Genomics in stroke
Fact or fiction?

Hugh Markus
How are my genes, doctor?
The possibilities are endless. According to scientists, it may soon be possible to identify people at risk of cancer and heart disease with a simple blood test.

Doctors may also be able to use the same test to find out how individual patients will respond to specific medicines and tailor their treatment accordingly.

Such possibilities are down to advances in gene research.

Thousands of scientists around the world have immersed themselves in studies aimed at identifying genes that cause disease. They are making steady progress.

Some of their findings are already being put into practice in the NHS.
Advances in genotyping

Candidate gene association study
  Single variant (SNP)

Genome wide association study
  Genotyping array
  Up to 5 million SNPs

1990-2005  2005-2013
Published Genome-Wide Associations through 12/2012
Published GWA at \( p \leq 5 \times 10^{-8} \) for 17 trait categories
deCODE your health

Calculate genetic risk – Empower prevention
your genes are a road-map to better health

Discover your ancestral roots
your genetic relationship to world populations

Introductory price
from $195 USD
Go To Store

"Now we have the ability to test someone’s genetic risk for certain disease states and then make clinical decisions based on that genetic backdrop"
Amy Donen, Nurse Practitioner

News > Discussing Genetic Risk Testing on Martha Stewart Show
Genome-wide association study identifies a variant in HDAC9 associated with large vessel ischemic stroke

The International Stroke Genetics Consortium (ISGC) and the Wellcome Trust Case Control Consortium 2 (WTCCC2)

b

Large vessel stroke
HDAC9 9p21.3

-10*10^11, OR=1.42

P = 1.87x10^-11, OR=1.42
GWAS in stroke only just beginning
Science enters $1,000 genome era

By Paul Rincon
Science editor, BBC News website

The HiSeq X Ten is capable of sequencing five human genomes a day, Illumina claims.

The ability to sequence a human genome for just $1,000 has arrived, a US genetics company has announced.

San Diego-based Illumina says it is to release a new sequencing machine that can deliver five genomes in a day.

The race to unlock a human’s genetic blueprint for $1,000 has been underway for more than a decade.

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Cost of sequencing the genome

- **Cost per Genome**
- **Moore’s Law**

The graph shows the decreasing cost of sequencing the genome over time, with a significant drop in the early 2000s and a continued downward trend towards the end of the observed period.
Welcome to Genomics England

We are a new company set up by the Department of Health to help deliver the 100k Genome Project first announced by the Prime Minister, David Cameron, in December 2012.

This project will sequence the personal DNA code – known as a genome – of up to 100,000 patients over the next five years. This unrivalled knowledge will help doctors’ understanding, leading to better and earlier diagnosis and personalised care. Based on expert scientific advice, we will start by tackling cancer, rare diseases and infectious diseases.

The company will manage contracts for sequencing, data linkage and analysis, and set standards for patient consent. Read more...
Conclusions

• Fact (although also some fiction)

• Genome sequencing will impact, before long, on clinical diagnosis

• Genetics offers one of the few ways to discover really novel pathogenic information which can potentially lead to new treatment approaches