

Health implications and counselling for paternity testing

MJ Kotze¹, CL Scholtz¹ and P Opperman²
Genecare Molecular Genetics (Pty) Ltd.,¹ Cape Town and² Pretoria

Correspondence to: Dr Maritha Kotze, PO Box 15743, Vlaeberg, 8018
Tel: 021 424 6504, Fax: 021 422 5539, E-mail: mjk@genecare.co.za

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Introduction

Identification and parentage testing using DNA technology has become an important tool in both the legal and medical fields. It is used by the legal fraternity to resolve will and maintenance disputes, criminal investigations and immigration decisions. Identity testing in the clinical field involves twin studies, maternity determination after hospital mix-ups, as well as gamete and embryo identification after possible laboratory errors.¹ When two baby boys were inadvertently exchanged at birth by hospital staff in a provincial hospital in South Africa in 2004, DNA testing of the mothers and babies were essential to make the correct matches.

In a study by researchers in Liverpool, performed in men and women wanting proof of paternity from testing as well as studies based on genetic health screening, it was found that rates of cases where a man was not the biological father of his child was on average one in 25, ranging from 1% in some studies to as much as 30%.² Increasing paternity testing and use of DNA techniques in clinical and judicial procedures implies that paternal discrepancy will be identified increasingly.

Paternity test

Paternity testing is based on the fact that a child inherits half of his/her DNA from the mother and half from the father.³ The fundamental basis of paternity exclusion is that if there are a sufficient number of paternal "obligate" alleles (more than 2) absent from the DNA profile of the alleged father, this would provide evidence against paternity.

Currently, the most popular DNA technology used in parentage testing concerns short tandem repeats (STRs) which consist of short, repetitive DNA elements 3 to 7 base pairs in length. These highly polymorphic repeats are widely distributed throughout the human genome and are easily detected using polymerase chain reaction (PCR) technology. After amplification, the alleles are differentiated by the number of copies of the repeat sequence in the amplified region and are distinguished from one another using fluorescence detection (Promega Technical Manual, D012). Multiplexes of 15 loci are sufficient to distinguish significantly between related and unrelated individuals.¹

Procedure

1. Cheek swab sample collection and completion of test request form.
2. Seal samples and request form in envelope and sign off.
3. Samples securely transported to laboratory.
4. Delivery and information documentation

at the laboratory.

5. DNA isolation and profiling using state-of-the-art technology.
6. Reliable test results reported confidentially in a supportive environment.

Accuracy of DNA testing

Although numerous techniques have previously been employed to establish parentage or kinship the use of DNA testing has become the benchmark since it is highly accurate (99.9%) and has improved significantly over the past 20 years.⁴ The genetic results are evaluated by using standard statistical parameters to compare the likelihood that the alleged father is the biological father of the child with the likelihood that the alleged father is not the biological father of the child.

Although the genetic material of both parents and the child are usually obtained for paternity testing, paternity can also be excluded with great accuracy when only the alleged father and child are tested. Exclusion is then based on the finding that DNA markers present in the father's genetic profile cannot be found in the profile of the child. However, since false inclusion is possible when the mother's sample is not tested, analysis of all parties involved are preferred.

Interpretation of results

In cases where the "obligate" paternal allele differs at only one or two loci, caution should be used in interpreting the results since a mutational event could have occurred. Mutational events can represent false evidence that two people are not related. It is well known that STR mutation rates differ in men and in women and that most (>90%) are insertions or deletions of one tetrameric repeat sequence (rarely two or three). It is therefore important to incorporate mutation rates into the calculation of paternity ratios to negate false evidence of exclusion.

A common problem faced by laboratories is the fact that personal information that may affect the interpretation of the results is not brought to their attention. For example, if the mother had sexual relations with the brother of the alleged father and this information is revealed, statistical methodologies can be adjusted to take into consideration the fact that the alleged father is related to the biological father. Brothers share 50% of their genetic material and have the same Y-chromosome.

Legal implications

Establishing paternity may be easy enough with one quick genetic test, but issues around the number and relationship of the individuals being tested is not so simple. When paternity testing is requested without the knowledge or consent of both parents, the person

requesting the test must be a legal custodian of the child. In other words the couple should still be married or custody provided to the father in a court case. For the results to stand up in court, the sample collection should preferably be done in the presence of a commissioner of oath and an affidavit provided which states that the correct procedure has been followed for sample collection and analysis of the samples. This approach is important as deliberate mislabelling of a specimen is possible; for instance in an attempt to avoid the obligations of paternity.

Paternity testing made easy

- Painless sample collection procedure using cheek swabs
- Application of internationally approved DNA profiling method
- Accurate calculation of paternity index to confirm or exclude paternity
- Confidential test results reported within 7 days
- Post-test counselling provided in a supportive environment

Conclusions

The cheek sample collection procedure is quick and painless and the paternity test kits containing the swabs and test request forms can be ordered online (www.genecare.co.za). Sample collection can be conducted at a clinic, the doctor's consulting room, or in the comfort of the client's home, provided that at least two witnesses are present and sign the test request form to verify that the origin and labelling of the specimens are correct. However, if the results are required to resolve legal disputes, the samples have to be taken in the presence of a commissioner of oath. DNA testing provides an accurate answer but no-one should have to deal with the results without any support. In our experience, explaining the results and how the test was performed in simple terms, can substantially release the stress experienced by those who have to cope with the outcome of a paternity test.

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