“An Extended Family with a Dominantly Inherited Speech Disorder” (1990), by Jane A. Hurst et al. [1]

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In 1990, researcher Jane Hurst and her colleagues published "An Extended Family With a Dominantly Inherited Speech Disorder," in which they proposed that a single gene was responsible for a language disorder across three generations of a family. Affected individuals of the family, called the KE family, had difficulty producing, expressing and comprehending speech. Hurst and her team studied the KE family and the disorder at the Department of Clinical Genetics at the Great Ormond Street Hospital for Children in London, England. Their report was subsequently published in the journal Developmental Medicine and Child Neurology in 1990. The authors' conclusions helped researchers better describe and explain language as a developmental and biological phenomenon and led later researchers to discover the proposed gene, mutations to which caused the language disorder.

In the late 1980s Hurst, a geneticist, worked for Michael Baraitser at the Department of Clinical Genetics when Elizabeth Auger, a special needs teacher, contacted Baraitser. Auger worked at the Lionel Primary School in Brentford, England, where she first met the KE children in her speech and language class. She noted that the KE children could not coordinate the muscle movements required for speaking, describing the children as if parts of their faces were frozen. As a result, according to Auger, their speech had limited vocabulary, long pauses, and unintelligible sounds. She presented the KE case to Baraitser as a potential study for a heritable language disorder. Hurst then began the study with Baraister, Auger, and Auger's colleagues at the primary school.

In "An Extended Family With a Dominantly Inherited Speech Disorder," Hurst and her colleagues reported the results of their examination of six of the KE children, all from the third affected generation of the KE family. They noted the frequency of symptoms among the children to determine if the family had a heritable language disorder. In their case histories of the children, the researchers noted each child's age, early developmental milestones such as first words and first steps, intelligence level, and common disorder symptoms. The research team concluded that the KE family's disorder pointed predominantly to a genetic factor, not to a combination of genetic and environmental factors.

"An Extended Family" begins with a brief description of the disorder in relation to the family. Hurst and her colleagues classify the condition as developmental verbal apraxia, a motor speech disorder in which the affected individual cannot coordinate the muscle movements to form words. The researchers created a family pedigree diagram showing that the grandmother, four of her five children, and eleven of her twenty-three grandchildren had verbal apraxia. About half of the grandchildren of the KE family had difficulty pronouncing words, using correct tenses, and understanding complex sentences. Based on that pattern, the researchers confirmed that the disorder is dominantly inherited, meaning that an individual needs only one copy of the gene to display the effects of that gene. When one parent carries a single copy of the dominant gene, scientists predict that about half of the parents' children will inherit the gene and therefore have the condition.

Following the introduction, the authors summarize case studies from six of the affected children. The six children, introduced by initials and number from the family pedigree, came from three different families in the larger KE family. Their ages ranged from five-years-old to sixteen-years-old, with three boys and three girls. The case histories enabled the researchers to compare developmental markers such as height and early motor development and to assess whether or not any of the markers correlated with the children's speech impairments. If the children's condition was strictly a language disorder, then the case studies would not reveal developmental abnormalities like lower intelligence or smaller brain size.

The children in the first three case studies came from the same family. The researchers describe all three as having normal early development and average intellectual ability. The oldest sibling, around sixteen-years-old, did not begin speaking until he was eighteen-months-old. His speech was more fluent than his sisters' speech, which the researchers described as unintelligible. His sisters, at five and six-years-old, had difficulty stringing more than two words together, which the researchers compared to the same level as a toddler. Both sisters also had difficulty organizing and executing oral patterns that are necessary for speech.

The children in the fourth and fifth case studies were siblings, and cousins to the first three children. Educational psychologists reported that both siblings, a fifteen-year-old and six-year-old boy, had average intelligence. The researchers describe their expressive language as disordered and their speech as unintelligible. Though both showed normal developmental markers, the
researchers note that the reading level of the fifteen-year-old was at half the normal level for his chronological age, and the expressive language of the six-year-old was at a three-year-old's level.

In the last case study, the researchers describe an eleven-year-old girl and cousin to the other children. Like the others, the girl exhibited normal developmental milestones except in regards to language. She did not speak until the age of three and still had difficulty speaking at age eleven despite having attended a speech and language class for six years.

Because all of the children showed normal development and average intelligence, the researchers discounted a more widespread, complex developmental disorder. Instead, the researchers identified common symptoms of the KE family disorder and predicted the processes that are disrupted by a mutated gene.

Regarding the speaking abilities of the KE children, the researchers found that all of the children had physical problems that prevented them from speaking normally, and that the children had issues processing the meaning of words and sentences. For example, some children produced the sound "bu" instead of the word "blue," or omitted consonants in words like "table." Because the omissions were not consistent and the children could sometimes pronounce one of two similar words, the researchers hypothesized that the children did not struggle with individual movements. Instead, the children had difficulty with combined movements and making the combinations quickly, especially tongue and mouth movements required for complex words.

The authors report that many of the children also had difficulty comprehending sentences and assigning meaning to objects and phrases. The children often used approximate words for objects, such as "sky" instead of "star" or "tea" for the image of a cup. That implied that the language disorder in the KE family not only affected the mouth movements required to speak, but also impacted cognitive processes required to develop and use language normally.

Following their discussion of the KE children's symptoms, Hurst and her team conclude that the KE children share an inability to coordinate complex sequences of movement required for speech and for constructing grammatically correct sentences. They note that the children could not use sign language to express themselves. Because sign language requires the use of coordinated gestures to communicate, the children's inability to use it implied a problem with both coordinating movements and with expressing meaning. The authors propose that the responsible gene influences developmental processes required for language acquisition.

In 1990, the journal Developmental Medicine and Child Neurology published "An Extended Family With a Dominantly Inherited Speech Disorder." At the time of its publication, linguist Myrna Gopnik had written a short article claiming that the gene responsible for the KE family's disorder affected their grammatical skills, not their ability to construct facial movements. Gopnik, a professor of linguistics at McGill University [5] in Montreal, Canada, conducted her own field interviews with the KE family, and she further noted the children's difficulty with conventional grammar rules. She argued that the children, therefore, could not construct grammar rules and instead had to learn each word as an individual item. Steven Pinker, a linguist at Harvard University [6] in Boston, Massachusetts, supported Gopnik's view in his 1994 book The Language Instinct.

Researchers from various institutions in England, including the University of Oxford in Oxford, England, disputed Gopnik's claims in separate short commentaries. They argued that Gopnik's grammar theory did not account for the KE children's other impairments, and was therefore an incomplete description of their disorder. The disagreement over the impact of the gene continued until 2001, when geneticists Simon Fisher and Cecilia Lai at the University of Oxford [5] discovered the specific gene. At the Wellcome Trust Center for Human Genetics of the University of Oxford [8], Fisher and Lai determined that a mutation in the FOXP2 gene was responsible for the KE family's disorder and that the mutation affected language acquisition, not grammar acquisition, thus ending the decade-long debate.

The discoveries in "An Extended Family With a Dominantly Inherited Speech Disorder" resulted in subsequent investigations on how language acquisition occurs during human development, and how human language acquisition differs from vocalization and vocal learning in other animals. The attribution of language disorders to particular genes [6] led researchers to research the genetic detection of developmental language disorders and potential gene therapies for those disorders.

Sources

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