British Society for Paediatric Endocrinology & Diabetes

RESEARCH and INNOVATION AWARD REPORT

As per the agreed conditions of the Grant, the BSPED requires a written report from the award winner demonstrating how the grant benefited the applicant/home institution and how the funding has enabled advances in endocrinology/science/medicine as a whole. The final written report is due two years after the grant is awarded detailing final findings, whether positive or negative. The report should include details of any publications resulting from the funding and will be published on the BSPED website.

Max 1000 words in total.

Name	Dr Katherine Hawton
Title of project	Rapid-onset obesity, hypoventilation, hypothalamic dysfunction and autonomic dysregulation (ROHHAD) BPSU Study
Year of award commencement	2022
Date of report	10/11/2024
Method	This study is being conducted as British Paediatric Surviellance Unit (BPSU survey) through the Royal College of Paediatrics and Child Health (RCPCH). All consultant paediatricians (or equivalent) are sent a monthly reporting e-card every month to report any cases fulfilling the below criteria: **Reporting instructions** Please report all existing and new cases you have seen in the last month fulfilling this case definition of ROHHAD (in the UK and Republic of Ireland) not previously notified. **Case definition** Please report any patients under the age of 15 years with: 1. Rapid onset of obesity in childhood after 12 months of age in a previously healthy child a. with evidence of crossing three major centile lines for weight within a 12-month period (eg 9th to 75th centile) b. or family photographic evidence of highly significant weight gain in 12 months, in cases with no baseline weight measurement AND 2. Sleep-disordered breathing requiring overnight respiratory support Exclusion, any of:

	• Evidence of structural abnormality of hypothalamic-pituitary axis on MRI • Congenital structural brain abnormality eg, septo-optic dysplasia • Acquired brain injury e.g., craniopharyngioma • Other causes identified for obesity eg, Prader Willi syndrome, monogenic obesity • PHOX2B variant associated with congenital central hypoventilation syndrome There will be an initial 13-months surveillance period followed by a follow-up questionnaire being sent out to each clinician 12 months after the initial case identification. The primary aims of the study are: • To measure how many new and existing cases there are of ROHHAD in the UK/RoI • To identify the clinical features of patients diagnosed with ROHHAD in the UK/RoI • To identify how patients with ROHHAD are being diagnosed and managed in UK/RoI The study has been approved by the Research Ethics Committee and the Health Research Advisory Group (England and Wales) (REC Reference 22/SW/0040; IRAS project ID 277788), the Public Benefit and Privacy Panel (Scotland), the Privacy Advisory Committee (Northern Ireland) and has been granted Section 251 HRA-CAG permission (CAG reference: 22/CAG/0040). The study website can be found below: https://www.rcpch.ac.uk/work-we-do/british-paediatric-surveillance-unit/ROHHAD
Results	Unfortunately, there have been a number of delays with the study being commenced. The phase 1 application had been already approved by the BPSU at the time of applying for the BSPED prize, and it took a long time for a response to the phase 2 application. In addition the BPSU methodology has recently changed with the development of a new online data-reporting platform and this has led to further delays. The study commenced surveillance in November 2024 (which will capture cases see in October 2024 onwards) for 13 months and clinicians have begun reporting cases. Therefore, no results are available yet, but results from initial surveillance will be available by the end of 2025, and from follow-up data by the end of 2026.
Conclusion	Conclusions will be made once results have been analysed.
Publications	Hawton K, Hilliard T, Langton-Hewer SC, Burren C, Crowne EC, Hamilton-Shield JP, Giri D. Rapid-onset obesity, hypothalamic dysfunction, hypoventilation, and autonomic dysregulation syndrome - neuro-endocrine tumours (ROHHAD-NET): case series

	and learning points. J Pediatr Endocrinol Metab. 2023 Jan 25;36(4):418-423. doi: 10.1515/jpem-2022-0376. PMID: 36696572. Hawton K, Giri D, Crowne E, Greenwood R, Hamilton-Shield J. The Enigma That Is ROHHAD Syndrome: Challenges and Future Strategies. Brain Sciences. 2024; 14(11):1046
Benefit to applicant	This research study has provided the basis of an MD into ROHHAD syndrome. I have had the opportunity to develop a range of research skills through this study, including development of protocols, applying for ethical approvals, patient and public involvement in research and writing of scientific papers. I am committed to further developing my research skills through studying for an MD and contributing to paediatric endocrine research throughout my career. I am also developing some useful clinical expertise in ROHHAD syndrome.
Benefit to department/ institution	For the Paediatric Endocrinology and Diabetes department in Bristol, this grant has enabled for a national study led by department to be conducted. The grant has also enabled clinicians in Bristol to develop expertise in ROHHAD syndrome. For BSPED, we hope that this study will generate useful findings for clinicians seeing children and young people with ROHHAD, or suspected ROHHAD, in the future. This study may provide information to drive improvements in clinical practice by identifying the prevalence, clinical features, diagnostic process and management of ROHHAD. Information provided by this study could help to lead to changes to standardise the diagnosis and management of ROHHAD in order to eventually improve patient outcome in this potentially devastating condition. We also hope to raise awareness of the condition, and by doing so, enable earlier diagnoses to be made.

Please email all reports to bsped@endocrinology.org