Genomics delivers

FDA approves new cholesterol lowering drug
New drug targets

Tailored therapies
Precision medicine

Predictive and preventive medicine

The 'Angelina Jolie' effect on breast cancer and BRCA
The virtuous cycle of Genomic Medicine

Individual | Population

- Governance and ethics
- Volunteers / Patients
- Clinical data / samples
- Biobank
- Genomic / *omic analyses
- Big data
- Environmental data
- New database
- New knowledge
- Clinical trials / Pharmas
- External database
- Clinical utility
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- Feedback to patients and family
- Preventive medicine
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Primary objective:
Identify genome associations with common cardiovascular, psychiatric and metabolic conditions:
→ New drug targets

Method:
• Population-based design
• 6'000 adult participants
• Extensive phenotype
• GWAS (n = 6000) + exome (500)

Output:
→ New genomic regions associated with a variety of conditions
→ 12-year follow-up (funded by SNF)
→ 250+ publications
From Genomics to Genomic Medicine

- Biobank
- Clinical data / Biological samples
- Volunteers / Patients
- Governance and ethics
- New drugs regulatory approval
- Preventive medicine
- Tailored therapies
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Overcoming The Last Mile Challenge: Distributing Value To Billions
CoLaus and the Lausanne Institutional Biobank (BIL)

CoLaus: Population-based cohort

BIL: Hospital-based cohort

Observation cohort

Intervention cohort
Lausanne Institutional Biobank (BIL)

A state-of-the-art bioresource for Genomic Medicine:

- General consent: includes possibility to perform whole genome sequencing + recontact participants
- Repository for DNA samples and data;
- 1st systematic hospital-based biobank in Switzerland
As per September 12th 2016

A clear expression of solidarity:

- 34’220 patients contacted;
- 25’212 patients signed the general consent (74%);
- 85% wish to be contacted again for clinically actionable incidental findings.
The Clinical Research Support Service (SRC)

An integrated infrastructure to accelerate research and knowledge transfer from lab to patient;

Regroups under the same roof:
- BIL
- Pre-analytical laboratory
- Clinical research center
- Dedicated DW (under construction)
A datawarehouse dedicated to clinical research (under construction)

Clinical data → DWH RC
- Patient ID
- Clinical data
- Genetic data

Depersonalization

Genetic data → Data shared with investigators

Clinical data

Genetic data

Data shared with investigators
Proof-of-concept Study:
Familial hypercholesterolemia (FH)

Biobank participants: n = 17760

Subjects willing to be re-contacted:  
 n = 13094

Subjects with at least 1 value of  
 total/LDL-cholesterol: n = 5111

Subjects with CAD before  
 55/60 (M/F) and  
 TC > 7.5 mM: n = 25

Subjects without CAD, TC > 7.9 mM  
 and/or LDL > 3.6 mM: n = 69

Sequencing LDLR, PCSK9 and ApoB  
 genes, n = 94

3 mutations known to cause FH

APOB
PCSK9
LDLR

Subjects (1-94)
Translating Genomics into the clinic:
Planned applications

- Prevalence of disease-causing variants
- Penetrance of disease-causing variants
- Biomarker discovery and validation
- Phenotypisation of Human KO
- Genetically enriched clinical trials
- Prevention - MedOmics
Translating Genomics into the clinic:
Sustainable health

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<th>Prevalence of disease causing variants</th>
<th>CAD / FH</th>
<th>T2DM</th>
<th>Cancer</th>
<th>AMD</th>
<th>AD/PD</th>
<th>Others</th>
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<td>Penetrance of disease-causing variants</td>
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- Unmet medical needs;
- Partly heritable;
- Actionable;
- Amenable to monitoring;
- Strong expertise locally
Genomics: back to patients and their families

• Task force on restitution of genomic information to participants, funded by Vaud Ministry of Health → robust and ethical framework;

• Development of a new multidisciplinary genomic medicine clinic (MedOmics) : pilot phase;

• Development of a Health 2030-funded training and continuing education on genomics clinical implications.
Conclusion

• Genomic medicine = strategic priority at CHUV/UNIL
• Pioneering initiatives - unique combination of population and hospital based cohorts;
• One-stop-shop for translation of genomics into the clinic and society, anchored in an hospital environment ;
• Resource to be constructed with, and made available to, Health 2030 and Swiss Personalized Health Network community.
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