WORLD SCIENCE FORUM
RIO DE JANEIRO

Inequalities as Barriers for Global Sustainability: DNA and the Human Variome Project

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Medical Director, QuantumDx group

World Science Forum
Rio de Janeiro
25st November 2013

Newcastle University

Beeld

Jou wêreld, Jou koerant•• Woensdag 15 Mei 2013 R7,50

Ekstra bylae oor jagbedryf **Binne**

Eugene de Kock moet weer wag **2**

Aussie-l Cheeta

“

Die lewe he uitdagings. ons nie ban te maak nie wat ons ka waaroor on beheer kan

– ANGELINA JOLIE BESLUIT OM 'N D MASTEKTOMIE T

2 aspirin tablets/day for 2 yrs reduced heredi rs at 5 yrs

Long-term effect sk in carriers of hereditary colore s from the CAPP2 randomised cont

John Burn, Anne-Marie Gerdes, Finlay M ne Olschwang, Diane Eccles, D Gareth Evans, Eamonn R Maher, Lucio Bertario, Marie- V Hodgson, Annika Lindblom, Jan Lubinski, Patrick J Morrison, Victoria Murday, Raj Hans F Vasen, Gail Barker, Gillian Crawford, Faye Elliott, Mohammad Movahedi, Kirs ynch, John C Mathers, D Timothy Bishop, on behalf of the CAPP2 Investigators

Lancet 378
December 11th 2011

43 CAPP2 recruiting centres in 16 countries recruited 1009 mismatch repair gene defect carriers

UK

Aberdeen, Edinburgh, Glasgow, Newcastle, Leeds Sheffield, Manchester, Liverpool, Birmingham, Cardiff, Belfast,Oxford, Bristol, St Marks, St Georges, Guys-London Southampton, Exeter, Guildford, Worthing,

Rest of Europe

Finland, Sweden, Denmark, Germany, Belgium, Poland, Netherlands, France, Hungary, Switzerland,Portugal, Spain, Italy

Australia

Melbourne
Brisbane
Newcastle
Perth

Human Genomics Strategy Group report 2012

- Government response
  - 100,000 whole genomes
  - £100 million investment
  - Genomics Strategy Board (NHS now HEE)

Genomics england

Functional classification of JDW SNPs

SNP Type	Novel SNPs
Missense	2421
Nonsense	5
Synonymous	2261
UTR	7102
Intron	381924

August 2010 China National Highway 110 traffic jam, China, world's worst traffic jam ever….
more than 100 kilometres from August 14 - 26, inc. at least 11 days of total gridlock.

We must collect, store and interpret 3 million variants per person….

THE HUMAN VARIOME PROJECT

LOVD

An online reference database which will provide free access to all variants which have been assessed and their functional significance validated

www.thehumanvariomeproject.org

Nat Genet, 2007. **39**(4): p. 433-6

HVP country nodes and support

Family at risk of breast cancer due to p53 mutation

Diane Carol

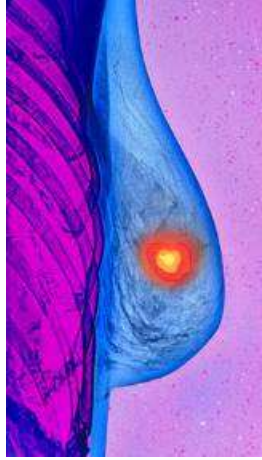
Varley et al Oncogene 1998

KEYNOTE LECTURE PRESENTATION Open Access

Hereditary cancer syndromes: opportunities and challenges

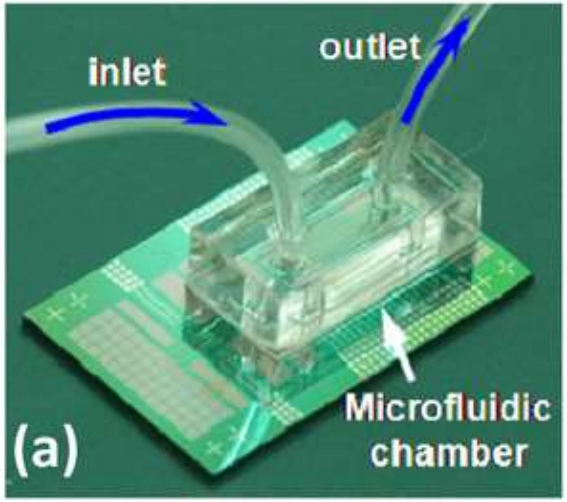
Patricia Ashton-Prolla

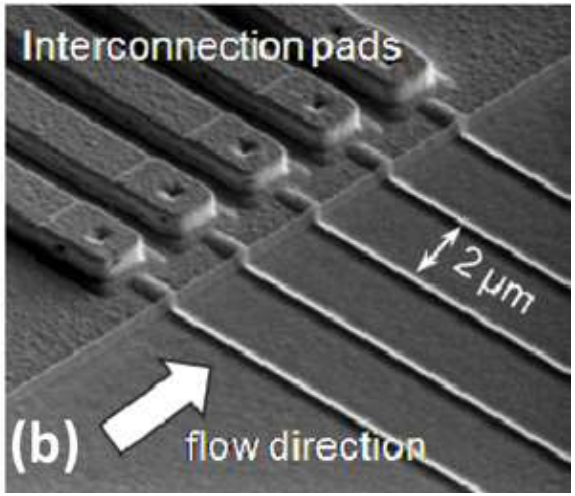
From São Paulo Advanced School of Comparative Oncology
Águas de São Pedro, Brazil. 30 September - 6 October 2012



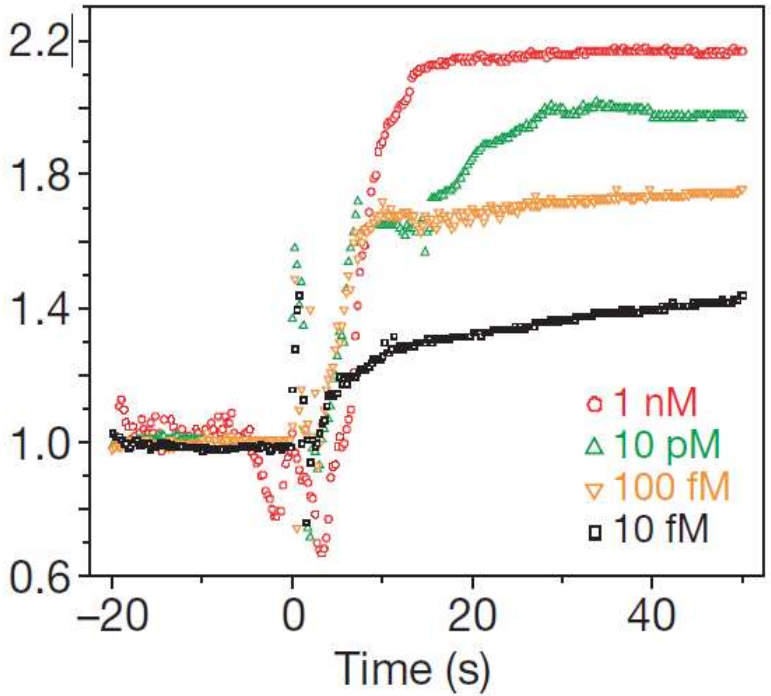
1 in 300 women in South Brazil carry p.R337H founder mutation in tp53 – a single DNA letter change causes 1 in 12 cases of all breast cancers

The much higher rate of childhood adrenal cancers has led to understanding how assembly of the protein is pH dependent

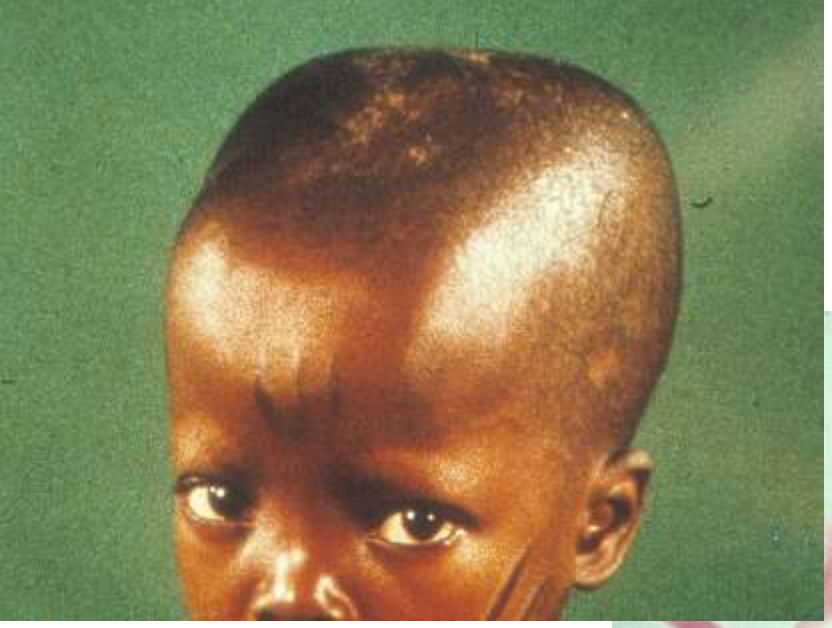




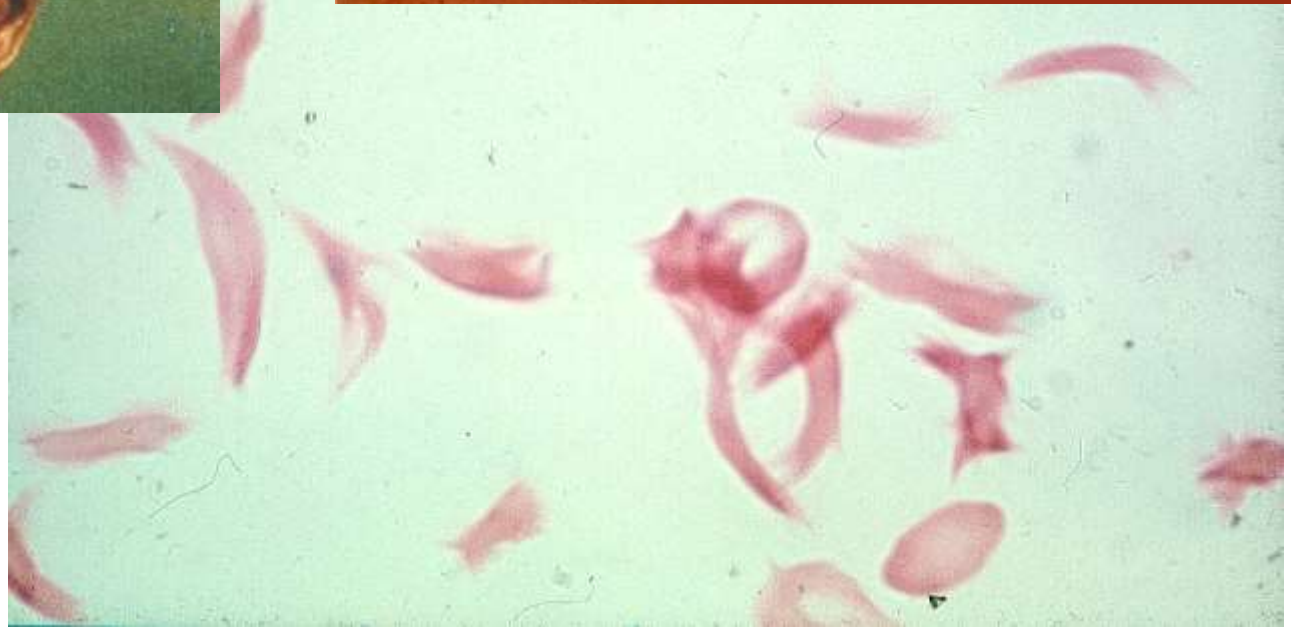
QuantuMDx



We will produce a cheap handheld device capable of analysing DNA at the point of care in 10 minutes



Expansion of bone marrow spaces to correct haemolytic anaemia



Sickle Cell Disease
Homozygous for a missense mutation

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So

In memory of Fred Sanger

UNESCO DECLARATION 1997

Article 12 a) Benefits from biology, genetics and medicine, concerning the human genome, shall be made available to all, with due regard for the dignity and human rights of each individual.

Double Nobel laureate

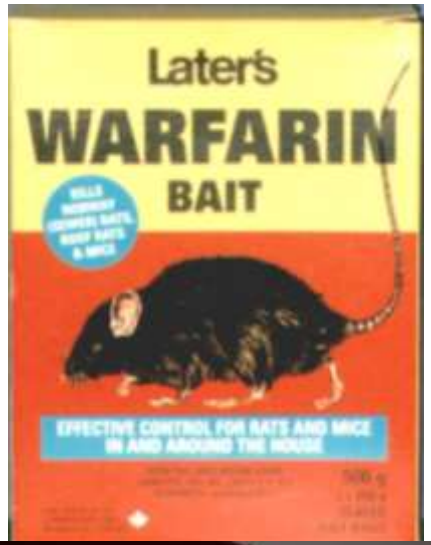
1918-2013

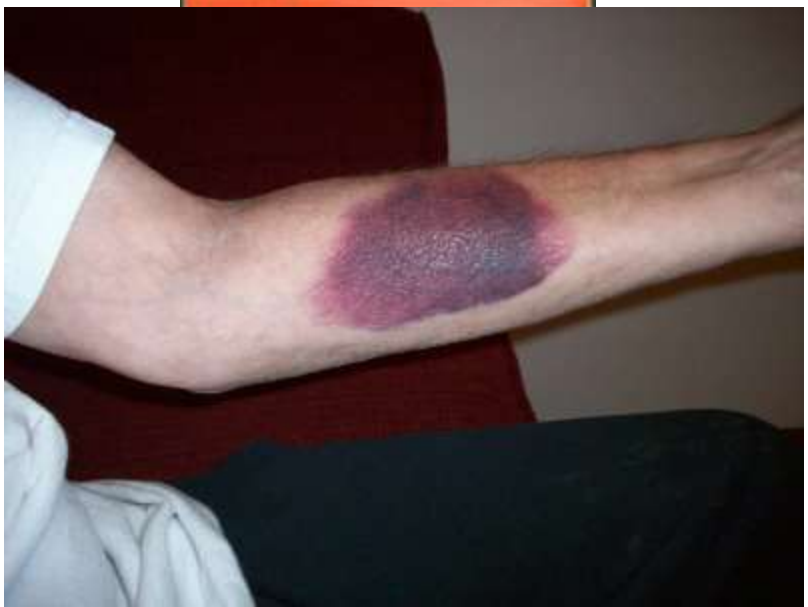
The man who showed us how to sequence DNA

pharmacogenetics

3 SNPs predict loading dose and likely dose needed to achieve INR

Population varies 20 fold yet we Treat everyone the same





Sequencing 2005

Using AB 3730 XL*

Washington University
Genome Sequencing Center

Baylor College of Medicine
Human Genome Sequencing Center

*fluorescent Sanger chemistry
on capillary sequencers