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The Role of Genetics in Speech, Language, and Reading Disorders among Children

Speech, language and reading disorders are among the most common disorders in the United States, affecting one of every ten people. In 2001-2002, more than one million students in special education programs in public schools in the US were identified as having a speech or language impairment.[1]

In children, speech, language and reading disorders may have significant short and long term effects on well-being. Students with communication disorders often encounter academic difficulties with language, reading and spelling; but research also suggests that these disorders can lead to general academic difficulties that may be long lasting.[2] However, early identification of communication disorders and appropriate intervention can greatly improve a child's future outcomes.[3]

There are many types of speech, language and reading disorders; among children, speech sound disorders are the most prevalent. They occur when there is a significant delay in the acquisition of articulate speech sounds and may be associated with a limitation in the motor skills needed to produce speech sounds or with phonological errors (i.e. how speech sounds are represented in the brain). Speech sound disorders occur most often in young children, with a prevalence of about 16% in children at age three.[4] By age six, this number has declined to 3.8% but of these children, about half will have later academic difficulties.[5]

Childhood speech sound disorders may have life-long effects. As adults, individuals who report a history of childhood speech sound disorders continue to do worse on measures of communication and language skills than do adults without this history. Moreover, adults with a history of these disorders complete fewer years of schooling and require more remedial services. Speech sound disorders can also affect employment because jobs often require communication skills and value the use of information and communication technologies. Research has shown that adults with a history of speech sound disorders often hold jobs that require fewer of these types of skills.[6]

Speech Sound Disorders (SSD): A significant delay in the acquisition of articulate speech sounds. Estimated prevalence of 3.8% in 6-year-old children, with higher rates in younger children.[5]

Language Impairment (LI): A developmental language disorder that can affect both expressive and receptive language and impairs the ability to understand and/or use words in context. The estimated prevalence of LI at kindergarten is 7.4%.[7]

Reading Disorders (RD): A learning disorder that involves significant impairment of reading accuracy, speed, or comprehension to the extent that the impairment interferes with academic achievement or activities of daily life. The prevalence rate of RD is estimated at 5% in school-age children.[8]

It is critically important to gain a better understanding of these disorders so early identification and intervention can take place. While communication disorders may be related to other disabilities or syndromes, in most cases the causes are unknown. Their long term social, educational and economic consequences, combined with unknown etiology, are strong justifications for the pursuit of research including family and genetic studies. Such studies aim to understand more about the underlying risk factors that may influence these complex disorders. Researchers at Case Western Reserve University are exploring the role of genetics and heredity in communication disorders such as speech sound disorders.

The Role of Genetics in Speech, Language, and Reading Disorders among Children

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Dr. Barbara Lewis is interested in the causes of child speech and language disorders with a focus on genetic, medical and neurological conditions that impact speech and language development. Her research has focused on the genetic basis of speech sound disorders. With funding for the last 19 years from the National Institute on Deafness and Other Communication Disorders (NIDCD), she and colleagues have been able to follow children with speech sound disorders from preschool to school age to adolescence to determine long-term outcomes for children with early childhood speech and language disorders.

A growing body of evidence suggests an underlying genetic basis for speech sound disorders, the most common speech and language disorder in children. Despite this, researchers have not identified a particular gene which predisposes children to this condition; however, several candidate genes are presently being studied. Dr. Lewis' earlier research demonstrated that speech sound disorders tend to aggregate in families. In a recent study, Dr. Lewis and a group of colleagues from Case Western Reserve University further explored these issues by looking at speech and language disorders in the parents of children diagnosed with speech sound disorders.

STUDY AIMS

Dr. Lewis and colleagues were interested in comparing parents with and without a history of speech sound disorders in order to further understand the familial nature of speech sound disorders and their long term effects.

Both speech sound disorders and language impairment have been shown to aggregate in families, a finding which provides justification for the consideration of the influence of genetic factors in both disorders. Because of this, Dr. Lewis and colleagues looked at the two disorders together. **Their primary study questions were:**

- To what extent do speech sound disorders, with or without language impairment, aggregate within the families of children who have speech sound disorders, and to what extent do these disorders aggregate within an individual?

- To what extent does language impairment, with or without speech sound disorders, aggregate in families of children with speech sound disorders, with or without language impairment?

STUDY DESIGN

The study population included parents of children with speech sound disorders. Many parents of children with speech sound disorders report also having experienced these types of disorders themselves. Therefore, studies of parents can shed light both on the familial aspects and adult outcomes of these conditions.

One hundred and forty seven parents from the greater Cleveland, Ohio area participated in this study (58 fathers and 89 mothers). Parents were assessed for speech, language and reading disorders. Of the parents who participated, 36 had a known history of speech sound disorders, while 111 did not.

Participants were tested on their ability to produce speech sounds, phonological processing skills, spelling, reading decoding, oral motor skills and language. These tests included the repetition of multisyllabic words, tongue twisters, Pig Latin, written spelling, and other tests of communication. Family history of speech sound disorders was tested directly when possible, and through interviews when direct testing was unfeasible. Data collected from the tests were recorded, reviewed and analyzed by licensed speech-language pathologists. Data were also collected about

the presence of language impairments, reading disorders and spelling disorders in the nuclear family.

The authors divided the parents into three groups for comparison; parents with a history of speech sound disorders, parents with a history of speech sound disorders and language impairment, and parents without a history of either condition. Results of assessments of language, speaking, spelling and reading were compared for these three groups.

KEY FINDINGS

Overall, parents without a history of speech sound disorders performed better on measures of language, spelling and reading than did parents with a history of these disorders. In particular, parents with a history of speech sound disorders performed less well on all measures except on Pig Latin and measures of oral motor skills. Parents with a history of speech sound disorders and language impairment did worse than those parents with a history of speech sound disorders alone. There were no significant differences in education or occupational level between parents with a history of both speech sound disorders and language impairment and parents with a history of speech sound disorders alone. Additionally, there were no differences between mothers and fathers across any of the groups.

The Familial Nature of Speech Sound Disorder

Dr. Lewis and colleagues found that there is significant co-occurrence of speech sound disorders and language impairment within individuals. Within families, there was significant aggregation of both speech sound disorders and language impairment, although the aggregation was stronger for language impairment. Specifically, the likelihood of experiencing language impairment increased by a factor of more than four for each additional family member with language impairment, while the odds of speech sound disorders increased more than twofold for each additional family member with speech sound disorders. This suggests that the risk for these disorders increases as more relatives are affected and illustrates that speech sound disorders and language impairment aggregate in families and often occur together.

The Long Term Effects of Speech Sound Disorders

Dr. Lewis and colleagues compared adults with a history of speech sound disorders to those without a history of these disorders. They found that adults with and without a history of speech sound disorders had similar occupational levels and academic attainment. However, adults with a history of speech sound disorders continue to perform worse on measures that test speech sound production than do adults without this history.

Dr. Lewis and her colleagues (Dr. Sudha Iyengar and Dr. Catherine Stein from the Department of Epidemiolgy and Biostatistics and Dr. H. Gerry Taylor from the Department of Pediatrics) have been involved in research examining the genetic basis of speech, language and reading disorders. With funding from the National Institutes of Health (NIH) and the National Institute on Deafness and Other Communication Disorders (NIDCD), Dr. Lewis and the team at Case Western Reserve University have reviewed the literature and tested models of spoken and written language. This research has added to the understanding of the interrelationships among SSD, LI, and RD, with the goal of advancing the development of more effective clinical interventions appropriate for subgroups and at a particular stage of development.

Regions of chromosomes 1, 3, 6, 7 and 15 that were identified in previous studies of reading and language disorders were examined. Findings showed that these same regions were linked to speech sound disorders. These chromosome regions are known to harbor genes that influence neural development prenatally. Currently, Dr Lewis, along with Dr. Jean Tkach in the Department of Radiology, is conducting studies employing functional imaging technology to determine how individuals with genetic differences process speech sounds.

Primary conclusions from this research include:

- Many genes contribute to SSD.
- The team hypothesize that some genes contribute to SSD disorders alone, whereas other genes influence both SSD and other written and spoken language disorders. These genes have broad effects on how the brain develops.
- They postulate that underlying common cognitive traits, such as phonological memory and awareness, are responsible for shared genetic influences of spoken and written language; thus explaining the high rates of co-morbidity of these disorders in children.
- The combination of environmental as well as genetic influence determines whether the individual will demonstrate a speech, language and/or reading disorder.

IMPLICATIONS FOR POLICY AND PRACTICE

The identification of underlying genetic factors for speech sound disorders has implications for researchers, clinicians and families. Perhaps most importantly, knowledge of genetic factors may improve diagnosis and early identification of children at risk of speech sound disorders. This early identification will allow for timely environmental intervention.[9] Early intervention is crucial because of the potential of communication disorders to lead to social and educational isolation.[1] In addition, communication and language skills are most easily learned before the age of five. While initial speech or language difficulties can be characterized as 'baby talk', if children do not outgrow these speech patterns as expected, they may become disorders that can impede learning.[1]

Children who exhibit such disorders may benefit from the assistance of a speech-language pathologist. Speech-language pathologists work with children, families and schools to facilitate a child's communication. This is done through individual therapy with the child and the development of goals and techniques to be used at home and in the classroom.[1] Therapy can be useful to children throughout their educational trajectory as the understanding of vocabulary, language and reading become more complex. Further, speech-language pathologists can provide long term assistance to children in the transitions from school to employment.

Research has demonstrated the efficacy of early intervention with speech-language pathologists on the communication of children with speech sound disorders.[2] The amount of treatment that a child receives is also associated with better outcomes. This progress is even more significant given that, untreated, communication disorders have the potential to become worse and to lead to deficits in reading, writing and spelling.[2] Despite this, in Ohio and across the US shortages of speech-language pathologist exist. More efforts (like those highlighted in the text box) are needed to ensure that children receive timely and appropriate intervention.

The research being conducted by Dr. Lewis and the team at Case Western Reserve University is vital. Increased understanding of the role of family risk and the genetic pathways of communication disorders is important for researchers and clinicians. For clinicians, this information may validate existing diagnostic categories or stimulate the development of new diagnoses. Genetic studies may also provide additional possibilities for the treatment of communication disorders. The co-occurrence of language, reading, and spelling disorders may be illuminated by new knowledge of the role of genetics in these disorders. For researchers and clinicians, understanding genetic factors helps to bridge gaps between different disciplines and may lead to a more comprehensive understanding of communication disorders.

Efforts in Ohio to ensure that early identification and proper intervention are available to all children

Ohio schools face persistent shortages in available, qualified speech-language pathologists (SLPs). A number of organizations are working collaboratively to address this shortage:

The Ohio Department of Education (ODE) and **Ohio State Board of Education** recently approved $5 million ($2.5 million per year for two years) to fund a proposal to address the shortage of speech-language pathologists in the schools. The program includes initiatives aimed at meeting three key goals: increasing the number of graduate-level students, encouraging more SLPs to work in the schools, and improving retention of school-based SLPs. http://www.ode.state.oh.us/

The Ohio Master's Network-Initiatives in Education (OMNIE) Project is a collaborative endeavor to ensure the recruitment, retention, and professional development of qualified speech-language pathologists in Ohio schools. http://www.omnie.org/

The Ohio Board of Speech-Language Pathology and Audiology was established by the Ohio General Assembly to protect the health and promote the welfare of Ohioans by licensing and regulating the practices of speech-language pathology and audiology. http://slpaud.ohio.gov/

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