Feasibility study of breath ammonia device to manage children with urea cycle defects (AmBeR). Industry collaboration

Key Words : Children and Young People; urea cycle defects, ammonia, biomarker device

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Abstract

Urea cycle defects (UCDs) are a group of inborn errors of metabolism that result in the absence or reduction in a number of enzymes or cofactors involved in the urea cycle by which protein breakdown ammonia is made into harmless urea. Ammonia is a toxic substance and is normally kept at blood levels below 30-50 µM. However, in UCD sufferers, it can reach several hundred micromolar. At elevated concentrations, ammonia has severe effects. Principally, it crosses the blood-brain barrier where it causes neurotoxicity. Recently, a device (AmBeR) for measuring ammonia in breath has been developed and used at the clinical point of care. Such a device may have the potential to enhance the management of UCD patients if it can be shown to correlate with blood ammonia and that this can be correlated with patient wellbeing on a daily basis. This study is a very early phase intervention designed to test whether children with UCD and associated conditions aged >3 years, can use a breath device satisfactorily to generate breadth to adequately measure ammonia that correlates with blood ammonia taken at routine clinic visits. Patients/parents/physicians will also complete a short questionnaire on user experience.