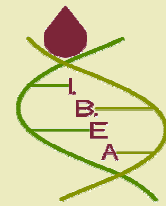


# I.B.EA – The Italian bioBank for Alternating Hemiplegia a tool for the promotion of the research on a rare disease



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## Summary

The Italian bioBank for Alternating Hemiplegia I.B.EA is an open repository containing the clinical data and the blood samples of the Italian patients affected by Alternating Hemiplegia (AHC), a very rare neurological disease.

The architecture and the protocol of I.B.EA are illustrated here to clear up how, through the direct management of a patient association, the following important goals can be achieved:

- support to the research on a rare disease like AHC, by providing non exclusive access to the clinical data and the biological samples of a large number of validated cases
- safeguard of the privacy of the participating patients and of their rights to the information about the results of the research projects using their data and samples
- creation of a network of patients, physicians, researchers and experts for the sharing of the knowledge and for the promotion a better patient diagnosis and care.

## Background

**Alternating Hemiplegia (AHC)** is a very rare disorder characterized by early onset, recurrent episodes of hemiplegia affecting alternatively both sides of the body, occurrence of paroxysmal phenomena such as tonic and dystonic attacks, oculomotor and autonomic disturbances. It is a highly chronically debilitating suffering with deleterious effects on the quality of life of the affected patients.

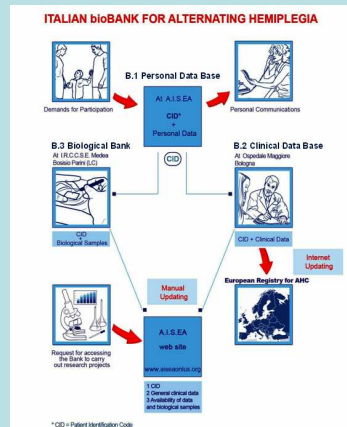
**A.I.S.EA**, the Italian Patient Association for AHC, was created in 1999 with the main goals to support the families, spread the knowledge about the disorder, promote and support the research.

Since the beginning, the association realized that the best way to achieve this last goal was to provide the research groups with an easy, non exclusive access to the clinical data and blood samples of as many AHC cases as possible.

At the same time, the patients of the associations wanted to safeguard their rights to the privacy, to the correct use of their data and samples and to the information about the results of the research projects using the Bank.

Therefore **I.B.EA** - the Italian bioBank for AHC, has been created, a project coordinated by A.I.S.EA, in collaboration with its Scientific Committee.

## I.B.EA Architecture



The Architecture of I.B.EA.

The three sections B1 – B2 – B3 are physically separated but logically joined by means of the CID, the Patient Identification Code

### The Italian bioBank for Alternating Hemiplegia, I.B.EA is composed of three sections:

#### B.1 Personal Data Base - managed by A.I.S.EA

It contains the personal data of the patients and the link between such data and the related CID, the anonymous, numeric code used to reference the information kept in the following two sections.

#### B.2 Clinical Data Base – managed by Dr Giuseppe Gobbi, Child Neuropsychiatry Unit, Maggiore Hospital, Bologna.

It contains the clinical data, the video recordings and the photographs of the participating patients, to be used for clinical and therapeutic studies. All these data are identified only by the CIDs.

#### B.3 Biological Bank – managed by Dr. Maria Teresa Bassi, Laboratory of Molecular Biology, Scientific Institute E. Medea (LC).

It contains the biological samples of the participating patients (DNA, RNA and cellular lines) and of their parents (DNA), to be used for genetic research. The samples are identified only by the CIDs.

A subset of the Data in the Clinical Data Base is also copied in the European Registry for AHC, under the management of ENRAH, the European Network for Research on AHC.

## I.B.EA Protocol

The protocol of the Italian bioBank for Alternating Hemiplegia I.B.EA consists of the following steps:

### 1. Enrolment of the patients, their parents and their treating physicians

A.I.S.EA collects the consent forms from the participating patients and parents and from their treating physicians, for the processing of their data (Italian National Law 30.06.2003 Nr 196). A CID is assigned by A.I.S.EA and notified to each participant. Only A.I.S.EA can communicate personally with the participants.

### 2. Collection of the Clinical Data and the Blood Samples

The treating physicians receive from A.I.S.EA the questionnaires to fill with the clinical data of their patients and to send to the Clinical Data Base. The questionnaires are labeled only with the CIDs of the patients, thus protecting their anonymity. With the collaboration of the physicians, also the blood drawing is organized and the samples, labeled by the CIDs, are sent to the Biological Bank. A.I.S.EA updates the list of the available CIDs on its web site: this way, the patients can easily check their presence in the Bank.

### 3. Validation of the Diagnosis

At least once a year, A.I.S.EA organizes a video-session, during which new suspected AHC cases are presented by their physicians and discussed by the Scientific Committee of the association and all the physicians participating to I.B.EA.

If the diagnosis is validated, the patient and the presenting physician are enrolled in the Bank.

### 4. Access to the Bank

Any research group can request to use the Bank, by filling a form and sending it to A.I.S.EA.

The request is evaluated by the Scientific Committee of the association and by the Ethic Committee of the Scientific Institute E. Medea, within the following two months.

A Material Transfer Agreement (MTA), based on the evaluation comments and proposed by A.I.S.EA, must be signed by the research group, before accessing the Bank.

## Results

At present, I.B.EA contains the complete clinical documentation and the biological samples of more than 30 Italian patients.

Three clinical studies and two genetic research projects are currently using the Bank: A.I.S.EA also agreed with their managers, through the signing of the MTA, that the results will be delivered to the patients, kept in the Bank and shared with the scientific community through publications.

I.B.EA is also the central node of a network inside which a better circulation of information and ideas is fostered, thus further promoting the start of coordinated and collaborating research projects and activities.

In particular, through the collegial validation sessions and the active participation of the treating physicians, correct and early diagnoses and a better care of the patients have been promoted.

## Conclusions

New enhancements for I.B.EA have already been planned. In particular, a new project **I.B.EA on-line**, has already started, to build a Clinical Data Base accessible via Internet in a secure and controlled way.

This will allow a more efficient data entry and management and an easier data retrieval.

Also interfaces to the ENRAH European AHC Registry and to the Public National Registry for Rare Diseases will be developed for the automatic data transfer.

I.B.EA on-line will be available also in English, to facilitate the access by international research groups.

## Acknowledgements

A.I.S.EA wants to thank the members of its Scientific Committee, its advisors and all the treating physicians of I.B.EA, for their active participation to this important project.

Many thanks to the Azienda ASL – Ospedale Maggiore, Bologna for hosting the Clinical Data Base and to the Scientific Institute E. Medea, Bosisio Parini (LC) for hosting the Biological Bank. Thank you also to the Managers of these two sections of I.B.EA.

A special thank to the Ethic Committee of the Institute E. Medea for their participation to I.B.EA and for their precious advice to the association.

### For additional information, please contact:

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Year	Consent	Year of CID	Gender	Diagn.	Diagn.	Diagn.	Diagn.	Diagn.	Diagn.
2002	171001	1999	M	Complete	YES	0	Not in Bank		
0003	171002	1999	M	Complete	YES	0	Not in Bank		
0004	171003	1999	M	Complete	YES	0	Not in Bank		
0005	171004	1999	M	Complete	YES	0	Not in Bank		
0006	171005	1999	M	Complete	YES	0	Not in Bank		
0007	171006	1999	M	Complete	YES	0	Not in Bank		
0008	171007	1999	M	Complete	YES	0	Not in Bank		
0009	171008	1999	M	Complete	YES	0	Not in Bank		
0010	171009	1999	M	Complete	YES	0	Not in Bank		
0011	171010	1999	M	Complete	YES	0	Not in Bank		
0012	171011	1999	M	Complete	YES	0	Not in Bank		
0013	171012	1999	M	Complete	YES	0	Not in Bank		
0014	171013	1999	M	Complete	YES	0	Not in Bank		
0015	171014	1999	M	Complete	YES	0	Not in Bank		
0016	171015	1999	M	Complete	YES	0	Not in Bank		
0017	171016	1999	M	Complete	YES	0	Not in Bank		
0018	171017	1999	M	Complete	YES	0	Not in Bank		
0019	171018	1999	M	Complete	YES	0	Not in Bank		
0020	171019	1999	M	Complete	YES	0	Not in Bank		

The updated availability in the Bank is displayed on the website of A.I.S.EA. On the website, the list of the projects currently accessing the Bank is also available.