Innovaciones en la Información de la Salud
El papel de la biblioteca hospitalaria en la Medicina de Precisión

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Challenging times....

A transformation is required, with multiple dimensions

European Healthcare systems are no longer sustainable...
Data-driven innovation

Prevention and early risk detection
New therapies and diagnostics
Personalised Medicine
New health and care models

High-performance computing
Cloud computing
Trust
Artificial Intelligence
Advanced Data-Analytics
Internet of Things (IoT)
Wearables
Telehealth

User driven-innovation
Personalised medicine

"Personalised medicine refers to a medical model using characterisation of individuals’ phenotypes and genotypes (e.g. molecular profiling, medical imaging, lifestyle data) for tailoring the right therapeutic strategy for the right person at the right time, and/or to determine the predisposition to disease and/or to deliver timely and targeted prevention. Personalised medicine relates to the broader concept of patient-centred care, which takes into account that, in general, healthcare systems need to better respond to patient needs."

"Council conclusions on personalised medicine for patients" of 7 December 2015 (2015/C 421/03)
Recent history of EU activities on Personalised Medicine

2011: European Perspectives conference
2013: Commission Staff Working Document on "use of '-omics' technologies in the development of personalised medicine"
2015: Council conclusions on Personalised Medicine
2016: Personalised Medicine Conference & Launch of International Consortium of Personalised Medicine (ICPerMed)
2018: ICPerMed 1st Conference in Berlin
40 members, including:

- Research funders
- Health ministries
- Science and Education Ministries
- Regional authorities
Actions in support of ICPERMed

**HEcoPerMed**: To addresses a research need identified by ICPERMed in their Action Plan, namely to support health economics research and assessments of available and newly developed PM approaches.

**EULAC-PerMed**: To support the international dimension of ICPERMed, reaching out to new potential partners; promoting international standards and underpinning the EU-CELAC political dialogue.

**SAPHIRE**: To establish an inclusive network and support interregional collaborations, identifying needs and activities in line with ICPERMed and smart specialisation strategies.

**Regions4PerMed**: To support the regional dimension of ICPERMed and promote personalised healthcare through cooperation and coordination, creating a participatory approach and channelling investments towards Personalised Health.
**Personalized medicine** refers to an approach to patients that considers their genetic make-up but with attention to their preferences, beliefs, attitudes, knowledge and social context.

**Precision medicine** is "an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person."
Obama anuncia un plan para financiar la “medicina de precisión”
El objetivo de la Casa Blanca es crear una base de datos genéticos de un millón de ciudadanos

Medicina personalizada y de precisión
Antonio Alarcó, portavoz del PP en la Comisión de Sanidad del Senado

La creación de Rediex va a potenciar la medicina personalizada en España

La medicina de precisión, más allá de la teoría...
ESTRATEGIAS DE INNOVACIÓN

Estrategia para una transferencia eficiente en ISPA/ FINBA
Health & Healthcare Innovation

- Evidence-Based Medicine
- European Alliance for Personalised Medicine
- Genomic Medicine
- Precision Medicine
- Stratified Medicine
- P4 Concept

Regenerative Medicine:
- Tissue Engineering
- Biomaterials
- Stem Cell Therapy

Advanced Therapies in Healthcare

Drug Discovery Today: Therapeutic Strategies
Health & Healthcare Innovation
e Health, Data Science & Advanced Analysis Platform

- Applied Bioinformatics
- Cybersecurity
- Big Data
- Personalised Precision Medicine (PPM)
- Internet of Things (IoT)
Genome Sequence

Massive Analysis of SNPs

Reference assembly human genome (build 39)

5'-TGCACGCTGATCATC..........AGATAGCTATACATCGC-3'

SNPs

5'-TGCACGCTGATCATC..........AGATAGCTATACATAGCG-3'

3096 MGB

36299 GENES

1% (3 millions) 85% of known diseases

750 megabytes
Estimations: 20,000 to 100,000.
Now: 24,500? (3000 pseudogenes?)
50% mRNAs do not code for proteins (mouse)
50% display alternative splicing
25%-60% unknown

Genes in the DNA...

...code for the structure of proteins...
That undergo post-translational modifications

From genotype to phenotype...

...which accounts for the function...

(Only the genetic component...)

...conforming complex interaction networks...

A typical tissue is expressing among 5000 and 10000 genes

...providing they are expressed in the proper moment and place...

Each protein has an average of 8 interactions

...in cooperation with other proteins...

...whose final effect can be different because of the variability.

More than 3.5 million SNPs have been mapped

Adapted from J. Dopazo
**Systems biology** is the study of biological systems as collections of networks at multiple levels, ranging from the molecular level, through cells, tissues and organisms, to the population level.

**Systems medicine** is the application of systems biology to human disease by using high throughput technologies – such as DNA and RNA sequencing – to produce global data sets tracking multiple dimensions of dynamic network interaction.
Three converging megatrends driving the transformation of healthcare

- Systems biology and systems medicine
- P4 medicine
- Digital revolution
- Consumer-driver healthcare and social networks
**Personalized medicine** refers to an approach to patients that considers their genetic make-up but with attention to their preferences, beliefs, attitudes, knowledge and social context.

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Reactive healthcare: based on limited measurements from relatively small test cohorts

Present-day omics studies: complex data sets consisting of many measurements of relatively small test cohorts

P4 healthcare: based on billions of data points about multiple dimensions of health of each member of the population

Number of people

Number of data dimensions (e.g., genomic and proteomic, among others)
GENES → PROTEINS → FUNCTIONS

P4 Medicine
- PREDICT
- PREVENT
- PERSONALIZE
- PARTICIPATE

PREDICTION → PRECISION
Data origen

GENES ➔ PROTEINS ➔ FUNCTIONS

SNPs-GWAS
WES
Microsatellites
Epigenomics
Microbiome

RNAseq
Arrays
microRNA

Proteome Pathways
Interactome Metabolome
DNA PROTEINS

FUNCTIONS

Next Generation Sequencing (NGS) + Bioinformatics

- Personal Genomics
- Personal enviromics
- Personal genetic modifiers
- Pharmacogenomics
- Nutrigenomics
- Microbiome
Bioinformatics

Coronary artery disease

Obesity

Osteoarthritis
1. Familial Dilated Cardiomyopathy (FDCM) n=6

2. Familial Generalized Vitiligo and Autoimmune Comorbiditis (FGVAC) n=6

3. Familial Anterior Cruciate Ligament injury (FACL) n=4

4. A “healthy” family, n=6

Clinical status of family members. Circles indicate females; squares, males; and a slash, deceased. Filled symbols indicate affected status and open symbols unaffected.

Exploring Personal Genomics In Familial Multifactorial Genetic Diseases Analyzed By Integrative Targeted Whole Exome Sequencing Analysis
Primary audiences (PA) (more important) and secondary audiences (SA) (less important). End users: As shown in following table there are different ultimate dissemination end users

<table>
<thead>
<tr>
<th>End users</th>
<th>Utility of results</th>
<th>Promoting users interest</th>
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<td>SA</td>
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<td>Health care providers</td>
<td>PA, Interpretation, Communication</td>
<td>PA</td>
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<td>Ethical and Legal professionals</td>
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<td>Patients</td>
<td>PA, Comprehensive personal understandin g</td>
<td>PA</td>
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<td>PA</td>
</tr>
<tr>
<td>Individuals (society)</td>
<td>SA, Comprehensive Social understandin g</td>
<td>PA</td>
<td>SA</td>
<td>PA</td>
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</tbody>
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The Precision Medicine Growing Ecosystem

Patients <-> Clinicians

Electronic Health Records

Biospecimens

Family History
Exposomes
Envirome
Genotypes
Phenotypes
Outcomes

Curated Database

Researchers <-> Research Data <-> Clinical Labs

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Researchers ➔ Research Data ➔ Clinical Labs

Data Management Roles for Hospital Library

Data management for informed decision making
Scholarly research product
Exploring data use and data support

MUCHAS GRACIAS