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Letter

Screening sperm donors for cystic fibrosis

Ian Findlay, **Howard Cuckle**, **Richard J Lilford**, **Anthony J Rutherford**, **Philip Quirke**, **Steven Lui**

Author Affiliations

EDITOR,—Cystic fibrosis is the most common serious autosomal recessive condition in white populations, affecting about 1 in 2500 live births, and until recently life expectancy rarely exceeded 30 years. The most common cystic fibrosis mutation (δF508, accounting for about 80% of two million British carriers), is a 3-bp deletion in a transmembrane protein cystic fibrosis transmembrane regulator gene. The next most common three or four mutations account for a further 5% of carriers.

One in 25 white people carries cystic fibrosis. As carriers are unaffected, individuals are often unaware until they have an affected child.

Sperm donors are currently questioned for a famly history of genetic and other disorders (including cystic fibrosis) and tested for a variety of diseases. However, since few carriers have a family history of cystic fibrosis [1] and as donors are currently not screened for cystic fibrosis, there is a potential problem.

If a donor is a cystic fibrosis carrier there is a high risk of this leading to at least one offspring being affected with cystic fibrosis and several being carriers—as a maximum of 10 can be fathered by each donor, the risk is 1 in 3 that a child will be affected (probability=1-(24/25)^{10}). The risk of an individual pregnancy being affected is increased 25-fold (from 1 in 2500 to 1 in 100). If sperm were screened for the most common cystic fibrosis mutations and only proved non-carriers used, the risk would be reduced sixfold (to 1 in 14000).

To determine if cystic fibrosis screening for donors is effective, we retrospectively tested sperm samples from our donor sperm programme. Since δF508 accounts for 85% of carriers in the Yorkshire region, we screened only for this mutation. Although screening is normally performed on blood or mouthwash samples, [2] we tested the sperm samples themselves. To avoid the remote possibility that sperm preparation would preferentially select unaffected sperm, samples were treated in a similar manner (using Percoll gradients) to those used in in vitro fertilisation procedures. The DNA amplification methods were similar to those used in sexing and cystic fibrosis diagnosis of single cells using fluorescent polymerase chain reaction. [3]

The sperm from 22 prospective and current donors were tested. Two donors were found to carry cystic fibrosis δF508. Both were prospective donors and were removed from stock.

Screening for cystic fibrosis in sperm donors is currently inadequate; we recommend that donors be routinely tested for cystic fibrosis. This testing is both effective and inexpensive (under pounds sterling25 for δF508 or about pounds sterling50 for the commonest four mutations). One other consideration is possible medicolegal implications—that is, the exposure of parents to an unnecessary high risk of having a child affected with cystic fibrosis.

References

1. Holloway S, Brock DJH. Cascade testing for the identification of carriers of cystic fibrosis. *Journal of Medical Screening* 1994;1:159–64. [Medline]
2. Lewis FA, Cross P, Sehmi I, Cuckle H, Quirke P. Population screening for the cystic fibrosis gene using fluorescent PCR. *J Pathol* 1993;170: 34A.
3. Findlay I, Ray P, Quirke P, Rutherford AJ, Lilford R. Allelic dropout and preferential amplification in single cells and human blastomeres: implications for preimplantation diagnosis of sex and cystic fibrosis. *Human Reproduction* (in press).

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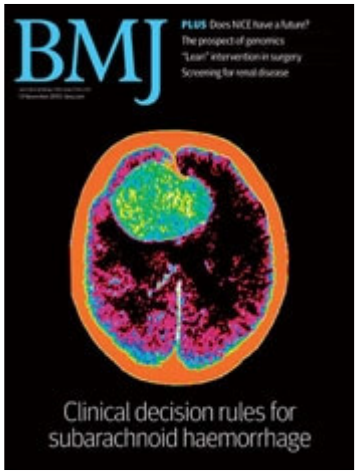
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