

# THE INDEPENDENT



70p

Thursday 7 June 2007  
www.independent.co.uk  
... NUMBER 6,440

## Tracey Emin

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**Bipolar disorder**  
Also known as manic depression, it affects 100 million people around the world

**Coronary heart disease**  
The most frequent cause of death in Britain, with 100,000 victims every year. By 2020, it will be the biggest killer in the world

**Hypertension**  
High blood pressure affects 16 million people in Britain. Can lead to stroke, heart disease and kidney failure

**Rheumatoid arthritis**  
Nearly 400,000 people in Britain are afflicted with this auto-immune disease of the joints

**Type 1 diabetes**  
Diabetic condition in which sufferers have to inject insulin. Affects 350,000 people in UK

**Crohn's disease**  
Up to 60,000 people are affected by this debilitating bowel condition which can cause distress and pain for a lifetime

**Type 2 diabetes**  
Almost 2 million Britons are affected by this late-onset disease, which is linked with the growing obesity epidemic

# THE GENETIC REVOLUTION

DISCOVERY OF GENES RESPONSIBLE FOR SEVEN OF THE MOST COMMON ILLNESSES OFFERS HOPE TO MILLIONS OF SUFFERERS

FULL STORY, PAGE 2

# Millions offered hope by discovery of the genes behind killer diseases

By Steve Connor  
Science Editor

A groundbreaking study into the genetic basis of disease has opened the door to new ways of understanding and treating common illnesses affecting millions of people – from manic depression to heart disease.

Scientists have announced the first results of the biggest and most comprehensive investigation into the genes behind seven medical disorders, using a revolutionary approach for analysing vast tracts of the human genome.

The findings have been described as an unprecedented tour de force for British science involving 50 research groups and 200 scientists who pioneered the approach of studying common diseases by analysing the DNA of thousands of people.

The two-year, £9m study took DNA samples from 17,000 people across the UK and built up a database handling 10 billion items of genetic information. It will lead to a new understanding of illnesses as varied as high blood pressure, bipolar disorder and rheumatoid arthritis.

Initial findings from the study released yesterday identified a dozen genes or tiny “point mutations” in the human genome that appear to increase the risk of someone developing a particular disorder during his or her lifetime.

One unexpected result

## In numbers

10 billion

Items of genetic information used for the research, collected from the DNA samples of 17,000 people

was finding the first genetic link between type 1 diabetes and a bowel condition called Crohn's disease – both were associated with a gene known as PTPN2.

Scientists involved in the study said the research promised to open the way to an era of “personalised” medicine in which doctors routinely analyse the DNA of patients to find out which drugs their genes are best suited for – rather than the existing approach of “one size fits all”.

In addition, the methodology of the study could ultimately tease apart the role of nature and nurture in the creation of a person's psyche, making it possible to understand why some people are prone to developing mental illness such as manic depression or schizophrenia.

“Many of the most common diseases are very complex, part of ‘nature’ and ‘nurture’, with genes interacting with our environment and lifestyles,” said Professor Peter Donnelly of Oxford University, the leader of the scientific consortium behind the study.

“By identifying the genes underlying these conditions,

our study should enable scientists to understand better how disease occurs, which people are most at risk and, in time, to produce more effective, more personalised treatments.

“The new approach works well and reliably. Our understanding of the genetics of common diseases will change enormously over the coming years. I think we are just scratching the surface.”

The study, published in the journal *Nature*, came out of the mammoth effort during the 1990s to decode the entire three billion “letters” of the human genome. It was co-ordinated by the Wellcome Trust Case Control Consortium, and funded by the Wellcome Trust, the world's biggest medical research charity.

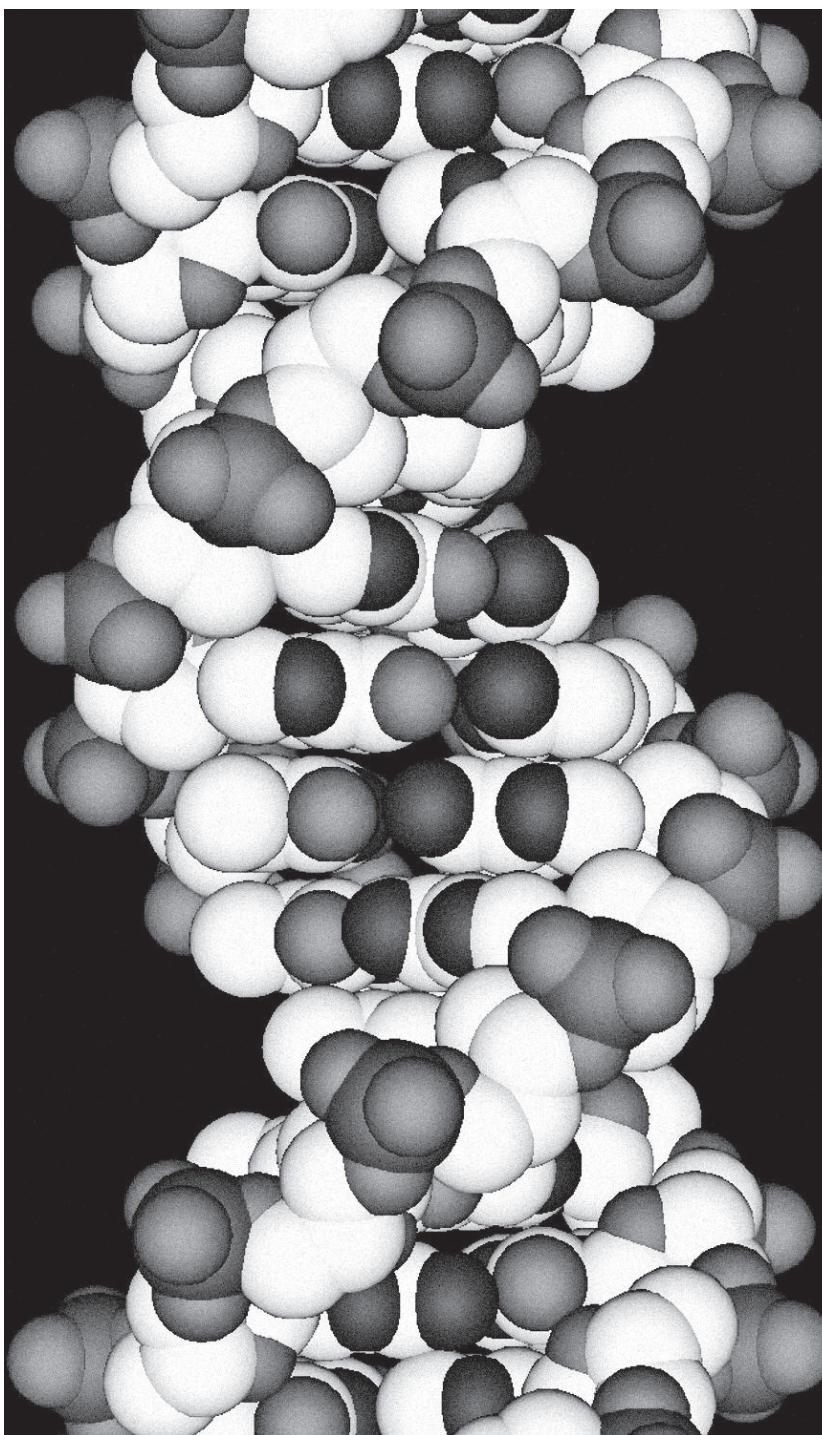
By analysing the smallest differences in the genetic sequences of patients with a disease and comparing them to the DNA of healthy people, the scientists were able to identify point mutations strongly associated with increasing someone's risk of that illness. The scientists found, for instance, three new genes that each appeared to increase the risk of someone developing Crohn's disease by between 20 and 40 per cent, compared with the risk within the general population.

Thousands of people affected by at least one of the seven diseases gave blood samples that were analysed by sophisticated DNA “chips”. These can rifle through huge regions of a person's genome to locate point mutations that are different from the general population.

Mark Walport, director of the Wellcome Trust, said: “Just a few years ago it would have been thought wildly optimistic that it would be possible in the near future to study a thousand genetic variants in each of a thousand people.

“What has been achieved in this research is the analysis of half a million genetic variants in each of 17,000 individuals, with the discovery of more than 10 genes that predispose to common diseases.”

Britain is in the process of establishing a vast databank of human DNA called UK Biobank – the biggest of its kind in the world – which will store the genes



Scientists studied the DNA of 17,000 people in a study costing £9m WELLCOME TRUST

of half a million people who will be closely monitored for the rest of their lives for correlations between genes and disease.

Dr Walport said the findings of the latest study were proof that the UK Biobank approach of analysing DNA on such a large scale will work in terms of finding out why some people fall seriously ill. “This research shows it is possible to analyse human variation in health and disease on an enormous scale,” Dr Walport said.

“It shows the importance of studies such as the UK Biobank, which is

seeking half a million volunteers aged between 40 and 69, with the aim of understanding the links between health, the environment and genetic variation,” he said.

The latest findings come just weeks after scientists used a similar approach to identify four new genes that play a small but significant role in increasing a woman's risk of developing breast cancer, and an earlier discovery of a gene that confers a 70 per cent higher risk of becoming obese.

Professor Donnelly said that it was possible to search for the genetic

basis of disease in a fundamentally different way than in the past when the emphasis was on finding the cause of single-gene disorders in families affected by disorders such as cystic fibrosis and Huntington's chorea.

“We are now able to effectively scan most of the common variation in the human genome to look for variants associated with diseases. This approach will undoubtedly herald major advances in how we understand and tackle disease in the future,” Professor Donnelly said.

LEADING ARTICLE, PAGE 30

## An atlas of the human body

### ANALYSIS

BY STEVE CONNOR

When the human genome was fully decoded at the turn of the century, one scientist described it as the biggest advance since the invention of the wheel. It has taken some years for research to live up to the expectations attached to the deciphering of the human genome. Today's announcement goes some way to addressing the rationale for decoding the 3 billion “letters” of the human DNA code.

It is now possible to use the framework of the human genome as an atlas to find our way around the individual genomes of real people with real medical problems. Knowing that a fault in a gene is associated with a disorder could help scientists to understand the molecular basis of that condition, leading to better treatment and possibly even a cure.

Until recently, medical genetics had to focus on often rare conditions resulting from defects in single genes. Now the emphasis is on more common diseases where the genetic element may be small but the effect is still significant.

We are all born with genetic predispositions. The science of mass DNA analysis promises to tease apart the inherited traits. The hope is that it may be possible to analyse our genomes at a fraction of the current cost. However, this is a long way off. Nevertheless, scientists are on the cusp of great discoveries about the nature of human disease.

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