Executive Summary

Birmingham Women and Children and UCB pharma Ltd: Adult testing for rare epilepsies

Collaborative Working Project Executive Summary

Birmingham Women and Children and UCB Pharma Ltd, are embarking on a collaborative working project to optimise the identification of adult patients with undiagnosed, unidentified, or misclassified rare genetic epilepsies also classified as DEE's (developmental and epileptic encephalopathy). The parties have agreed that the project should be undertaken in accordance with the principles and guidance relating to collaborative working between the NHS and the pharmaceutical industry.

An estimated 1:15,000 people have Dravet syndrome in the UK, if this figure is correct, then current figures suggest that there are less than a quarter of the people with Dravet identified in the locality. The project will facilitate the service and if the incidence is as high as suspected then the project will support identifying patients by adding additional resources and expertise, including clinician hours and service development.

The current NHS England epilepsy genetics panel has over 400 genes on it, but the most common positive answer is SCN1A (80% of patients have a genetic mutation of this variant), the cause of Dravet syndrome. It is expected that with an increase in genetic testing potentially half of all results will result in a clinical change of treatment.

The primary objective of the project is to uncover and identify rare epilepsy patients by consenting and testing historical and future adult patients and capture different ways that DEE patients present and manifest within clinic. For people with Dravet this may include stopping certain medications (such as sodium channel drugs) or unlocking the ability to prescribe new medications only licensed for Dravet. There is a benefit for both patients and carers and additionally an understanding of genetic cause helps with diagnosis, prognosis and provides the fullest possible explanation of why someone has a complex epilepsy.

The resource commitments from the parties are similar.

The project commences in September 2023 and is an 18 -month commitment.

Intended benefit:

For the patient	• Important correct diagnosis and all implications associated with disease. • Ending diagnostic odyssey • Aligned treatment decisions with NICE guidelines for historically misclassified patients. • Parental relief of diagnosis • Surveillance • Trial access when appropriate – when genetic entry inclusion is required. • Prognostication • Disease related community support • Better experience of the healthcare system
For the NHS	• Provision of resources for genetics • Treatment in line with national guidelines leading to less hospital admissions with seizure burden. • Support improvement in future diagnosis and reductions in long term healthcare costs. • Higher quality care • Services configured around patient needs
For UCB	• Accurate diagnosis of rare epilepsies may include the opportunity to use UCB therapies.

	• Better understanding of patient profiles for adult patients for future indications and to further inform future UCB products and service developments. • Deeper relationships / partnership working with NHS. • Potential expansion of the relevant and eligible patient population because of the activity • Increase in the appropriate use of medicines aligned to local or national guidance • Better understanding of the challenges faced by the NHS in delivering high-quality patient services and care • Faster implementation of NHS policy which may be relevant to an organisations business.

It is anticipated that a report on the outcomes from this collaborative working project will be published April 2024

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