When should NHS contact patients over faulty genes?

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The NHS and health services worldwide need to develop policies on when patients should be "re-contacted" about faulty genes, as the current lack of guidance creates a dilemma for health services, experts have warned.

Re-contacting happens when new information about a patient's genes comes to light—such as when a certain gene mutation is reclassified as a cancer risk.

A policy brief by Professor Susan Kelly and Dr. Naomi Hawkins, of the University of Exeter, says genomic medicine is changing rapidly, and new discoveries create a dilemma for health services whose patients are affected.

People are also sequencing their genes when they take personal genetic tests to determine their ancestry, and genetic testing is increasingly being ordered by non-genetic specialists such as oncologists and paediatricians.

"Some people would want to be told if they are at risk of a serious illness, while others may not want to know," said Professor Kelly.

"Currently, if advances in genomic medicine reveal that a particular patient is at risk of cancer or another disease, the response is decided by whichever doctor or geneticist is involved.

"There is no NHS guidance on what to do, and the same applies in most countries."

The policy brief says it is "essential that the NHS develop policy for re-contacting in clinical genetics," and says patients and clinicians must be involved in forming the new policy.

It says patients with different conditions may have varying views, so policies should be "sufficiently flexible to accommodate these differences."

Dr. Hawkins said: "The NHS is adopting genomic medicine rapidly and clinicians are concerned that there is no NHS policy yet on re-contacting.

"Re-contacting raises important ethical and legal issues regarding the management of genomic information about patients and families."

Situations which might give rise to re-contacting include: re-evaluation of a genetic variant that has been analyzed in the past; new information about the patient's condition has come to light; or new treatment guidelines for the patient's condition have been issued.

Re-contacting occurs when the patient is not under the ongoing care of a healthcare professional, and it is distinct from a routine follow-up.

Cases examined by the academics include "Barbara," seen by geneticists in 2004 because of a family history of breast cancer. At the time, genetic testing identified no explanation for her family history, but now a new mutation has been found which does. Her medical records do not document any expressed wishes for follow up. The genetic service decided to test a stored sample on the basis that Barbara's original consent was for any genetic test that would explain her family history and Barbara was found to have the gene mutation. Geneticists didn't get a response when they contacted Barbara by letter and phone, but did inform her GP as the positive gene results impacts on the cancer screening she has been attending since 2004.

Provided by University of Exeter