Prevalence and outcome of treatable lysosomal storage diseases in children from north-easter part of Libya

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Background: Lysosomal Storage Diseases (LSDs) are important inherited metabolic disorders with major healthcare concern. LSDs occur in 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 LSDs, 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 these treatable LSD.

Methods: 27 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 with Gaucher disease, having a 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 1, 4 of them on enzyme replacement, 2 brother and sister with mild form on regular follow up without treatment. Four cases of type three all receiving cerozyme as replacement therapy and 1 child diagnosed as Gaucher type 2 died after an 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 3 years, male to female ratio 1.2:1, 4 have severe MPS1, 3 were moderate to severe form and 1 was mild form. All of them received alldrozyme replacement therapy; one had bone marrow transplant of severe form, 1 died and 1 lost follow up, 3 brothers, 2 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. None of them received enzyme replacement therapy till now, 3 children were diagnosed with Pompe disease, having a 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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