Prevalence of Lysosomal Storage Diseases in Malaysia

Affandi Omar\(^1\), Norashareena Mohd Shakrin\(^1\), Nor Azimah Abdul Aziz\(^2\), Siti Aishah Abdul Wahab\(^2\), Lua Seok Hian\(^3\), Yusnita Yaakob\(^2\) and Julaina Abdul Jalil\(^1\)

\(^1\)Biochemistry Unit, Specialised Diagnostic Centre, Institute for Medical Research
\(^2\)Molecular Diagnostic and Protein Unit, Specialised Diagnostic Centre, Institute for Medical Research

Abstract

Lysosomal storage diseases (LSD) are a group of genetic disorders resulting from defective lysosomal metabolism and subsequent accumulation of substrates. It can be categorised into a group of lipidoses, mucopolysaccharidoses (MPS), cholesterol ester, glycogen and mucolipidoses. In Malaysia, information about the incidence of LSDs is relatively limited. The aim of this study was to determine the prevalence of LSD in Malaysia and compare them with other countries. A retrospective epidemiological study was conducted by covering the period between 2000 and 2017. The birth prevalence of LSD in Malaysia was calculated based on 83 confirmed cases by enzymatic assay and/or mutation analysis. The combined birth prevalence for all LSD in Malaysia is 0.93 per 100,000 live births compared to the Netherlands (Europe) and United Arab Emirates (Asia) which were 14 and 26.9 per 100,000 live births respectively. Within the group of lipidoses, Gaucher disease is the most frequent LSD with calculated birth prevalence of 0.31 per 100,000. Metachromatic leukodystrophy (MLD) was diagnosed in 18.07% of all diagnosed cases and the calculated birth prevalence was 0.20 per 100,000 live births. Within the group of MPS, MPS IVA has the highest calculated birth prevalence (12.2% of all cases of MPS diagnosed) of 0.33 per 100,000 live births. The birth prevalence of MPS II was 0.23 per 100,000 live births (0.45 per 100,000 male live births), representing 36.59% of all cases of MPS diagnosed. The combined birth prevalence for all MPS was 0.59 per 100,000 live births. In conclusion, this study revealed that LSD is not rare in Malaysia as compared to other countries; therefore awareness of LSD should be promoted among health care provider in Malaysia since specific therapies are available for most of the cases diagnosed.

**Keywords:** Prevalence; Lysosomal Storage Disease; incidence

*Author for Correspondence*