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I.B.AHC

Biobank and Clinical Database for Alternating Hemiplegia
www.ibahc.org

A Proposal for Networking European Biobanks on AHC

Filippo Franchini

I.B.AHC Project Manager



A.I.S.EA Onlus

Italian Association for Alternating Hemiplegia
Via Sernovella, 37 – I - 23878 Verderio Superiore (LC)
www.aiseaonlus.org

Abstract

- Alternating Hemiplegia (a rare disease)
- The I.B.AHC Biobank and Clinical registry
 - Structure description and key issues
 - The architecture
 - The solutions
 - The outcomes
- The situation in Europe
- A proposal for networking
- Closing remarks

Alternating Hemiplegia

A (very) Rare Disease

- Average incidence **1/1.000.000** (EU rare disease definition $< 5/10.000$).
- Less than 200 cases in Europe, about 500 worldwide.
- Widely undiagnosed or misdiagnosed (no data for China, India, Africa; poor data for other countries).
- Clinical characterization strongly suggests a genetic cause -> **Biobanking required!!!**
- Scarce clinical and genetic research.

The I.B.AHC Project

Biobank and Clinical Registry on AHC

- Project **started October 2001**, still fully operational.
- Multilanguage **clinical database** containing the data of 38 AHC patients.
- **Biobank** containing the samples of 40 AHC patients and their parents.
- **Entirely funded and actively supported by A.I.S.EA Onlus** in close collaboration with its Scientific Committee.

The I.B.AHC Project

Structure description

- **Personal database:** personal information about I.B.AHC participants (patients, relatives, treating physicians), used for coding and information exchange.
- **Clinical database:** structured collection of clinical data of AHC patients, validated and constantly updated.
- **Biobank:** repository of biological samples (DNA, cell lines) belonging to AHC patients and their parents.

The I.B.AHC Project

Key issues

- **Operational issues**

Standard practice at any step of the process (subscription, data collection, blood samples collection, follow-ups)

- **Privacy and security issues**

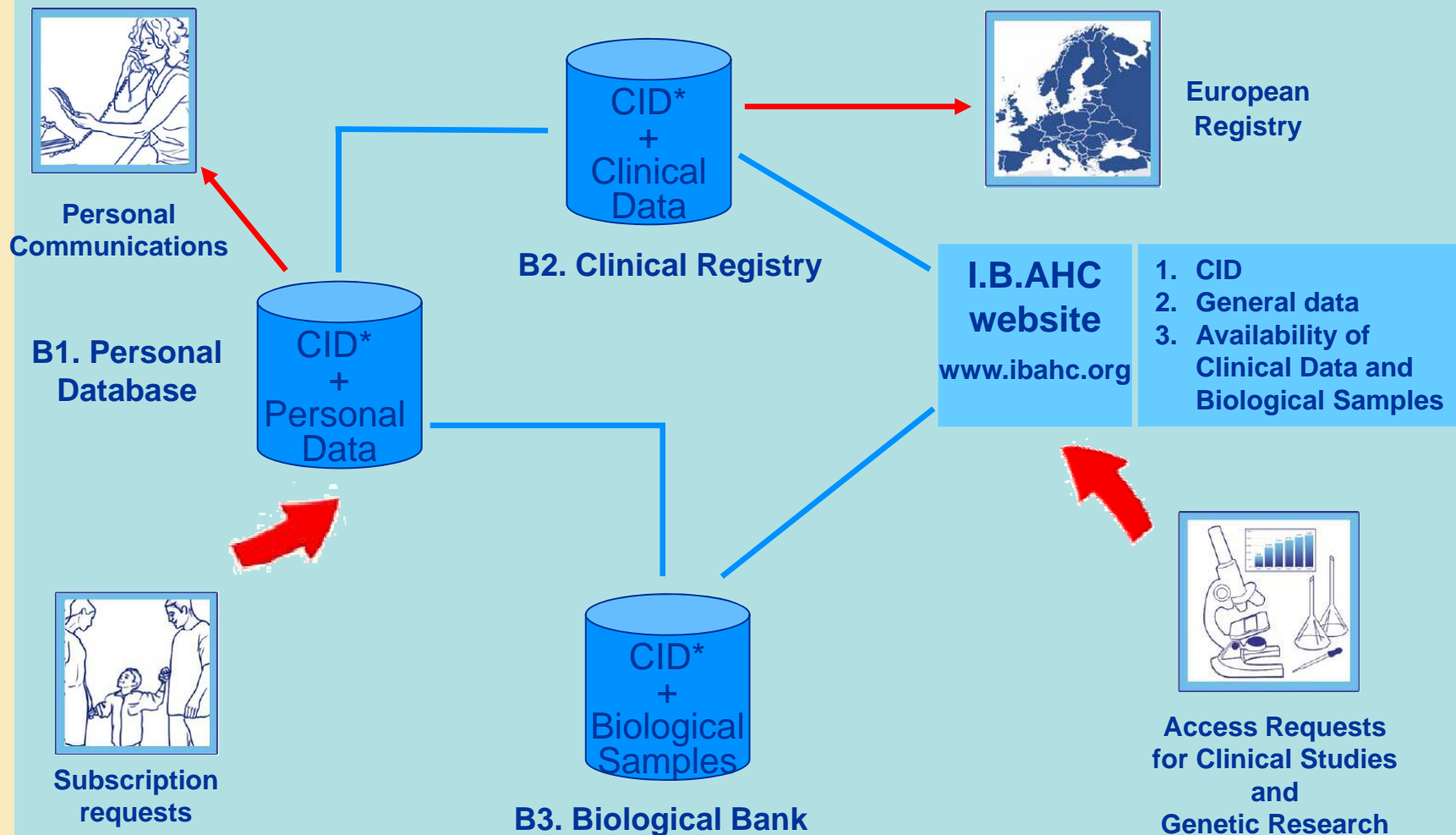
Unique coding scheme, anonymous data and samples, preservation of integrity and flow of information

- **Ethical issues**

Guarantee that patients' data and samples are treated according to strong ethical principles

The I.B.AHC Project

The Architecture



* CID = Patient Identification Code

The I.B.AHC Project

The solutions

- **Operational issues**

*A **detailed and flexible protocol** clearly establishes what each player (patients, parents, doctors, researchers, etc.) has to do at any step of the process. **The Personal Database** is the only entry/exit point of both the Biobank and the Clinical Database*

- **Privacy and security issues**

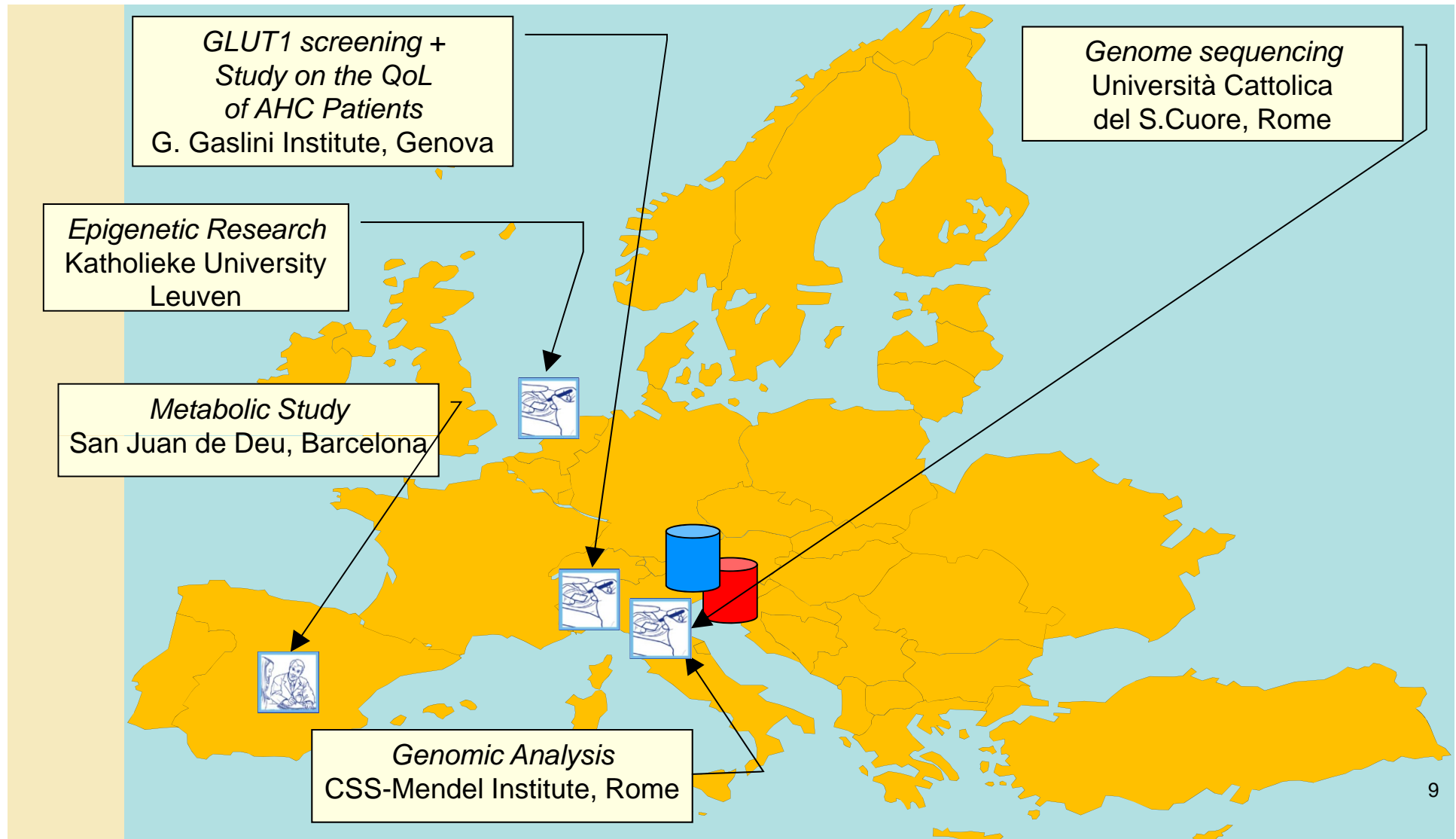
***The CID** is the only connection between personal identities, data and samples. **A.I.S.EA** is the only player able and allowed to link CIDs and personal identities*

- **Ethical issues**

***An official agreement** has been arranged between **A.I.S.EA** and the **Ethical Committee of I.R.C.C.S. E. Medea** (hosting the samples)*

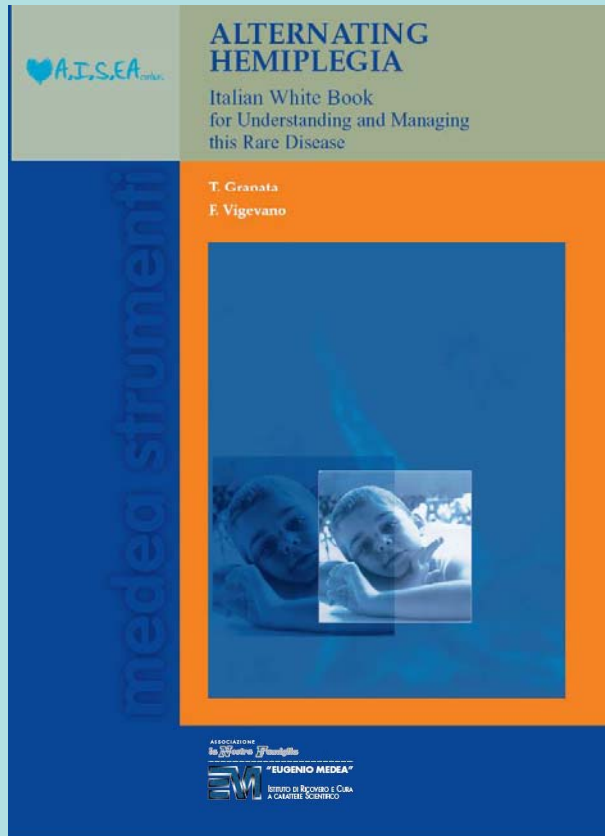
The I.B.AHC Project

The outcomes - 1



The I.B.AHC Project

The outcomes - 2



The Italian White Book for Understanding and Managing Alternating Hemiplegia



National Guidelines for Assistance to Persons Affected by Alternating Hemiplegia and Their Families

The Situation in Europe

| Country (pts – estimated nr) | Biobank | Clinical Registry |
|------------------------------|---------|-------------------|
| Italy (48) | Yes | Yes |
| France (65) | Yes | No ^(a) |
| Spain (25) | Yes | No ^(a) |
| Germany (35) | No | No ^(a) |
| Great Britain (20) | No | No ^(a) |
| Holland/Belgium (8) | Yes | No ^(a) |
| Denmark (8) | No | No ^(a) |
| Czech Rep. (11) | Yes | No ^(a) |
| Others ^(b) (26) | n.a. | n.a. |

(a) Clinical data available (local ENRAH Registry) but not linked to Biobanks

(b) Include Ireland, Iceland, Slovenja, Poland, Israel, Turkey

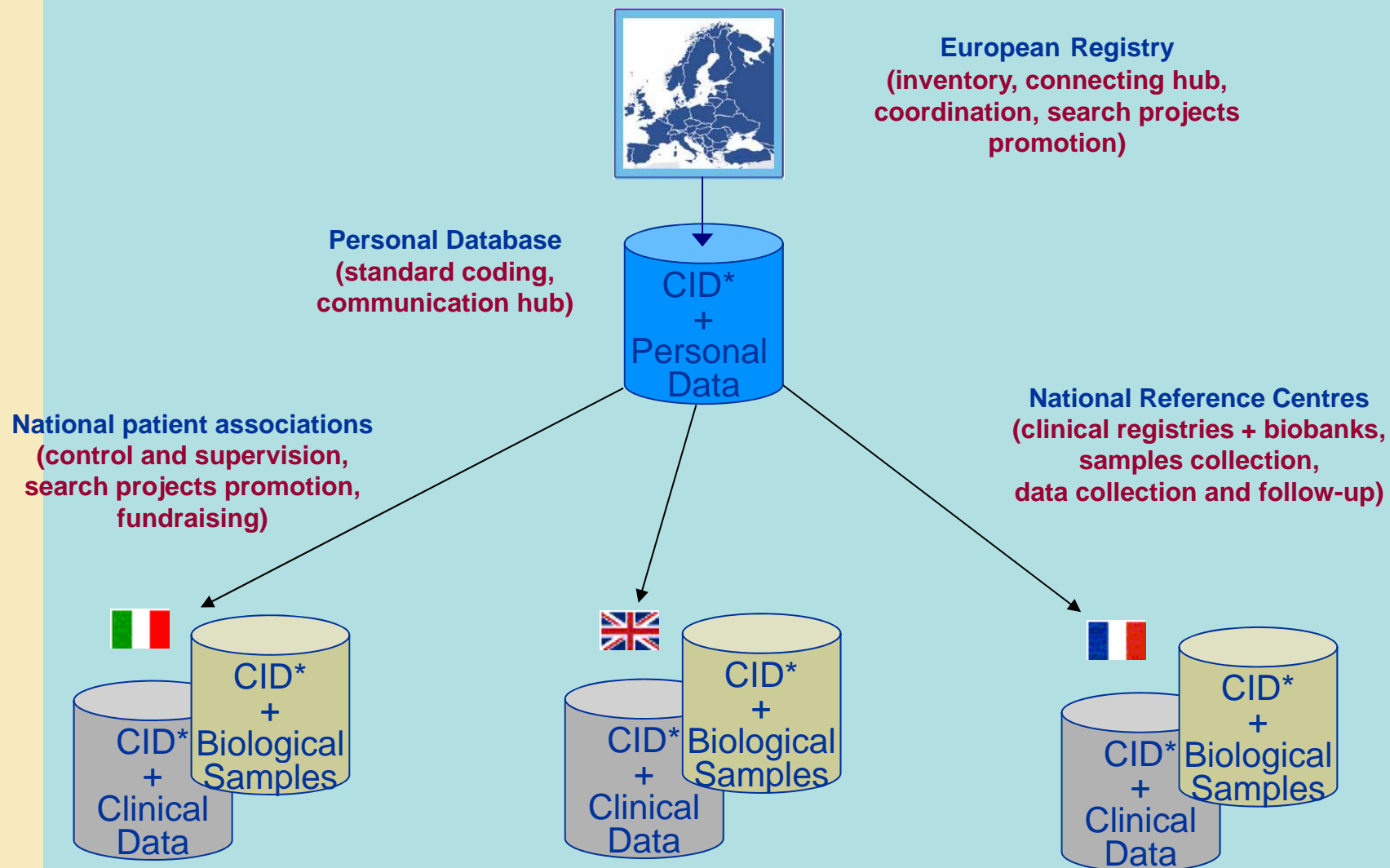
A Proposal for Networking

1 - Background

- **General agreement** among all AH patient representatives in Europe.
- Adoption of the **I.B.AHC model** as a standard template for clinical registries.
- Adoption of a **common coding scheme** (CID).
- Population of national clinical registries.
- Creation of national biobanks (where missing) and connection of biobanks with national clinical registries.
- **Orphan countries' patients** free to access any national biobank and clinical registry.
- Setup of a **European AH BioNetwork** connecting all national repositories under a central administration.

A Proposal for Networking

2 – Scheme and functions



Closing Remarks

- Making a rare disease less rare:
 - European inventory of as many cases as possible
 - A larger number of clinical data and biosamples available to researchers
 - A larger number of interested and actively involved persons
 - Improved visibility and acknowledgement
 - Lobbying actions (both national and European)
- A feasible model for other rare and non rare diseases
- AH patients' best answer to **identifying challenges and opportunities for biorepositories in the next 5 years!!!**

The I.B.AHC Project

Contacts and References



I.B.AHC
Biobank and Clinical Database for Alternating Hemiplegia
www.ibahc.org – info@ibahc.org

Maria Rosaria Vavassori

President of A.I.S.EA and *I.B.AHC Project Coordinator*

vavassori@aiseaonlus.org - (+39) 339 771 4620

Filippo Franchini

I.B.AHC Project Manager

franchini@aiseaonlus.org - (+39) 329 072 7957

Thank you for your attention