Speaking the same language: genetics and communication disorders

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This commentary is on the article by Simpson et al. on pages 346–353 of this issue.

Professionals working in the area of genetics and communication disorders have challenges in communicating about disorders that may have a genetic basis. Simpson et al. found that sex chromosome aneuploidies were present at a much higher rate (about 3%) in individuals with language disorders than the rate (about 0.25%) in individuals without any noted problems in language skills. Their results suggested that some individuals with a diagnosis of specific language impairment may, instead, have a genetic disorder requiring medical intervention. The authors recommended determining the chromosome type of individuals with language disorders and/or dyslexia so that individuals with sex chromosome aneuploidies could be identified earlier and receive appropriate medical intervention. The potential impact of this paper is interesting to consider from the fields of both speech-language pathology and genetics.

Despite the title of the article (Increased prevalence of sex chromosome aneuploidies in specific language impairment and dyslexia) some of the probands did not have a diagnosis of specific language impairment. ‘Speech and language disorders’ are mentioned in several places in the article, and the authors’ genetic screening recommendation was not limited to individuals with specific language impairment, but included individuals with ‘language problems’. Mixing the terms ‘speech’ and ‘language’ can create confusion of interpretation among communication disorder professionals. Speech is the process where the respiratory, larynx, and articulatory (e.g. tongue, lips, teeth) systems are used to say words. Language is understanding and producing words based on ideas, thoughts, and feelings. The confusion may arise from the fact that language can be expressed through speech or written words. ‘Specific language impairment’ is a language (not speech) disorder that has no obvious cause and has been found in about 7% of elementary-age children.

With an increasing number of communication disorders being identified as having a genetic component, more speech-language pathologists and audiologists are exploring the field of genetics. Basic genetics education is included in some curricula and most clinicians are familiar with some of the basic terms and facts needed to understand this type of research. These facts are that humans typically have 46 chromosomes (22 matching pairs and 1 pair of sex chromosomes, [XX for a female or XY for a male) and that extra or missing parts of a chromosome can cause a problem. ‘Proband’ is the term that refers to the individual being tested. ‘Aneuploidy’ is the term for the wrong number of chromosomes – either too few or too many. If the problem is with the sex chromosome pair, the resulting problem is a sex chromosome aneuploidy. If a presumed pair of chromosomes is found to include a third chromosome, the resulting trio is called a ‘trisomy’. ‘Karotype’ has at least two meanings: (1) the testing for the chromosomes an individual has and (2) the resulting picture of an individual’s chromosomes, typically shown as matched chromosomes with numbers 1–22 and the sex chromosomes.

As genetic information plays an ever increasing role in many health issues, however, basic knowledge of genetics is inadequate. Much of the genetic literature on communication disorders has focused on syndromes with speech, language, and/or hearing difficulties. Some of these sex-linked chromosomal disorders have not been associated with language disorders until recently, or the underlying language disorder may be the same but the causes of the language disorder are different. In addition, the literature is scattered across discipline-specific and multidisciplinary journals, which can hinder the awareness and uptake of genetic findings into clinical practice.

Pressing questions exist on how to disseminate and evaluate genetic information among professionals and lay stakeholders. For example, easily-accessed databases listing genetic disorders with their relevant characteristics, diagnostic criteria, and treatment options are needed but require constant monitoring and updating as new information is discovered and older information disproven. Policy decisions regarding genetic screening (e.g. who to screen, what to screen, when to screen, and payment for screening) will also need to be monitored and updated. Research recommendations, such as those by Simpson et al., highlight the complicated interactions among genetic research, clinical practice, and public policy.
The development of social strengths in children with cerebral palsy

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The study by Tan et al. is of interest because it shows that children’s development is not necessarily delayed because of disability, specifically cerebral palsy (CP).1 CP is one of the most common childhood-onset disabilities and a condition frequently researched. PubMed, for example, reports 13 885 hits for CP compared with conditions such as developmental delay (6079) and myelomeningocele (602). As CP causes activity limitations, which can lead to restrictions in socially or culturally influenced areas of life, the study by Tan et al. provides insights into the social strengths that children with CP have. The results, indicating that many of the children diagnosed with CP can develop to the same level of social participation as children without disability (even though it may take more time), should have a positive impact on the adults who are around children with disabilities.5,6 Child participation, as a determinant of well-being and life satisfaction, may also decrease mental health problems.7 In addition to the abilities needed for social interactions, social participation includes an experience of belonging and inter-subjective interaction that leads into acts of acknowledgment.8 Because adults provide ‘scaffolds’ for the experiences of children with disabilities, parents and teachers have the responsibility to encourage the children to start interacting with peers and to introduce them to potential friends. The results reported in this study in terms of the development of children’s abilities for interactions, should influence the adults’ attitudes to the children’s social roles in a positive way.

When receiving support from adults, it is likely that the children’s understanding of and adaption to social demands improve. For children with CP, participation restrictions are most often associated with their physical impairments related to environmental barriers, such as reduced access to venues and events.9,10 However, negative social attitudes to disability may also constitute barriers to participation. As children with disabilities attend community activities less frequently than typically developing children,11 adults must make efforts to introduce the children to such activities in addition to introducing them to those peers sharing the activity.

For persons with severe CP or persons with additional intellectual disability, the result of the study shows there is no specific age where development ceases. This should be

REFERENCES