Genomic research and precision medicine

Jacques Fellay
EPFL School of Life Sciences
Swiss Institute of Bioinformatics
Lausanne, Switzerland
Genomic medicine: a revolution in the making

Eric Green et al., *Charting a course for genomic medicine from base pairs to bedside*, Nature 2011
Human genomics beyond the clinic

“Invention of the year” 2008:

1. The Retail DNA Test

Press Releases

23andMe Genotypes One Millionth Customer

Direct-to-Consumer Genetic Testing Company Reaches Milestone; Increases Potential for Genetic Research

Mountain View, Calif. – June 18, 2015 – 23andMe, Inc., the leading personal genetics company, today announced it has genotyped more than one million people worldwide. By accessing their own DNA to learn more about themselves, 23andMe customers have helped
The Vatican updated the traditional seven deadly sins with seven new social sins, to bring the list into line with the temptations of the modern world. The additions: bioethical sins, morally dubious experiments that harm human embryos, drug abuse, polluting, social injustice, accumulating excessive wealth and creating poverty.

Man makes life! Or almost. (…) It will be possible generate an endless list of organisms that can perform all sorts of molecular magic, from turning sugar into fuel or digesting oil spills in oceans to even churning out cures for disease.
Genomic medicine

TODAY

• Mendelian diseases (rare functional variants)
• Pharmacogenetics: 170 gene-drug pairs in the FDA “Table of Pharmacogenonomic Biomarkers in Drug Labels”, but only 40 genes involved
• Cancer genomics
• Non-invasive prenatal testing (maternal blood sequencing)
Genomic medicine

TOMORROW

• “Universal sequencing”
• Complex trait genomics (genome data in every health record) – will require an in-depth understanding of functional genomic variation
• DTC genomics brought to doctors
Perspective

• Genomic-based medicine is around the corner
• Genomic-based medicine is only the beginning of “big-data-based” personalized healthcare