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# Predictive testing for pre-malignancy as a prelude to adoption? An English case

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**Abstract** Genetic testing as a preliminary to adoption has been discussed in the literature [BAAF/BSHG Statement on the use of DNA testing to determine racial background], together with the medical benefits derived from such testing ASHG/ACMG Statement [Am J Hum Genetics, 66: 761–767, 2000]. But specific cases that reach court are rare. Such a case was recently discussed on national radio [http://www.bbc.co.uk/radio4/science/ethicscommittee_s4_tr3.shtml], disclosing sufficient facts to allow analysis of the ethical and legal issues encountered. Furthermore, the outcome of the case, a court order to test the child in infancy, can be reviewed in the light of the current law, together with the geneticist's response to this order.

**Keywords** Adoption · Genetic · Testing · Court order

As genetic science has developed, the potential for its application to children who are being put up for adoption has increased. Questions concerning genetic health of the birth family are included in the standard forms provided by the British Association for Adoption and Fostering [1]. Genetic testing could give the opportunity to be certain of the racial origins of the child's parents, and to confirm clinical suspicions of heritable disease, which are present at birth. Additionally, it would be possible to predict the development of genetic disease that manifests in adult life, by presymptomatic testing. This, however, does not provide a uniform level of certainty. Whilst a positive presymptomatic test for Huntington's disease confers a near-certainty of the disease's later development, a positive test for the BRCA1 gene (predisposing to breast cancer) is associated with far less certainty that the disease will develop. Furthermore, any genetic testing is best done in the context of family testing, not least because this may allow the relevant mutation to be identified as a preliminary to testing the index patient. In adoption cases, the biological family may not be available, reducing the chances of identifying the mutation.

For this and other reasons, geneticists avoid presymptomatic testing in children until the child is competent to participate in the consent process. The obvious exception to this delaying policy is where presymptomatic testing will result in clinical surveillance, from which may flow prophylactic treatment during childhood. An example of this would be presymptomatic genetic screening for familial adenomatous polyposis (FAP). In this condition, a family history of FAP (which manifests as innumerable intestinal polyps, that will develop, if untreated, into fatal malignancy) will lead to genetic testing of the asymptomatic child. If the FAP gene abnormality is detected, the child will need to undergo frequent and onerous screening procedures to enable the prompt detection of polyp development; which will then lead to excisional surgery.

It is because of the long-term benefit of presymptomatic testing, i.e. avoidance of bowel cancer, that the normal presumption against pre symptomatic testing is rebutted.

However, there are obvious benefits to both prospective adopters and adoption authorities to define as clearly as possible the potential medical problems that may arise in the adoptee. The efforts to investigate the risks of future physical or mental ill-health are often predicated upon the notion that if the adoptive family can anticipate serious disease as a result of predictive testing, the family can prepare itself for the eventuality, and make appropriate

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advance provisions, including financial ones. As will be demonstrated, this is not a universally accepted approach.

The ability to make some predictions as to the likelihood of developing mental illness or dependency on alcohol are cited as examples of presymptomatic tests that are likely to deter prospective adopters; paradoxically, for no good reason, since many of these 'predictive tests' have, in reality, little predictive value. There is, nonetheless, access available to commercial genetic testing that purports to provide detailed risks for common medical conditions.

The case, transmitted on BBC Radio, related to a 15-month boy who was being prepared for adoption by the local authority, who was acting as the adoption agency. Members of his birth family had Multiple Endocrine Neoplasia type 1 (MEN1) [2], and this conferred a 50% risk that he might inherit the condition.

At issue was whether the local genetics service was prepared to perform a presymptomatic test on the child.

The most obvious advantage to the test would be that if the child did not carry the specific mutation identified in his parents, the diagnosis of MEN1 would be excluded. If a test showed that he did have the diagnosis, his prognosis would be difficult to determine, since the significance of a positive test is uncertain. Some 'positive' patients never manifest the disease; others present in their twenties, thirties and forties; and there is a small group who present in their second decade of life. The severity of the phenotype is also highly variable, with some patients presenting in their 50s with mild hypercalcaemia, whilst others presenting with tumours of the pituitary, pancreas and parathyroids in the third decade; and no patterns of disease can be predicted within a particular family.

There is no doubt that in patients who do develop the disease, the consequences are severe, their lives are shortened and they require excisional surgery to either cure (where possible) or ameliorate (where not) the tumours that develop as a consequence of the diagnosis.

The normal approach to presymptomatic testing in this condition would be to delay the test until the age of 8–10 years, in order to allow the child to participate in decision-making. The geneticists involved in the case were therefore reluctant to agree to the local authority's request. Their reluctance was heightened by the recognition that a negative gene test for MEN1 did not exclude the diagnosis. The facts of the case did not reveal whether the precise gene mutation had been identified in the parents, and this would have a bearing on the likelihood of successful testing in the child, since positive identification of the MEN1 mutation is more difficult if the family's mutation is unknown. Presymptomatic testing almost invariably depends upon knowing which mutation to test for. The results of any potential test were thus uncertain, and the uncertainty was compounded by the unpredictability of the phenotype within the family.

An alternative to genetic testing is biochemical screening. Although this is in no way comparable to genetic testing, since it will not provide a diagnosis of MEN1, yearly screening of the serum calcium would identify early evidence of hypercalcaemia, indicative of hyperparathyroidism. This indicator of tumour formation in the parathyroid glands is an almost invariable manifestation of MEN1. Biochemical screening thus alerts the clinician to the development of tumour formation, although does not provide the genetic confirmation of the diagnosis.

The genetics service opposed the pre symptomatic testing of the 15-month child. Although the court initially ordered that a test should be performed, the order was subsequently withdrawn, possibly because his long-term fosterer indicted that she wished to adopt him irrespective of the results of the gene test. Subsequently, the local authority approached the court 18 months later, a second order was obtained, and the genetic test was performed.

Presymptomatic testing in preparation for adoption has been extensively reviewed. The advantages to the prospective adopter are acknowledged [3], but it is argued that genetic testing is different from other forms of preplacement assessment. Notwithstanding the vigorous debate on whether presymptomatic testing in children for diseases that manifest in adulthood is ever justifiable [4], the interests of the child, the adopters, the adoption agency and the birth parents have been considered.

That the best interests of the adoptive child (throughout life) is paramount is enshrined in the current English [5] and American [6] legislation. This places a duty on the geneticist to treat all children equally, whether in preparation for adoption or those living with their birth family, unless the adoptive process alters the substance of what is in the best interests of the child. If it were the case that knowledge of the MEN1 genotype would confer benefit on the child awaiting adoption, these particular circumstances may justify presymptomatic testing that would not otherwise be clinically appropriate. However, if the best interests of the adoptive child are not furthered by genetic testing, two classes of children are created, and those being tested as a preliminary to adoption suffer only the disadvantages flowing from the test, whilst accruing no clinical benefit. They are contrasted with the class of children 'protected' by the geneticists from presymptomatic testing, until they are of an age where the decision to test reflects their best interests.

What disadvantage will flow from a positive test? It is notable that in one series, only 15% of people with a parent diagnosed as having Huntington's disease elected to have a presymptomatic test [7]. The prognosis in Huntington's disease is more certain, and perhaps more devastating, than that of MEN1. Nevertheless, the data provide an indicator that from the patients' perspective, predictive testing is not universally welcomed. From the perspective of the child

awaiting adoption, a diagnosis of MEN1 may deter prospective adopters. This in itself could indicate that their future commitment as loving parents may be lacking. Alternatively it may only reflect that they have considered their resources insufficient properly to care for a child (and adult) with a serious systemic disease, and that they have served the best interests of the child by allowing a better-equipped family the chance to adopt him. Ironically, such a decision may lead to a thoughtful family, determined to act in the child's best interests, ruling themselves out of the chance to adopt a person with MEN1 whose phenotype dictates that he will be asymptomatic for the majority of his adult life.

Disadvantage may also be incurred from being successfully adopted with an established genetic diagnosis of MEN1. But the adoptee with the confirmed diagnosis is at risk of being considered 'different', from the moment he enters the family. He has a far greater potential for developing serious systemic disease when compared with siblings sharing a population risk; and it could be argued that he will have his share of the families 'goods'…. Whether emotional, temporal or financial, reduced accordingly [8]. Accordingly, together with discrimination, there is the possibility that the child's self esteem and aspiration may suffer, consistent with the family's (incorrect) belief that his future is 'preordained' [9].

This is to be distinguished from an adopted child who arrives in a family as an equal; spends years in the family, acquiring and earning his position in the family unit; only to be affected by serious disease unexpectedly.

It is conceded that an untested 3 year old who enters a family with a potential diagnosis of MEN1 may eventually have it confirmed.

Additionally, the requirements for biochemical screening will remain, for early determination of tumour formation. This will require periodic visits for blood tests and clinical review.

But given the natural history of the disease, these infrequent reminders of a potentially latent disease will be insufficiently intrusive to weaken the bonding with his adoptive family to the point where he is considered an unequal member.

Although county court judgements are not reported, and thus not available for analysis, the decision to order presymptomatic genetic testing for MEN1 will have been based upon the statutory checklist of factors [10] which must be taken in to account by the court. On this second occasion the child was now about 3 years of age. Given the widespread presumption against presymptomatic testing in these circumstances by the genetics community, the judge had a duty to weigh these concerns against evidence that failing to perform the test will have contravened the child's best interests.

The child was now older; the judge may have considered it unlikely that waiting until the age of eight would necessarily result in a definitive consent from the patient; assent would be highly likely, and since substantial benefit was unlikely to accrue from delay, the test might as well proceed without it. He might also have reasoned since the clinical need for genetic confirmation would be more pressing in 5 years time, it would then be performed irrespective of an 8 year olds refusal, although it has to be immediately recognised that this has never been examined in the common law.

The little boy was still with his original long- term foster mother, and the bond between them must have become dense. His foster mother was applying for the adoption. This was not a situation where the relationship between an adoptee and his new parent was forged on the basis of his 'certain' abnormality, causing the seeds of discrimination to be sown. It does beg the question as to why, if the family had looked after him for so long, were they again wishing to obtain a test, since it seems unlikely that if they were willing to adopt him, they would be influenced by its outcome. Presumably, they reasoned that a negative test (assuming that the birth parents' mutation had been identified) would allow his biochemical screening to stop, whereas a positive result would simply assert the inevitability of the screening programme.

A final, and interesting, aspect of the case was the willingness of the genetics department to accede to the order to perform the test. No English court will require a clinician to perform an elective intervention contrary to their clinical judgement [11]. In the circumstances where this conflict occurs, the health authority's duty is to approach another clinician, who may or may not be prepared to comply with the order. It appears from the transcript that the genetics service tested the toddler whilst still unconvinced that it was in the child's best interest. One must sympathise with what must have been a very uncomfortable position, but nevertheless, the option to refer the case to another centre was not discussed. If such a referral had been made, and again refused on the grounds of clinical best interests, both the court and the local authority would have been given some pause for thought, and potentially, deferred the decision to a more appropriate date.

As geneticists' ability to predict peoples' future health is further refined, the pressure from adoption agencies and prospective adopters to test children awaiting adoption will increase. At the time of writing, a case is being pleaded on the basis that it is in the best interests of the adoptee to test for FAP in infancy, rather than at the age of 10 years. If geneticists are convinced that such tests are premature, the appropriate reasoning needs to be in place when providing evidence to the family court. Faced with a court order to

test, a consistent approach should be taken. If a genetic test is clinically inappropriate before the court hearing, it should remain so afterwards; although this will obviously merit renewed scrutiny.

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