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DIAGNOSIS OF ORGANIC ACIDAEMIAS IN A SELECTED PAEDIATRIC POPULATION: SIX YEAR EXPERIENCE IN A SINGLE CENTRE

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INTRODUCTION

Organic acidaemias (OA) are a group of heterogeneous inherited metabolic disorders. The early diagnosis improves the efficacy of treatment.

OBJECTIVES

To describe the clinical spectrum of patients with OA and the diagnostic experience of OA at Lady Ridgeway Hospital for Children (LRH).

DESIGN, SETTING AND METHOD

We conducted a descriptive study of patients referred to department of chemical pathology for metabolic screening from 2010 January to 2016 January. Screening was by the acyl carnitine and amino acid profiles of whole blood performed by tandem mass spectrometry and biotinidase activity by immunoassay. Positive screening tests were confirmed by analysis of urine organic acids (OAA), biotinidase activity by spectrophotometry and mutations for biotinidase deficiency. Above investigations were performed in reputed overseas laboratories.

RESULTS

A total of 459 patients were screened. Among the 39 patients with a positive screening test for OA, 14 patients have been confirmed as having OA. In the group with confirmed diagnosis, the mean age of appearance of symptoms and the age at biochemical diagnosis were 18.2 and 33 months respectively. Biotinidase deficiency was the most frequent disorder (6). Other OA detected were propionic acidaemia (2), glutaric aciduria type-1 (2), isovaleric acidaemia (2), methylmalonic aciduria (1) and beta-ketothilase deficiency (1). The most frequent symptoms were seizures (42.8%), altered level of consciousness (35.7%), respiratory distress, repeated vomiting and global developmental delay (28.5%). Rest of the 25 patients in whom screening was positive for OA, have not been further investigated due to unaffordability or death before further investigations.

CONCLUSION

All the patients who had a positive screening were confirmed to have an OA. Out of the 39 patients with a positive screening test, only around one third of patients were able to undergo confirmatory testing. This highlights the need to improve diagnostic facilities for OA.