An epidemiological study of Lesch-Nyhan Disease (LND) in the UK

Study Start Date: ***June 2006 to June 2008***

Research Worker: ***Dr Gillian T McCarthy, Honorary Consultant Paediatrician, Chailey Heritage Clinical Services***

This was an epidemiological study to find out more about LND in the UK.

Data was gathered during 2006-2008. The findings were analysed and written up for publication.

Developmental Medicine & Child Neurology published the findings in 2010.

- McCarthy GT, Green EM, Ogunbona O, Simmonds HA, Fairbanks L, Pountney T, Bryant E. (2010). *A population study of Lesch-Nyhan disease in the UK*. Developmental Medicine and Child Neurology 53: 34-39.

ABSTRACT

Aim: The aims of this study were to identify all people with Lesch-Nyhan disease (LND) born in the U.K. between 1988 and 2008, and to obtain a clinical profile including age at diagnosis, genetic background, family-history, neurological signs and medications.

Method: Potential participants were contacted through the British Paediatric Neurology Surveillance Unit. Questionnaires were sent to each child's paediatric neurologist or primary consultant. Two purine laboratories provided metabolic information.

Results: Twenty-three live males with LND were identified in the 0- to 20-year age band, and eight live males over the age of 20 years were identified. Thirty-one live people with LND were identified in the UK in 2008, giving a prevalence of 1 in 2 million people. Over the 20 years of study, there was a mean incidence rate of 0.18 per 100,000 live births, range 0 to 0.5.

Interpretation: To our knowledge, this study is the first to provide details of the prevalence and incidence of LND in the UK. The data highlight that clinical profiles, at the time of diagnosis, and management of the disease are variable. There is the need for ongoing monitoring of allopurinol dosage and metabolic screening.

Acknowledgements
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