EPD/23/13 - Epilepsy: Precision Investigation, Stratified Care, and Outcomes of Therapy (EPI-SCOT)

Epilepsy is the most common serious neurological condition. One in 100 people is diagnosed with epilepsy during childhood. Children with epilepsy have a significantly increased risk of learning, behavioural, and psychological difficulties and they are seven times more likely to die during childhood than their peers. Through new anti-seizure medicines continually come onto the market, outcomes for children with epilepsy have not improved in the past 30 years. My research has taken a different approach to talking this problem. Through this proposal I aim to understand epilepsy in terms of causes and risk factors. My vision is that through targeting causes and risk factors we will transform outcomes for people living with epilepsy.

Until recently, for most children with epilepsy no underlying cause for their seizures could be identified. Recent advances in genetics and brain imaging have dramatically changed this. A cause can now be found for most young children with epilepsy. Because there are so many different causes, each of which may follow a different course, we now talk about “the epilepsies”, a collection of rare conditions, rather than “epilepsy” as a single disease.

Despite the advances in our understanding of underlying causes, many children and young people with epilepsies have difficult to control seizures and experience challenges with learning and behaviour. Up to one third of children with epilepsy continue to have seizures no matter how many treatments we try. Developmental, behavioural and psychological outcomes are as important for people with epilepsy and their families as seizure frequency/control, and are strongly associated with quality of life.

In this study we will follow children with epilepsies more closely than ever before. All children in Scotland diagnosed with epilepsy before their 16th birthday will be included. We will ensure that children are consistently investigated for underlying causes, regardless of which region within Scotland they live. We will use up-to-date diagnostic genetic testing techniques. We will gather data from multiple sources on each participant. Parents will be asked to keep regular seizure diaries with a user-friendly online tool. Families will be supported here by the research team. Validated Questionnaires will be used to assess development, behaviour and the impact of epilepsy on the quality of life of children. Research team field workers will help parents to complete questionnaires. We will obtain fully informed consent to access clinical data from parents/guardians, and in the case of older children from patients with epilepsy themselves. We will capture in depth information about the participants at baseline – i.e. as soon as they have been given a diagnosis of epilepsy – and again one year after diagnosis. Between baseline and one year follow-up we will ensure that accurate records of every epilepsy treatment used are kept so we can

investigate which treatments are associated with better seizure control. We will carefully study relationships between the baseline and follow-up data so we can learn about which factors are associated with which outcomes for children and young people with epilepsy. The outcomes we will look at include seizure control, development, behaviour, and quality of life.

Analysis of our data will enable us understand what factors determine important outcomes for patients with epilepsy and their families, and identify which of these we might be able to influence to improve outcomes. This research will provide important data on how common genetic forms of epilepsy are, and what these look like in terms of seizures, development, and associated medical problems. This will allow families to be given accurate information early on in diagnosis, and will be invaluable to scientists developing new therapies so they can assess whether any new treatments are effective