

## Scientists hail genome breakthrough

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The pattern of mutations in cancer could eventually be used to tailor treatments to particular patients. Scientists have reconstructed the biological history of two types of cancer in a genetic tour de force that promises to transform medical treatment of the disease. The feat, a world first, lays bare every genetic mutation the patients have acquired over their lifetimes that eventually caused healthy cells in their bodies to turn into tumours. The procedure gives doctors a profound insight into the biological causes of a patient's cancer and marks a major milestone in progress towards personalised anticancer therapies and strategies to prevent the disease. "This is a really fundamental moment in the history of cancer research. We have never seen cancer revealed in this way before," said Mike Stratton, a co-leader of the [Cancer Genome Project at the Wellcome Trust Sanger Institute](#) near Cambridge. The researchers took diseased cells from a 45-year-old man with a type of skin cancer called malignant melanoma, and from a 55-year-old man with small cell lung cancer. They then used advanced genetic sequencing machines to read the full genomes of both the cancer cells and healthy tissues taken from the same patients. By comparing the genetic makeup of the diseased and healthy cells, the scientists created catalogues of all the mutations found only in the cancerous tissues. Most of these genetic glitches are harmless, but every once in a while a mutation causes major damage that pushes a cell closer to becoming cancerous. The scientists focused on skin and lung cancer because the environmental causes are well known. Most melanomas are triggered by overexposure to ultraviolet rays in sunlight as a child, while almost all small cell lung cancer is caused by smoking. In the case of the lung cancer patient, scientists discovered 23,000 mutations that were exclusive to the diseased cells. Almost all were caused by the 60 or so chemicals in cigarette smoke that stick to DNA and deform it. "We can say that one mutation is fixed in the genome for every 15 cigarettes smoked," said Peter Campbell, who led the lung cancer part of the study. "That is frightening because many people smoke a packet a day." Lung cancer accounts for one in seven deaths in the UK and is almost untreatable. Fewer than 10% of patients in the UK survive more than five years after being diagnosed. The risk of developing the disease falls dramatically in smokers who have quit for more than 10 years. Genetic sequencing of the skin cancer cells revealed 33,000 mutations caused by exposure to direct sunlight. Humans have 23 pairs of chromosomes that carry all of our genetic material in the form of three billion pairs of letters. In both patients, scientists saw a variety of mutations. The most common were point mutations, which flip one letter of the genetic code into another. More complex mutations involved missing or extra sequences of DNA. Occasionally, chromosomes had broken apart or fused together in the wrong way. "It's like doing archaeological excavation. You've got traces and imprints of all these processes that have been operative for decades before the cancer arose," said Stratton. The work is reported in two studies published in the journal *Nature*. The rapid advance of genetic technology is likely to make the technique a routine procedure for cancer patients within 10 years. The Sanger Institute scientists costed the procedure at \$100,000 per person a few months ago, but they expect that to fall to \$20,000 in the next 18 months. "In the long term, every cancer patient will have this done in a clinically relevant timeframe, so in the six weeks it takes to be seen, biopsied and taken into the clinic," said Stratton. The research is the first to emerge from a global consortium that is analysing the genetic makeup of 50 different types of cancer. The 10-year project will help cancer specialists unravel the particular mutations that drive each variety of tumour. By understanding the genetic flaws behind common cancers, scientists hope to develop more powerful and precise anti-cancer drugs. In the near term, researchers expect to develop blood tests that pick up signs that a cancer is returning in patients who have already had surgery or chemotherapy. [Cancer Genetics Medical research Health Health & wellbeing Smoking Lung cancer Skin cancer \(melanoma\) Ian Sample guardian.co.uk](#) © Guardian News & Media Limited 2009 | Use of this content is subject to our [Terms & Conditions](#) | [More Feeds](#)

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