



ISLAMIC REPUBLIC OF IRAN



Shahid Beheshti  
University of Medical Sciences

**In the name of GOD**

**Research Affairs**

**Disease Registry Unit**

**Annually report of Disease Registry Program**

|  |   |
|--|---|
| <b>Title of Registry Program</b>                         | Iranian registry of neurometabolic patients                             |
| <b>Main director of registry</b>                         | DR. PARVANEH KARIMZADEH   |
| <b>Organization/Research center/Hospital/Department:</b> | <b>PEDIATRIC NEUROLOGY RESEARCH CENTER OF MOFID CHILDREN'S HOSPITAL</b> |
| <b>Accept Date</b>                                       |   |

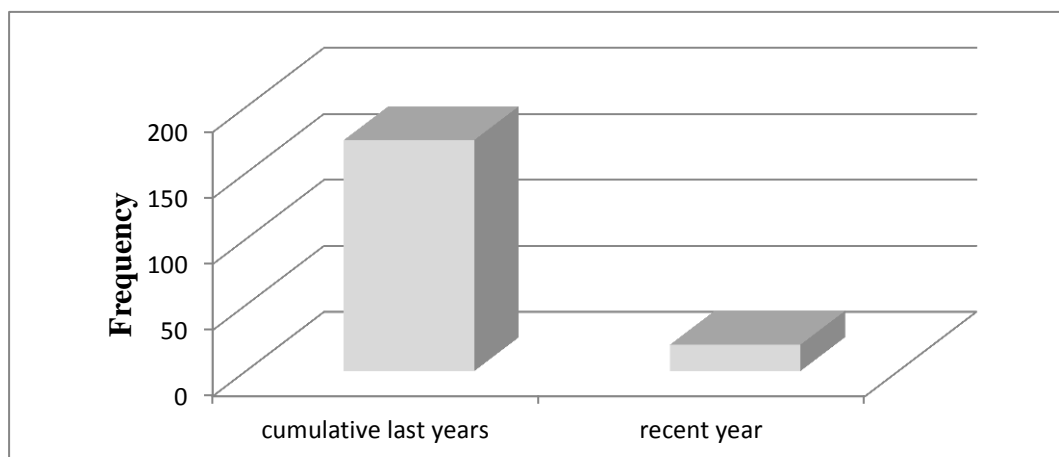
1. How many annual reports? 1
2. Report date: 2019-07-29

3. Steps taken so far (according to the action plan): (Specifically for deployment and commissioning centers)

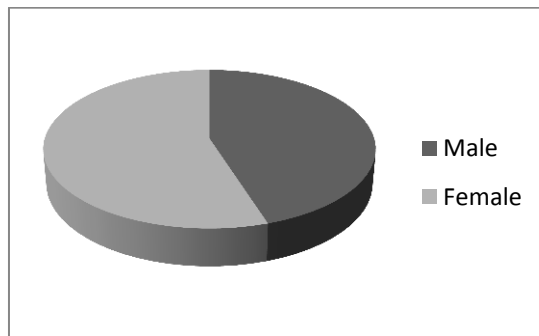
**– THE NEUROMOTOR PATIENT REGISTRATION SITE HAS BEEN LAUNCHED SINCE 2010.**

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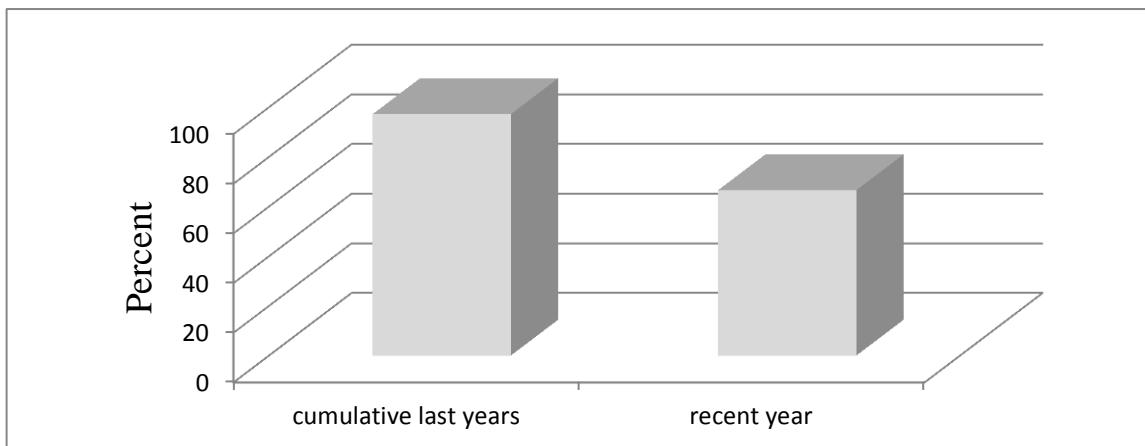
**4. Absolute Frequency Reporting of recorded cases in recent years**



### 5. Sex distribution of recorded cases



### 6. expected rate recorded Reporting in recent years



$$\text{cumulative last years} = \frac{\text{number of reported cases in last years}}{\text{number of expected records in last years}} \times 100 = \frac{175}{180} \times 100 = 97.2$$

$$\text{recent year} = \frac{\text{number of reported cases in recent year}}{\text{number of expected records in recent year}} \times 100 = \frac{20}{30} \times 100$$

## 7. Published articles

| authors  | Journal / Congress                           | Published year | Title of article  | Number |
|--|--|----------------|---|--------|
| A. Nikkhah, P. Karimzadeh, M.M. Taghdiri, M.M. Nasehi, M. Javadzadeh, E. Khari   | Iranian Journal of Child Neurology           | 04/2019        | <i>Hyperkinetic movement disorders in children: A brief review</i>  | ۱      |
| Parvaneh Karimzadeh  | DOI:10.1002/ccr3.1998                        | 01/2019        | <i>3-Hydroxyisobutyryl-CoA hydrolase deficiency in an Iranian child with novel HIBCH compound heterozygous mutations</i>              | ۲      |
| Abbas Masserrat, Fatemeh Sharifpanah, Leila Akbari, Seyed Hasan Tonekaboni, Parvaneh Karimzadeh, Mahmood Reza Asharafi, Safoura Mazrouei, Heinrich Sauer, Massoud Houshmand  | DOI:10.3892/br.2018.1156                     | 10/2018        | <i>Mitochondrial G8292A and C8794T mutations in patients with Niemann -Pick disease type C</i>  | ۳      |
| Parvaneh Karimzadeh  | International journal of preventive medicine | 08/2018        | <i>Risk Factors of Pediatric Arterial Ischemic Stroke; A Regional Survey</i>  | ۴      |
| Sepideh Tabarestani, Pediatric Neurologist, Parvaneh Karimzadeh, Farzad AHMADABADI Md  | DOI:10.31031/RPN.2018.01.000524              | 2018.01        | <i>PEX12 Novel Mutation: A Case Report on Iranian Girl with Childhood Onset Peroxisomal Disorder: Pseudo ALD?</i>                     | ۵      |
| Mohammad Ghofrani, Mohammad Mehdi Nasehi, Sasan Saket, Mohsen Mollamohammadi, Mohammad Mahdi Taghdiri, Parvaneh Karimzadeh, Seyed Hassan Tonekaboni, Mohsen Javadzadeh, Narjes Jafari, Azadeh Zavehzad, Masoud Hasanvand Amouzadeh, Mahsa Beshrat, Meysam Babaei | Iranian Journal of Child Neurology           | 07/2018        | <i>Prediction of Response to Treatment in Children with Epilepsy</i>  | ۶      |
| Parvaneh Karimzadeh, Hamid Nemati  | Iranian Journal of Child Neurology           | 06/2018        | <i>Summer Causes and Factors Associated with Neonatal Seizure and its Short-term Outcome: A Retrospective Prognostic Cohort Study</i> | ۷      |
| Parvaneh Karimzadeh Md, Tonekaboni, Narjes Jafari Md, Masoud Hasanvand Amouzadeh Md, Mahsa Beshrat Md, Meysam Babaei   | Iranian Journal of Child Neurology           | 06/2018        | <i>Prediction of Response to Treatment in Children with Epilepsy</i>  | ۸      |
| Abbas Masserrat, Fatemeh Sharifpanah, Leila Akbari, Seyed Hasan Tonekaboni, Parvaneh Karimzadeh, Mahmood Reza Asharafi, Safoura Mazrouei, Heinrich Sauer, Massoud Houshmand  | DOI:10.3892/br.2018.1095                     | 05/2018        | <i>: Mitochondrial G8292A and C8794T mutations in patients with Niemann -Pick disease type C.</i>                                     | ۹      |
| Narjes Jafari, Karl Golnik, Mansoor Shahriari, Parvaneh Karimzadeh, Sayena Jabbehdari  | DOI:10.4103/jovr.jovr_242_16                 | 01/2018        | <i>Ophthalmologic Findings in Patients with Neuro-metabolic Disorders. Journal of Ophthalmic &amp; Vision Research</i>                | ۱۰     |

|   |                                    |          |  |    |
|---|------------------------------------|----------|--|----|
| Parvaneh Karimzadeh, Samaneh Naderi, Farzaneh Modarresi, Hassan Dastsooz, Hamid Nemati, Tayebeh Farokhashtiani, Bibi Shahin Shamsian, Soroor Inaloo, Mohammad Ali Faghihi | DOI:10.1186/s12881-017-0417-4      | 12/2017; | <i>Case reports of juvenile GM1 gangliosidosis type II caused by mutation in GLB1 gene. BMC Medical Genetics</i> | ۱۱ |
| Parvaneh Karimzadeh, Toktam Moosavian, Hamidreza Moosavian  | Iranian Journal of Child Neurology | 01/2018  | <i>Recurrent stroke in a child with TRMA syndrome and SLC19A2 gene mutation</i>                                  | ۱۲ |

## 8. published Books

| authors   | Published year       | Title of book                     | Number |
|---|----------------------|-----------------------------------|--------|
| <b>Parvaneh Karimzadeh, MD</b><br>Ali Nikkhah, MD<br>Narjes Jafari, MD<br>Sepideh Tabarestani, MD   | ۲۰۱۹<br>(در حال چاپ) | Atlas of neurometabolic disorders | ۱      |
| <b>Parvaneh Karimzadeh,</b><br>Mohammad ghofrani,<br>mohammad m. Taghdiri<br>Idin Tabrizi, Marjan Shakiba,<br>Roxana Azma,<br>Alireza Tavassoli, Mahmoudreza<br>Ashrafi, Mahshid mehdizadeh,<br>Ghalamreza Zamani | ۲۰۱۹                 | Adrenoleukodystrophy(ALD)         | ۲      |
| دکتر پروانه کریم زاده   | ۱۳۸۵                 | تکامل و اختلال تکاملی کودکان      | ۳      |
| دکتر پروانه کریم زاده   | ۱۳۸۹                 | ۳۶۵ روز اول زندگی کودک            | ۴      |

## 9. Workshops held

| Number | Title of Workshop | Participants | Organizer | Hold time | Hold location |
|--------|-------------------|--------------|-----------|-----------|---------------|
|--------|-------------------|--------------|-----------|-----------|---------------|

|  |                                   |  |   |   |                |
|--|-----------------------------------|--|---|---|----------------|
|  | Monthly Neurometabolic Commission | in Pediatric Neurology, Pediatric Endocrinology, Pediatric Oncology, Physical Medicine Specialists, Pediatric Radiology, Neurology, Neuropathology and Nutrition Specialists | Pediatric Neurology Research Center, Iranian Neurometabolic Association | )Last Thursday of every month( Since 2011 | Mofid hospital |
|  |                                   |  |   |   |                |
|  |                                   |  |   |   |                |

### 10.Seminars / conferences held

| Number | Title of Seminars / conferences           | Type of Seminars / conferences |          |          | Participants   | Organizer                           | Hold time        | Hold location  |
|--------|---|--------------------------------|----------|----------|--|-------------------------------------|------------------|----------------|
|        |   | international                  | national | regional |  |                                     |                  |                |
| 1      | 1 <sup>th</sup> Neurometabolic Conference |                                | *        |          | in Pediatric Neurology, Pediatric Endocrinology, Pediatric Oncology, Physical Medicine Specialists, Pediatric Radiology, Neurology, Neuropathology and Nutrition Specialists | Pediatric Neurology Research Center | 19Jan2011        | Mofid hospital |
| 2      | 2 <sup>th</sup> Neurometabolic Conference |                                | *        |          | //   | //                                  | 20 december 2012 | //             |

|   |   |  |   |  |    |    |                  |    |
|---|---|--|---|--|----|----|------------------|----|
| 3 | 3 <sup>th</sup> Neurometabolic Conference |  | * |  | // | // | 12 december 2013 | // |
| 4 | 4 <sup>th</sup> Neurometabolic Conference |  | * |  | // | // | 20 Nov 2014      | // |
| 5 | 5 <sup>th</sup> Neurometabolic Conference |  | * |  | // | // | 26 Nov 2015      | // |
| 6 | 6 <sup>th</sup> Neurometabolic Conference |  | * |  | // | // | 29 december 2016 | // |
| 7 | 7 <sup>th</sup> Neurometabolic Conference |  | * |  | // | // | 21 december 2017 | // |
| 8 | 8 <sup>th</sup> Neurometabolic Conference |  | * |  | // | // | 17 Jan 2019      | // |
| 9 | 9 <sup>th</sup> Neurometabolic Conference |  | * |  | // | // | 19December 2019  | // |

Number of annual reports submitted to the University Disease Registry:1

Name and surname of the Main director of registry: DR. PARVANEH KARIMZADEH

Date: 2019-07-29