Specific Treatment for Lysosomal Storage Disorders:
Enzyme Replacement Therapy, Bone Marrow Transplant and Others

SYR Lee, STS Lam, DKK Ng, KY Chan, KW Ng

Abstract
In this article, we review specific therapies that tackle the basic biochemical defects of lysosomal storage diseases. These include bone marrow transplantation, substrate deprivation therapy, enzyme replacement therapy and enzyme enhancement therapy. We particularly update the progress of development of enzyme replacement therapy, which plays a major role in the treatment of lysosomal storage diseases. Nowadays, enzyme products have been developed and marketed for treatment of Gaucher disease, Fabry disease, mucopolysaccharidosis I and currently there are ongoing trials of enzyme replacement for the treatment of glycogen storage disease II, mucopolysaccharidosis II and VI and Niemann-Pick B disease. Enzyme replacement therapy has a definite role in treatment of lysosomal storage diseases as it can ameliorate the signs and symptoms of the diseases. However, there are certain limitations. Enzyme replacement therapy is ineffective in improving or preventing neurological involvement. Response to treatment is slow in some situations, for example, bone involvement in Gaucher disease. It may also be unpredictable in other situations, for example, lung involvement in Gaucher disease. Hence, there is room for incorporation of other treatment modalities. One example is provided by mucopolysaccharidosis type I, in which bone marrow transplantation has a definite role as it prevents psychomotor retardation when carried out before significant brain damage occurs. (HK J Paediatr (new series) 2004;9:231-239)

Key words: Bone marrow transplant; Enzyme enhancement therapy; Enzyme replacement therapy; Lysosomal storage diseases; Substrate reduction therapy