinct clinics of the 11 responding
- Despite the large minority population of Arizona, PGD/PGS requests by these populations were few
- One clinic reported a large proportion of their patient base was Hispanic white and no clinics reported a Native American patient
- Overall, the clinics significantly differed in regard to patient numbers using PGD
- Patient use of PGD and PGS was disproportionately distributed across the clinics responding, a fraction of the total clinics serve as the largest providers of PGD within Arizona
- Well established clinics, as determined by founding date, were not consistently associated with greater PGD/PGS patient use
- The age range for Arizona patients was heavily shifted toward mothers of advanced maternal age. (35+ years old)
- The types of PGD diagnostics:
 - Most clinics employ sex selection (for medical and non-medical reasons) but one clinic explicitly stated avoidance of non-medical sex selection
 - Chromosomal and single gene analysis were reported by a majority of clinics
 - HLA and adult-onset disorders were not reported by any clinics
- Most clinics recognize their dependence on public patient education via the Internet and on primary care to refer their patient base
 - Most wanted to see greater public education of PGD/PGS testing
- Most clinics did not believe additional sources of regulation would improve practice, however, size of patient population correlated with desire for greater regulation

Age groups	% pts in Arizona
18-25 yr old	2.6%
25-30 yr old	2.8%
30-35 yr old	14.5%
35-38 yr old	22.6%
38-40 yr old	20.4%
40+ yr old	36.8%

Table 1: Total Percentages of Arizona PGD/PGS patients

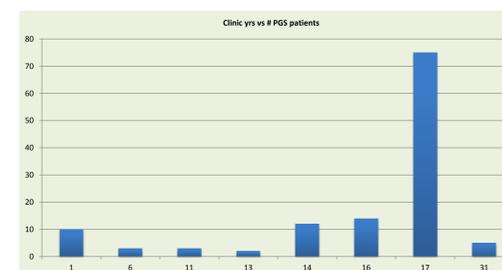


Figure 1: Comparison of years of clinic experience with # of PGS/PGD patients

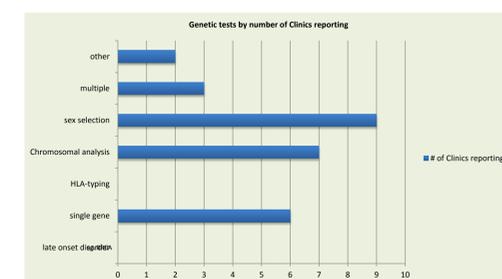


Figure 2: Types of Genetic Tests Used by Clinics

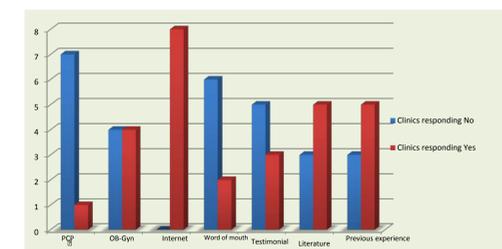


Figure 3: From eight of nine clinics responding, the initial information sources used by the clinics' current PGD/PGS patient base

Preimplantation genetic testing to be more widely advertised?	Yes	No
By Physicians?	6 clinics	3 clinics
Other healthcare providers?	6 clinics	3 clinics
Public media, press, internet?	5 clinics	4 clinics

Table 2: Clinics' suggestions for increased advertisement for PGS

Would IVF clinics benefit from increased regulation of PGD?	Number of clinics responding
Yes-very much	1 clinic responding
No-not at all	4 clinics responding
Minimal benefit or no opinion	4 clinics responding

Table 3: Clinic views on increased regulation for IVF clinics offering genetic testing

DISCUSSION

- The response of clinics largely correlated with current literature and expert opinion regarding regulation and growth of PGD/PGS testing

CONCLUSION

- Overall, the study elucidated the use of PGD and PGS within Arizona, many of the demographic statistics correlated well, considering the intended purpose for preimplantation genetic diagnostics, they demonstrated the patient population base identity and its ties to supply-demand, economic, and information causes
- From the study, the opinions of the clinics are explained more fully in the context that the survey provides for their clinical experiences
 - Such insight is invaluable when approaching any public discussion of changes to referral sources, monetary adjustments, patient education, or governmental regulation

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- This survey highlighted the relationship of the referring physician with the potential PGD/PGS patient, and elucidated the source of the majority of patient referrals, emphasizing that most patients who utilize PGD testing have utilized for previous pregnancies
- By better identifying patients utilizing PGD/PGS, this survey may assist both clinics and the national PGD community in focusing their efforts to promote realistic and understandable information about what PGD offers as well as the limitations of the techniques used
- Thus, even if the primary sources of referral continue to be Internet and the media, a concerted effort to better inform the patients' Internet sources with accurate information would be beneficial
 - This in itself, would provide further internal regulation of the extent of PGD practice and how it is utilized
- Regulation and discussion of sex-selection for non-medical reasons are addressed by each clinic differently
- Evident in the results from these clinics, is the need to let clinic experiences and opinions inform the discussion
 - A larger survey of clinic opinion, asking in what ways sex-selection should be standardized and when, would be helpful and could lead to agreement about standard procedure
 - If this did occur, arrived at outside governmental guidance, the ability to continue internal regulation might be preserved, despite pressure from states to have legislated standards
- Finally, socio-economic limitations to access of PGD/PGS testing were illustrated again in this study, however, patient population mirrored local population in one large clinic
 - The large patient population at this clinic affected overall numbers
 - Yet, obvious economic inhibition remains, and may only change as PGD procedure itself morphs and grows in efficiency, thereby reducing cost