

Joint Committee on Medical Genetics

The Royal College of Physicians The British Society for Human Genetics The Royal College of Pathologists

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The (Rt. Hon. the) Lord Warner
House of Lords
London
SW1A 0DW

Dear Lord Warner

Re: Human Tissue Bill

Legal means to allow the use of bodily material (including the extraction and testing of DNA) where consent has been expressly refused.

The Joint Committee for Medical Genetics (JCMG), in conjunction with the British Society for Human Genetics and the Genetic Interest Group, welcomes the changes that have been made to the Human Tissue Bill in its passage through Parliament, in particular the amendment of the Bill to allow DNA analysis for the benefit of a third party where it is not reasonably possible to trace the person from whose body the DNA has come, (Schedule 5, part 2 section 9). However, the JCMG wishes to bring the attention of the Lords an outstanding issue that may adversely affect patient care.

The JCMG is concerned that the absolute veto of living persons over the use of their bodily material (i.e. material from a human body which consists of or includes human cells) may jeopardise the clinical care of other family members. We would wish in every possible case to respect the right of a person to make an individual choice about testing. However, we are concerned that in such cases there are no mechanisms to adjudicate between the competing claims of individuals and none that will allow that bodily material to be lawfully used for the benefit of another family member, even where the failure to use that bodily material (i.e. to extract DNA from it and analyse it for the benefit of a third party) could result in serious consequences for relatives of that person.

Where the health of individuals is at stake, it runs counter to good clinical practice not to allow a forum for the competing claims of those individuals to be heard. We propose that the Bill be amended to include a provision allowing the High Court to determine the issues and make a decision about the legality of DNA analysis in such circumstances. The experience of our members suggests that this is only likely to be relevant to a very small number of cases each year satisfying a limited set of circumstances such as where the only source of DNA is from a histological sample from a tumour or biopsy and where one person (A) prevents that use of his or her tissues for the benefit of another (B) even where it is likely to result in risk of serious harm or death to the applicant or to his or her children or future children. We would be happy to assist in the formulation of appropriate criteria, which could be included in codes of practice in due course.

We attach an appendix to this letter setting out a number of clinical situations in which we believe that an application to the High Court would be justified where one person refuses to consent to the use of their bodily material (and thus their DNA) for the benefit of another and ask your Lordship to take the necessary steps to enable such action to be taken.

Yours sincerely

Dr Heather Skirton
Chair, Joint Committee for Medical Genetics

Appendix 1

Circumstances in which an application for a Court Order may be justified

1. Familial breast cancer

B is a 30 year old woman whose mother (C) developed breast cancer when she was 39, and died from the condition three years later. B knows that her aunt (A), her late mother's sister, was also diagnosed with breast cancer sometime in her thirties and had a mastectomy. A is still alive, but she and B are estranged.

B wants to know whether her family has the inherited form of breast cancer due to a mutation in either the BRCA1 or BRCA2 gene, which is highly likely given this family history. If it does, she feels that the appropriate way of reducing the risk of developing breast cancer for her is to have a bilateral prophylactic mastectomy. The surgeon she has discussed this with is only willing to perform this major operation if there is proof that she has indeed inherited a mutation of one of these genes.

There are a number of genes with many potential mutations that could be causing a predisposition to breast cancer in this family. It is only practicable to test an unaffected family member after identifying the gene mutation in a person who is known to be affected. This necessitates testing a sample from the tumour of either her mother or her aunt. Tissue blocks from her late mother are not available (she was treated abroad) and her aunt (A) puts the phone down when she rings and does not reply to her letters.

2. Hereditary Non-Polyposis Colon Cancer (HNPCC)

Multiple members of B's family have suffered from a variety of different cancers including colorectal, endometrial and gastric cancer. B's doctor has suggested that she may have inherited a familial cancer-predisposing syndrome such as Hereditary Non-Polyposis Colon Cancer (HNPCC). B's mother and father have both died in their 40's from breast and gastric cancer respectively.

There are a number of genes with many potential mutations that could be causing a predisposition to cancer in this family. It is only practicable to test an unaffected family member after identifying the gene mutation in a person who is known to be affected. The only living relative that B has is an aunt (A) who has had surgery for colon cancer in her late 30's. A refuses to give a DNA sample for testing. She also refuses to allow B's doctors to test a sample of tumour tissue that was retained following surgery. This could be used for microsatellite instability testing (a form of testing that provides an indication as to whether the cancer is due to an inherited mutation) and subsequent mutation testing for HNPCC mutations. B wants to know whether she should undergo the recommended regime of screening. This involves screening of many body systems, including regular colonoscopy, pipelle for endometrial cancer, ultrasound of renal tract and ovaries, gastroscopy, and mammography. Some of the screening tests are invasive with a morbidity and mortality risk.

3. Reproductive choice - X-linked recessive condition

B is a woman whose brother (A) has Duchenne muscular dystrophy, a serious neuromuscular disease that usually results in death by the early 20's. B is planning a family, and would not wish to have a son with this condition.

A had a muscle biopsy sample taken when he was a child. The tissue was stored but DNA was not extracted nor tested using molecular laboratory methods.

Now B asks if A's sample could be tested to detect any mutation. This result could be used to try and identify if she is a carrier, and to detect an affected fetus through a prenatal test. Other available tests (such as creatine kinase testing and exclusion testing) have not helped to clarify B's carrier status. A refuses to allow his DNA to be used to detect the familial mutation, even though his diagnosis is known. This decision might result in the birth of an affected fetus or the termination of a normal male fetus.

4. Reproductive choice - dominant condition

B is a young man whose father (A) has a progressive neurodegenerative condition. B has not had a good relationship with his father for 5 years, since he left home because of his father's outbursts. Both A's parents died young.

B and his wife wish to have a family, but B does not wish to have a family if he has the same neurodegenerative condition as his father. He wants to have a genetic predictive test, but has no access to his father's results. His father refuses to speak to him or about him.

The only source of DNA from his father is a tissue biopsy taken 8 years ago. B cannot be tested unless the laboratory is aware of the mutation for which the sample should be tested.