

Precision Medicine

Ferran Sanz



RESEARCH
PROGRAMME
ON BIOMEDICAL
INFORMATICS



Institut Hospital del Mar
d'Investigacions Mèdiques



Universitat
Pompeu Fabra
Barcelona

What is Precision Medicine?

According to the Precision Medicine Initiative, 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."

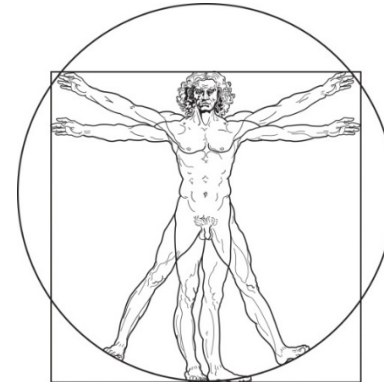
This approach will allow doctors and researchers to predict more accurately which treatment and prevention strategies for a particular disease will work in which groups of people. It is in contrast to a one-size-fits-all approach.

Reproduced from <https://ghr.nlm.nih.gov/primer/precisionmedicine/definition>

Knowledge management in disease genomics



genotype



phenotype



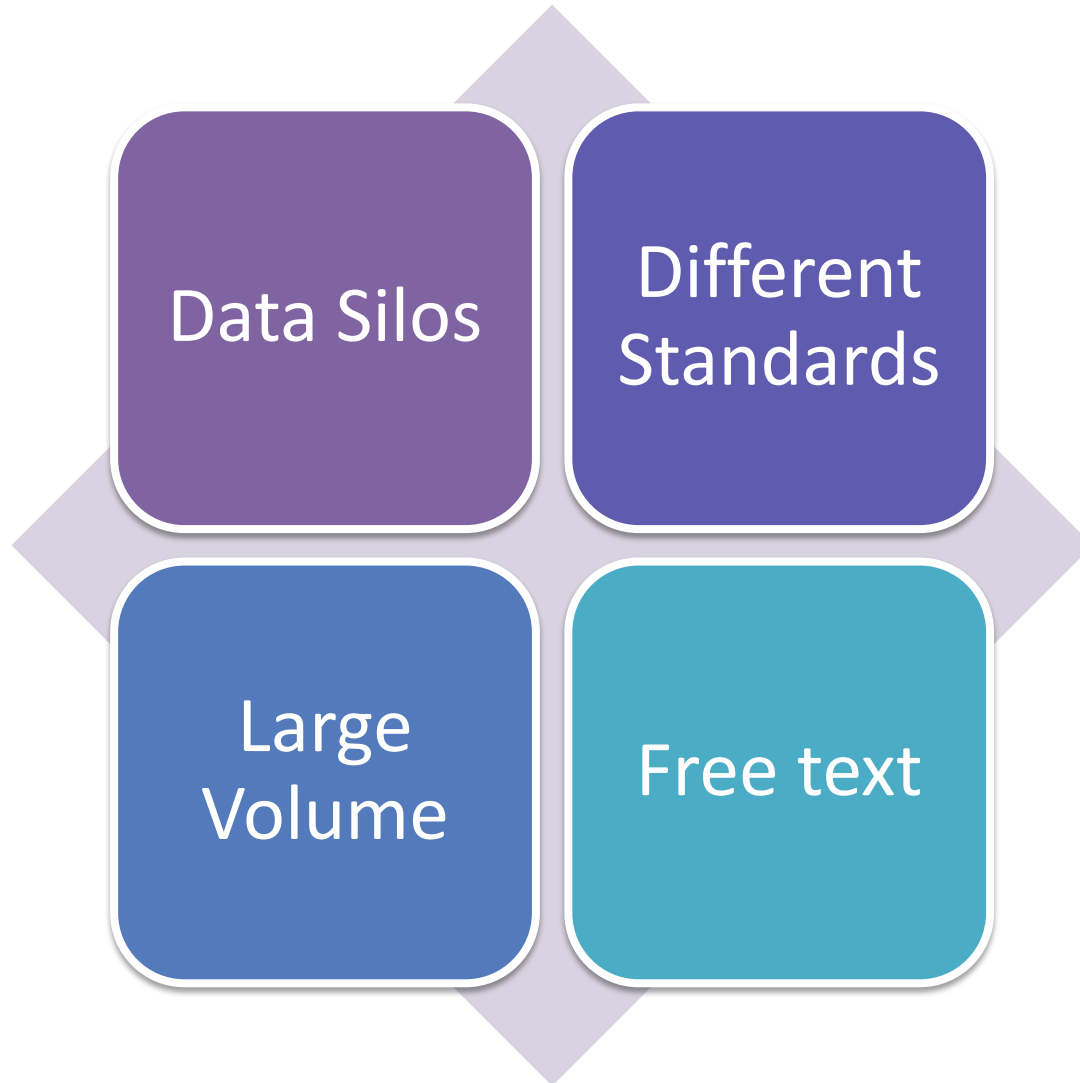
2,000 genes, 1,700 mutations
4,000 publications

800 genes, 600 mutations
1,500 publications

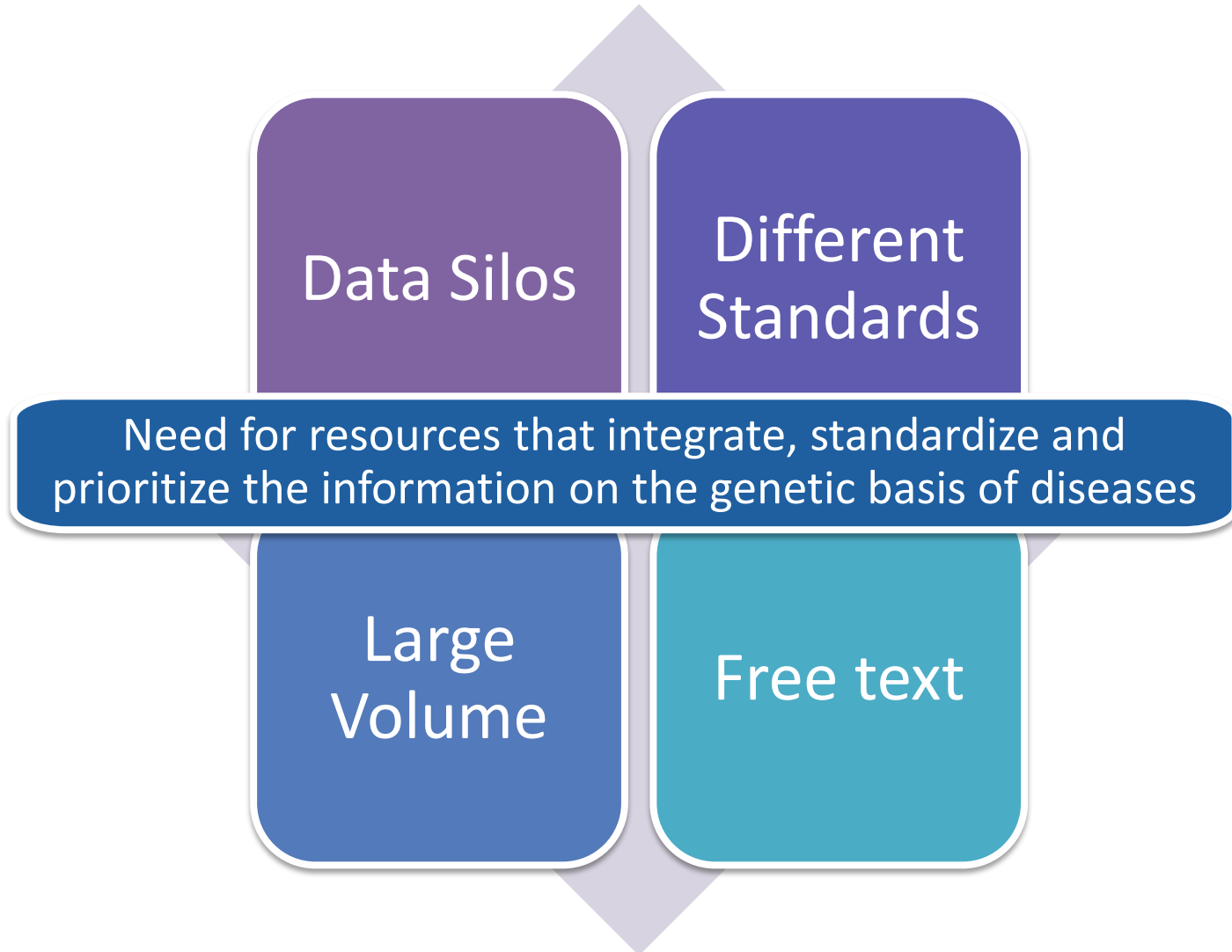
Schizophrenia

Bipolar disorder

Knowledge management in disease genomics



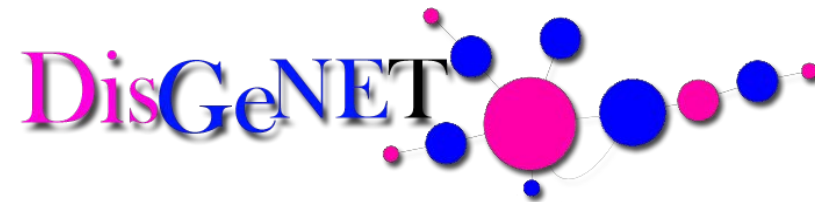
Knowledge management in disease genomics





Knowledge platform on disease genomics

- The most comprehensive catalog of human disease genes and variants
 - Covers the entire spectrum of human diseases: rare (Mendelian) and complex
 - 600,000+ associations between genes and diseases
 - 100,000+ associations between variants and diseases
 - Running for about 10 years (six versions released)
- State-of-the-art text mining approaches
 - 70% data extracted by text mining and unique to DisGeNET
- Information standardization using controlled vocabularies and ontologies
 - Allows automatic linkage and integration with other data sources
- Tools to prioritize and filter the information
- Community recognition:
 - 20,000+ users per year
 - 50,000+ visits per year
 - 300+ bibliographic citations



<http://www.disgenet.org/home/>



RESEARCH
PROGRAMME
ON BIOMEDICAL
INFORMATICS



Institut Hospital del Mar
d'Investigacions Mèdiques



Universitat
Pompeu Fabra
Barcelona