

THE ROLE AND IMPACT OF GENETIC COUNSELING IN FAMILY BUILDING: DISABILITY CONSIDERATIONS

Dominique Alexander, MS, Rutgers Microbiology and Molecular Genetics Program, LEND Genetic Counseling Trainee
Gina Davis, MS, CGC; Claire Shepherd, MS, CGC; Christine Seymour, MS, CGC
A Collaboration Between Advocate Genetics and Rutgers University

BACKGROUND

- Genetic counseling was a profession born as a response to the eugenics movement, aimed at promoting individual reproductive autonomy (32).
- However, tensions between disability communities and genetic counselors continue through debates surrounding carrier screening, gamete donor exclusion criteria, and prenatal diagnosis (18, 23, 33).
- Genetic counseling is recommended when a patient is working with a gamete donor, yet the implementation of this service varies (4-7).
- Moreover, little is known about the perspectives and concerns of gamete donation recipients as they pursue this path to family-building (15, 16).
- The few studies that have explored these topics previously take a retrospective approach and highlight the complexity of recipient values, suggesting recipient risk perception may align or differ from that of established guidelines (15, 16-19).

STUDY QUESTIONS

- What is the perceived utility of genetic counseling for gamete recipients as they actively navigate their family-building journey?
- What are the experiences and attitudes of gamete recipients, and how does their perception of risk compare to the ASRM guidelines for gamete donation?

METHODS

- A prospective pilot survey was given to 11 gamete recipients after their genetic counseling appointment between June 20, 2024, and December 31, 2024.
- A chart review was conducted to abstract non-identifiable demographic and clinical information.
- Descriptive statistics were generated for recipient survey responses, and a thematic analysis was conducted to extract commonalities in recipient open-ended responses.

RESULTS

- The response rate of the survey was 14.5%, with most respondents identifying as white (64%) and female (82%) at an average age of 37.3 years old.
- 90.9% of participants indicated that discussing the donor's family history and genetic risk assessment was useful for their decision making, with all feeling this history would be important for their future child.
- 81.8% of participants shared that the genetic counseling session made more confident in their decision to pursue gamete donation.
- Participants expressed that the most important factors in their decision to work with a gamete donor are the donor's medical and psychological history and the donor's family and genetic history.
- Risk perceptions of donor family history between gamete recipients and genetic counselors as informed by ASRM guidelines demonstrated discordance.
- 90.9% expressed that they planned to proceed with the gamete donor(s) they discussed during their match consultation, with none feeling that a substantial genetic issue had been identified.

Table 1. Purpose and value of genetic counseling services to participants considering gamete donation

Common Theme in Survey Responses	Illustrative Quote
Value of reviewing potential genetic risks revealed by testing results and family history intake in decision-making regarding working with a gamete donor	"We wanted more clarity on the specific genetic disorders that the donors were a carrier for. Genetic counseling helped us understand the level of risk."
Appreciation of opportunity for nuanced discussion regarding donor anonymity and sharing child(ren)'s genetic origins	"I appreciate how the counselors have made us think about the implications of using a donor. How to tell our children, especially early so that we normalize it."
Significance of knowledge and education as recipients navigate their fertility journey and prepare for the future	"[GC] ensures as much as possible is known about the donor to give potential offspring the best chance at a happy and healthy life."
Recognition of importance of genetic counseling, including as a part of standard protocol for gamete recipient donor selection	"I would recommend this to everyone who is in the infertility and surrogacy journey."

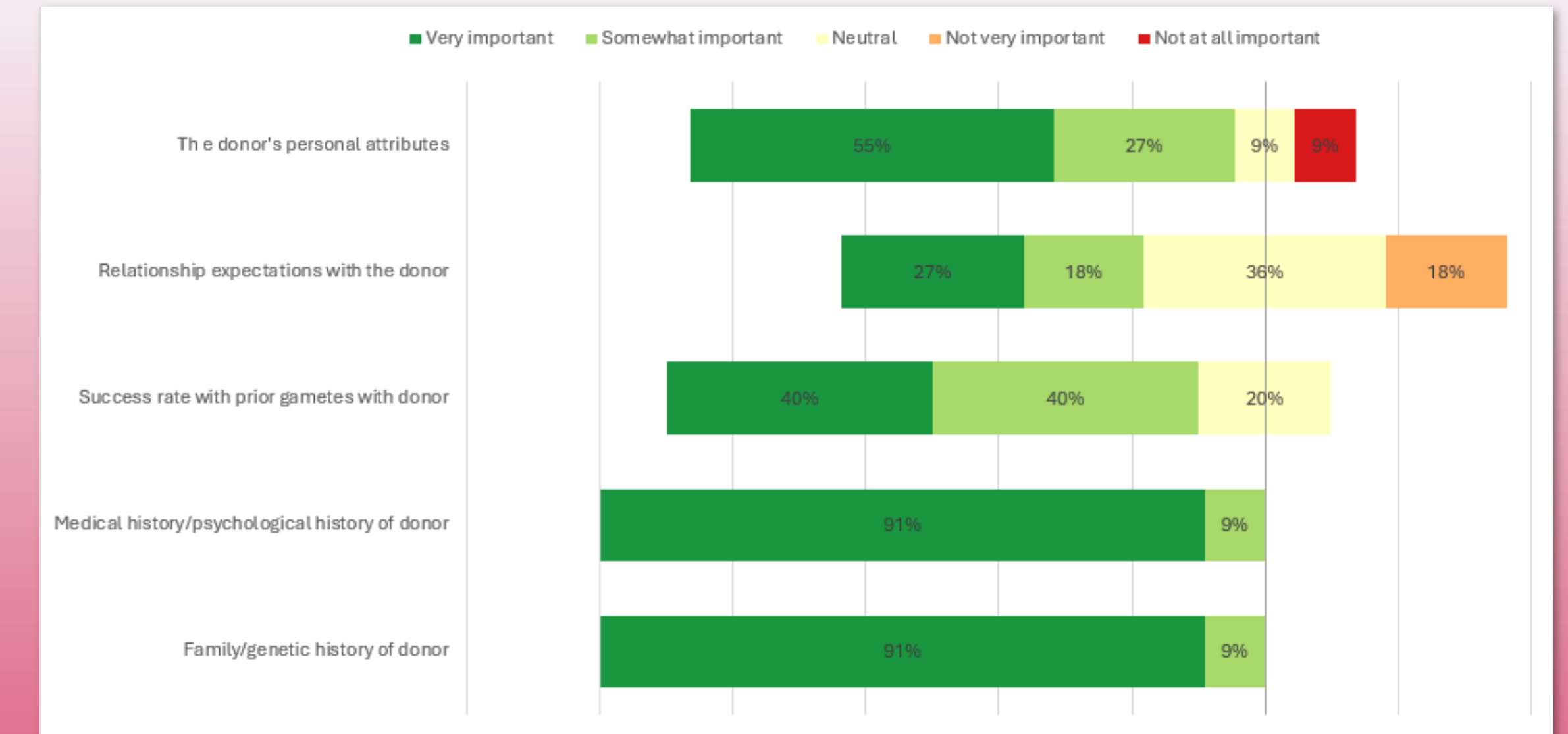


Figure 1. Gamete recipient values when considering gamete donor selection

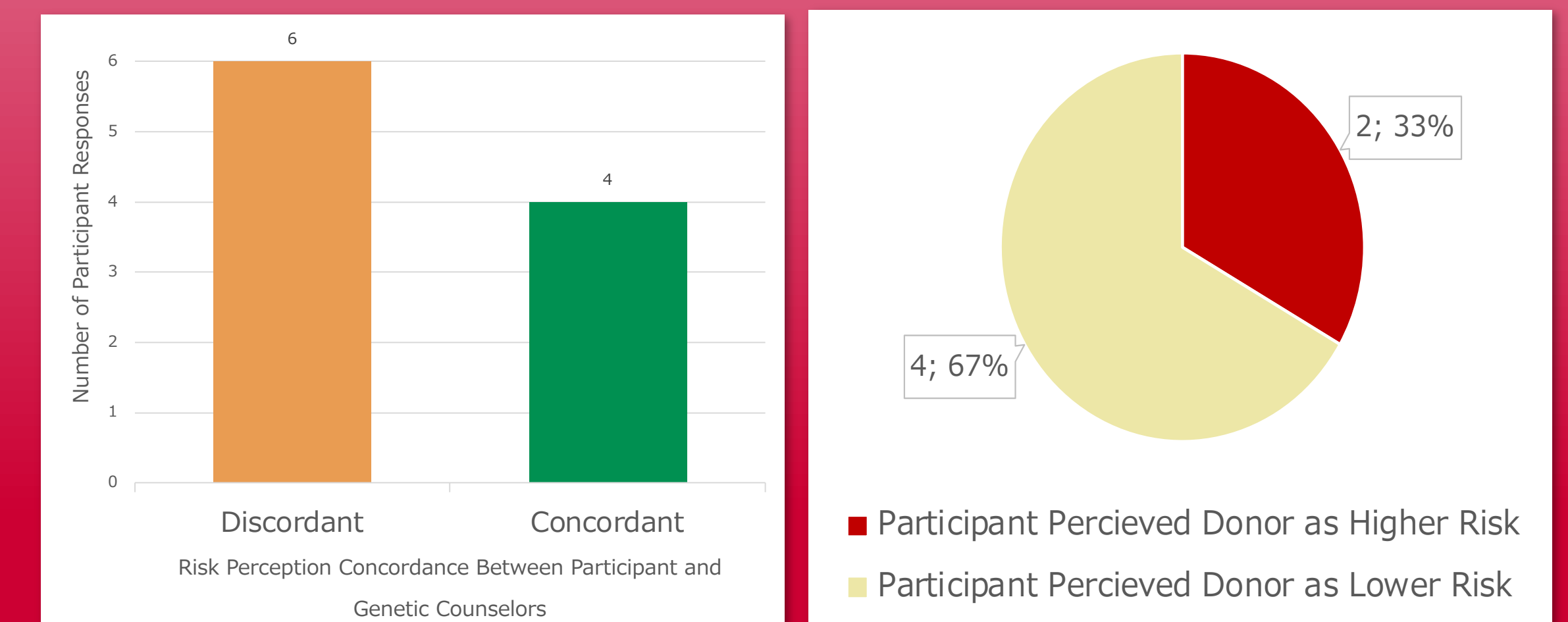


Figure 2. Donor risk assessment concordance between gamete donor recipients and genetic counselors

DISCUSSION

- The results of this pilot study highlight the benefits of genetic counseling as a component of standard fertility practice.
- This study also underscores the complexities of implementing strict exclusion criteria for gamete donation due to differing perceptions of severity.
- Strict exclusion criteria for gamete donation also likely exacerbate existing disparities in the diversity of the gamete donor pool (27-31).
- Exclusion criteria may not be in alignment with ethical principles of genetic counseling such as autonomy and anti-discrimination.

