

Neurometabolic Registry Site in Iran

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The Neurometabolic disorders are generally applies to monogenic diseases with a deficient activity in an enzyme, or a transporter in a pathway of cellular metabolism. This defects cause accumulation of the primary substrate or deficiency of product and lead to neurologic or developmental symptoms.

Inborn errors of metabolism are individually uncommon but important diseases and many of them have neurologic manifestations. In some cases enzyme replacement therapy or diet may be effective.

Noticed in their incidence(15.7/100000 live birth) and difficulties in their diagnosis, confrontation with them is not possible for all physicians and medical students and for this reason some of disease keep unknown for mentioned group.

We try to instigate a site for registration of Neurometabolic cases with their history, signs and symptoms, lab tests and imaging's.

Only clinical and biochemical well-defined diseases are registered and exclusively data of patients with confirmed diagnosis are being processed.

This is a collaborate project of all pediatric neurology centers in Iran and the main reason for initiating it is the fact that,in daily practice it is very difficult to find patients with metabolic diseases.

There is no disease specific registration for Neurometabolic diseases, making it impossible to give reliable incidence rates for them in Iran.

Keywords: Neurometabolic;Registry site; Inborn errors of metabolism

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