



University of HUDDERSFIELD

University of Huddersfield Repository

Findlay, I., Cuckle, Howard, Lilford, Richard J., Rutherford, Anthony J., Quirke, Philip and Lui, Steve

Screening sperm donors for cystic fibrosis

Original Citation

Findlay, I., Cuckle, Howard, Lilford, Richard J., Rutherford, Anthony J., Quirke, Philip and Lui, Steve (1995) Screening sperm donors for cystic fibrosis. *British medical journal*, 310 (6993). p. 1533. ISSN 0959-8138

This version is available at <http://eprints.hud.ac.uk/5514/>

The University Repository is a digital collection of the research output of the University, available on Open Access. Copyright and Moral Rights for the items on this site are retained by the individual author and/or other copyright owners. Users may access full items free of charge; copies of full text items generally can be reproduced, displayed or performed and given to third parties in any format or medium for personal research or study, educational or not-for-profit purposes without prior permission or charge, provided:

- The authors, title and full bibliographic details is credited in any copy;
- A hyperlink and/or URL is included for the original metadata page; and
- The content is not changed in any way.

For more information, including our policy and submission procedure, please contact the Repository Team at: E.mailbox@hud.ac.uk.

<http://eprints.hud.ac.uk/>

[Home](#) > [Volume 310, Number 6993](#) > [BMJ 1995;310:1533](#) (Published 10 June 1995)

BMJ 1995;310:1533 (Published 10 June 1995)

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 ($\delta F508$, 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 family 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 fibrosis¹ 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 $\delta F508$ 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 $\delta F508$. 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 sterling 25 for $\delta F508$ or about pounds sterling 50 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).

This article has been cited by other articles:

Dornase alfa for cystic fibrosis

BMJ 1995;311:813

[\[Full text\]](#)

What's new

Last 7 days

Past weeks

Current print issue

Rapid responses

Blogs

Podcasts

- [Richard Smith: How to turnaround a failing hospital](#) (11 Nov 2010)
- [Martin McShane: Climate change](#) (11 Nov 2010)
- [Julian Sheather: Is happiness a mental disorder?](#) (11 Nov 2010)
- [Emily Arthurs: Five year survival](#) (10 Nov 2010)
- [Daniel Palazuelos: Grassroots fertiliser](#) (9 Nov 2010)

Keep updated

Get email alerts



Get RSS alerts



[Latest from BMJ](#)

[BMJ simple search](#)

[Follow BMJ on twitter](#)

[Watch on YouTube](#)

Services

- [Submit an article](#)
- [Subscribe/Activate](#)
- [Request permissions](#)

Tools

[Print this page](#)

BMJ Group portals

- [Diabetes](#)
- [Oncology](#)
- [Junior doctors](#)
- [Clinical trials](#)

Resources

- [Readers](#)
- [Authors](#)
- [Reviewers](#)
- [Media](#)
- [BMA members](#)
- [Advertising and sponsors](#)
- [Subscribers](#)

Print issues

- [Current issue contents](#)
- [Current cover image](#)
- [Past issues](#)
- [Subscribe](#)



