Iranian Registry of Neurometabolic Disease

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The metabolic disorders are the group of diseases in them an enzymatic defect or a coenzyme deficiency lead to a failure in metabolism of a substrate and then accumulation of a toxic product or deficiency of a needed product.

When this process involves central nervous system result in neurometabolic disorder.

The overall incidence of this group of disease is 15/100000 live birth (worldwide).

Genetic transmit ion of them is generally Autosomal Recessive and the patients usually are products of consanguineous marriage.

We decide to activation of a system for registration of diagnosed cases of neurometabolic diseases, collect the clinical symptoms and signs and laboratory or neuroimaging findings of these patients, considering their scarcity, differences in clinical findings and importance of these group of diseases.

Goals of commence of these site are:

1. Registeration of all of neurometabolic cases in Iran.
2. To get a rough count of the neurometabolic diseases in Iran.
3. Make a wide range data bank for neurometabolic disorders in national level.
4. Make a Educational site (E Learning) concerning neurometabolic disorders.

During the 1 years from activation of IRNMD, 74 cases are submitted in our site(Table 1).

We hope to develop our system and data bank with your collaboration.

Keywords: Nerometabolic disease; Registry site; Iran

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Table 1. Distribution of submitted cases in IRNMD

Diagnose	Number of approved cases	Percentage
Propionic acidemia	2	2.7 %
Methylmalonic acidemia	5	6.8 %
methylmalonic aciduria and homocystinuria	6	8.1 %
Biotinidase deficiency	16	21.6 %
isovaleric acidemia	1	1.4 %
D-2-Hydroxyglutaric aciduria	3	4.1 %
L-2 Hydroxyglutaric aciduria	1	1.4 %
phenylketonuria	5	6.8 %
Homocystinuria	1	1.4 %
MCAD glutaric aciduria type ll / ethylmalonic - adipic aciduria	3	4.1 %
Adrenoleukodystrophy	1	1.4 %
Tay-sachs disease	1	1.4 %
Sandhoff disease	1	1.4 %
Niemann-pick disease	1	1.4 %
Niemann-pick type C disease	1	1.4 %
Krabbe disease	2	2.7 %
Maple syrup urine disease(MSUD)	1	1.4 %
Propionic acidemia	2	2.7 %
Canavan	2	2.7 %
Zellweger	6	8.1 %
Glutaric aciduria type l	5	6.8 %
Miscellaneous	8	10.8 %