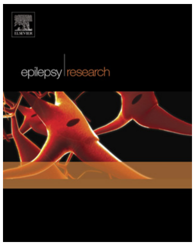
Epilepsy Research (2015) **112**, 64—71

ELSEVIER

journal homepage: www.elsevier.com/locate/epilepsyres



‘‘It’s good to know’’: Experiences of gene identification and result disclosure in familial epilepsies

Danya F. Vears[a,b], **Karen L. Dunn**[b], **Samantha A. Wake**[b,c], **Ingrid E. Scheffer**[a,b,d,*]

[a] *Epilepsy Research Centre, Department of Medicine, University of Melbourne, Austin Health, Melbourne, Australia*
[b] *Department of Paediatrics, University of Melbourne, Royal Children’s Hospital, Melbourne, Australia*
[c] *Victorian Clinical Genetics Services, Murdoch Childrens Research Institute, Royal Children’s Hospital, Melbourne, Australia*
[d] *The Florey Neurosciences Institutes (Austin), Melbourne, Australia*

Received 15 January 2015; accepted 9 February 2015
Available online 16 February 2015

KEYWORDS
Epilepsy;
Genetics;
Qualitative research;
Genetic testing;
Genetic counselling

Summary Recognition of the role of genetics in the epilepsies has increased dramatically, impacting on clinical practice across many epilepsy syndromes. There is limited research investigating the impact of gene identification on individuals and families with epilepsy. While research has focused on the impact of delivering genetic information to families at the time of diagnosis in genetic diseases more broadly, little is known about how genetic results in epileptic diseases influences people’s lives many years after it has been conveyed.

This study used qualitative methods to explore the experience of receiving a genetic result in people with familial epilepsy. Interviews were conducted with individuals with familial epilepsies in whom the underlying genetic mutation had been identified. Recorded interviews underwent thematic analysis. 20 individuals from three families with different epilepsy syndromes and causative genes were interviewed. Multiple generations within families were studied. The mean time from receiving the genetic result prior to interview was 10.9 years (range 5—14 years).

Three major themes were identified: 1) living with epilepsy: an individual’s experience of the severity of epilepsy in their family influenced their view. 2) Clinical utility of the test: participants expressed varying reactions to receiving a genetic result. While for some it provided helpful information and relief, others were not surprised by the finding given the familial context. Some valued the use of genetic information for reproductive decision-making, particularly in the setting of severely affected family members. While altruistic reasons for participating

---

[*] Corresponding author at: Epilepsy Research Centre, Austin Health, Heidelberg, Victoria 3084, Australia. Tel.: +61 3 90357112; fax: +61 3 94962291.
E-mail address: scheffer@unimelb.edu.au (I.E. Scheffer).

http://dx.doi.org/10.1016/j.eplepsyres.2015.02.011
0920-1211/

in genetic research were discussed, participants emphasised the benefit of participation to them and their families. 3) 'Talking about the family genes': individuals reported poor communication between family members about their epilepsy and its genetic implications. The results provide important insights into the family experience of genetic epilepsies and communication within families. This information can be used to inform the development of guidelines for genetic result disclosure and genetic counselling for individuals and families with epilepsies.

Introduction

Technological advances in genetics have lead to increasing integration of disease gene mutational analysis into clinical practice for many disorders including cystic fibrosis and hereditary cancers (Half et al., 2009; Massie et al., 2010; Weitzel et al., 2011). There has been considerable success with gene discovery in the epilepsies, predominantly in monogenic epilepsy syndromes. While genetic results often provide answers to the cause of a patient's disorder, little attention has been paid to the positive and negative outcomes from disclosing these results in epilepsy.

In particular, research into the impact of receiving genetic information for individuals and families with epilepsy, both in clinical and research settings, has lagged behind with the focus often on heritable cancers where positive mutation testing indicates the individual is at-risk of developing the condition rather than confirming a diagnosis (Forrest et al., 2008). In these studies, adverse psychological responses to genetic findings are uncommon and often transient (Marteau and Croyle, 1998; Palmer et al., 2002). It is not known whether these outcomes can be extrapolated to other conditions, with different clinical features, prognoses, health implications, inheritance patterns, and in different contexts, including the research setting.

Since discovering the first gene for epilepsy in 1995, the recent burgeoning in epilepsy gene identification has meant we need to understand the impact of gene discovery on patients and their families. In cases where genetic causes have been identified, no research has been conducted to assess the benefits and harms of disclosing this genetic information to individuals and their wider family with epilepsy, although the critical nature of gaining this information has been emphasised (Ottman et al., 2010).

The purpose of this study is to investigate the experience of receiving a genetic cause for epilepsy in the research setting and the impact of this information on the individual with epilepsy and their relatives.

Materials and methods

Recruitment

Potential participants were identified from three families who previously participated in the Epilepsy Genetics research program at The University of Melbourne at Austin Health. As our research program was commenced almost 25 years ago, we were able to study individuals who had received a genetic cause for their epilepsy a mean of 10 years prior to study. This provided an opportunity to evaluate the long term impact of their genetic research finding after many years.

Two methods of sampling were used to gain a broad range of perspectives. First, purposive sampling was used where potential participants were selected from families with an identified genetic mutation. Individuals were sent letters of invitation to participate and then provided with further information. Second, snowball sampling was utilised, whereby potential participants suggested family members interested in participating (Liamputtong and Ezzy, 2005).

The epilepsy syndromes and genetic mutations in the three families studied have been reported (Marini et al., 2003; Scheffer et al., 1995, 2007; Steinlein et al., 1995). Genetic results were delivered according to the following protocol: a letter was sent to participants informing them a genetic mutation was found in their family; they were offered an opportunity to contact the research group for further information about their mutation status. Where participants made contact, results were given to them or their doctor, and genetic counselling was offered.

Efforts were made to study the impact of genetic findings on the wider family by including the spouse of affected individuals who also had affected children with a gene mutation.

Individuals were excluded if English was not their first language or if they were less than 18 years of age.

Interviews

Qualitative methods, commonly used in genetic counselling research, were employed. Semi-structured interviews were conducted to gain detailed information about the participants' experiences of gene identification and disclosure. A list of questions aimed to explore (i) how individuals recalled receiving their genetic results; (ii) what additional information about their results they would have liked; (iii) the impact genetic results had on themselves and their families; and (iv) their seizures and experiences of epilepsy. Interviews were undertaken by one researcher (DV) in participants' homes and conducted one-to-one or with two family members if requested. The average interview length was 58 min (range 40—120 min). Interviews were audio-recorded, transcribed and de-identified.

Analysis

The dataset underwent thematic analysis using an inductive rather than deductive approach where codes were developed from the interviews (Braun and Clarke, 2006; Parker