GENETIC RESEARCH REGARDING COMMUNICATION DISORDERS

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A communication disorder is an inability to understand and/or use speech and language to relate to others. For the majority of communication disorders, we do not understand the cause. We know that many result from hearing impairment, intellectual disabilities, cerebral palsy, mental retardation, and cleft lip and/or cleft palate.

The presence of a genetic component of a disease can be difficult to identify. Evidence supporting a genetic component includes familial clustering of cases, increased incidences of consanguineous mating (i.e., mating between closely related individuals), increased prevalence that exists within genetically segregated communities, increased risk that exists for the children or siblings of affected individuals, and concurrence of identical twins with the disorder.

Scientists have declared several syndromes with a known genetic cause (and many more with both a genetic and environmental etiology) that are seen in many speech-language pathologists' places of practice - Down syndrome, fragile-X syndrome, Pierre Robin sequence, and Prader- Willi syndrome. Genetic research is being conducted on a host of other common genetic conditions that are relevant to speech-language pathologists, which include stuttering, autism, apraxia of speech, speech sound disorder and dyslexia

Many health professionals lack confidence in the area of genetics due to a lack of education in the area of genetics. This lack of confidence and or knowledge among health professionals regarding genetics and genetic disorders, early detection of disturbance and stimulation indicates a need for further investigation and identification language disorder and dislexia genes.

KEY WORDS: communication disorder, genetic cause, genetic conditions.

1. INTRODUCTION

Genetic research is being conducted on a host of other common genetic conditions that are relevant to speech-language pathologists, which include stuttering, autism, apraxia of speech and dyslexia. Scientists have declared several syndromes with a known genetic cause (and many more with both a genetic and environmental etiology) that are seen in many speech-language pathologists' places of practice.

To understand how the genes for language disorders are identified, it is essential to understand the types of studies geneticists utilize. This can be visualized as a multi-step process of increasingly narrow scope, starting with heritability studies, proceeding to classical karyotype analysis, then to genetic linkage analysis followed by high-resolution genetic association studies in a process termed "positional cloning" (Collins, 1992), and ending with functional assays of candidate genes.

Many health professionals lack confidence in the area of genetics due to a lack of education (Neils-Strunjas, Guerdjikova, et al. 2004) in the area of genetics. Many professionals may feel that the idea of more knowledge regarding genetics could be overwhelming for the therapists' (speech-language pathologists') work load/case load (Guttmacher, Porteous et al., 2007). This lack of confidence and or knowledge regarding genetics and genetic disorders among health professionals indicates a need for further investigation and identification language disorder and dislexia genes.

Speech-language pathologists working in hospitals and clinics will often ask if other family members presented with communication disorders during their development. Case history forms may include specific questions regarding when siblings reached developmental milestones or how the milestones of other family members were reached in comparison to the individual being assessed. School speech-language pathologists may also benefit from knowing the family history of a child and whether or not he or she may be predisposed to a communication disorder. However, whether or not speech-language pathologists are making a connection between the family history (which is inherently genetic information) and its significance to the clinical picture is largely unknown. By possibly knowing more information about the genetic contributions to communication disorders and how the disorder may develop and progress, (as it may have among other family members) will the speech-language pathologist be able to contribute to the professional team. The speech-language pathologist will then be able to determine the best treatment plan for individuals with communication disorders (with or without additional, complicating medical issues).

2. GENETIC SYNDROMES SEEN IN PRACTICE

The idea that differences in language and reading abilities are partially attributable to genetics is not new. As early as the 19th century, for instance, educators and physicians described families in which more than one member had difficulty learning to read (Hinshelwood, 1917). With the evolution of more sophisticated techniques of genetic analysis, our understanding of the biologic basis of these language disorders continues to grow.

Scientists have declared several syndromes with a known genetic cause (and many more with both a genetic and environmental etiology) that are seen in many speech-language pathologists' places of practice. Examples include: speech-language impairment, autism, reading impairment, dyslexia, Down syndrome, fragile-X syndrome, Pierre Robin sequence, and Prader-Willi syndrome. A survey of allied health professionals suggests that speech-language pathologists are not confident in their abilities to talk to patients and or family members about the genetics of these syndromes (Neils-Strunjas, Guerdjikova et al. 2004). Each of the aforementioned genetic conditions have been, or are currently, the subject of ongoing genetic research. Genetic research is being conducted on a host of other common genetic conditions that are relevant to speech-language pathologists, which include stuttering, autism, apraxia of speech, speech sound disorder and dyslexia.

2.1 Stuttering

Hegde (2001) claimed, "*Stuttering* is a disorder of fluency with excessive amounts, or excessively long durations of dysfluency, which are combined with tension, struggle, and related behaviors,". Research has indicated that some individuals who stutter have a different organization/dominance within their cerebral hemispheres as well (Ambrose & Cox, 1996). Five in 100 preschool-aged children and one in 100 school-aged children stutter (Ambrose & Cox, 1996). A strong genetic predisposition is more likely, however, in children whose stuttering persists beyond elementary school (Felsenfield, 2002). Many genes have been implicated in stuttering, including genes on chromosomes 1, 12 (Riaz, Steinberg et al., 2005), and 18 (Shugart, Mundorff et al 2004). Additional genes that have implications regarding whether a child will recover from stuttering have also been identified (Ambrose & Cox, 1996). As the underlying etiology of stuttering becomes clearer, it may be possible for speech-language pathologists to make more accurate prognoses for affected children and their families.

2.2 Autism

Autism is a disorder that may be present at birth in a child, but is usually noticed sometime during the first three years of age. As *Hegde* (2001) observed, a child with autism has a lack of wanting to relate to others in addition to other "peculiar verbal and non-verbal behaviors,". Current data indicates for parents with one child affected by autism, the risk of another child with autism is 5%. When more than one sibling of a child with autism is affected, the risk increases to 25%. Males are three times more likely to be affected than females. Immediate relatives of individuals with autism may develop characteristics that also fall within the autism spectrum (Whitelaw, Flett et al, 2007). A group of researchers at Cold Spring Harbor Laboratory in Long Island, New York looked for genetic mutations associated with autism in 528 families. Some of the families had multiple children diagnosed with autism, and other families had only one child diagnosed with autism. Families with no history of autism comprised the control group. The results from the study showed that genetic mutations were ten times more likely to be present in families with a child with autism, when compared to the control group. The results also indicated autism was five times more likely to occur among the families with multiple affected children. The rate of autism, however, was found to be highest when only one family member was affected with autism (Swaminathan, 2008a). The study mentioned at Cold Spring Harbor Laboratory still leaves many unanswered questions for clinicians and researchers. The clinical significance of this research from Swaminathan (2008a) states, "Although 90% of autism cases are sporadic, heritable/familial forms of autism also occur," By knowing more information regarding genetics, the speech-language pathologist genetic disorders and autism.

Scientists studying autism agree the disorder is influenced by environmental, as well as genetic components. Some studies have linked autism to genes located on chromosomes 16 and 20, among many other possible genes (Swaminathan, 2008b). Various organizations are struggling with the speculations and uncertainties from current research (Swaminathan, 2008b). Current studies have also examined parents' knowledge of risks regarding autism. A recent study in Canada revealed that a majority of parents of children with autism overestimated the chance of having another child with autism. Fewer than half of these parents reported that the recurrence rate had been explained to them by a professional (Hurley, Losh et al. 2006). Conversely, a study performed in Tasmania and Australia, indicated a majority of the parents were informed of the recurrence rate of

autism among family members and siblings as well as the fact that autism is more common in males than females. The study also discussed fertilization and pre-implantation to select female embryos to reduce the risk of autism (Whitelaw, Flet et al., 2007), which is a controversial topic within the field of genetics. When considering autism, specific aspects of cognitive impairment may be important for genetics research, and may be considered by the speech-language pathologist. In addition, face recognition, emotion recognition, and theory of mind are frequently impaired among individuals with autism (Iarocci, Yager et al. 2007). Three-quarters of the autism population have an IQ below 70. Another form of autism contains a profound form of mental retardation (Starr, Berument et al. 2001).

2.3 Childhood Apraxia of Speech

Childhood Apraxia of Speech is a severe developmental speech disorder which includes characteristics of articulatory struggle, an awareness of speech errors, difficulty in perception, decreased expressive language, delays in literacy, and decreased speech performance abilities with that of an increase in speech complexity (Gillon & Moriarty, 2007). Other characteristics also may include the following: inconsistent speech performances, limited phonetic inventory, problems with imitation, poor or slow response to treatment, oral apraxia and in-coordination, prosody disturbances, as well as vowel and diphthong errors (Shriberg, Campbell et al., 2003).

2.4 Speech Sound Disorder

A speech sound disorder is described as a significant delay in a child's acquisition of articulated speech sounds (Shriberg, Tomblin et al., 1999). Speech Sound Disorder (SSD) has been described as having multiple genetic etiologies, and numerous genes contributing to the disorder (Stein, Millard et al., 2006). Some genes affected may be causing SSD to occur, while other involved genes are impacting an individual's reading and writing (Lewis, Shriberg et al, 2006) SSD is thought to have life long impacts in some cases. By being able to identify SSD early on, intervention is much more successful in a child's overall development with specific regards to speech and language (Fisher, Francks et al., 2002). Research has indicated the prevalence of speech sound disorder, learning impairment, and reading disorder within a family are greater than their prevalence in the overall population (Pennington, 1997). Speech-language pathologists in clinical practice should be aware that a family history of speech sound disorder,

learning impairment, and reading disorder increases the risk for these disorders in families where two or more family members in the immediate family are affected (Lewis, Shriberg et al. 2006).

2.5 Dyslexia

Dyslexia is a complex disease with a strong genetic component, affecting at least 4% (Lewis, 1994) of all schoolchildren. It is characterized by extreme difficulties in acquiring skills in reading and writing, causing severe problems for children, parents, and teachers. Dyslexia (or specific reading disability) is defined as a specific and significant impairment in reading ability that cannot be explained by deficits in either intelligence, learning opportunity, motivation or sensory acuity. Dyslexia is the most common childhood learning disorder (Grigorenko et al., 1997; Fisher et al., 2002). A recent survey has assessed the prevalence of dyslexia to be 3.6% of all primary school children in the Netherlands. In the same survey, a M/F ratio of around 2:1 was found (De kovel et al., 2004). The available evidence from family and twin studies further suggests that dyslexia is a significantly heritable trait (Fisher et al., 2002; Francks et al., 2004]. Numerous linkage studies for dyslexia have been carried out and at least eight loci have been repeatedly linked to dyslexia: *DYX1* on 15q15-q21 (Grigorenko et al., 1997), *DYX2* on 6p21.3-p22 (Grigorenko et al., 1997), *DYX3* on 2p11-p16 (Fagerheim et al., 1999; Francks et al., 2002; Anthoni et al., 2007; De kovel et al., 2008), *DYX5* on 3p12-q13 (Nopola-Hemmi et al., 2001), *DYX6* on 18p11.2 (Fisher et al., 2002), *DYX7* on 11p15.5 (Hsiung et al., 2004), *DYX8* on 1p35-p36 (Rabin et al., 1993; Grigorenko et al., 2001; Tzenova et al., 2004; Miscimarra et al., 2007; De KoveL et al., 2008), and *DYX9* on Xq26-q28 (Fisher et al., 2002; De Kovel et al., 2004). Additional significant multipoint linkage findings on chromosome regions 6q12 *(DYX4)* (Petryshen et al.,2001), 7q32.2 (Kaminen et al., 2003), 13q22.1,18q22.2-q22.3, and 21q21-q22 (Fisher et al., 2002) have been reported, but these results have not been replicated yet. In 2003, *Taipale* et al. reportedthe identification of a candidate gene for dyslexia *(DYX1C1)* on 15q21. *DYX1C1* was disrupted by the 15q21 translocation breakpoint in four dyslexic members of an earlier described family, carrying a translocation between chromosomes 2 and 15, t(2;15)(q11;q21) (Nopola-Hemmi et al., 2000). By applying a similar ''positional cloning'' approach, the *ROBO1* gene was recently found to be disrupted at the 3p 12 translocation breakpoint in an individual with both dyslexia and a translocation between chromosomes 3 and 8, t(3;8) (p12;q11) (Hannula-Jouppi et al., 2005). In addition, a cluster of five genes

in the *DYX2* locus (*VMP, DCDC2, KIAA0319, TTRAP,* and *THEM2)* has been consistently reported to be associated with dyslexia (Londin et al., 2003; Deffenbacher et al., 2004), although it is currently not clear which of these five genes would be (a) dyslexia candidate gene(s) (Francks et al., 2004; Cope et al., 2005; Meng et al., 2005; Paracchini et al., 2006; Schumacher et al., 2006; Brkanac Z., 2007; Luciano et al., 2007).

Based on twin studies (Stevenson,1991; Olson et al., 1994), genetic influence is estimated at 60–70%. Several genomic regions were identified which may contain genetic variants related to dyslexia (Grigorenko et al., 1997; Schulte-Körne et al., 1998). Therefore, further studies investigating specific genes within these regions appear to be a very promising approach leading to a better understanding of dyslexia. The aim of this study was to verify and refine recent findings from Anglo-Saxon studies in a German case-control cohort because it is crucial to regard the influence of different languages in dyslexia. Although both, English and German, belong to the Indo-Germanic languages, there are strong differences in the regularity of the grapheme-phoneme correspondence. So the same genes could have different consequences for dyslexia in both languages. In genome scans, the best replicated regions concerning dyslexia are located on chromosomes 6 and 15 (Nöthen et al., 1999; Müller-Myhsok and Grimm, 1999; Schulte-Körne et al.,1998). From region 6p22.2, several genes have been studied previously (Cope et al., 2005; Schumacher et al.,2006; Harold et al., 2006; Brkanac et al., 2007; Luciano, 2007). Of these, it is examined *DCDC2* at the single nucleotide polymorphism (SNP) level. Additionally, a 2,445bp deletion in *DCDC2,* which was initially described in an American cohort (Meng et al.,2005).

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ГЕНЕТСКА ОСНОВА ГОВОРНО-ЈЕЗИЧКИХ ПОРЕМЕЋАЈА

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РЕЗИМЕ

Поремећај комуникације подразумева немогућност да се разуме и /или користи говор и језик. Етиологија ових поремећаја је често непозната. Оно што знамо јесте да они могу бити последица оштећења слуха, церебралне парализе, интелектуалног инвалидитета, менталне ретардације, расцепа усне и /или расцепа непца. Присуство генетске компоненте болести није увек лако утврдити. Докази који могу подржавати генетску компоненту су чести случајеви у породици, присутност честих бракова између лица у сродству, повећана преваленца поремећаја у генетски одвојеним заједницама, повећани ризик за децу и рођаке афектираних особа, као и одвајање идентичних близанаца са поремећајем.

Научници описују неколико синдрома често присутних у пракси клиничких логопеда, за које се зна одређена генетска основа (и још много више оних који су последица удружених одређених фактора средине и одређеног генотипа), као што су Даунов синдром, синдром фрагилног X, Пјер-Робинов и Прадер-Вилијев синдром. Генетска истраживања се спроводе и за низ других говорно-језичких поремећаја који се и најчешће срећу у клиничкој пракси, а то су: муцање, аутизам, развојна дисфазија, поремећај изговора гласова и дислексија.

Многи стручњаци немају увид у могуће генетске основе комуникацијских поремећаја због неадекватне едукације, као ни поверење у могућност њихове ране детекције и стимулације. Овај недостатак поверења и/или знања међу стручњацима из области генетике и о говорно-језичким поремећајима, њиховој раној детекцији и стимулацији указује на потребу за даљим истраживањима и идентификацијом гена одговорних за поремећаје говора и језика, као и гена за дислексију.

КЉУЧНЕ РЕЧИ: поремећаји комуникације, генетска основа, услови генетских истразивања