THERAPEUTIC POTENTIAL OF ENZYME SUPPLEMENTATION IN GENETIC DISORDERS: A SYSTEMATIC REVIEW

SKRIPSI

AMADITA JACINDA DIVA
10016224222

PROGRAM STUDI S1 FARMASI
FAKULTAS FARMASI
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Diajukan sebagai salah satu syarat untuk memperoleh gelar Sarjana Farmasi

AMADITA JACINDA DIVA
10016224222

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ABSTRAK

Therapeutic Potential Of Enzyme Supplementation in Genetic Disorders: A Systematic Review

Amadita Jacinda Diva

Program Studi S1 Farmasi, Universitas Bakti Tunas Husada

Abstrak

Penyakit genetik akibat defisiensi enzim, khususnya *Lysosomal Storage Diseases* (LSDs) seperti *Pompe Disease, Gaucher Disease, Fabry Disease dan Mucopolysaccharidoses* (MPS) merupakan penyakit langka yang berkembang secara bertahap dan berdampak pada penurunan kualitas serta harapan hidup pasien. *Enzyme Replacement Therapy* (ERT) menjadi terapi utama yang banyak digunakan. Penelitian ini bertujuan menganalisis potensi dan efektivitas ERT serta mengidentifikasi tantangan, keterbatasan dan faktor yang memengaruhi keberhasilan terapi. Penelitian menggunakan metode *Systematic Literature Review* terhadap artikel ilmiah tahun 2020 – 2025 dari basis data PubMed dan ScienceDirect yang dianalisis secara deskriptif kualitatif. Sebanyak 45 artikel yang dianalisis menunjukkan bahwa ERT efektif memperbaiki fungsi organ, meningkatkan kemampuan motorik dan fisik, menurunkan akumulasi substrat patologis serta meningkatkan kualitas dan harapan hidup pasien, terutama bila terapi dimulai sejak dini. Namun, ERT memiliki keterbatasan berupa kebutuhan terapi seumur hidup, biaya tinggi pembentukan antibodi anti-enzim, keterbatasan distribusi enzim ke jaringan target serta ketidakmampuan menembus sawar darah otak. ERT memiliki potensi terapeutik yang signifikan sebagai terapi standar pada penyakit genetik akibat defisiensi enzim. Keberhasilan terapi sangat dipengaruhi oleh waktu inisiasi, kondisi klinis awal, status imun, variasi genetik serta kepatuhan terapi jangka panjang

Kata Kunci : ERT, Pompe, Gaucher, Fabry, MPS

Abstract

Background: Genetic disorders caused by enzyme deficiencies, particularly Lysosomal Storage Diseases (LSDs) such as Pompe Disease, Gaucher Disease, Fabry Disease, and Mucopolysaccharidoses (MPS), are rare disorders that progress gradually and lead to reduced quality of life and life expectancy. Enzyme Replacement Therapy (ERT) is the primary treatment widely used for these conditions. This study aims to analyze the potential and effectiveness of Enzyme Replacement Therapy (ERT) and to identify the challenges, limitations, and factors influencing treatment success. This study employed a Systematic Literature Review of scientific articles published between 2020 and 2025 from the PubMed and ScienceDirect databases, analyzed using a qualitative descriptive approach. A total of 45 analyzed articles demonstrated that Enzyme Replacement Therapy (ERT) is effective in improving organ function, enhancing motor and physical abilities, reducing pathological substrate accumulation, and increasing patients' quality of life and life expectancy, particularly when therapy is initiated early. However, ERT has several limitations, including the need for lifelong treatment, high treatment

costs, the development of anti-enzyme antibodies, limited enzyme distribution to target tissues, and the inability to cross the blood–brain barrier. Enzyme Replacement Therapy (ERT) has significant therapeutic potential as a standard treatment for genetic disorders caused by enzyme deficiencies. Treatment success is strongly influenced by the timing of therapy initiation, baseline clinical condition, immune status, genetic variability, and long-term treatment adherence.

Keywords: ERT, Pompe, Gaucher, Fabry, Mucopolysaccharidoses