An old problem in a new age: Revisiting the clinical dilemma of misattributed paternity☆

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A B S T R A C T

Clinical genetics has wrestled with the problem of misattributed paternity for decades. While there are no clear directives on policy, surveys suggest that genetics professionals are inclined to avoid disclosure when possible. Changes associated with the increased use of genomic testing will alter the context and may limit the benefits of non-disclosure. Multi-site testing will preclude the uncertainty often associated with single-gene testing. Increased use of genetic testing in clinical and non-clinical settings will create new opportunities for the subsequent unmasking of misattributed relationships, as will the presence of test results in the electronic medical record. Family health history information will become more valuable as it is used more often and to better effect in risk assessment, diagnosis, treatment and reproductive decision-making. These changes associated with genomic testing increase the risks and decrease the benefits associated with the nondisclosure of misattributed paternity. For ethical and practical reasons, genetics professionals, and those who advise them, should consider a greater emphasis on the value of carefully planned disclosure.

1. Introduction

There is nothing new about the problem of misattributed paternity; it is biblically old. The unmasking of it via genetic testing produces a clinical dilemma that pits the value of truthfulness and our duty to inform against our profound disinclination to disrupt relationships within a family. While this conflict remains, its context is changing rapidly due to developments on at least three fronts. First, multi-site and genomic tests identify misattributed relationships with greater certainty than the single-gene tests that predate them. Second, a steady increase in the use of genetic testing in and out of clinical settings makes it more likely that misattributed relationships will be uncovered, and more likely that they will be uncovered subsequently if not revealed at the time of testing. Third, improvements in our ability to use family health history information in risk assessment, diagnosis, treatment and reproductive decision-making raise the stakes on nondisclosure. All of these changes force us to take a new look at an old problem as we transition into the post-genomic era.

2. The status quo: a de facto policy of non-disclosure

The discovery of misattributed biological relationships as a result of clinical genetic testing can take several forms. For example, single-nucleotide polymorphism (SNP) arrays can reveal parental consanguinity, which may be unknown to members of a family (Helm et al., 2014). Revelations about misattributed maternity may also increase in frequency as more couples opt to conceive using donor eggs or embryos (CDC, 2012). However, the discovery of misattributed paternity—in which the presumed biological father of a patient is discovered not to be the father—is still the most common scenario, with rates of occurrence estimated to be between 0.8 to 30% (Bellis et al., 2005). This paper discusses misattributed paternity specifically; similar arguments would apply to alternative scenarios such as unanticipated consanguinity or misattributed maternity, as has been documented in a case involving an IVF error.

There is no formal consensus on how to handle the discovery of misattributed paternity. The few guidelines that exist are contradictory. In 1983, the President’s Commission for the Study of Ethical Problems in Biomedical and Behavioral Research recommended that misattributed paternity be disclosed to both partners; eleven years later, an Institute of Medicine (IOM) committee suggested that: “on balance… information on misattributed paternity be communicated to the mother, but not be volunteered to the woman’s partner.” (Commission et al., 1983). A recent opinion paper from the American Society of Human Genetics (ASHG) on the ethical, legal and psychosocial implications of genetic testing in children and adolescents gave a nod to truthfulness but came down on the side of nondisclosure: “While honoring their broad responsibility to be truthful with patients and their families, we recommend that health-care providers avoid...
disclosure of misattributed parentage unless there is a clear medical benefit that outweighs the potential harms.” (Botkin et al., 2015).

From available surveys, it would appear that most genetic professionals agree with the IOM. In a survey of genetic counselors in 1989, over 95% said they would not disclose misattributed paternity to the father (Pencarinha et al., 1992). A survey of medical geneticists the following year by Wertz et al. found an equally resounding 96% would not tell the father when recessive disease testing indicated misattributed paternity, most of them adding that they would opt to tell the mother in private (Wertz and Fletcher, 1991; Wright et al., 2002). Case reports suggest that decisions are made on a case-by-case basis, taking into account pertinent specific details like the state of existing relationships within the family and potential medical ramifications of nondisclosure (Soderdahl et al., 2004). However, these studies provide strong evidence that absent compelling reason to do otherwise, clinicians in practice default to not telling the father as the safer alternative.

Evidence is less conclusive regarding patient preferences and expectations. The handful of studies that exist suggest that patients are more likely than doctors to say that a doctor should disclose if a man inquires about paternity. Lyn Turney, in a 2005 article in Qualitative Health Research, reported that Australians showed a “higher-than-average” level of comfort with disclosure of misattributed paternity to the presumptive biological father (Turney, 2005).

Modern technology has revitalized interest in this age-old question. In a recent article for the Hastings Center Report, Amulya Mandava and colleagues note that the growing number of genomic research studies necessitates an ethics framework to determine if researchers ever have a duty to disclose the incidental discovery of misattributed parentage to study participants (Mandava et al., 2015). Using a framework they propose, the authors conclude that there is typically no such duty, because a researcher’s role-specific commitment to avoid the hypothetical harms of disclosure is stronger than any duties they have to realize its potential benefits. In other words, providing a research participant with accurate information about a diagnosis or reproductive risk may or may not be valuable, but it is not a researcher obligation.

In the medical realm, where a clinician’s duties of beneficence and honesty to patients are considerably stronger, recent reflection on the disclosure of misattributed relationships has amounted to a doubling down on the status quo. In a 2014 article in Pediatrics, Marissa Palmor and Autumn Fiester acknowledge the limitations of informed consent in the pediatric setting, when parents’ attention “is not fully on the ramifications of non-parentage but on the health of their child”, and contend that it is unlikely to provide a thoughtful and trustworthy measure of parents’ interest in paternity testing. Instead, the authors propose, all informed consent forms for genetic testing of minors should contain boilerplate language indicating an ironclad policy of nondisclosure: “we advocate the incorporation of a new clause into the informed consent forms for pediatric genetic testing that clearly states that any incidental information about parentage will not be revealed, regardless of the result (Palmor and Fiester, 2014)”

3. New contextual considerations in favor of disclosure

Palmor and Fiester’s argument in favor of nondisclosure as a policy is based on the assumption that the potential harms of disclosure outweigh the potential benefits. They argue that increasing use of genetic testing will lead to more cases of misattributed paternity, and overwhelm our current case-by-case model. “The incidental discovery of nonparentage either burdens individual providers with the agonizing and near-impossible task of weighing the pros and cons of disclosure in the particular case or prompts the providers to call a consult with the institution’s ethics service so that the ethics committee can engage in an assessment of those risks and benefits. Case-by-case decision-making...is not a satisfactory solution to this ever-increasing clinical occurrence because it undermines consistency, transparency and uniformity across the institution or practice.”

Although this blanket stance may seem like an attractive option in that it simplifies a complex situation, new circumstances enabled by modern technology warrant a careful reconsideration. Consider the classic scenario Turney presented to her focus groups and survey participants: a mother, a father and a child with recessive disease, where tests results showed one deleterious allele in the child that matched the mother and a second deleterious allele in the child that did not match the father. Among the challenges that have been associated with such cases is the vanishingly rare but real possibility of a new mutation. It is not a good thing to tell people they are mistaken about the paternity of their child; it is an even worse thing to make that suggestion and be wrong. Presumably, the possibility of a new mutation, however rare, gave clinicians another reason not to reveal misattributed paternity.

Modern tests that interrogate multiple sites throughout the genome, or the genome as a whole, offer no such room for temporizing. Many new tests, including cell-free fetal DNA (cfDNA) testing and parental follow-up testing to determine the clinical significance of a variant of uncertain significance identified in a child regularly require maternal and paternal DNA for comparative purposes and can reveal much more information about the degree of genetic relationship between two individuals. Clinical use of whole exome sequencing (WES) is also enhanced by the use of maternal and paternal DNA. In a UCLA report on their initial experience with WES, diagnostic yield was 22% without parental DNA, and 31% for trios (mother, father and child) (Levenson, 2015). Indications long known to produce findings of misattributed paternity, such as recessive disease carrier testing and haplotype testing for organ donation, are all increasing in frequency. We are entering an era of both greater certainty in and greater opportunity for the discovery of misattributed paternity as the number and type of tests available and the use of those tests continue to rise year over year.

At the same time, a universal policy of nondisclosure presumes that disclosure is reliably in the hands of the clinician. It is not. By federal law, patients (or their parents and guardians) have a right to all test results. It is not safe to assume that patients or family members will not be able to deduce the possibility or even the certainty of misattributed biological relationships on their own. If it does not happen at the time of testing, it may happen down the road. Strategies for nondisclosure often rely heavily on limited genetic literacy. In Wertz’s 1990 study, 20% of geneticists said that they would “fudge the issue,” and 13% said that they would tell the couple that they were “both genetically responsible.” Lies, evasions and artful, tactful lack of communication may or may not succeed in keeping the secret at the time of testing. New information, new suspicions, more time to think, access to the internet, and possibly even a much hoped-for increase in genetic literacy could all undermine the effectiveness of that approach over time.

The idea of universal nondisclosure assumes a passivity on the part of patients that is backward-looking, and not appropriate to an age where many people are actively engaged in their healthcare decision-making and have access to all the resources of the internet. If an individual realizes what the test indicates despite nondisclosure, the downside to nondisclosure is not only a potential loss of trust, but also a loss of any discussion, counseling or support the clinician might have been able to offer.

Genetic testing is no longer confined to the clinical realm. As Dena Davis points out in her 2014 article, “The Changing Face of Misidentified Paternity,” the use of direct-to-consumer paternity tests has risen steeply worldwide, as cost has decreased and availability widened (Davis, 2007). Recreational testing can reveal misattributed paternity incidentally as well as by design. In the U.S. alone, millions of people have used genetic testing from companies such as Ancestry.com and 23andMe that provide information on genealogy and ethnicity. ‘Relative-finder’ services that indicate degree of kinship can expose family secrets, as can SNP data that a variety of companies make available to their clientele.
Direct comparisons between paternal and child DNA can indicate misattributed biological relationships, and so can unexpected results among other members of the family, such as individuals raised as siblings who turn out to be biological half siblings, or presumptive cousins who are biologically unrelated. Anecdotal reports have surfaced with the rising popularity of genealogy testing. Attention was drawn to the topic in 2014 by a Vox media article entitled, “With genetic testing, I gave my parents the gift of divorce (Vox Media, 2014).” Private reports complement this very public example. In 2013, a geneticist in Long Island got a strange question from a patient interested in confirming his own interpretation of a report he had received from 23andMe. The company had identified a woman as his granddaughter, based on a 25% match in variable sites. Problematically, he was in his early thirties, and the woman was only one year his junior. Since his mother had done 23andMe testing as well, he was able to determine that all the sites where they matched were paternal in origin. The young woman, enthusiastic at first about exploring the connection, had ceased to return his emails. “Could this individual be his half sister?” the patient asked (Personal communication, 2014).

Today, in the event of nondisclosure, it is reasonable to anticipate that either recreational testing or future medical testing could reveal a misattributed biological relationship at a later date, and might lead to some Monday-morning quarterbacking about the justifiability of a clinician’s decision not to disclose. Now and in the future, information about misattributed relationships will not remain buried in the clinician’s own files as it would have years ago. Rather, it will be enshrined in the electronic health record (EHR). Most physicians now use the EHR (Furukawa et al., 2014), with government penalties in the form of docked reimbursements for those who do not; studies show a five-fold increase in the use of electronic health records in hospitals from 2008 to 2014 (Office of the National Coordinator for Health Information Technology). The information is therefore permanently available for anyone accessing the EHR, and it cannot be flagged or marked to warn future medical caregivers of what has not been communicated. A decision not to disclose information about a misattributed biological relationship today rests uneasily on the assumption that discovery will not take place at a later date, and essentially places a time bomb in the medical record.

A greater risk of subsequent discovery provides several arguments for disclosure. The first is that it raises a potential new sort of liability for genetic professionals. It is easy to imagine that some men, perhaps most men, finding out that they are not the biological father of their child and deducing that this information had been withheld, would be angry. The fact that their anger is both foreseeable and understandable may constitute in itself an argument that nondisclosure is unethical. At a minimum, it compromises the perception of medical professionals as truthful and neutral providers of information. Finally, the risk for later discovery diminishes the value of nondisclosure, since the consequences for the family are more likely to be delayed than avoided.

Most significantly, nondisclosure blinds families to information about biological parentage that has medical significance. An affirmative promise as suggested by Fiester and Palmor that information relevant to parentage will never be revealed sets up a conflict in those cases where clear medical ramifications would tip the balance in favor of disclosure. There are many clinical situations in which disclosure could be mandated on medical grounds. It may be in the best interests of the child to find and test his or her biological father. It may be that either one or both parents are using these test results to guide decisions about whether or not to have more children, or what testing is required for a pregnancy. It may be that other family members are undergoing expensive or potentially hazardous risk assessments that are not necessary, and/or that biological relatives are not getting surveillance they should be getting. Clinicians may attempt to engineer the correct outcome without giving enough explanation to reveal a misattributed biological relationship, but misdirection and lack of transparency are hardly bedrocks on which to base policy for clinical practice.

The full medical implications can go far beyond the immediate ramifications. Discussions of misattributed paternity consistently dismiss the medical value of information on parentage (Christenhusz et al., 2013; Jackson et al., 2012), but this is contrary to the direction in which medicine has been moving, and contrary to the most recent thinking in comparable areas such as adoption and gamete donation, where there is increasing emphasis on the need for accurate family health history information. Many countries, including Sweden, Norway and the United Kingdom, have restricted or forbidden the use of anonymous gamete donation because of what they recognize as every child’s ‘right to know’ his or her genetic heritage (Ravelingien and Pennings, 2013).

The expanded use of genetic testing across multiple medical disciplines increases the already-important role that family health history information plays in risk assessment, diagnosis, prognosis and reproductive decision-making. In specialties as diverse as cardiology, cancer and ophthalmology, family history is used to determine who gets testing and to qualify and interpret the results. Gaps in family health history are a subject of concern for adoptees. Molecular genetic testing has been shown to be of use for adoptees to fill in blanks, but with the understanding that genetic analysis is “unlikely to provide a replacement for family history when available (May et al., 2015).”

Misattributed paternity is far more problematic in that there it not only leaves a gap in family health history information on the paternal side, but fills that gap with misinformation. Inaccurate family health history can be falsely alarming or falsely reassuring, and it can affect the child and other family members. Clinicians making the argument that there is no compelling reason to reveal misattributed paternity imply that a lifetime of living with and imparting inaccurate family health history will not impact medical care. This is an uncomfortable position for genetics professionals. It should give them pause.

4. Arguments against disclosure: the need for evidence

The most compelling reason not to reveal information about misattributed relationships is to avoid the potential harms that may result from disclosure. Undeniably, revelations about misattributed paternity will create short-term upheavals. What the long-term effects may be are more difficult to parse. It is fair to speculate that disclosure may harm the child by harming the family unit; it is also fair to speculate that disclosure might have other downstream consequences, such as allowing the child a relationship with their biological parent. The point is, we don’t know how this information will affect the child’s life, only that it will affect the child’s life. In a pediatric setting, the primary objective is the best interest of the child. A common and credible argument for nondisclosure in the case of misattributed paternity is that it is necessary to preserve the family unit and the child’s relationship with the father (assuming such a relationship exists). This argument makes a couple of assumptions, neither of which is completely reliable. One, it assumes that disclosure would destroy the family, and that non-disclosure would preserve it. Two, it assumes that this family and this relationship, as constituted, represent the child’s best chance at a happy home life.

Another frequently cited reason to refrain from disclosure is to respect patient confidentiality. In the Wertz survey, a majority of medical geneticists agreed with the statement that disclosure would ‘violate the mother’s expectation of confidentiality’. The misattributed paternity scenario, however, is not what we typically think of when we discuss doctor-patient confidentiality. In fact, if the medical caregivers are struggling to decide who to tell, it implies that either the mother knew and chose not to confide in her medical team or that she did not know, in which case she could not confide. Either way, she has not confided anything and therefore cannot have an expectation of confidentiality in the traditional sense. Nor are they revealing her test results without permission, since it is the father and the child’s test results that are at issue. Using the word ‘confidentiality’ is laying claim to
something we acknowledge as a right, and one that can be waived only in extraordinary circumstances. But both the mother and the father have rights in this scenario, and there is no obvious reason to prioritize her right to keep a secret over his right to know.

The decision to err on the side of protecting the status quo is obviously more comfortable for many medical professionals, and may be the easier option, but it has a cost for the family as well. Current policy is based on assumptions and could benefit from empirical research on the aftereffects of disclosure in a variety of settings. Not only would such data reveal what is morally at stake for patients and their families as a consequence of disclosure, it would furnish clinicians with an evidence base to rely on when judging the likelihood that disclosure will result in harms that outweigh its benefits.

5. The necessity of a nuanced approach

The transition into the genomic era brings together a number of trends: better tests, more tests, and more of a role for genetic information (including family health history information) in optimizing medical care. All of these changes argue against a default assumption that clinicians should not disclose a misattributed biological relationship if they have a choice. But the changes, while profound, are not always decisive. Additionally, we are aware that to a large extent the arguments we make here are grounded in culturally specific legal and social norms, and may not adequately reflect the extent to which a revelation of misattributed paternity poses a risk to mother and child in other settings.

A single, universal policy for handling the discovery of misattributed biological relationships is an alluring ideal, providing a quick, clear answer to a thorny and complex problem. Unfortunately, the weight of specific details, such as immediate medical consequences of nondisclosure or the potential for serious and imminent physical harm, requires the exercise of clinical judgment informed by evidence, and is likely to resist any attempt to preempt our current case-by-case model of decision-making. No dilemma worth its salt can be solved by a single line in a standard informed consent document. The discovery of misattributed biological relationships is a clinical challenge, and it will continue to be a clinical challenge.

The likelihood of later discovery and the increasing use of family health history information in clinical care suggest that the clinician’s impulse to default in favor of nondisclosure is a less viable strategy in the genomic age. Though it is well intended, medicine is likely to move脉 to default in favor of nondisclosure is a less viable strategy in the

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