



ISLAMIC REPUBLIC OF IRAN



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Research Affairs  
Disease Registry Unit

## In the Name of GOD

### REGISTRY PROPOSAL: (1-2 PAGES excluding references)

#### A. Title of Registry

Iranian registry of Neurometabolic patients

#### B. Brief Intro to Registry –Literature Review (1-2 paragraphs)

Neurometabolic diseases refer to a group of disorders that are characterized by a lack or dysfunction of an enzyme or vitamin necessary for a specific chemical reaction in the body.

The field of pediatric Neurometabolic Disorders discuss about diagnosis and treatment in children with metabolic disorders with neurological sign and symptoms, including:

- Central nervous system white matter disorders (leukodystrophies)
- Disorders of Aminoacids
- Disorders of organic acid metabolism
- Disorders of Lysosomal storage diseases
- Mitochondrial disorders
- Peroxisomal disorders
- Lipid metabolism disorders
- Carbohydrate metabolism disorders
- Neurodegenerative disorders with gray matter involvements

We try that in Neuromabolic registry recording the history of the disease, signs and symptoms, laboratory tests and neuroimaging findings of these patients Collect and record in this site.

It should be emphasized that only clinically and biochemically approved patients will be registered.

In this registry system we try to find out the prevalence and incidence of neuromabolic diseases in Iran. It needs the collaboration between the Pediatric Neurological Research Centers and all pediatric neurologic departments in Iran, in order to register all different types of neurometabolic disorders.

And because consanguineous marriage is more common in Iran we expect more cases in our country for registry in this system.

Since these patients need to be monitored by their neurologists, neuropathologists, neuroradiologists, neurogeneticists, and dietitians throughout their lives, it is important to record their information and follow up the trend of their disease.

C. Registry Objectives (what is it you are specifically looking at, trying to reach?)

- Use of information collected in all research studies of neurometabolic patients.
- Collect Clinical and laboratory data to evaluate the prevalence of these diseases.
- Genetic testing to prevent the birth of a child with these diseases.
- Observing the disease changes during the treatment process.

D. Registry Design (participants, data collection, statistical analysis plan, etc.)

Participants are all children 1 to 18 years of age that refer to pediatric neurology departments with a variety of neurometabolic disorders.

According to statistics, 360 patients with a variety of neurometabolic diseases are registered at the neurology clinic of Mofid children's hospital. As there are 15 other active centers in the country, there are about 3,000 samples registered annually.

The registry process is that, at the neurometabolic registration site, patients' information enters the site after genetic testing is approved.

E. Timeline for your Registry project (project deadlines set by you and your mentor)

Proposal edition and confirmation takes 4 months.

Registry duration set 10 years and it will be continue more.

Each 10 years data will be evaluated and results use in researchers and articles to improve patient's conditions.

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