

## In the Name of GOD



**Shahid Beheshti  
University of Medical Sciences**

### **Research Affairs Disease Registry Unit**

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

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

Neurocutaneous syndromes are disorders that affect the brain, spinal cord, organs, skin, and bones. The diseases are lifelong conditions that can cause tumors to grow in these areas. They can also cause other problems such as hearing loss, seizures, and developmental problems. Each disorder has different symptoms. The 3 most common types of neurocutaneous syndromes are: Tuberous sclerosis (TS), and Neurofibromatosis (NF), including NF1, NF2, and Sturge-Weber disease but there are also other diseases in this group such as incontinentia pigmenti, hypomelanosis of Ito, Von Hippel-Lindau disease, and others. This registry, will pay attention only to the first 3 diseases; i.e. NF, Tuberous sclerosis, and Sturge Weber syndrome.

The model of transmission of these diseases is usually Mendelian inheritance. Tuberous sclerosis and Neurofibromatosis have autosomal dominant transmission. Neurocutaneous diseases (or phakomatoses) are lifelong conditions that can cause tumors to grow in different tissues of the body. They can also cause other problems such as hearing loss, seizures, and developmental problems; therefore, they have a great impact on the general wellbeing of the individual, and a huge burden on the health services for diagnosis, treatment, and follow up of the patients.

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

In the first place, it is of utmost importance to have data about the prevalence of these diseases among Iranian population. Some of the medications used in the treatment of these diseases are relatively new and expensive drugs- such as mTOR inhibitors, (with different brand names like rapamycine, everolimus, tacrolimus, sirolimus, or afinitor) and in order to have a good evaluation about the amount of needed drug in the country we should know the total number of patients. Also, by having a registry, we will know the prevalence of different complications among

these patients (tumors, refractory epilepsy, etc.), and better anticipate their occurrence.

Besides, to be able to judge about the effectiveness of different therapeutic methods and drugs which are being used by clinicians throughout the country, we need a good registry system for surveillance of the patients.

With institution of this registry system, we will be able to follow these patients efficiently and have a better understanding of the prevalence of the complications and anticipate their occurrence.

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

The cases will be collected from the patients who present or are referred to pediatric neurology clinics. Statistical analysis will be done with the help of the statisticians.

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

Proposal edition and confirmation takes 1 month.

Registry duration set 5 years and it will be continue more.

Each 10 years data will be evaluated and results are analyzed by theresearchers and will be used to improve the patient's conditions.

E. Who will provide support and feedback and how often will this occur?

All of the members of the neurocutaneous disorders registry team will work on the project.

**Pediatric Neurology Research Center**

**Mofid Children's Hospital**

**Dr.Mohsen Javadzadeh**