COLLABORATIVE WORKING EXECUTIVE SUMMARY: ADULT TESTING FOR RARE EPILEPSIES: SHEFFIELD TEACHING HOSPITALS NHS FOUNDATION TRUST AND UCB PHARMA LTD

Project Partners: Sheffield Teaching Hospitals NHS Foundation Trust (STHT) and UCB Pharma Ltd

Project Summary: The project aims to optimise the identification, diagnosis, and management of adults with undiagnosed, misclassified, or unidentified rare genetic epilepsies, including Dravet syndrome and other developmental and epileptic encephalopathies (DEEs). Many adults currently receiving care transitioned from paediatric services before routine access to genetic testing was available, resulting in under-diagnosis and sub-optimal treatment pathways.

Key deliverables from the project include:

- **Increased identification and confirmation of rare genetic epilepsies** in adult patients through systematic consenting and access to NHS England epilepsy genetic testing.
- **Improved clinical decision-making**, including treatment optimisation in line with NICE and national guidelines (e.g. discontinuation of contraindicated medicines and access to condition-specific therapies where appropriate).
- **Generation of real-world evidence (RWE)** to improve understanding of adult rare epilepsy phenotypes and care pathways.
- **Knowledge sharing and capability building** across participating clinical sites through peer-to-peer education, Steering Group oversight, and multidisciplinary collaboration.
- **Dissemination of outcomes**, including an end of project outcomes report, poster presentations, and peer engagement

Key Responsibilities:

Joint Responsibilities (UCB and STHT)

- Deliver the project in accordance with the agreed objectives, timelines, and ABPI collaborative working principles.
- Cooperate on protocol development, reporting, and publication of outcomes in a transparent and compliant manner.
- Ensure compliance with data protection legislation, confidentiality obligations, and freedom of information requirements.

Sheffield Teaching Hospitals NHS Foundation Trust

- Ensure suitable clinical expertise, facilities, and governance structures are in place.
- Employ and support a genomic practitioner to deliver patient finding, consenting and genetic testing activity.
- Generate anonymised real-world data and contribute to dissemination in conjunction with other participating centres.

UCB Pharma Ltd

- Provide financial and non-financial contributions to enable delivery of the project.
- Contribute neurological expertise, alongside training, education, and peer-to-peer support.
- Review and comment on publications and outputs prior to dissemination, ensuring compliance with the ABPI Code and applicable law.

Summary of expected outcomes/benefits for each party:

For the patient

- The ability to receive the correct diagnosis and all the implications associated with optimal management of their disease
- The ability to align treatment decisions with NICE guidelines for historically misclassified patients
- The potential to provide relief for parents by gaining a diagnosis
- The ability to be entered into appropriate clinical research when genetic diagnosis is part of the inclusion criteria
- Gaining a better experience within the healthcare system

For the NHS

- The provision of resources and development of pathways for genetic testing
- The ability to provide optimal treatment in line with national guidelines which has the potential to reduce hospital admissions and seizure burden
- Support improvement in future diagnosis and the potential to reduce longer term healthcare costs
- Provision of optimal care for patients
- The opportunity to configure services around patient needs

For UCB

- Increased, accurate diagnosis of rare epilepsies increases the number of patients identified who may be eligible for treatment with UCB therapies
- The opportunity to better understand patient profiles for future indications and to further inform future UCB products and service developments
- The opportunity to increase understanding of the challenges faced by the NHS in delivering complex patient services and care and to develop strong relationships with the NHS.
- The ability to optimize uptake of innovative medicines in appropriate patients.

Funding Contributions: The project will use the pooling of skills, experience and resources from both parties:

UCB will fund 0.4WTE Band 5 genomics practitioner for 2 years (Direct costs), and UCB employee time (Indirect cost) for training support, project management and project publication contributing to a total cost of £51,882

The NHS will contribute a total cost of £50,859 for project delivery, including HCP time, education and training, increased genetic testing and associated ancillary tests, as well as publication write up. The NHS contribution to the project does not include any transfer of monies, it is based on the approximate costs of resource allocation.

Transfers of value will be disclosed on Disclosure UK in accordance with the ABPI Code.

Summary of project outcomes and lessons learned will also be published within 6 months of completion on the UCB website

Project commenced October 2024

Anticipated completion date December 2026

For more information, please contact: partnerships@ucb.com

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