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The role of genes in the etiology of specific language impairment

D.V.M. Bishop*

Department of Experimental Psychology, University of Oxford, South Parks Road, Oxford OX1 3UD, UK

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Abstract

Although specific language impairment (SLI) often runs in families, most pedigrees are not consistent with a single defective gene. Before progress can be made in molecular genetics, we need a better understanding of which aspects of SLI are heritable. Twin studies are useful in allowing us to distinguish genetic from environmental influences. This point is illustrated with a study in which twins were given tests of nonword repetition (regarded as an index of phonological short-term memory) and auditory processing. Children with SLI were impaired on both measures, but these deficits had different origins. Auditory processing problems showed no evidence of genetic influence, whereas the nonword repetition deficit was highly heritable. Future genetic studies of SLI may be most effective if they use measures of underlying cognitive processes, rather than relying on conventional psychometric definitions of disorder.

Learning outcomes: Information in this manuscript will serve to (1) equip readers with an elementary understanding of methods used in molecular genetic studies of language impairment; (2) familiarise readers with the logic of twin studies in behavioural genetics, using both categorical and quantitative methods; (3) illustrate the importance of phenotype definition for genetic research, and the usefulness of genetic methods in illuminating theoretical relationships between deficits associated with SLI; (4) show how genetically informative methods can be used to study environmental as well as genetic influences on impairment.

Keywords: Genetics; Specific language impairment; Twins

* Tel.: +44-1865-271-386; fax: +44-1865-291-255.
E-mail address: dorothy.bishop@psy.ox.ac.uk (D.V.M. Bishop).

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1. Introduction

It is unusual for specific language impairment (SLI) to make headline news. However, in October 2001, the media were buzzing with stories based on an article in Nature (Lai, Fisher, Hurst, Vargha-Khadem, & Monaco, 2001), which described how a small genetic mutation led to severe speech and language disorder in around half the members of a British family, the KE family. Most clinicians and researchers in the field of speech–language pathology are intrigued by this finding, but unfamiliar with the methods used by geneticists, and unclear about the implications for SLI in general. In fact, as we shall see, the family studied by Lai et al. (2001) is unusual in having such a straightforward relationship between genes and disorder. Nevertheless, the discovery is important in providing insights into how genes may influence the development of brain regions important for language learning. More prosaically, it provides a good starting point for introducing some of the core concepts in this field.

2. Identifying the gene causing SLI in the KE family

Geneticists make a distinction between the genetic make-up of an individual, the genotype, and the observable physical and behavioural manifestations of that make-up, the phenotype. By observing how a phenotype runs in a family, we can get clues as to the mode of inheritance. The first published account of the KE family (Hurst, Baraitser, Auger, Graham, & Norell, 1990) noted that in all three generations, around half the individuals had a severe articulatory dyspraxia (see Fig. 1). To understand how to interpret the KE pedigree, we need to recall some elementary genetic facts. Humans have 23 pairs of chromosomes, one member of

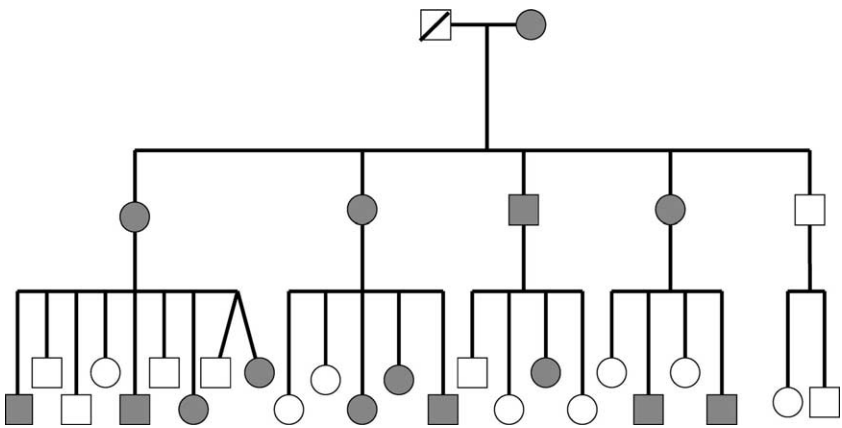

Fig. 1. Pedigree of the KE family. Females shown as circles and males as squares. A line through a symbol indicates deceased. Shaded figures are affected with speech and language disorder.