

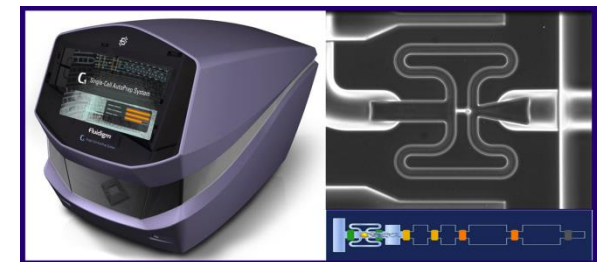
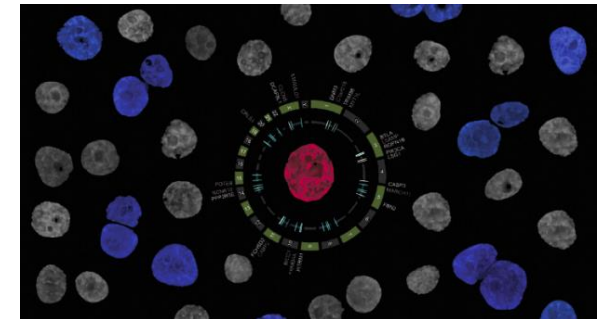
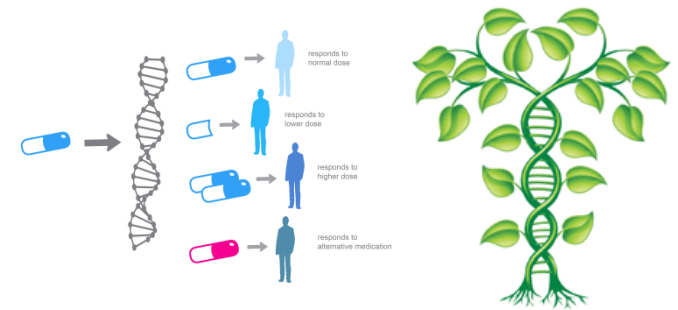


“Impacto de la bioinformática en las ciencias agrarias y biomédicas”

III Jornada de Biotecnología Situación actual y tendencias

Sobre bioinformática:

- Contexto histórico y definición
- Aplicaciones:
 - Medicina personalizada
 - Diagnóstico genético
 - Agrogenómica
- Perspectivas de futuro y retos



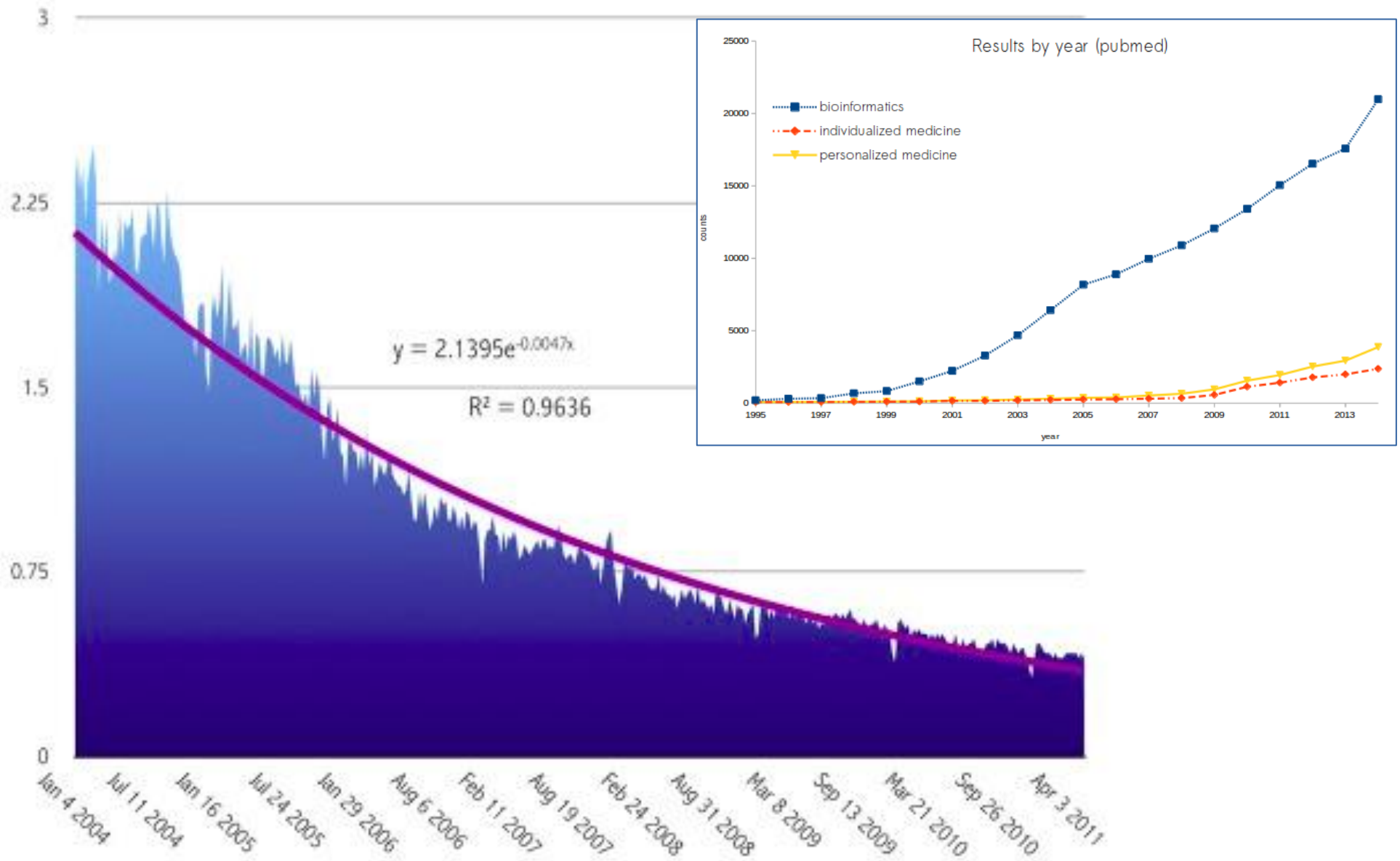
<http://www.msscience.com.au/view/affymetrix/affyagrinomics>

<http://www.alphagenomix.com/genetic-testing/empowering-personalized-medicine/>

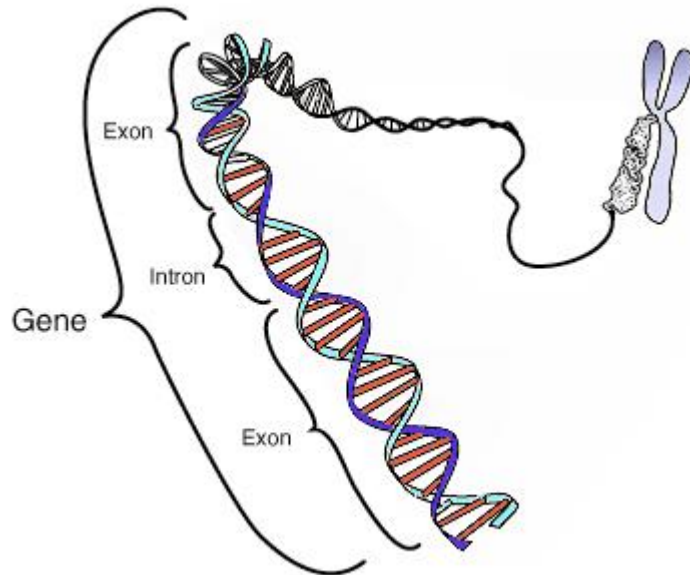
<http://qb3.berkeley.edu/qb3/fgl/images/c1systemhtml.png>

<http://blogs.biomedcentral.com/on-biology/wp-content/uploads/sites/5/2015/04/Single-cell-Leung-et-al-620x342.png>

Muerte y resurrección de la bioinformática



Datos e información

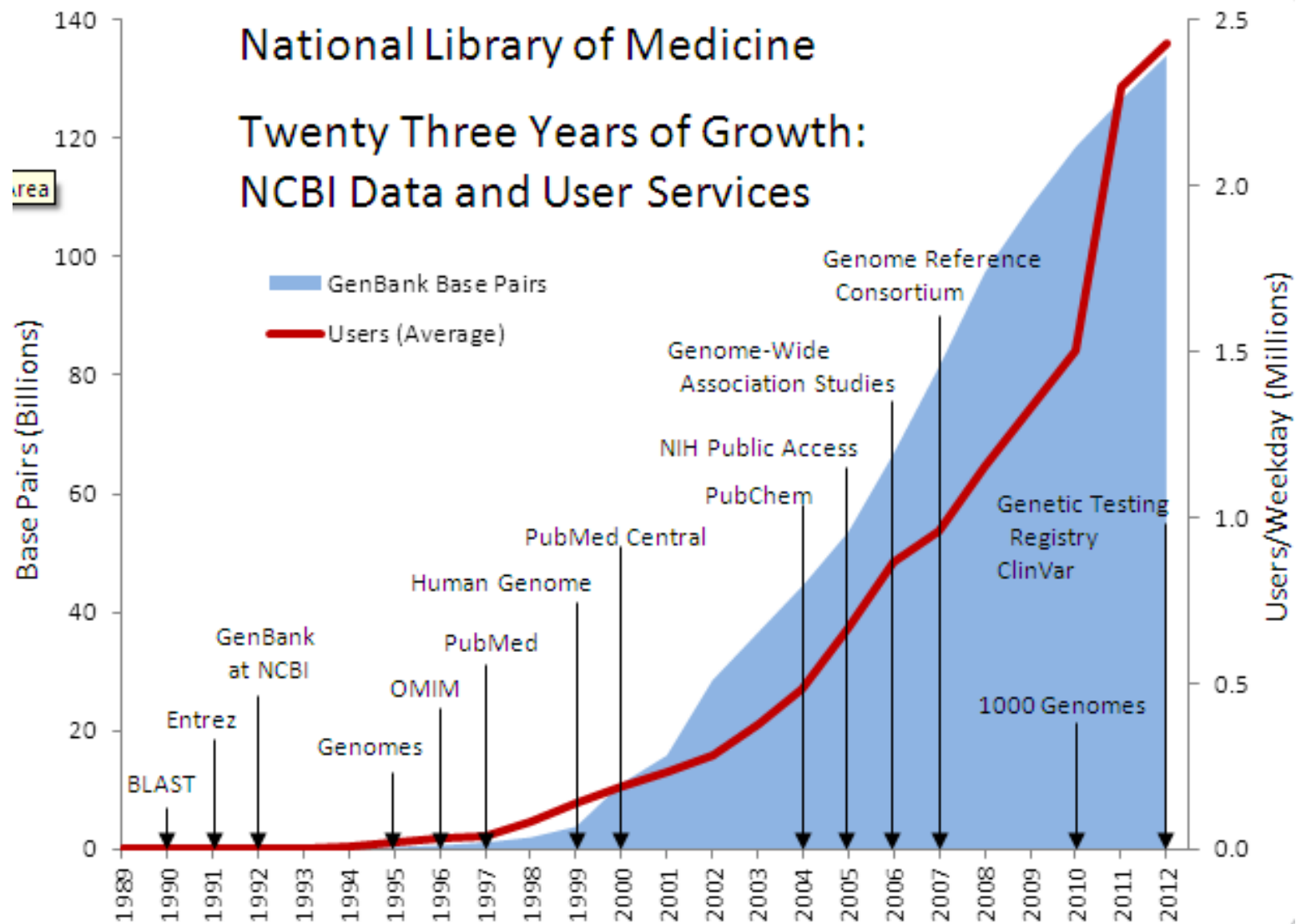


```

1   GGATCCTTTG CAAGAGCAGC CAGTGTCTT ACCCACTGAG CCCATCTCTA AGCCCCACAC CCAGCTTCTT B2
71  TGATACAAGG TCTGGTAGCT CAAACTGGAT ATGCAGCCGA GGAGGTTGAC CTGGTATTCC CTACCTACCC
141 TCTTCTCTCT ACCTTCCAAG TGCTGATATT ATACATPAGGC ATGGATAGTC ATGCCACCA GTTTGCCTTG
211 ATGGCACCAG AGTCAGGAAA GTCCAAACCT GGTAGTTGCA AACACAGCAA GAGGGTAGAG GCAGCCATTG
281 TCCTCTGGCT GCCTTGGATA CAGAGCTTCT GGGTTGGGTG GCCTTGGGTC AGTTTCCGA ATGGTTCACC
351 CTTGGGGAAA GGAACACTG CTGAAGAGGT GGGACCCTGG GAGGGCCGGG CCTCCAGCTG GGTCTCTCCA DR
421 GCCCTCGCCT TGGAACTAG GCTGGAGGGA GCCAACCCAG ATCCTGGACT GCTACAGTTA GGTGAACAGG
491 CTCTGCAGCC TCCCCTTCCC TTGGTAGCTG TGGTGGTGGT GGTGGTGGT GTGGTGGTGG TGGTGGTGGT
561 GGTGGTGGT GTAGTGGTGG TGGTGGTGGT GGTGGTGGT GTGGTGGTGG TGGTGGTGGT AGTGGTGGT
631 GTGGTGGTGG TGGTGGTGGT GGTGGTGGT GACTTTGGCT GTGACTCTGG AGCCCGACTC TGCTGCCTTA DR
701 CACAGCCAC ACAGCCTCTC TGAGCCTCAC TGACTGTGGA TAATCCCCTA TGTGAGGGA TCATTAGAAG
771 GCTTGCAGC GCCCAACCTC CTCACTTATC TCGTCTCTGT AAACCCCTTC TCCAGGCTCA GGCAGGGTTC
841 TGGGTAGCCT TGATGGAAAA GGTCTTCTG GCCCCATAT CAATGTTTAA CCGGATGAC AGGTCAATAT
911 TTACCAGCAC TGAACACAAG CTGAACGGGG GTTGGCTGAG AGCAAAGTTC CTGTGGGAGG AGAGAGCTAG
981 GCCCTGCTGG CTGCACTGGG GAGCGTGGGG AGCAAGCTGG TCCAGGAGAA CACCGTGTGC TGCGTgTCTg Exon 1
1051 CGCTAAGAAG CCTGAAGAG TAGGATGGAG ..... intron 1 (971 bp) ..... GAACTAACAC
2031 TCCCTGTTAG GAACATGGCA CTGCTCCGGG GGCTCCTCGT ACTCAGCTTG TCCTGCCTGC AAGGTCCCTG Exon 2
2101 TTCACG
  
```

Figure 3. Nucleotide sequence of the 5' end of the murine *AP* gene comprising approximately 1 kb of 5' flanking sequence, exon 1, intron 1 (shown in part) and exon 2. The ATG codon in exon 2 is underlined. The (TGG)_n repeat sequence, shown at its maximum observed length, is indicated with a dotted line. An 18-bp direct repeat (DR) is indicated by arrows below the sequence, and a B2 repeated element (B2), located on the antisense strand, is underlined. A conserved A box found in RNA polymerase III promoters and a putative TACA box are indicated. Comparison with the published murine *AP* cDNA sequence (GenBank AC Z36774) revealed a nucleotide insertion (G) at position 1046 (lower case + underlined) and a C → G substitution at position 1050 (lower case) in exon 1. The first 6 nucleotides of the published GenBank cDNA sequence (*EcoRI* site) were not considered in the comparison. The 5' end of exon 1, which was not identified, was left open (reproduced from ref. 36, with permission). The nucleotide sequence as shown in this figure has been submitted to the GenBank data bank with accession number Y12312.

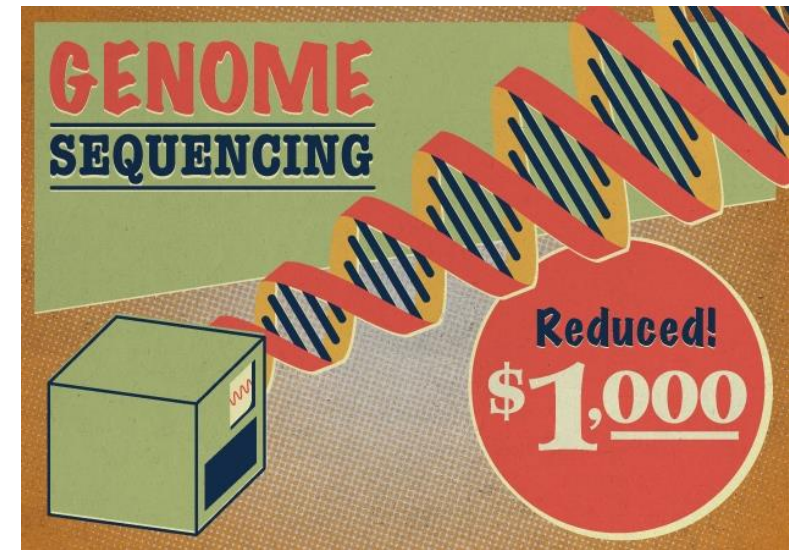
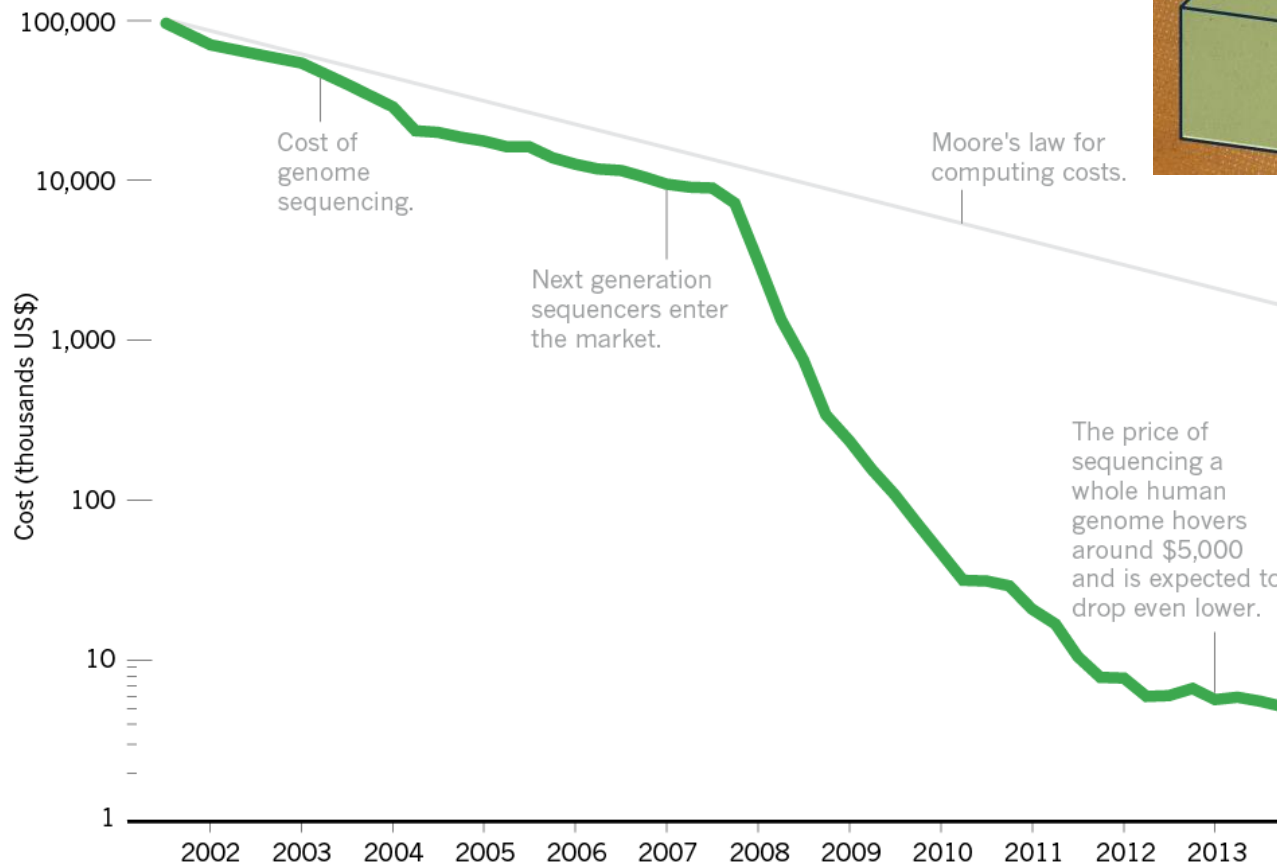
Datos y más datos



Coste de la secuenciación

Falling fast

In the first few years after the end of the Human Genome Project, the cost of genome sequencing roughly followed Moore's law, which predicts exponential declines in computing costs. After 2007, sequencing costs dropped precipitously.



Hayden EC. Technology: The \$1,000 genome. Nature. 2014 Mar 20;507(7492):294-5. doi: 10.1038/507294a. PubMed PMID: 24646979.

<http://www.nature.com/news/technology-the-1-000-genome-1.14901>

Big Data



Big data is a broad term for [data sets](#) so large or complex that traditional [data processing](#) applications are inadequate. Challenges include analysis, capture, [data curation](#), search, [sharing](#), storage, transfer, visualization, and [information privacy](#).

<http://www.amarulasolutions.com/wp-content/uploads/2014/08/HiRes.jpg>
https://en.wikipedia.org/wiki/Big_data

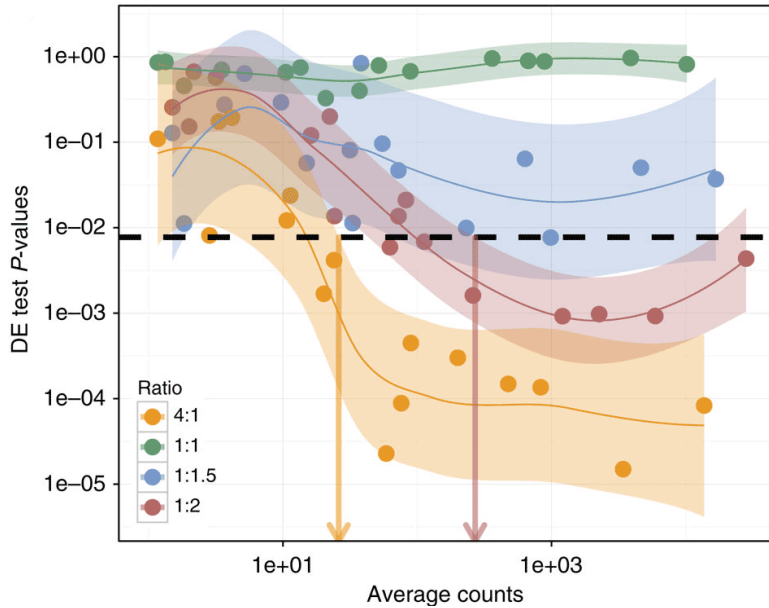
Business Intelligence



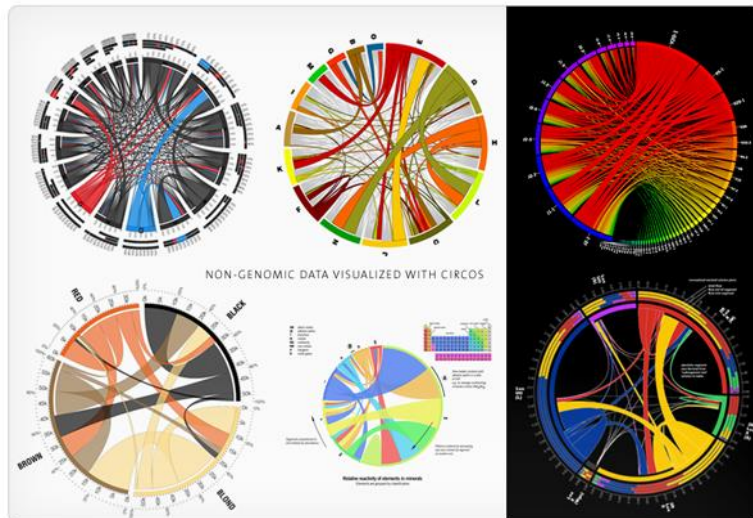
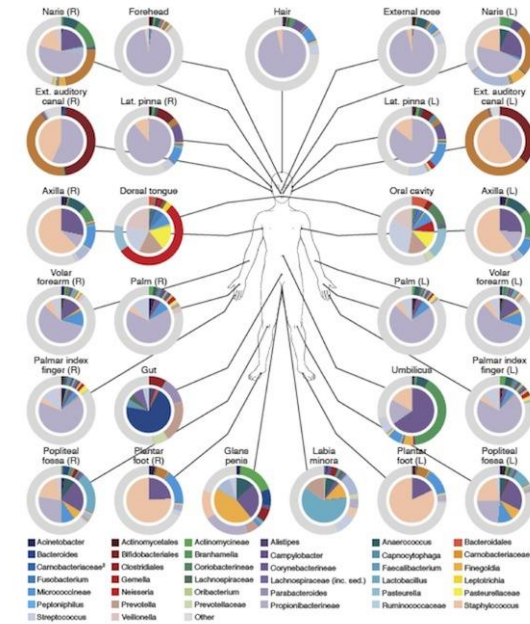
Business intelligence (BI) is the set of techniques and tools for the transformation of raw data into meaningful and useful information for [business analysis](#) purposes. BI technologies are capable of handling large amounts of unstructured data to help identify, develop and otherwise create new strategic business opportunities. The goal of BI is to allow for the easy interpretation of these large volumes of data

https://en.wikipedia.org/wiki/Business_intelligence
<http://www.vedamssoft.com/images/bi.jpg>

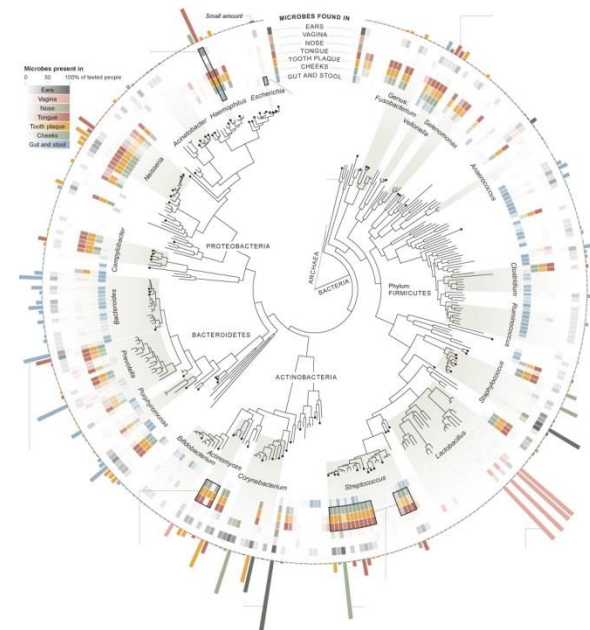
Visualización de los resultados (*Discovery tools*)



<http://www.rna-seqblog.com/wp-content/uploads/2014/10/ercr-feat.gif>



<http://circos.ca/images/published/>

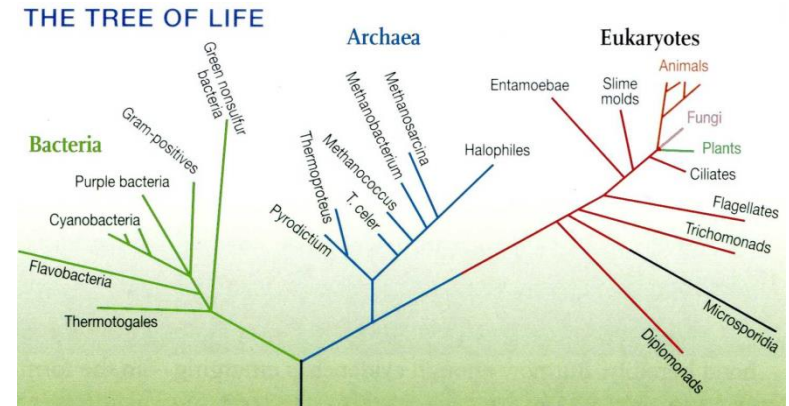
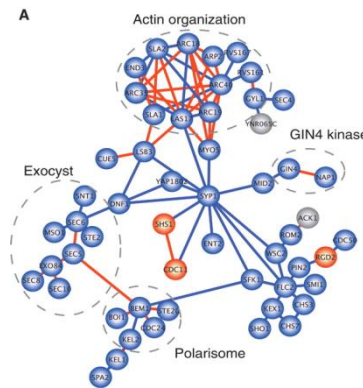
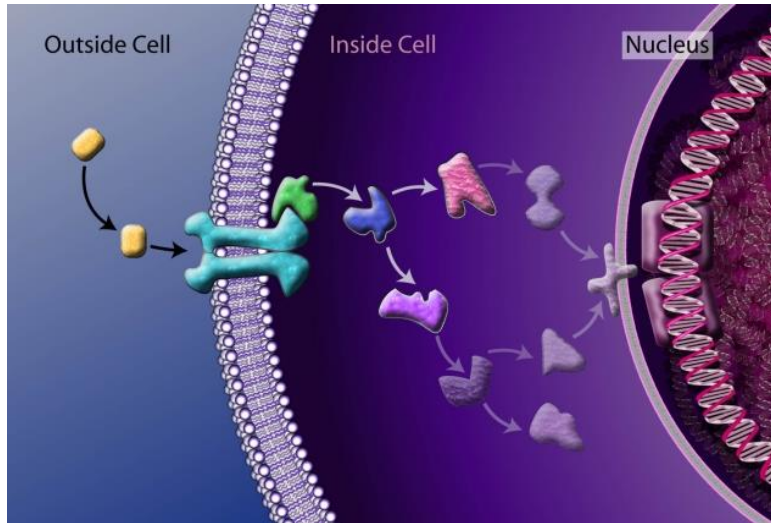


<http://www.itsokaytobesmart.com/post/25414129641/the-microbiome-the-presence-and-abundance-of-our-invisib>

Definición (intento de...)

Biology may be viewed as the study of **transmission** of information: from mother cell to daughter cell, from one cell or tissue type to another, from one generation to the next, and from one species to another. This **informational viewpoint** is termed *bioinformatics* .

Eisenberg D, Marcotte E, McLachlan AD, Pellegrini M. Bioinformatic challenges for the next decade(s). *Philos Trans R Soc Lond B Biol Sci.* 2006 Mar29;361(1467):525-7. PubMed PMID: 16524841; PubMed Central PMCID: PMC1609334.



http://news.mit.edu/sites/mit.edu.newsoffice/files/styles/news_article_image_top_slideshow/public/images/2011/20111116150237-1_0.jpg?itok=hyMAsDL_

<http://www.sciencemag.org/content/320/5882/1465/F5.large.jpg>

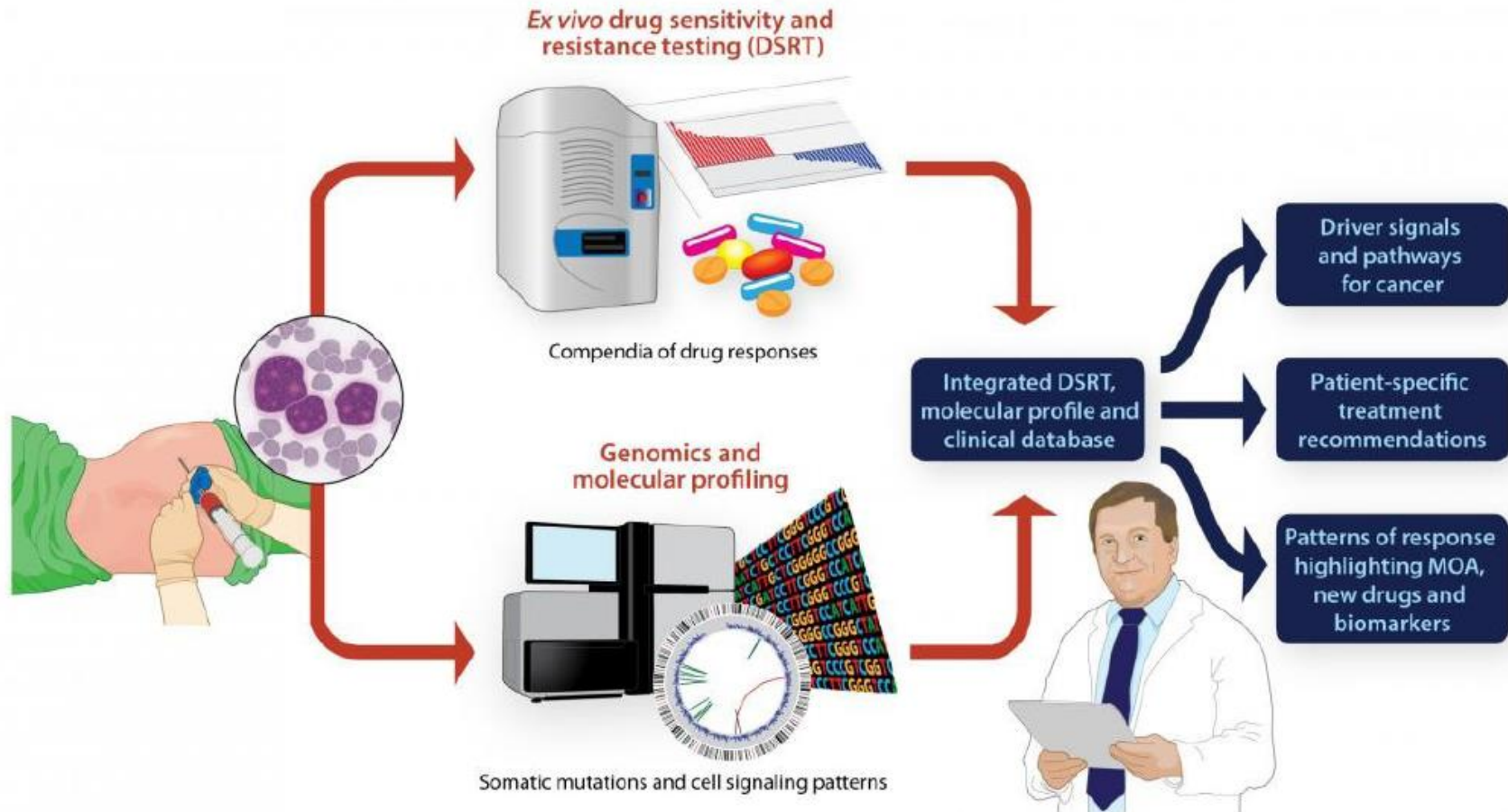
http://njsas.org/life/images/tree_of_life.jpg

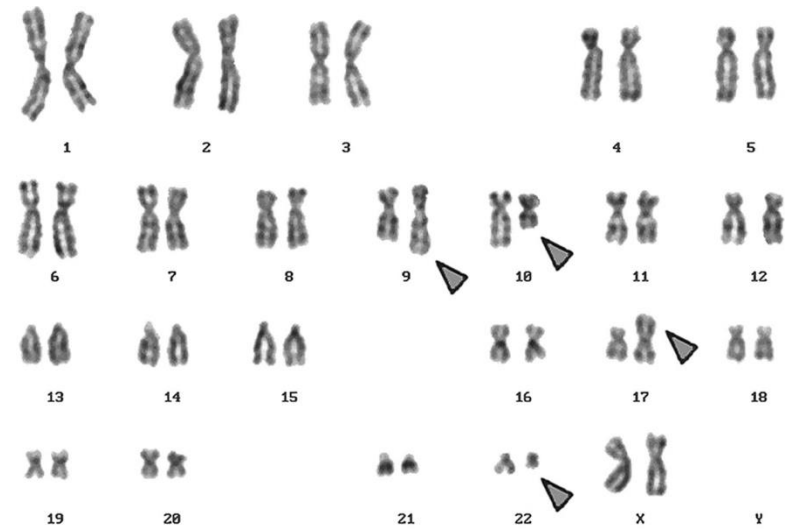
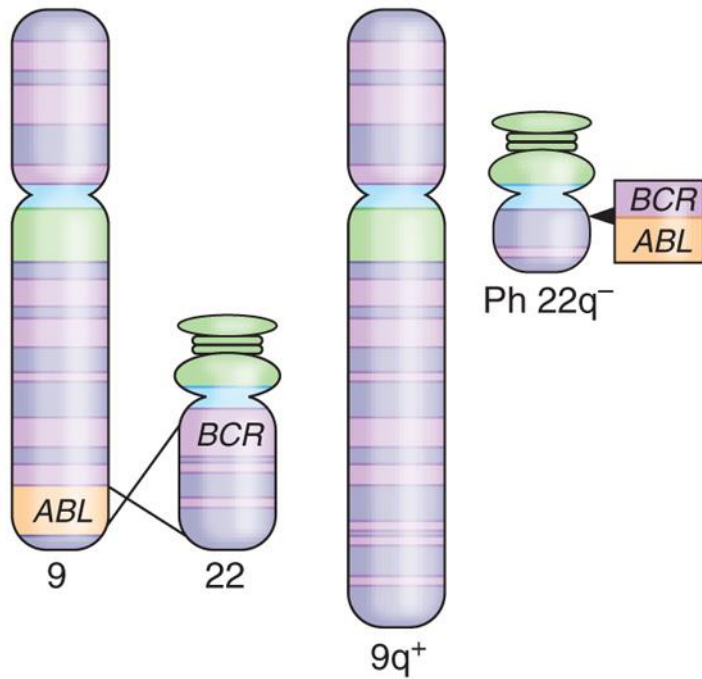
Medicina Personalizada

FIMM

Institute for Molecular Medicine Finland
Nordic EMBL Partnership for Molecular Medicine

Building a bridge from discovery to medicine



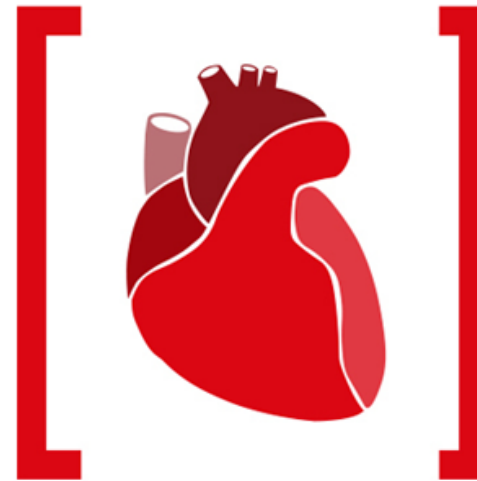


Consejo genético



Líder en genética cardiovascular

El servicio más completo en diagnóstico genético de enfermedades cardiovasculares.



La interpretación es esencial

Cardiólogos especialistas elaboran informes pormenorizados basados en información actualizada.

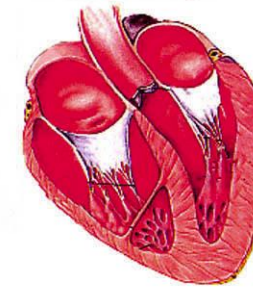


Consejo genético II

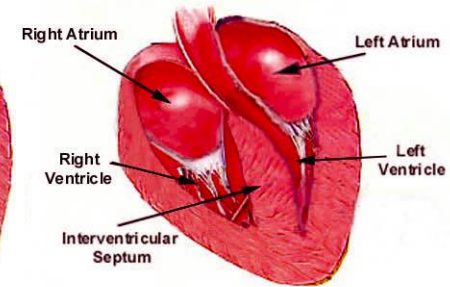
Paneles enfermedades específicas

- Miocardiopatías ▼
- Canalopatías y arritmias ▼
- Síndromes aórticos
- Cardiopatías congénitas
- Dislipemias y aterosclerosis
- Miopatías esqueléticas
- Enfermedades raras con afectación cardiovascular ▼

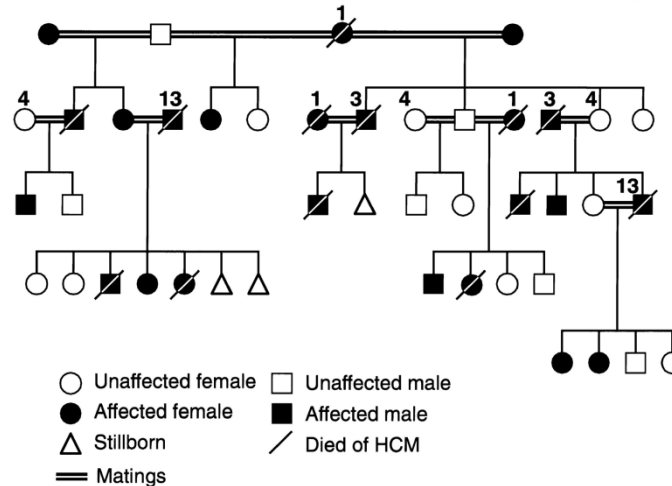
Hypertrophic Cardiomyopathy



Normal Heart



Hypertrophied Heart



Estudio recomendado

Panel de miocardiopatía hipertrófica

16 genes

Nuestro panel básico para MCH se encuentra indicado como primera aproximación diagnóstica

ante la sospecha clínica de MCH.

Incluye los 9 genes sarcoméricos principales que se han asociado a la enfermedad y a su vez, consta de 7 genes asociados a enfermedades cuya presentación clínica puede ser indistinguible de la MCH clásica (fenocopias) donde se incluye

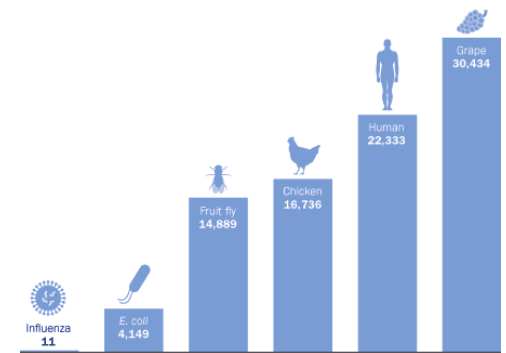
el gen GLA asociado a enfermedad de Fabry.

MYH7, MYBPC3, TNNT2, TNNI3, TPM1, TNNC1, MYL2, MYL3, ACTC1, PRKAG2, LAMP2, PTPN11, GLA, DES, TTR, FLNC

Agrogenómica

MicroWine
A Marie Curie Initial Training Network

This project is funded
by the European Union



To exploit the power of 'Next Generation' DNA sequencing approaches for the benefit of the wine industry, we have initiated MICROWINE, a 15 ESR Marie Curie Actions European Training Network.

- ESR1 - Deciphering the role of soil geochemistry in microbial community structure. University of Copenhagen, Copenhagen (Denmark)
- ESR2 - Contribution of wine microorganisms to the aroma composition of wine and its sensory impact. University of Zaragoza (Spain) (6 months in DLR-RLP)
- ESR3 - Deconstructing the origin and spread of resistance to Phylloxera. University of Warwick, England
- ESR4 - Genomics effects of grape plants on the differential microbial communities. INRA, Montpellier (France)
- ESR5 - Optimisation of microbial DNA profiling laboratory methods. University of Copenhagen, Copenhagen (Denmark)
- ESR6 - Soil and grape microbes and their relationship to wine. Aarhus University, Roskilde (Denmark)
- ESR7 - Soil microbes and plant health. Aarhus University, Roskilde, Denmark
- ESR8 - The dynamics of microbiomes in the winery. Dienstleistungszentrum Ländlicher Raum – Rheinpfalz (Germany, 12 months; UC Davis, USA, 3 months); Chr. Hansen (Denmark)
- ESR9 - Diversity and genomic characteristics of *Oenococcus oeni* strains in regional wines produced under organic or conventional methods. University of Bordeaux
- ESR10 - Microbiome variation as an effect of geography. Universidad Nacional de La Plata (Argentina)**

Wine grapes harbor a wide range of microbes originating from the surrounding environment, many of which are recognized for their role in grapevine health and wine quality. In the sixteenth century, the *Vitis vinifera* was introduced to America. In the “new world” the *Vitis* cultivars are cultivated in diverse environment.

- ESR11 - Post vinification micro-ecology of wines; the role of phenolic composition and the effects on quality. Escola Superior de Biotecnologia – Catholic University of Portugal / CBQF, Porto, Portugal
- ESR12 - Detection and treatment of esca infected grapevines. Instituto Superior de Agronomia, Lisbon (Portugal)
- ESR13 - Developing bioinformatics tools for wine fermentation, wine quality and wine health. Technical University of Denmark, Lyngby, Denmark
- ESR14 - Constraint based modeling of the grape/wine metagenome. VU University Amsterdam, Netherlands
- ESR15 - Modelling the role of microbes in wine productions. Inria & University of Lyon, France

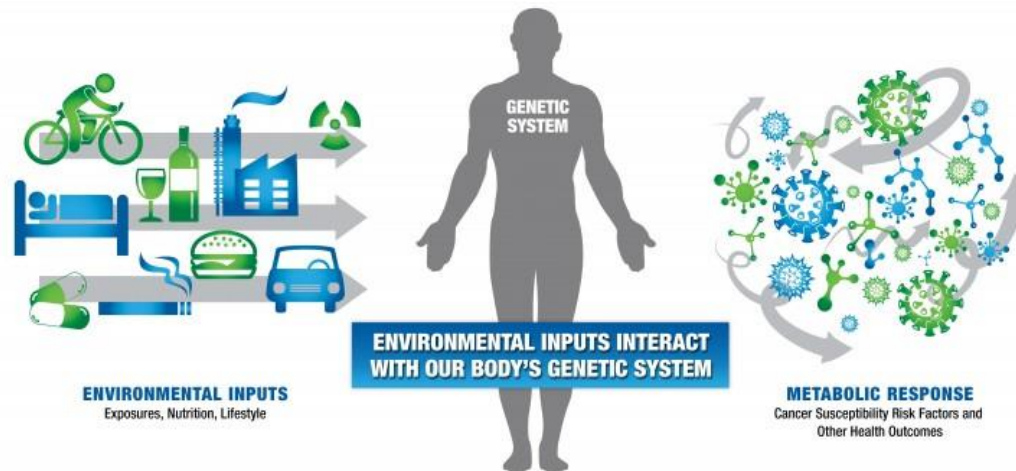
<http://www.microwine.eu/projects>

<http://www.genoscope.cns.fr/externe/GenomeBrowser/Vitis/>

<https://urgi.versailles.inra.fr/Species/Vitis>



Genótipo-Fenótipo

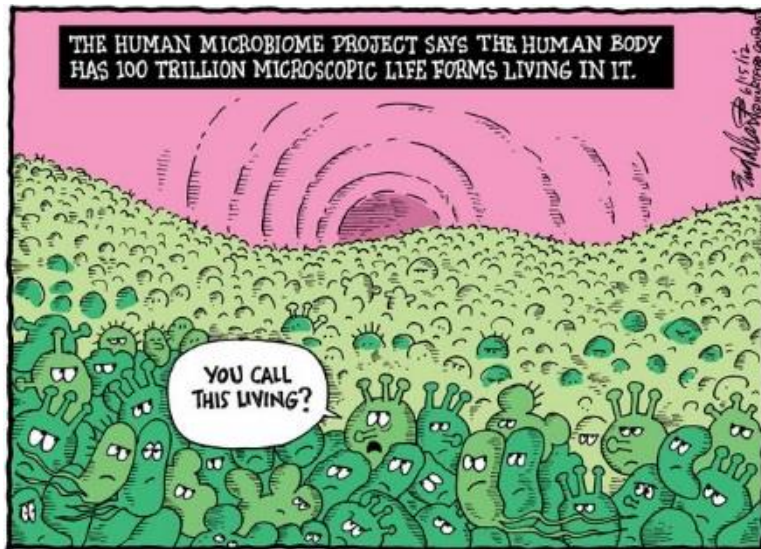
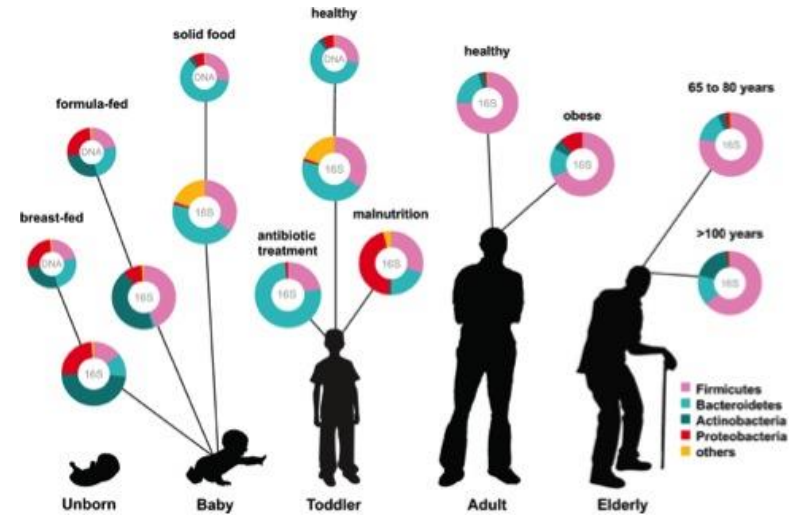
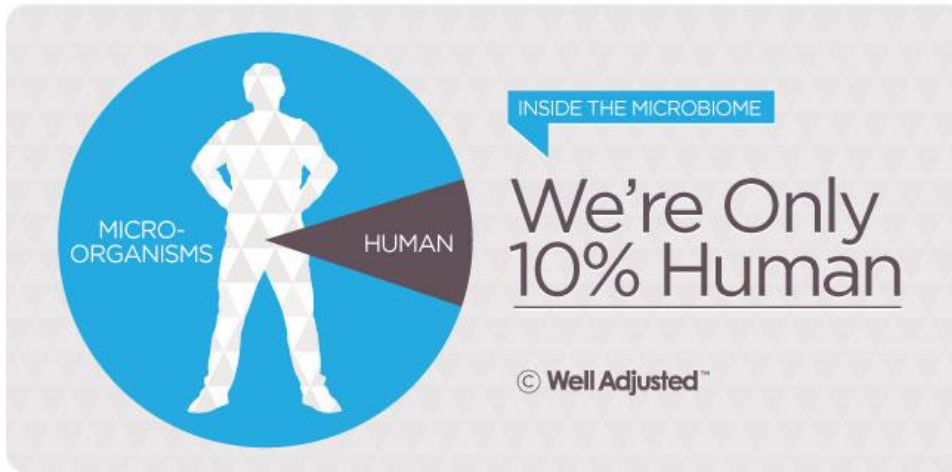


<http://evolution.berkeley.edu>

<http://blog-epi.grants.cancer.gov/wp-content/uploads/2013/03/image-GxE-interaction-e1364410175929.jpg>

<http://sanfranciscocrossfit.com/programs/biological-assessment/>

hasta que puntos somos humanos?

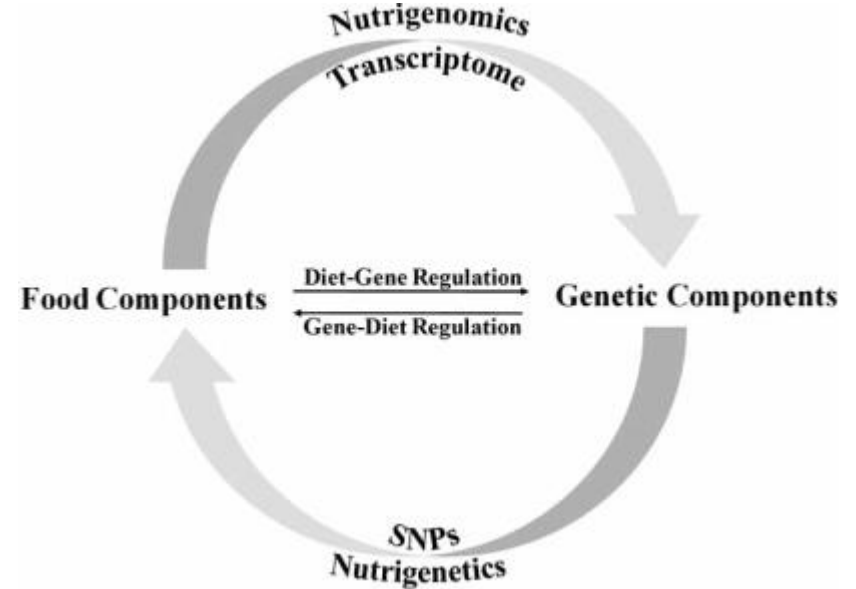


<http://welladjusted.co/blog/2013/08/07/research-reveals-were-only-10-percent-human/>

<https://studentaffairs.duke.edu/blog-entry/how-your-microbiota>

http://www.actionbioscience.org/images/microbes_over_time_sized.jpg

Nutrición personalizada

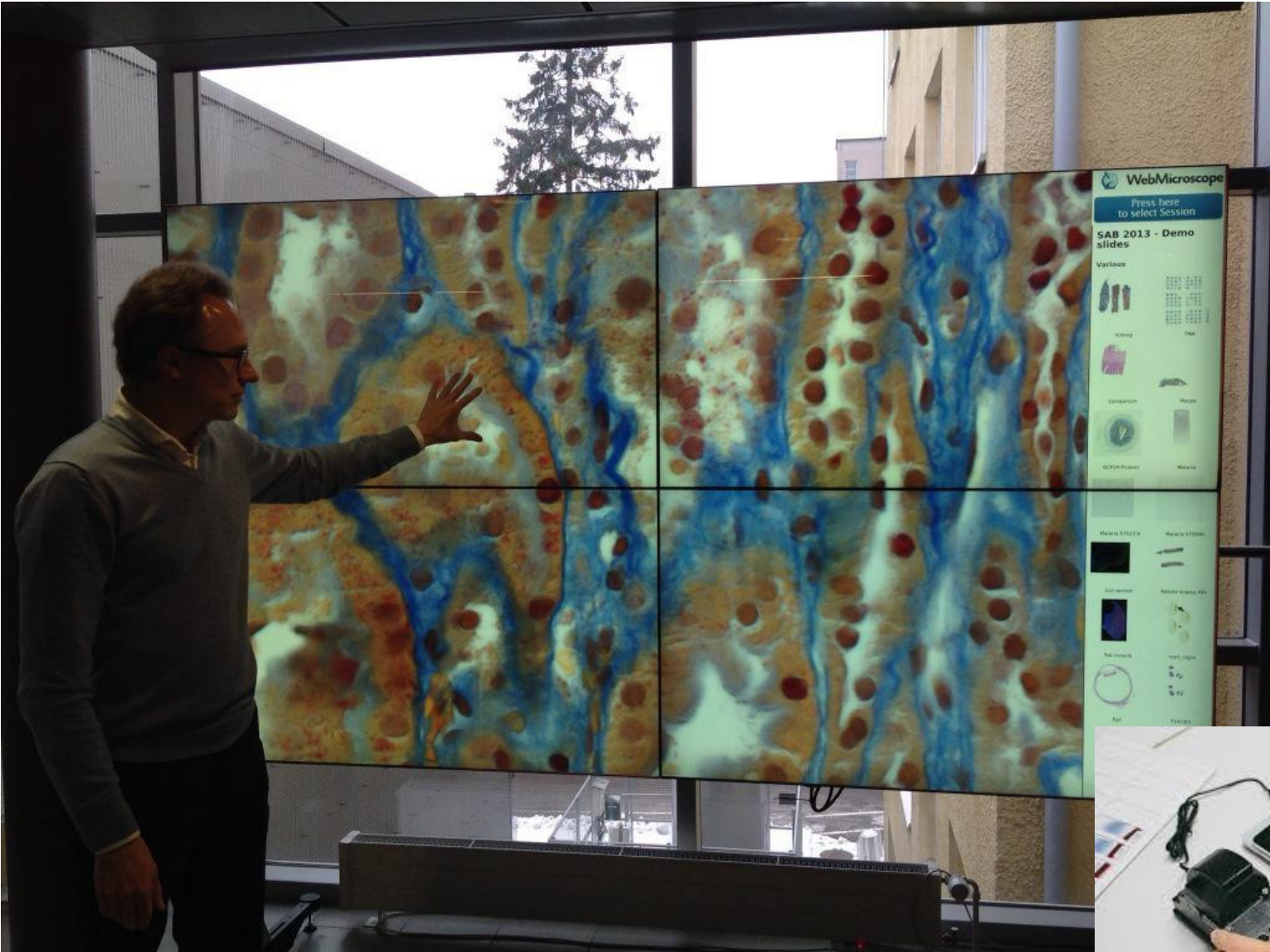


Nutrigenetics and nutrigenomics are defined as the science of the effect of genetic variation on dietary response and the role of nutrients and bioactive food compounds in gene expression, respectively.

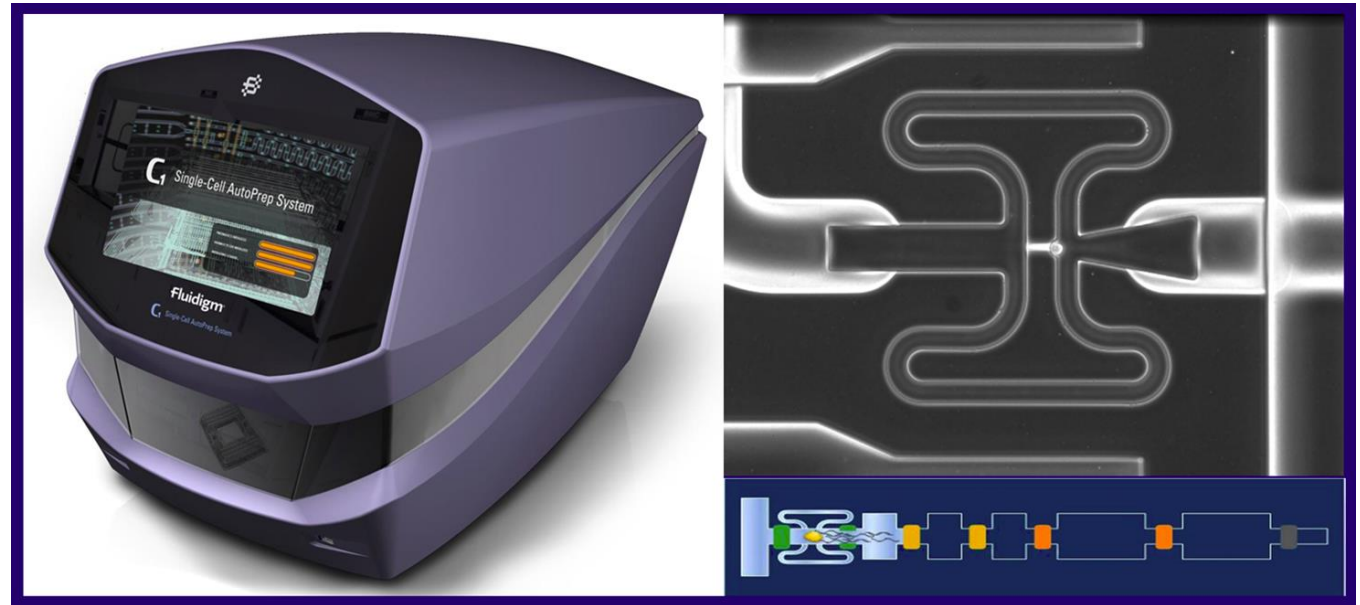
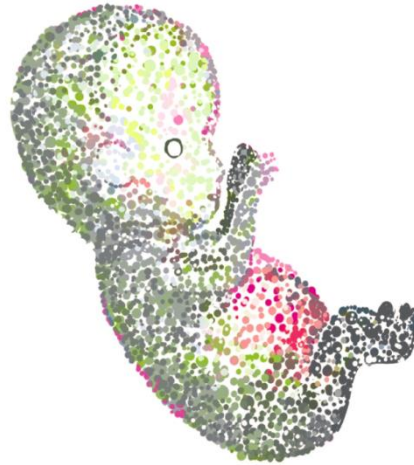
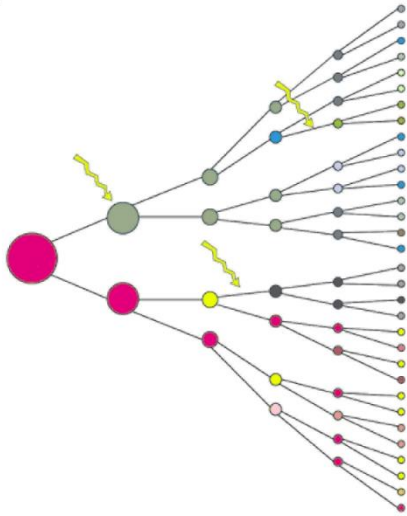
http://openi.nlm.nih.gov/detailedresult.php?img=2682937_CG-9-239_F1&req=4

<http://www.geneticliteracyproject.org/2015/04/15/nutrigenomics-can-diets-be-tailored-to-an-individuals-genetic-make-up/>

Mejoras tecnológicas



Mejoras tecnológicas II



<https://sangerinstitute.wordpress.com/2014/04/28/single-cell-genomics-thinking-small/>

<http://blogs.biomedcentral.com/on-biology/wp-content/uploads/sites/5/2015/04/Single-cell-Leung-et-al-620x342.png>

A quién madruga...

Integrating translational bioinformatics into the medical curriculum

Benedict Yan¹, Kenneth H.K. Ban², Tin Wee Tan²

(2014)

*The increasing adoption of genomic and other high-throughput 'omics technologies in clinical labs requires that healthcare professionals, in particular, physicians be equipped with the knowledge and skills to understand and interpret genomic and other high-throughput 'omics data for patient care. Indeed, this critical need was articulated in a report by the Human Genomics Strategy Group⁴ (established as part of the UK Government's response to the 2009 House of Lords Inquiry into genomic medicine) that recommended "**urgent action**" be taken by the Department of Health to "ensure that workforce developments do not lag behind service developments, and that an appropriately skilled workforce is available".*

...

Accordingly, there is a critical need to provide teaching and training for healthcare professionals, in particular physicians, and this need has been recognized internationally

Money money money

Education

A Quick Guide to Genomics and Bioinformatics Training for Clinical and Public Audiences

Michelle D. Brazas^{1*}, Fran Lewitter², Maria Victoria Schneider³, Celia W. G. van Gelder⁴,
Patricia M. Palagi⁵

(2014)

*Healthcare professionals recognize their limitations in evaluating genomic data and readily seek training opportunities, not to become bioinformaticians as through biomedical informatics programs, but to **become knowledgeable** users who can understand the output from **bioinformatic analyses of genomic data** and competently make datadriven medical decisions.*

*Since big biological data is also predicted to become **big business** [29], direct-to-consumer genomics services (e.g., 23andMe) are spending enormous resources on communicating complex scientific data to their web-savvy clients [30].*

Cuellos de botella

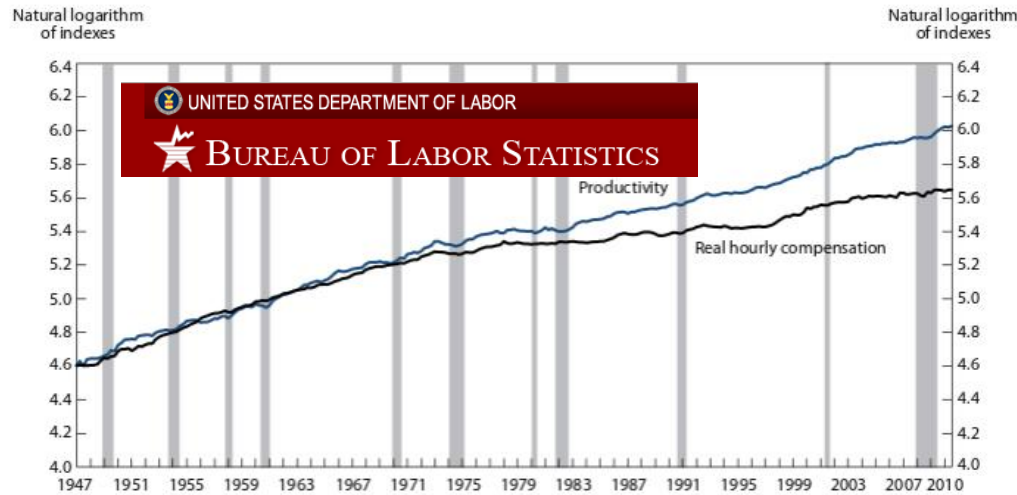
Needs Assessment for Research Use of High-Throughput Sequencing at a Large Academic Medical Center

Albert Geskin¹, Elizabeth Legowski¹, Anish Chakka^{1,2}, Uma R Chandran^{1,2}, M. (2015) Michael Barmada^{3,4}, William A. LaFramboise^{1,2}, Jeremy Berg³, Rebecca S. Jacobson^{1,2,*}

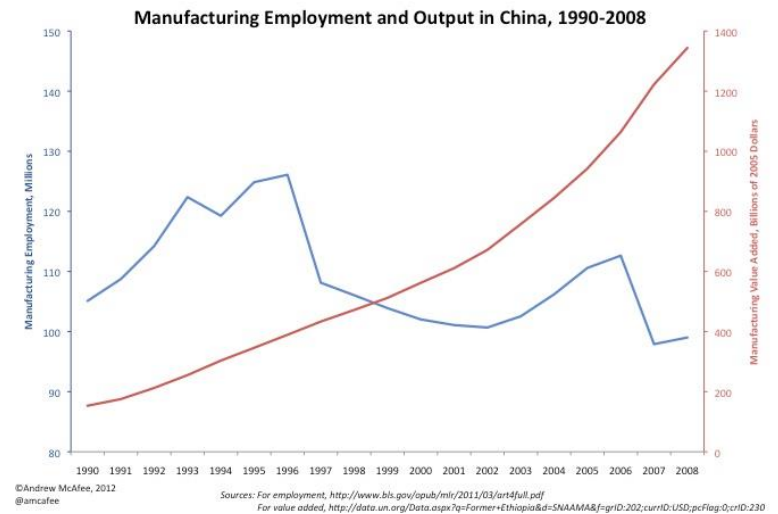
*NGS data offers unprecedented detail; however, the current rate of NGS data generation outpaces the rate at which we are able to analyze it. The complexity of NGS data analysis requires specialized interdisciplinary skills in biology, computing, information technology, and statistics [28, 29]. With the predicted increase in demand for data analysts and the lack of personnel with bioinformatics skills, NGS data analysis could become a significant **bottleneck** in biomedical research.*

Desempleo tecnológico

1. Productivity and real hourly compensation, nonfarm business sector, first quarter 1947–third quarter 2010



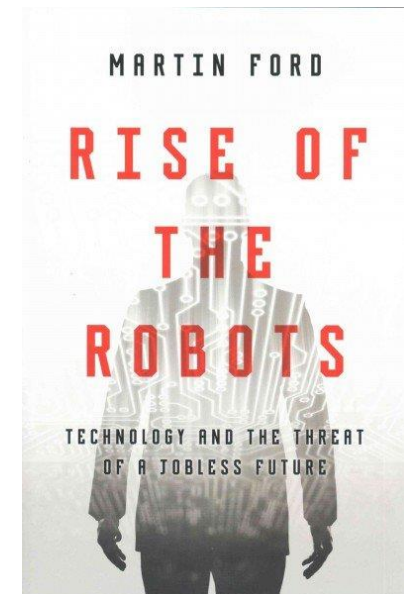
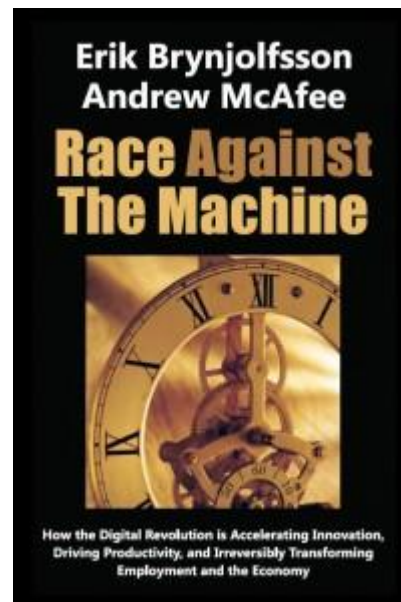
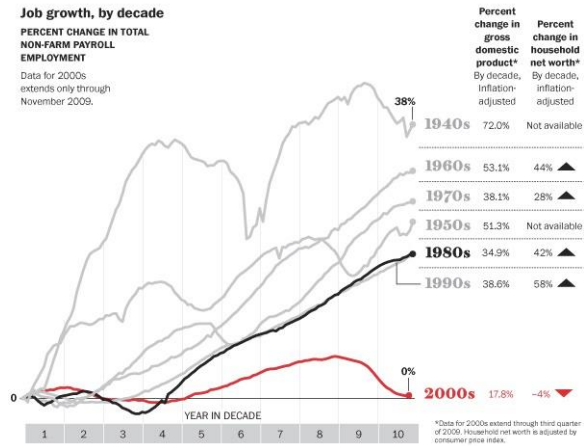
NOTE: The shaded bars denote National Bureau of Economic Research (NBER)-designated recessions.



Job growth, by decade

PERCENT CHANGE IN TOTAL NON-FARM PAYROLL EMPLOYMENT

Data for 2000s extends only through November 2009.



<http://www.bls.gov/opub/mlr/2011/01/art3full.pdf>

<http://www.forbes.com/sites/singularity/2012/07/19/could-automation-lead-to-chronic-unemployment-andrew-mcafee-sounds-the-alarm/>



The Venus Project

GRACIAS!

jmlopezmarti@hotmail.com