The Italian ALS Patient-driven registry and
The ALS Biobank: two new tools to promote
and stimulate the research
Christian Lunetta
ALS is rare and clinically heterogeneous disease
ALS is rare and clinically heterogeneous disease, and to better understand the demographic characteristics of people living with ALS (PALS), the natural history of ALS across all phenotypes, and disease management in the real-world setting, requires large-scale data collection and harmonization.

The model identified five groups with distinct survival times (median in months) from symptom onset:
- Very short 17.7;
- Short 16.5,
- Intermediate 32.2;
- Long 43.7;
- Very long 91.0.

Eight of the 16 candidate predictors were selected for the multivariable prediction model.

11 475 patients
14 Centers
Large-scale data collection and harmonization can be done via several mechanisms, including:
- population-based disease registries,
- voluntary direct reporting by patients (patient-driven registry),
- merged databases from individual clinical research efforts,
- clinic-based data repositories.

In Italy we have only 4 Regional population-based disease registries:
- Piemonte – Val d’Aosta
- Puglia
- Emilia-Romagna
- Liguria
Large-scale data collection and harmonization can be done via several mechanisms, including:
- population-based disease registries,
- voluntary direct reporting by patients (patient-driven registry),
- merged databases from individual clinical research efforts,
- clinic-based data repositories.

I believe that the most important requirement for the new knowledge network envisaged by the Precision Medicine report is that it be driven by patients. Indeed, it is patients who uniquely understand the potential value of a social contract in which patients both contribute personal clinical data and benefit from the knowledge gained through the collaboration.

Patients are also in the best position to demand the sharing of both data and professional credit that will be necessary to fully capture the value of this new collaborative approach to acquiring, synthesizing, and widely disseminating biomedical knowledge.
A web-based, patient driven registry for Angelman syndrome: the global Angelman syndrome registry

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Letter to the Editor

Open Access

CrossMark
The concepts of patient-centered care and patient-centered research have moved to the forefront of health care and research in recent years.

The definition of patient-centered care as “providing care that is respectful of and responsive to individual patient preferences, needs, and values, and ensuring that patient values guide all clinical decisions.”

The definition of patient-centered outcome research as “research that addresses the questions and concerns most relevant to patients.”
Table 1-1. Goals and benefits of patient-centeredness in research and medical care

<table>
<thead>
<tr>
<th>THEME</th>
<th>GOALS</th>
</tr>
</thead>
<tbody>
<tr>
<td>Funding</td>
<td>Research funding is directed optimally to address questions that are of high priority and relevance to patients</td>
</tr>
<tr>
<td>Research design</td>
<td>To align research questions with evidence gaps and patient needs as well as enhance research efficiency</td>
</tr>
<tr>
<td>Availability of evidence and dissemination</td>
<td>Dissemination of research findings to health care providers and patients is timely and transparent, and communicated in a manner that is clear and understandable to patients</td>
</tr>
<tr>
<td>Informed decision-making</td>
<td>Patients have the necessary information to make informed decisions about the health care choices available to them, linked to health outcomes that are important to them</td>
</tr>
</tbody>
</table>
| Desired benefits                     | • Improvement in health outcomes that are most meaningful to patients and clinicians  
                                        • Increased satisfaction of patients and health care providers with medical care |

**Patient-centered care** and **patient-driven clinical research** is intended to result in two primary outcomes:

1. **increased satisfaction of patients and health care providers with medical care**;
2. **improvement in health outcomes that are most meaningful to patients and clinicians**

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Patient registry is “an organized system that uses observational study methods to collect uniform data (clinical and other) to evaluate specified outcomes for a population defined by a particular disease, condition, or exposure, and that serves one or more predetermined scientific, clinical, or policy purposes.”

In comparison to other research designs, patient registries offer some unique features that may be particularly useful for patient-centered outcomes research (PCOR).

Goals for the PPRNs are similar to those for many disease registries, with each network being focused on a specific condition and/or community of interest with an objective of creating a standard database that can be used to address future patient-driven research questions.

A hallmark of the PPRNs is to include patients as partners in the governance structure of the network and to collect PROs relevant to the community they serve to support patient-prioritized PCOR questions.
ITalian ALS Registry: a pilot study to assess the feasibility of a web-based, patient driven registry for Italian people with Amyotrophic Lateral Sclerosis

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1. Collect information on the distribution of ALS patients in Italy;
2. Collect data on the clinical, genetic and demographic characteristics of patients diagnosed with ALS, allowing greater knowledge of the disease and constant monitoring of any changes;
3. Contribute to the design of clinical trials and help identify suitable patients for clinical trials;
4. Collect data on the assistance received for the implementation of care standards;
5. Align data collection with a repository of biological material from ALS patients;
6. Encourage the networking of Italian clinical reference centers for ALS
Participants

- Promoting and supporting partner: AISLA Association
- Coordinating Center: NEMO Clinical Center Milano (PI: Christian Lunetta)
- Participating Centers:
  - NEMO Clinical Center Arenzano (PI: Fabrizio Rao)
  - NEMO Clinical Center Roma (PI: Mario Sabatelli)
  - NEMO Clinical Center Messina (PI: Gianluca Vita)
  - S. Gerardo Hospital, Milano (PI: Lucio Tremolizzo)
  - S. Raffaele Hospital, Milano (PI: Nilo Riva)
- Technical supporting partner: Associazione del Registro Italiano dei pazienti con Malattie Neuromuscolari (PI: Anna Ambrosini)

Referral general population: **22.5 million inhabitants** (1/3 of Italian population)
Referral ALS population: **1.600 - 2.100 patients**
The caregiver / support administration has access to the data only after signing the informed consent.

Registry log-in

A personalized authorization allows online access to the data.

NeuroGUID

Demographic Data

Informed Consent

Physicians

Registry curator

Patient forms

Form 1: Familial and past pathological history
Form 2: ALS history & milestones
Form 3: Communication aids
Form 4: Clinical studies
Form 5: Level of satisfaction with the assistance received and the places of assistance
Form 6: Clinical data (El Escorial criteria, FVC, ALSFRS-R, Survival)

Request for access to data

Registry curator

Steering Committee

De-identified aggregated data
De-identified data queries
Clinical trial recruitment

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What is NeuroGUID™

The Neurological Global Unique Identifier (NeuroGUID™) is a universal patient identifier that allows researchers in Neurological and other Central Nervous System diseases to share data specific to a study participant without exposing personally identifiable information (PII). It also enables matching participants across studies and research data repositories. In other words, NeuroGUID™:

- Allows aggregation of data from the same patient from multiple studies, regardless of where and when that data was collected.
- Prevents a single patient from being created multiple times in the same research database, in case she/he participates in a study at different sites or in multiple studies at the same time.
- Enables a researcher to combine patient information with data originating from other sources.

What information is required to create a NeuroGUID™

The following information is required to create a NeuroGUID™:

- Complete legal given first, middle and last names of patient at birth
- Date of birth
- Physical sex of patient at birth
- Name of city/municipality in which patient was born
- Country of birth

http://www.NeuroGUID.org
According to the principles of **patient-centered care** and **patient-centered research** we have built **an integrated model**

Diagram:

- **Patient Community**
- **Registry**
- **Biorepository**
Main Aims

✓ To encourage research for the identification of genetic mutations and other causes of disease
✓ To support the collection of biological material coming from ALS people with genomic features useful for understanding the genetic basis of the disease
✓ To standardize the quality of biological material available
✓ To ensure the cryopreservation and availability for a long time of biological material suitable for the needs of scientific research according to the evolution of knowledge about the disease
Sla, nasce la prima biobanca nazionale: “Aperta a tutti, darà impulso alla ricerca”

È stata inaugurata dal presidente Conte al Policlinico Gemelli di Roma. Al suo interno, Dna, plasma e colture di cellule verranno catalogati in forma anonima e messi a disposizione di tutti i ricercatori italiani e del resto del mondo

di Chiara Daina | 21 Giugno 2019

Inaugurata a Roma la prima biobanca nazionale sulla Sla

Il presidente Conte alla biobanca sulla Sla al Policlinico Gemelli.
It will collect biological specimens from a nationally representative sample of PALS, in particular that is not tied to a specific Center.

It will help to collect sample from peripheral Centers that do not have facilities for a biobank.

It will be an open access biobank where researchers will be able to gain samples from ALS patients.

It will be open to all researchers.

Together with ALS registry, it will help to create a network of neurologist and researchers across the country.
What types of samples will be available?

- Blood
- Serum
- Plasma
- DNA
- RNA
- PBMCs
- Urine
- CSF
- Muscle
- Skin
- Human primary cells derived from PBMC of PALS with specific genetic background
Un gesto con il cuore per sostenere la ricerca contro la SLA

300.000 euro

DAL 6 AL 12 MAGGIO, L’1% SULL’ACQUISTO DEI PRODOTTI SELEX SARÀ DONATO AD AISLA ONLUS

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to be continued...