Genetic evaluation procedures at sperm banks in the United States

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Objective: To assess how genetic evaluations of sperm donor applicants are performed in the United States.
Design: A questionnaire was designed to assess: 1) the professionals involved in the family history evaluation and genetic screening; 2) the genetic testing, counseling, and informed consent processes; and 3) how the results of genetic evaluations and new risk information is communicated to donors.
Setting: Semen donor facilities.
Participant(s): Representatives of semen donor facilities.
Intervention(s): None.
Main Outcome Measure(s): Descriptive data.
Result(s): Thirteen responses were received. All of the facilities assessed donors' family histories; eight of the facilities (62%) routinely informed donors about the results of these evaluations. At the majority of facilities (10/13), informed consent for genetic testing is obtained as part of the overall contract to be a sperm donor. Genetic counselors are employed full-time at two facilities and part-time at five others.
Conclusion(s): There is variability in the education and informed consent processes for semen donor applicants, including variable communication about the limitations of genetic tests and the potential implications for the donors' own children. Further research into the best practices for education and consent for sperm donor applicants may be beneficial to ensure the well-being of the donors and their future offspring. (Fertil Steril® 2013;99: 1587–91. ©2013 by American Society for Reproductive Medicine.)
Key Words: Semen donor, sperm donor, genetic screening, informed consent

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Genetic screening for reproductive purposes is recommended before conception whenever possible, so that patients can benefit from the greatest number of options and time for decision making regarding genetic testing and pregnancy management (1–3). An underlying principle of genetic testing is informed consent (4–6). It is a process through which an individual makes a decision or takes an action based on his or her individual needs and preferences, given a full understanding of the options available and the consequences of each decision. As such, reproductive genetic testing is traditionally performed on the biologic parents of a pregnancy, once they have been informed of appropriate testing recommendations and options and the risks, benefits, and limitations associated with specific tests and had the opportunity to make the decisions that are best for their families.

Genetic testing of reproductive tissue donors differs because the donors provide gametes for offspring that they will not parent. Therefore, the donors' preferences regarding genetic testing may not be the predominant factor determining which evaluations are performed on these individuals. The genetic screening of a donor may, alternatively, be performed based on ethnic background and/or specific indications in his or her family history, tissue donor screening regulations (7, 8), gamete donor screening guidelines (9, 10), general population carrier screening guidelines (11–16), and/or an individual sperm bank's policies. In addition, the recipients of donor gametes are likely to have a greater interest in the genetic evaluations that are performed on the donors than the donors themselves may have. However, it is likely that these evaluations would identify specific genetic risks for some donors, because many reproductive screening

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tests are designed to detect individuals who carry mutations for common genetic disorders that are inherited in an autosomal recessive manner (11–16), and donors are just as likely as any other individual to test positive on these tests. Even though the donors may not have personal preferences regarding the genetic screening performed on them at the time of their participation in a donor program, the results of these evaluations can have significant implications for them and their family members and for their future reproduction. Therefore, it is important that they are informed of these possibilities before testing and educated about the results of their tests.

In the present study, we examine the processes by which genetic evaluations are performed on sperm donors in the United States to determine if the donors receive the same education and opportunities to provide informed consent that are recommended for those individuals who are planning their own pregnancies (4–6). Specifically, we evaluated: 1) the professionals involved in the family history evaluation and genetic screening of donors; 2) the genetic testing, counseling, and informed consent processes for sperm donors; and 3) if and how the results of genetic evaluations and newly acquired risk information is communicated to these men.

MATERIALS AND METHODS

A questionnaire was designed to evaluate the characteristics of the sperm donor programs, including the routine processes for genetic evaluation of sperm donor applicants, the informed consent and genetic counseling practices, and the procedures for managing test results.

Semen banks in the United States were identified by internet and literature searches. Twenty-six facilities were identified, including the employer of one of the authors (P.C.). All facilities were invited to participate in a study regarding genetic screening practices at sperm banks. Each facility was contacted by telephone and asked to provide a contact name and e-mail address for an individual to whom it was most appropriate to send an online questionnaire on the genetic screening practices at that facility.

The questionnaire (Supplemental Fig. 1, available online at www.fertstert.org) was distributed through an online survey tool. Recipients were asked to forward the survey to an appropriate staff member if they were not the individuals who could most accurately address the survey questions. Four weeks after the survey was distributed, the facilities were contacted by telephone to remind them of the opportunity to participate if they had not yet had the chance to do so.

Institutional Review Board (IRB) approval was not obtained, because data on human subjects or private identifiable information was not gathered; the data focused on policies and protocols.

RESULTS

Responses were received from 13 of the 26 facilities (50%). The individuals who responded to the study had a variety of roles at the semen banks, as presented in Table 1, and included both clinical and nonclinical professionals. The level of genetics education achieved by the respondents is presented in Table 2, and ranged from no formal genetic training to advanced degrees in genetics.

TABLE 1

Staff members who responded to the survey.

Staff member	n
Donor coordinator (nonclinical)	3
Lab manager/supervisor	2
Tissue bank director	2
Physician	2
Genetic counselor	1
Nurse practitioner	1
Registered nurse	1
Medical technologist	1

Isley. Genetic evaluation of sperm donors. Fertil Steril 2013.

Both anonymous donor and directed donor services were available at 12 of the 13 facilities (92%); one facility offered only anonymous donor services. All results presented below pertain to anonymous semen donation practices unless stated otherwise.

Donor Applicant Family History Risk Assessment

All facilities collect a three-generation family history from each of their donor applicants. At ten facilities (77%), the family history was collected as part of a consultation with the donor applicant either in person or over the telephone. At three facilities (23%), the family history was collected from the donor but evaluated separately, without the donor present for clarification. The applicants' family histories were reviewed by a variety of professionals, including nurse practitioners, reproductive endocrinologists, medical geneticists, genetic counselors, and medical directors. The level of genetics education achieved by these individuals was unknown; only two of the individuals who completed the questionnaires were the same staff members who performed the family history evaluations.

At eight out of 13 sperm banks (62%) it was routine practice to inform donors of the results of their family history risk assessments. Four facilities provided a consultation to donors only when they thought there was a specific indication to do so, and one facility did not inform donors of the findings from these assessments. These consultations were performed by genetic counselors at four centers and by medical directors at three other facilities. Medical technologists, donor

TABLE 2

Highest level of genetic education completed by respondents.

Extent of genetics training	n
No formal genetics training	4
Single genetics course during medical or graduate training	3
Continuing education in genetics	3
More than one course in genetics	1
Degree in genetics	2

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