



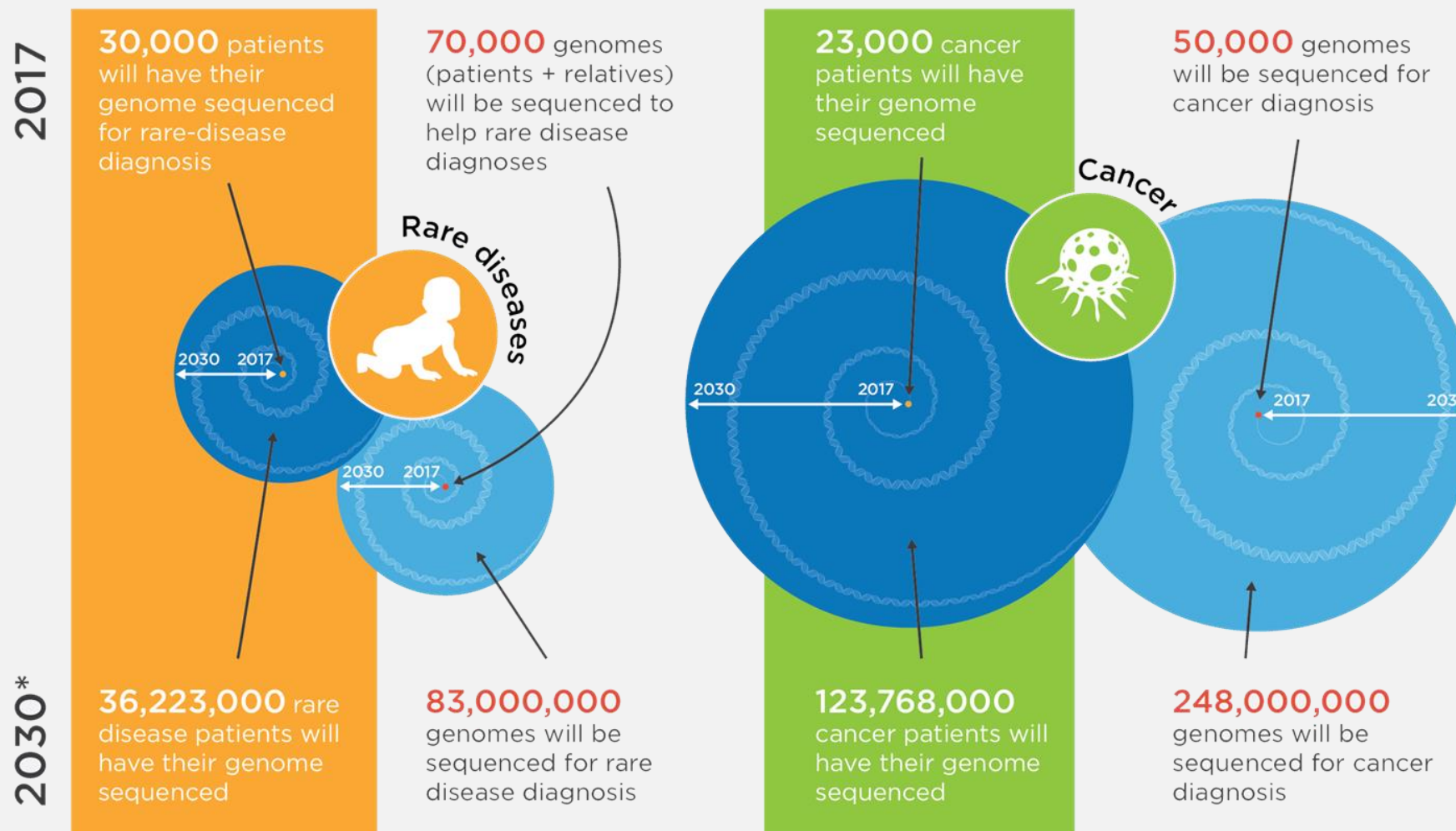
Genomics informatics and standardization from a Swedish point of view

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Nordic Workshop 11th of June, HIMSS Helsinki 2019



* Projected figures, based on current data and known status of genomics initiatives worldwide.

From Op-ed on BioRxiv: doi.org/10.1101/203554

Why standardize?

General benefits;

- Shows best practice
- A common solution to recurring problems.
- Increase patients safety
- Safer data sharing
- Common global terminology
- International frameworks and achitecture

Swedish, Nordic and European benefits;

- Sweden goals: e-Health 2025 and a Life science strategy
- The Nordic Alliance for Clinical Genomics
- European '1+ Million Genomes' Initiative

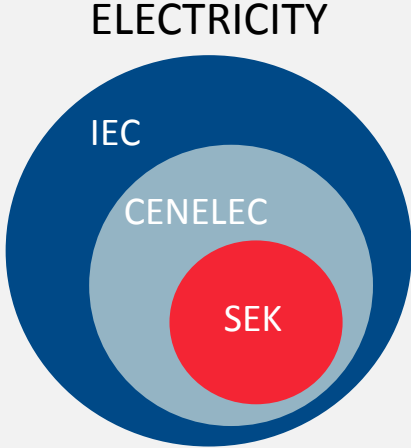
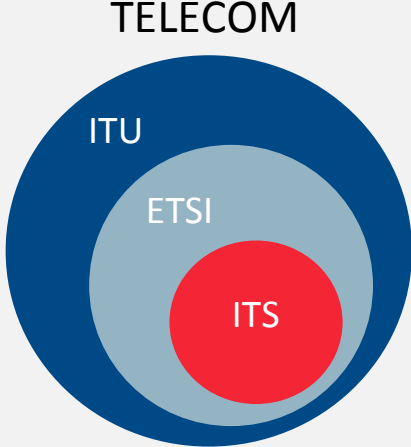
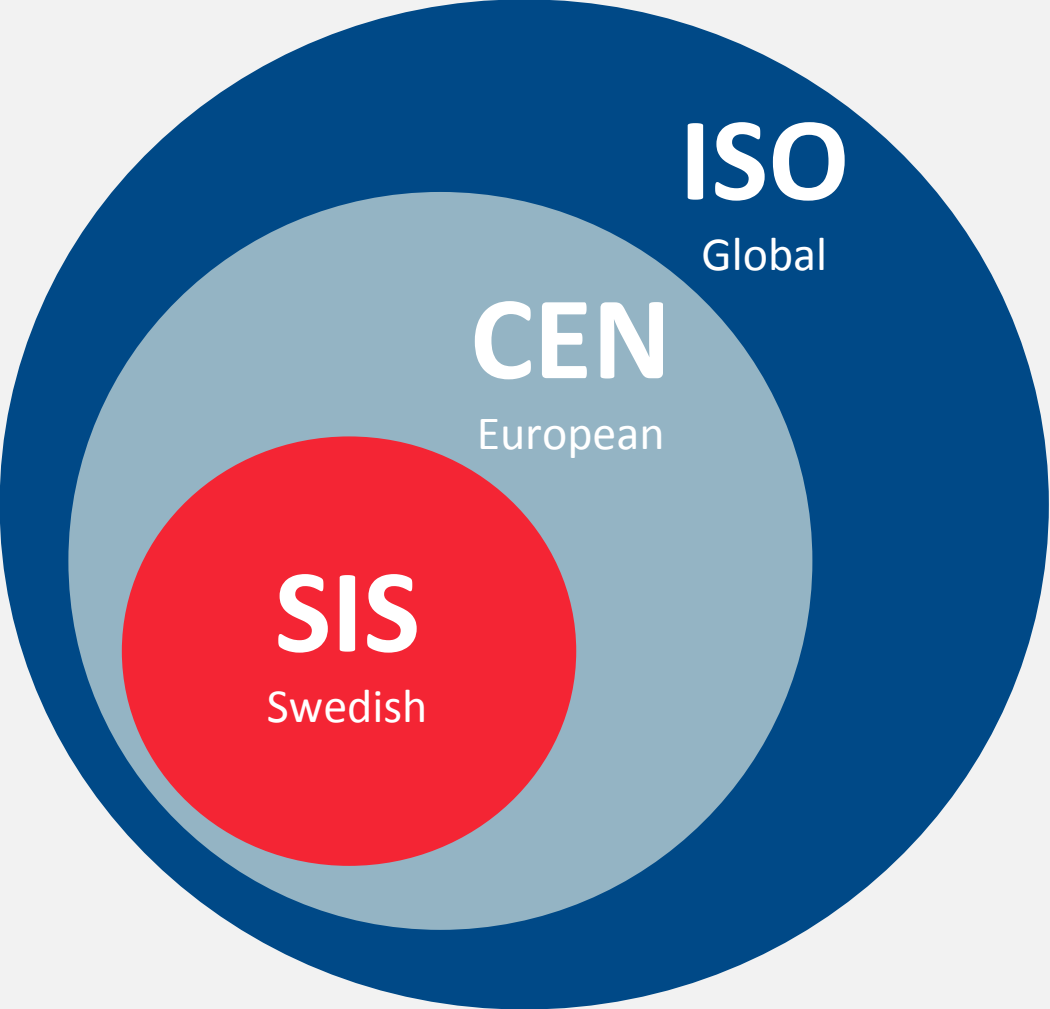
Medical Genomes



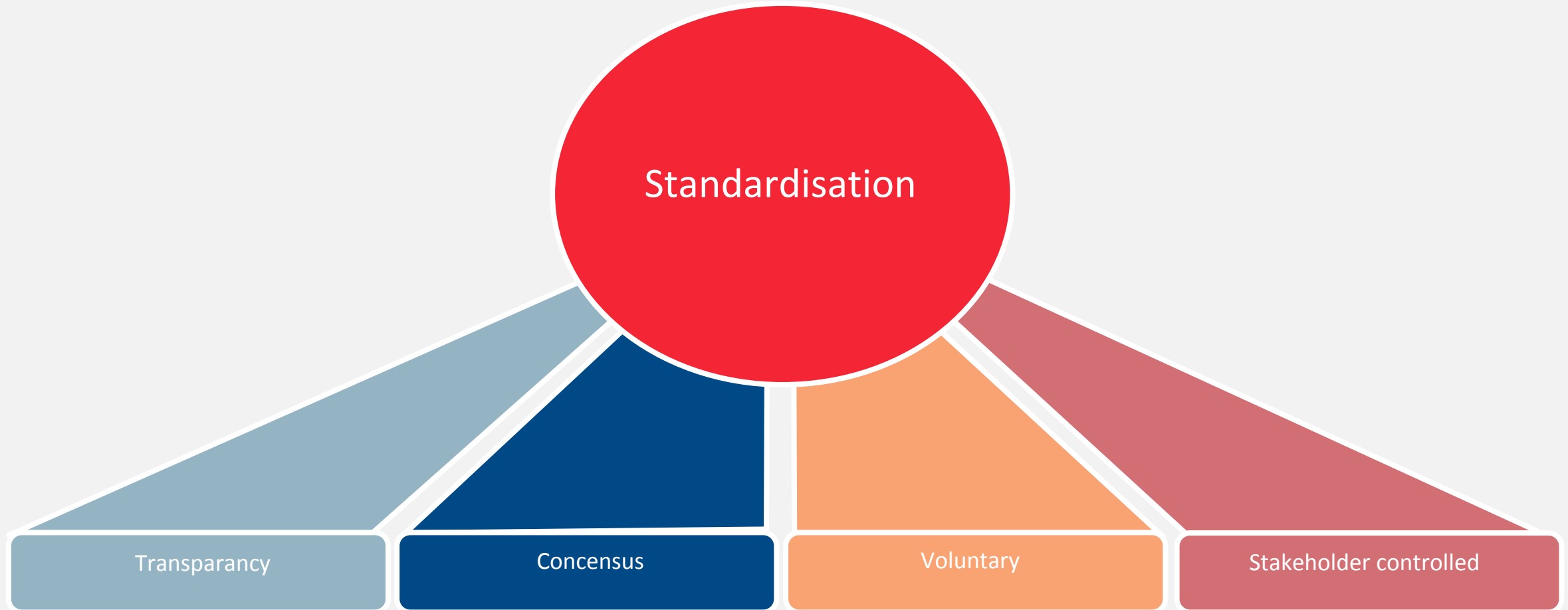
Global Alliance
for Genomics & Health

Countries with active national medical genome projects	Countries with some activity of medical genomes	Countries planning medical genome projects
Australia Denmark Estonia France Iceland Saudi Arabia UK USA Japan China Turkey	Canada Qatar Spain Netherlands Switzerland Finland Brazil Ireland Scotland South Africa Norway Sweden	India Jordan Kuwait Singapore South Korea United Arab Emirates Russia Argentina

Standardisation – local and global



Principles



ISO, CEN and the Nordic NSB's

- Have common terminologies
- Standards that collaborate within the field
- A system for new work proposals, revisions and publication
- Huge network
- Also collaborating with legislative systems as EU, FDA and others

Challenges

- Data is typically in silos: by type, by disease, by country, by institution
- Analysis methods are non-standardized; few at the scale needed for genomics
- Approaches to regulation, consent and data sharing limits interoperability
- Data volume for many millions of patients' genomes...
- There is an overwhelming mass of fragmented data, such as electronic medical records in many countries

Principles of data sharing

- International data sharing must be enabled; Global Standards are paramount
- Promote sharing across the translational continuum (basic science and discovery research, clinical trials, clinics, diagnostic labs, industry)
- Promote interoperability: scientific, technical, and ethical
 - **Adopt Global Standards**; have transparent documentation
 - **Standardized file formats**, bioinformatics & variant calling protocols, variant annotation
 - **Consent policies to enable data to be shared internationally**

ISO/TC 215 Health Informatics

Scope: Standardization in the field of health informatics to facilitate the coherent and consistent interchange and use of health-related data, information, and knowledge to support and enable all aspects of the health system

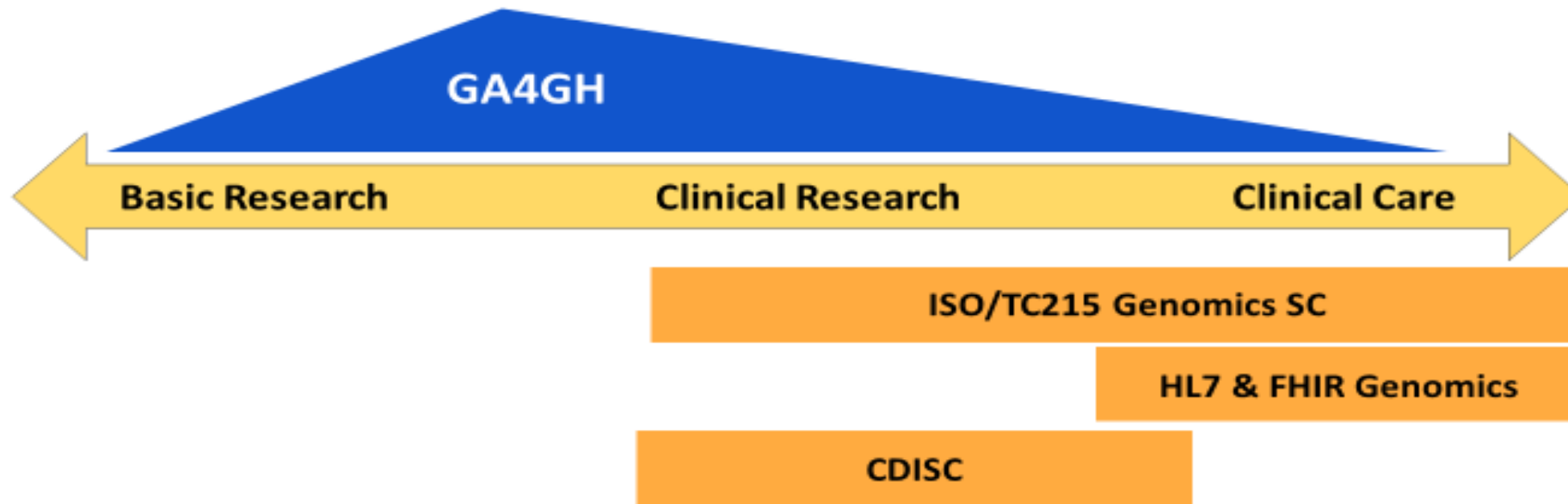
Membership

- ISO has over 200 members worldwide
- **31 Participating Countries**; One Country = One Vote
- SE, FIN, DK, NO
- 29 Observing Countries

Liaisons

- 32 ISO/TC liaisons
- 19 External liaisons
- **Sub committee : Genomics informatics starts during 2019**

Cross-SDO Genomics



ISO Genomics projects

Korea

- ISO/Technical Specification 20428 (Published): Data elements and their metadata for describing structured clinical genomic sequence information in electronic health records
- ISO/AWI TS 22693: Structured clinical gene fusion report in electronic health records
- ISO/DTS 22692: Quality Control Metrics for DNA Sequencing
- ISO/PWI TS 23357: Clinical genomics data sharing specification for next generation sequencing

Japan

- ISO/DIS 21393: Omics Markup Language
- ISO/AWI 21394: Whole Genomic Sequence Markup Language
- ISO/IS 25720: Genomic Sequence Variation Markup Language

China

- ISO/PWI 22690: Reliability assessment criteria for high-throughput gene-expression data



Thank you for the attention!

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