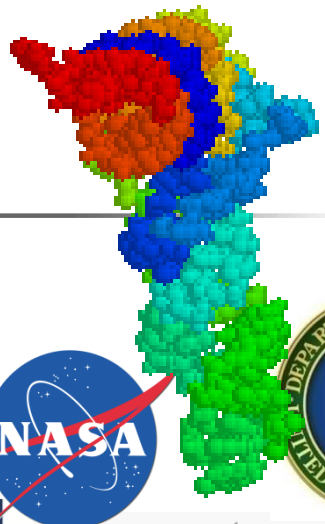
Information Technolgy Future of Medicine

11:13-11:20 AM 5-Jun-2012



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Thanks to:

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NIGMS

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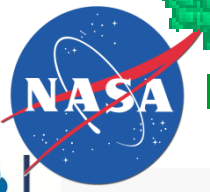
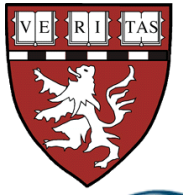
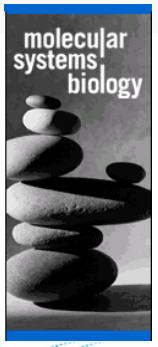






ArmRev.org

PERSONALGENOMES.ORG

Oppenheimer Foundation

Azco

Gen9

Read = = = = = = = = I/O = = = = = = = Write

IT Future of Medicine – Why now?

2008: GINA (HR 493) enables data sharing

2011: Batelle Report: Genome Project
ROI to U.S. economy of 140 to 1

2011: NAS Precision Medicine

2012: Million-fold drop in data costs continues

dels.nas.edu/Report/Toward-Precision-Medicine-Building-Knowledge/13284
battelle.org/spotlight/5-11-11_genome.aspx

US & EU Next-generation technologies → IT

1. Polonator MA
2. Roche-454 CT
3. AB-SOLiD MA
4. Illumina UK,CA
5. CGI CA
6. Helicos MA
7. Pacific Bio CA
8. IntelligentBioSys MA
9. Ion Torrent CT
17. LightSpeed CA
10. Genapsys CA
11. Electronic Biosci CA
12. Nabsys RI
13. OxfordNanopore UK
14. IBM-Roche NY
15. NobleGen MA
16. Genia CA
18. GnuBio MA
19. Bionanomatrix PA
20. Halcyon CA
21. ZS Genetics NH
22. Electron Optica CA
23. Genizon BioSci QC
24. LaserGen TX
25. GE Global NY
26. Stratos Genomics WA
27. Reveo NY
28. Firebird FL
29. Zeiss MA
30. Lucigen WI
31. Adv. Liquid Logic NC
32. Caerus Molec Diag CA
33. Nanophotonics Biosci CA
34. Network Biosystems MA
35. SeiraD NM
36. Affymetrix CA
37. Population Gen Tech UK
38. AQI Sciences AZ
39. Base4innovation UK
40. Li-Cor NE
41. U.S. Genomics MA
42. Mobious Genomics UK
43. Visigen TX
44. Starlight CA

http://arep.med.harvard.edu/gmc/nexgen.html

IT & Genomics impacts treatment & approval

Nic Volker: not intestinal surgery → cord blood
Beery twins: not cerebral palsy → Diet 5HTP
Ivacaftor: treat CFTR G551D → 3 months in FDA

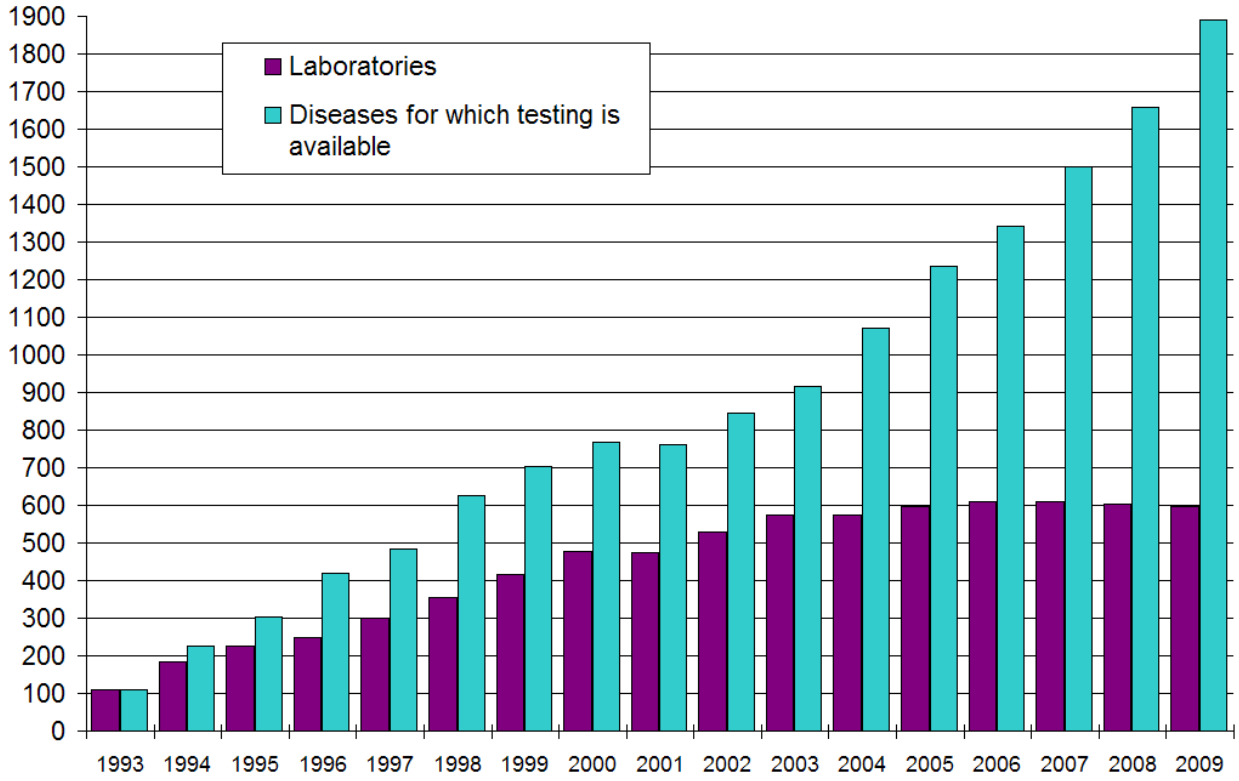
Individually Rare -- Collectively Common (10%)

2641 diseases (~6000 genes) are highly predictive & medically actionable. **Next: Education & IT.**

1963 PKU

1991 BRCA

2010 HCM



Genetests.org PGEd.org

Rare Protective alleles

- **MSTN** -/- Lean muscles <0.001%
- **LRP5** -/+ Extra-strong bones 0.001-8%
- **PCSK9** -/+ Lower coronary disease 3, 0.06%
- **CCR5** -/- HIV-resistant (Pox/Plague) ~0, 1%
- **FUT2** -/- Stomach flu resistant 20%

Embrace the extremes: informative, easy, powerful

blog.personalgenomes.org

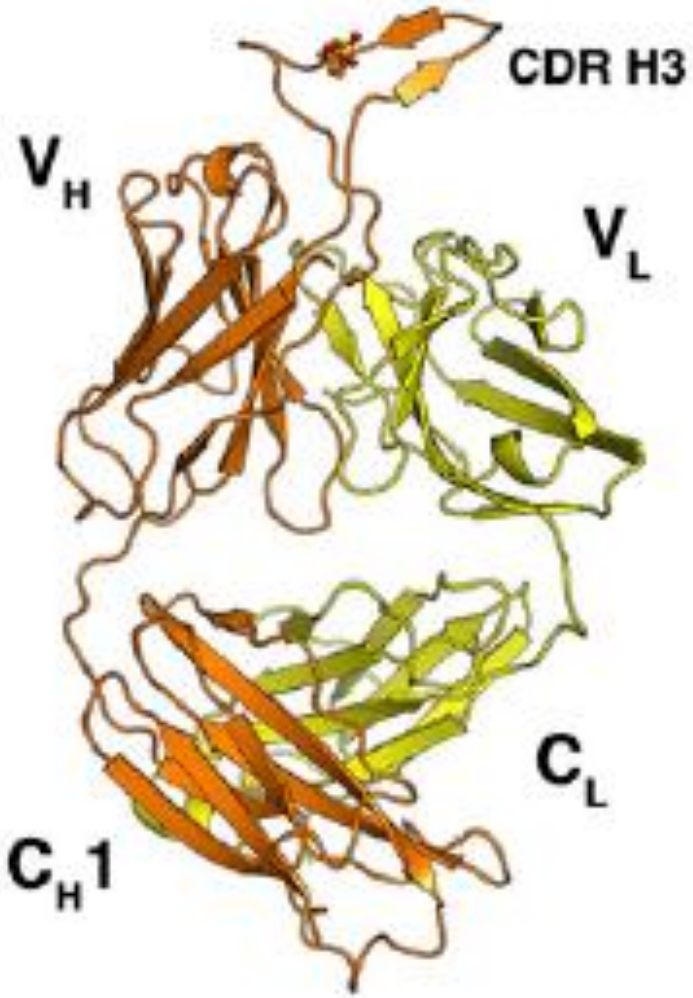
ITFoM Enviromental Data

Microbe tests: Detect Drug resistance spectrum
Earlier warning (e.g. meningitis) 2 to 8 hr

Immune tests: Focus on response to exposure
Longer times to detect exposure (e.g. HIV, TB)

Rare (therapeutic) antibodies

Broadly reactive antibody … potent neutralization of HIV-1 … unusually long, 28-amino acids (84 bp), CDR3… towers above the antibody surface. Pejchala et al. PNAS 2010

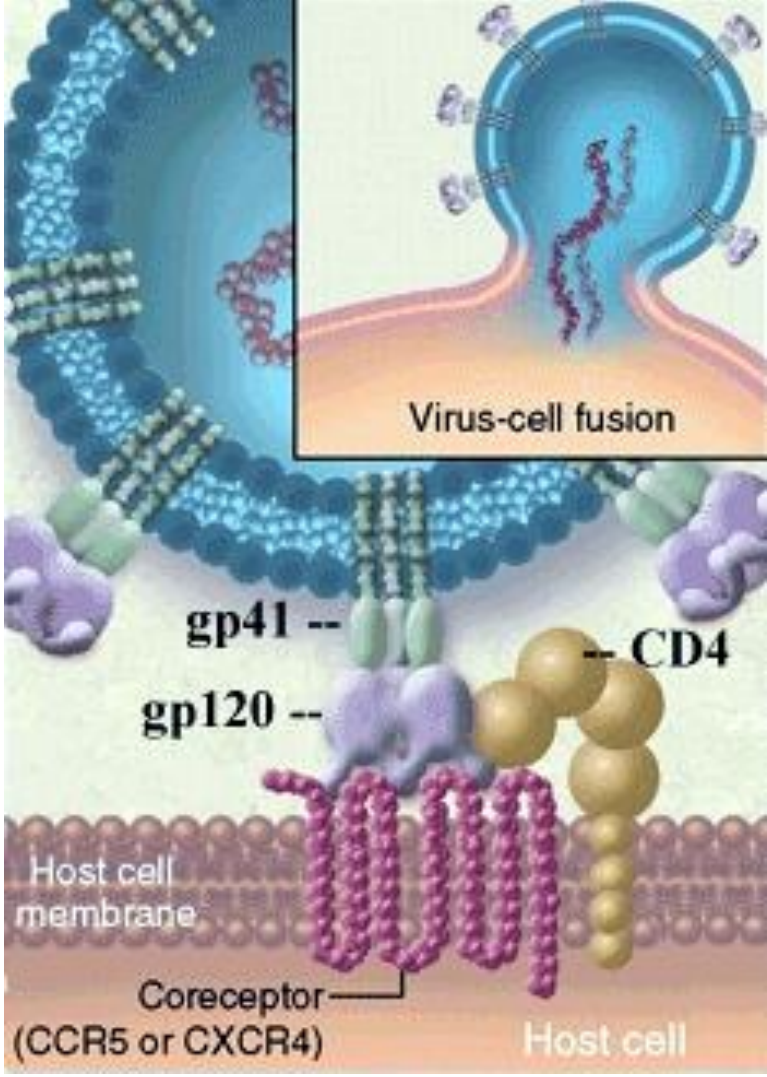


Antibody-based protection against HIV infection by vectored immunoprophylaxis Balazs, Baltimore et al. Nature 2012

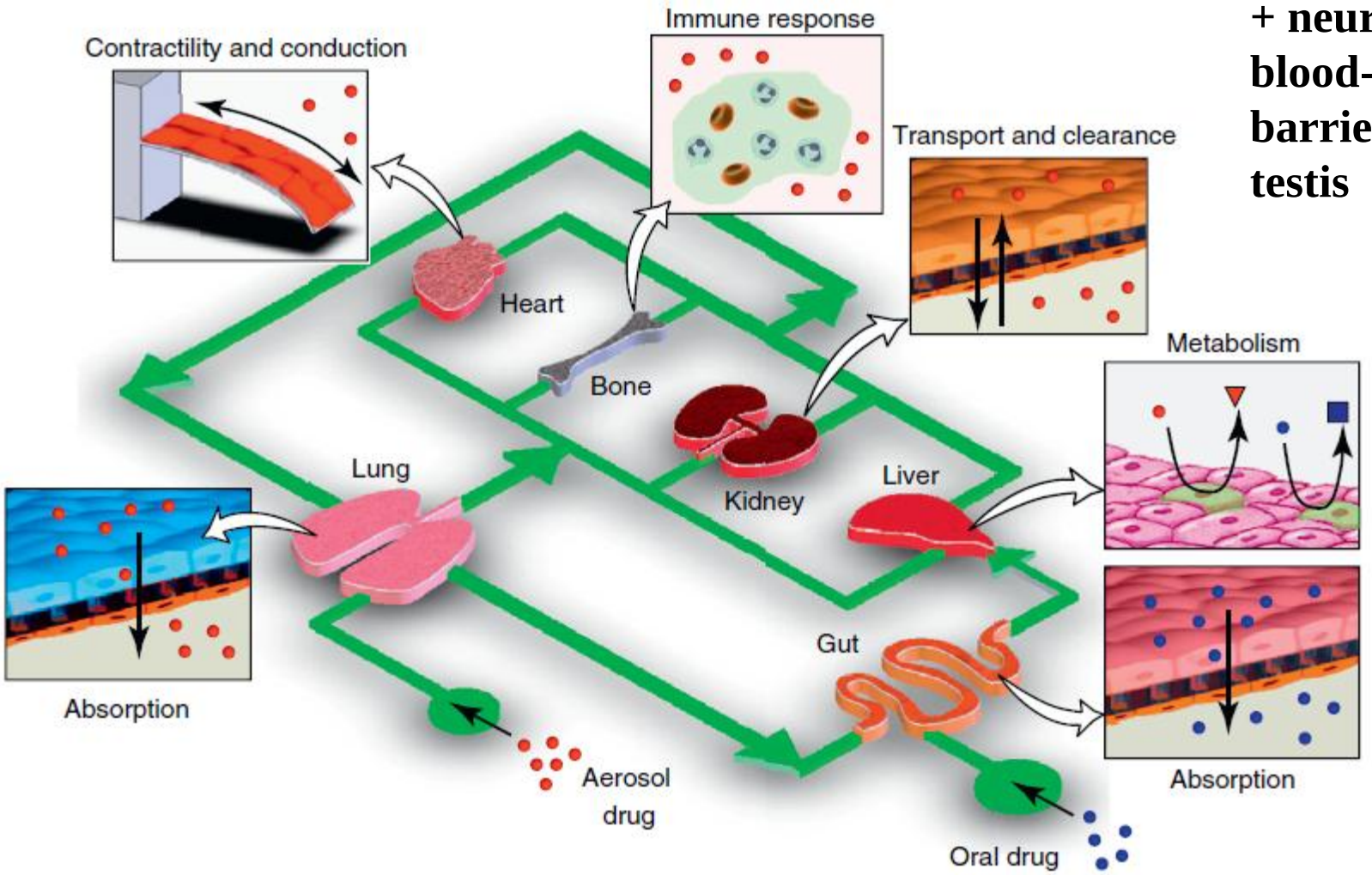
Precise Genome Therapy: prevent/cure HIV

"Long-Term Control of HIV by CCR5 Δ32/Δ32 Stem-Cell Transplantation" 2009
New England J Medicine

Sangamo Phase 2 clinical trial



New data sources: Personalized organs-on-chip



+ neural, blood-brain-barrier, skin, testis

WYSS INSTITUTE

Huh, Ingber et al. Science. 2010
Trends in Cell Biol 2011

Genomes = Traits

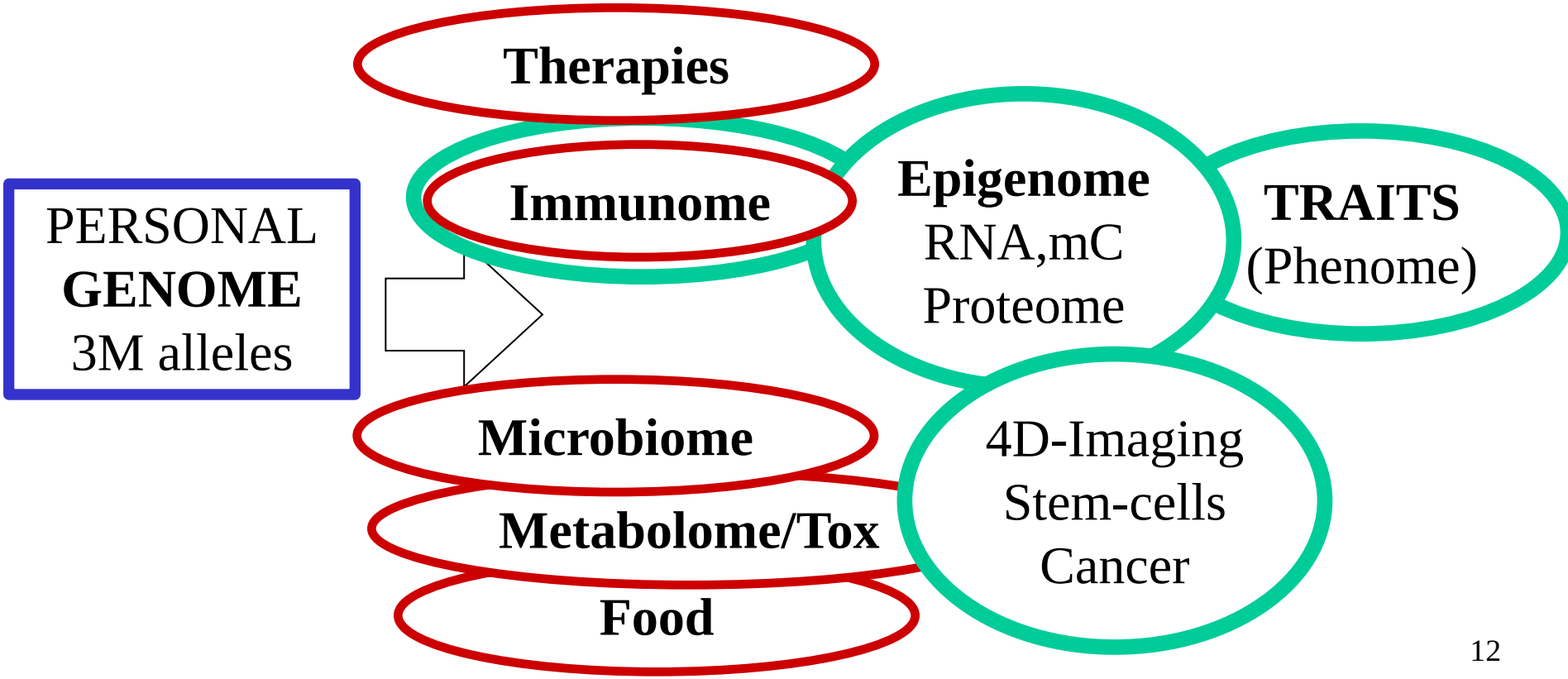
Personal Genome Project

Testing new technologies
Cohorts approved for sharing

PERSONAL
GENOME
3M alleles

TRAITS
(Phenome)

Genomes + Environments = Traits

Personal Genome Project

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