Title: Follow up evaluation of patients referred to Children`s Medical Center Laboratory for diagnosis of Mucopolysaccharidoses .Seven years' experience .

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Abstract

Background:

Mucopolysaccharidoses are a group of lysosomal storage diseases with deficiencies in specific enzymes which lead to accumulation of partially degraded Glycosaminoglycans (GAGs) in various tissues causing a wide variety of manifestations. It is a heterogeneous disease and a wide spectrum of severity is observed in patients. Most of the patients appear normal at birth, but in patients with a more severe phenotype, clinical symptoms initiate before age of 2 to 4 and they may die before the age of 10.

Up to date, nine enzyme deficiencies are diagnosed which lead to different types of MPS. The current average rate of incidence is 1 per 25000000 live births.

The accumulation of GAGs lead to excretion in urine, so one of the laboratory methods for MPS diagnosis is urinary GAG analysis.

Objective:

In the present study, data of 7-years experiences in the diagnosis of the MPS in laboratory of Children's Medical Center a referral hospital for metabolic patients in IRAN , are represented.

Methods:

All of the hospitalized and outpatients from March 2012 to March 2019 who requested a urine test for GAGs analysis were participated in this study.The urinary GAG analysis was performed through DMB and Berry spot tests. All patients with positive and trace results or disease-related symptoms were evaluated for enzyme activity assay or /and genetic study. We also followed up the patients in terms of the type of treatment received and possible death .

Results:

There were 248 positive and 244 trace(mild positive) results out of 1338 performed tests. The negative results mostly belonged to hospitalized newborns. Most of the patients with positive results were referred by Pediatric Endocrinologists, Cardiologist, Neurologist, Gastroenterologist, and Orthopedics. The number and percentage of each type of MPS,

patients who received ERT, and/or HSCT and the outcome for each patient will be discussed in details in the congress.

Conclusion:

Due to High frequency of consanguineous marriages, there is a high prevalence of MPS in Iran. Urinary GAG analysis helps screening and early diagnosis. In order to achieve maximum effect of available therapies, early detection and intervention are critical. More accessibility of enzymatic and genetic assays and PND would be effective in the reduction of morbidity and mortality of the disease.

BIOGRAPHY

Sedigheh Shams,PhD of clinical chemistry is the professor of in pathology department in Tehran University of Medical Sciences. She has more than 25 years' experience in children hospitals as lab director and academic staff. Her research interests include metabolic and genetic disorders especially the in MPS disease.In addition she is recently got a position in laboratory of Growth and Development Research Center Iran Metabolic Center .She collaborates in establishment The Newborn Screening Project for metabolic disorders by MS/MS and GC/MS in IRAN.

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