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Prevalence and outcome of Treatable lysosomal storage diseases in Children from north- eastern part of Libya

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Background: Lysosomal storage diseases are important inherited metabolic disorders with major healthcare concern. LSD occur at all ages and are clinically diverse, they differ greatly in their rate of progression and represent a large burden of illness in the population. The range of manifestations includes organomegaly, disturbed function of visceral organs, skeletal effects and neurological features. There is no specific or curative treatment for most lysosomal storage diseases, supportive and palliative treatment are nonetheless of great benefit .however recently recombinant DNA technology has led to the development of enzyme –replacement therapy for several lysosomal diseases. In this paper we present our experience from north-eastern part of Libya with some of this treatable LSD.

Methods: Twenty Seven cases with treatable LSD disorders were diagnosed and followed up in Benghazi children hospital during the period from (1997 – 2013) with Four different treatable LSD and general prevalence rate of 2.7/100.000. Initial work-up focused on clinical, laboratory and radiological evaluation. Lysosomal enzyme assay in peripheral blood leukocytes were performed according to standard techniques.

Results: Twelve children were diagnosed as Gaucher disease with prevalence rate of 1.2/100.000 The median age at diagnosis was 1 year, male to female ratio 1:1 , Six cases of Gaucher type one four of them on enzyme replacement two brother and sister with mild form on regular follow up without treatment . Four cases of type three all receiving cerozyme as replacement therapy and one child diagnosed as Gaucher type two died after one year of diagnosis he was also in Enzyme replacement therapy none of our patients has bone marrow transplant. Nine children were diagnosed as MPS1 with prevalence rate of 0.9/100.000 the median age at diagnosis was three years, male to female ratio 1.2:1, four have severe MPS1, three were moderate to severe form and one was mild form all of them received alldrozyme replacement therapy non had bone marrow transplant, of severe form one died and one lost follow up. Three brothers ,two of them were twin Diagnosed as MPS2 (Hunter) with low prevalence rate 0.3/100.000 all from Tobrok in the far east point of Libya ,non of them received enzyme replacement therapy till now . Three children were diagnosed as Pompe disease, with prevalence rate of 0.3/100,000. One patient was suspected as infantile pompe at one and half month of age died before result of enzyme assay. Two brothers diagnosed as juvenile pompe not on enzyme replacement therapy yet.

Conclusion: Lysosomal storage diseases, especially Gaucher and MPS1 disease, may represent important pathologies in our population and their prevalence rate was similar to the reported prevalence rate from other parts of the world .Specific diagnosis and follow-up is the key step in the accurate management and treatment of these patients.

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