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Appendix A: Minimal genetic screening for gamete donors

The American Society for Reproductive Medicine

Birmingham, Alabama

I. The Donor
   A. Should not have any major mendelian disorder. Mendelian disorders fall into the following categories:
      1. Autosomal dominant or X-linked disorders in which age of onset extends beyond the age of the donor, such as Huntington disease.
      2. Autosomal recessive inheritance (homozygous). Donors who are heterozygous need not necessarily be excluded if recipients are not also heterozygous.
   B. Should not have (or have had) any major malformation of complex cause (multifactorial/polygenic), such as spina bifida or heart malformation. A major malformation is defined as one that carries serious functional or cosmetic handicap. However, the definition of "major" is a matter of judgment.
   C. Should not have any significant familial disease with a major genetic component. This applies especially to first-degree relatives (parents, siblings, or offspring).
   D. Should not carry a known karyotypic abnormality that may result in chromosomally unbalanced gametes. Among healthy young adults, the chance of having a chromosomal rearrangement that could be transmitted in unbalanced form to offspring is small. For this reason, routine karyotypic testing of all donors is optional.
   E. A member of a high-risk group (see Table A1) should be tested routinely to determine heterozygosity for certain conditions. The list of tests may change as new tests for other disorders are developed. Heterozygosity need not necessarily exclude a donor, but certain donors may be inappropriate in a given case.
   F. New screening guidelines for cystic fibrosis in the general population have been developed recently by the American College of Obstetricians and Gynecologists and other organizations, and are applicable for gamete donors. Functionally, all gamete donors should be evaluated by the current tests recommended at the time of the donation.
   G. Donors should be generally healthy and young. Males 40 years and older are at increased risk for new mutations. Women 35 years and older are at increased risk for producing offspring with aneuploidy.

II. The donor's first-degree relatives (parents, siblings, or offspring) should be free of:
   A. Mendelian disorders as described in Section I.A.
   B. Major malformations as described in Section I.B.
   C. A chromosomal abnormality, unless the donor has a normal karyotype.
   D. If family history reveals a disorder for which definitive testing is available, and it is important to consider that candidate further as a donor, then it is appropriate to test for that specific disorder. Results will determine appropriateness of donor.

Guidelines
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TABLE A1

Genetic screening in various ethnic groups.

Ethnic Group	Disorder	Screening test
Ashkenazi Jews	Tay-Sachs disease	Decreased serum hexosaminidase-A or molecular analysis
	Canavan disease	DNA analysis to detect most common alleles
African American	Sickle cell anemia	Presence of sickle cell hemoglobin, confirmatory hemoglobin electrophoresis
Mediterranean people	β-Thalassemia	Mean corpuscular volume (MCV) $<$ 80%, followed by hemoglobin electrophoresis
Southeast Asians and Chinese (Vietnamese, Laotian, Cambodian, Filipino)	α-Thalassemia	MCV $<$ 80%, followed by hemoglobin electrophoresis
All ethnic groups In Caucasians of European descent and Ashkenazi Jews should be offered; in other ethnic groups (Asians, Hispanics, African-Americans) should be made available	Cystic fibrosis	DNA analysis of specified panel of 25 CFTR mutations (those present in $\geq$ 0.1% of the general U.S. population). Larger panel not recommended for screening.

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