Critical review

Fluency disorders in genetic syndromes

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Abstract

The characteristics of various genetic syndromes have included “stuttering” as a primary symptom associated with that syndrome. Specifically, Down syndrome, fragile X syndrome, Prader-Willi syndrome, Tourette syndrome, Neurofibromatosis type I, and Turner syndrome all list “stuttering” as a characteristic of that syndrome. An extensive review of these syndromes indicated clients diagnosed with these syndromes do show evidence of nonfluency patterns, but not all would be considered stuttering. Many of the syndromes are marked by degrees of mental retardation that probably contribute to a higher than average prevalence of stuttering, as well as a higher than average prevalence of other fluency disorders (when compared to the population at large).

An in-depth analysis of the available data indicates that some of these genetic syndromes show patterns of stuttering that may be indicative of only that syndrome (or similar syndromes) that can be differentially diagnosed from developmental stuttering. Among these patterns are the word-final nonfluencies noted in Prader-Willi syndrome; the presence of stuttering in the absence of secondary behaviors noted in Prader-Willi syndrome and; the presence of palilalia, word-final and word-medial nonfluencies, and word-medial and word-final nonfluencies in Tourette syndrome. Implications for future research are discussed in light of these findings.

Educational objectives: The reader will be able to: (1) describe the various different genetic syndromes that are associated with fluency disorders; (2) describe the types of nonfluencies that are associated with the major types of genetic syndromes that have fluency disorders; (3) describe the behaviors that may assist in differentially diagnosing different types of speech characteristics associated with various genetic syndromes.

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1. Introduction

It is well known that certain genetic syndromes are associated with communication disorders. For example, Adler (1976) suggested a strong link between specific syndromes and resulting speech and language disorders. Yet, the study of communication disorders in genetic syndromes has only gained significant interest in the past few years, most likely with the expansion of knowledge in the human genome project. Only recently have attempts been made to compile the available sparse information in order to gain a more comprehensive picture of the occurrence of communication disorders in genetic syndromes.

The most extensive overview to date is that of Shprintzen (1997) who compiled data on the major features, primary etiology, speech production, resonance, voice, language and hearing of 334 syndromes. Remarkably, the occurrence of stuttering or other fluency disorders received little mention on this list. The only two exceptions include Down syndrome and fragile X syndrome. In Down syndrome it said that “dysfluency” and “rapid bursts of speech” may occur (Shprintzen, 1997, p. 226). Information in fragile X syndrome is slightly more extensive: “dysfluency with classical stuttering and in some cases, ‘cluttering”; in more severe cases, nonsensical short bursts of perseverative or absent speech” (Shprintzen, 1997, p. 264). With these shortfalls in mind, the purpose of the present paper is to give a more detailed review of the occurrence and nature of fluency disorders in a number of genetic syndromes. In addition to Down syndrome and fragile X syndrome, Prader-Willi syndrome, Tourette syndrome, neurofibromatosis type I, and Turner syndrome will be discussed. It should be recognized that these syndromes vary with respect to their underlying cause and to the presence of any associated problems (in particular mental retardation). They were included in the present review because, as far as we could ascertain, they are the only ones for which some information on speech nonfluencies is available. This was established through searches of several major search engines, and follow-up review of the articles. In some cases, the terms “stuttering” and/or “disfluency” appeared in the citation or abstract, however, specific definitions or descriptions of these terms were not provided. For ease of understanding, the term “developmental stuttering” will be used throughout this review. This term will be used to refer to the most typical types of stuttering seen in children and adults, that is, the term will not be used to describe stuttering associated with acquired neurological disorders (Helm-Estabrooks, 1999), acquired psychogenic stuttering (Baumgartner, 1999), or cluttering (St. Louis, 1992). For clarification, acquired neurogenic stuttering refers to transient stuttering that begins with the onset of a specific, identifiable neurological event (ASHA, 1999); acquired psychogenic stuttering refers to stuttering that is clearly related to psychopathology (ASHA, 1999); and cluttering refers to a fluency disorder that is not stuttering and is characterized by rapid
and/or irregular speech rate, along with associated disturbances in language and/or attention. In addition to this terminology, the following terms will be used throughout this manuscript for ease in understanding and interpretation.

**Disfluency**: The non-stuttered breakdowns in fluent speech. The term *disfluency* will be used to identify the types on fluency breakdowns that are not considered to be stuttering and include interjections of sounds, repetitions of phrases, revisions, and incomplete phrases (Johnson, 1959; Silverman & Williams, 1967), as well as other forms of non-stuttered fluency breakdowns, such as circumlocutions, additions, and verbal mazes (Paul, Tetnowski, & Reuler, 2007).

**Dysfluency**: The stuttered breakdowns in fluent speech. These are the most common breakdowns in fluency that are perceived by listeners to be stuttering (Yairi, 1996) and those breakdowns in fluency that are most likely classified as stuttering according to experts in the field of stuttering (Ham, 1989). This term is synonymous with *stuttering* (ASHA, 1999), and following the lead of Yairi, will be referred to as *stuttering-like disfluencies* (SLD) throughout the remainder of this manuscript. The terms SLD and stuttering will be used to identify the part-word repetitions, single syllable word repetitions, prolongations, and blocks that are most consistently identified as stuttering.

**Nonfluency**: In many research papers, breakdowns in fluency are not clearly delineated as being either “disfluency” or “SLD”. Some of these same papers do not clearly describe whether they are referring to disfluency or SLD. The term “fluency” refers to the continuity of spoken language units (ASHA, 1999), and is the opposing term of “nonfluency”. Throughout this review, nonfluency will be the catch-all term used to refer to all breakdowns in fluency. This term will include non-determinate types of fluency breakdown that can include both categories (i.e., disfluencies and SLD).

2. Down syndrome

Down syndrome is a well-known condition resulting from trisomy 21. It is characterized by some typical physical characteristics such as a short stature, a flat occiput, small and slanting palpebral fissures, epicanthic folds (skin folds of the upper eyelid covering the inner corner of the eye), a depressed nasal bridge, a small nose, and an overlarge tongue relative to the size of the mouth. The syndrome is also associated with behavioral characteristics including the presence of speech and language problems. With an incidence of 1 in 600–700 births, Down syndrome is the most frequently occurring chromosomal abnormality (Lambert & Rondal, 1979). Quite understandably then, it is the syndrome in which the study of fluency problems has received most attention to date.

Schlanger and Gottsleben (1957) reported that speech problems occur more frequently in Down syndrome than in mental retardation of other origins. As far as stuttering is concerned, they found a prevalence of 33%; the estimated prevalence in mental retardation in general being 5%. Stuttering has indeed been reported to be unusually common in Down syndrome, with prevalence figures ranging between 15 and 48% (Bloodstein, 1995). An exception to this trend is in data reported by Martyn, Sheehan, and Slutz (1969). In their sample of 42 participants, stuttering was found in only 1 participant (1/42 or 2.3%). It is not clear if stuttering occurs more frequently in males than in females for the Down syndrome population (as is the case for stuttering in the non-mentally retarded population). According to Devenny and Silverman (1990), stuttering occurs to a similar degree in both sexes. There is some suggestion, though, that there is a gender difference in the proportional distribution of SLD. Evans (1977) found significantly more prolongations and blocks in male subjects than in female subjects and a similar but not significant trend for repetitions and interjections. This is an important issue when studying genetic syndromes and requires further
research. That is, the specific type of nonfluency must be noted and described when studying the speech of individuals with genetic syndromes.

A number of studies do discuss the nature of all nonfluencies in individuals with Down syndrome. Cabanas (1954), for instance, found blocks, hesitations, repetitions, and hurried speech in Down syndrome participants. Schlanger and Gottsleben (1957) report the presence of clonic, tonic, and secondary reactions but do not further specify these patterns. Willcox (1988) described repetitions of sounds, syllables, words, and phrases, and prolongations. According to Evans (1977), repetitions are most frequent, followed by interjections, prolongations, and blocks. These patterns signify a combination of both SLD and disfluencies, however, these patterns do not distinguish stuttering associated with Down syndrome from other types of stuttering not associated with Down syndrome.

It is not clear to what extent the occurrence of stuttering is associated with particular sounds. Cabanas (1954) claimed that for individuals with Down syndrome, stuttering occurs not predominantly on consonants, but mostly on vowels. Also, stuttering did not occur systematically on the same sounds, but might occasionally affect any sound. In this respect, the stuttering patterns demonstrated by individuals with Down syndrome would be different from developmental stuttering. Willcox (1988), however, examined the stuttering of five children with Down syndrome in relation to those of five developmentally normal subjects matched for language age. Willcox found no particular sounds in either group that occurred more frequently in SLD. Nevertheless, SLD on plosives were more common than on other sounds for both groups. Again, the stuttering patterns of Down syndrome individuals do not clearly distinguish them from other types of developmental stuttering.

Willcox (1988) also provides details on the frequency of repetitions, duration of prolongations and the locus of the SLD in the five children with Down syndrome she studied. All but one participant produced examples of three repetitions of a unit, while two participants demonstrated examples of four or more repetitions of a unit. Three of the five children produced prolongations of more than 1 s. The stuttering tended to occur on the first stressed element in the head, or on the nucleus of the tone unit, and they almost invariably occurred at the beginning of a word. This would be expected to be the case in most developmental stuttering children as well. A description of stuttering behaviors has not been shown to clearly distinguish stuttering associated with Down syndrome from typical developmental stuttering.

Opinions differ as to the presence of secondary stuttering behaviors in individuals with Down syndrome that have been diagnosed with SLD. Some authors maintain that secondary stuttering behavior is absent (Cabanas, 1954; Van Riper, 1971). According to Van Riper (1971) there may be some mildly feared speaking situations, but few word or sound fears. He noted that most of the individuals with Down syndrome would not be aware that they have stuttered or are stuttering (clearly he must have been writing about individuals with Down syndrome and associated cognitive deficits). Van Riper suggested that these individuals may be frustrated in trying to communicate, primarily because of word-finding and coding difficulties, but not because they cannot utter a given word when they try to. In other words, Van Riper was referring to word finding and/or other linguistic deficits seen in Down syndrome that were associated with causes other than stuttering. Other investigators, however, did observe secondary characteristics in individuals with Down syndrome. These secondary behaviors included avoidance and postponement behavior, and synkinesias (for instance, facial grimaces or blinking the eyes) (Devenny & Silverman, 1990; Preus, 1973; Stansfield, 1990). The presence or absence of secondary behaviors does not seem to differentially diagnose differences between stuttering associated with Down syndrome and developmental stuttering.
On a related issue, the absence of secondary behaviors is one of the arguments used by Cabanas (1954), Weiss (1964), among others, to argue that the SLD demonstrated by individuals with Down syndrome represents cluttering rather than stuttering. Weiss (1964) even claims that most of the “retarded individuals” with fluency breakdowns, in general, do not develop true stuttering. In his opinion, genuine stuttering develops as the result of concentrated effort to improve the fluency of speech and requires a keen memory of past failure. This would be beyond the capability of the severely mentally impaired (if this were the case with Down syndrome individuals). Otto and Yairi (1975) compared the spontaneous speech samples of 19 institutionalized, non-stuttering individuals with Down syndrome and 19 normally intelligent individuals. They agreed that “cluttering” more correctly characterizes speech in Down syndrome. Their conclusion is based, however, on the high speaking rate observed in many of their subjects. Preus (1973), on the other hand, studied 47 Down syndrome individuals in two Norwegian institutes for the mentally retarded and concluded that the stuttering or “stuttering-like” behaviors found in patients with Down syndrome may be classified as genuine stuttering. One third of his subjects, unexpectedly, exhibited secondary symptoms in the form of body movements, devices of masking (e.g. pretending to cough or changing word order when blocking on a sound), and other postponement and avoidance behaviors. According to Preus, these indicated proof of a “conscious experience of stuttering”. He added, however, that the presence of stuttering in the speech of individuals with Down syndrome does not rule out the possibility that cluttering may also be found. In his study, 27.7% of the subjects were diagnosed as pure stuttersers, 12.7% as pure clutterers, 19.2% as combined cases, and 40.4% displayed neither stuttering nor cluttering. The data on Down syndrome and cluttering is also unclear at this time. The uncertainty regarding the presence or absence of secondary symptoms prevents differentiation of developmental stuttering from stuttering associated with Down syndrome.

3. Fragile X syndrome

With an estimated prevalence of 1/4000 in males and 1/8000 in females, fragile X syndrome is the second most frequent cause of mental retardation of genetic origin after Down syndrome (Goldson & Hagerman, 1992; Turner, Webb, Wake, & Robinson, 1996). The condition finds its origin in a defect on the long arm of the X-chromosome and has as typical physical features a prominent mandible, a long and narrow face, large low-set ears, hyperextensible finger joints, and macro-orchidism (testicular enlargement) in post-pubescent males. Speech and language disorders are common. Since fragile X is an X-linked disorder and males have only one X-chromosome, they are usually more severely affected than females (Hagerman & Silverman, 1991).

Several authors report stuttered speech in individuals with fragile X syndrome and often a comparison is made with other populations. According to Hanson, Jackson, and Hagerman (1986), stuttered speech is a typical feature in impaired males with an IQ over 70. Belser and Sudhalter (2001) found more stuttered speech among those with fragile X compared to control groups with autistic disorder, and mental retardation not associated with fragile X syndrome. Other studies, however, suggest that stuttering occurs at a similar or even lesser degree in fragile X syndrome than in other populations. Paul et al. (1987) reported 2.88% stuttered syllables on average in the spontaneous speech of a group of adult males with fragile X, compared to 2.75% in a group with non-specific mental retardation and 2.00% in a group with autism. Ferrier, Bashir, Meryash, Johnston, and Wolff (1991) reported a mean percentage of stuttering of 4.9% in fragile X syndrome, 1.6% in autism and 6.1% in Down syndrome. Herbst (1980) recorded stuttering in only 3 out of 98 males with fragile X among 25 families. These figures should be interpreted
cautiously, however, as their definition of “dysfluent” (nonfluent) speech differed from study to study. Upon further review, Belser and Sudhalter defined “dysfluency” as “stuttered dysfluency” and “stuttered-like dysfluencies”. Paul et al. refer to their nonfluencies as rapid repetitions of syllable sounds, prolongations, and/or silent blocks. Ferrier et al. refer to their subjects’ behaviors as repetitions of sounds or syllables; repetition of whole words or phrases were not considered as “stuttering”. Finally, Hanson et al. defined their subjects’ speech as cluttering. With only one exception, the research indicates a higher degree of stuttering in individuals diagnosed with fragile X syndrome. This is an important point in that it had previously been unclear whether the nonfluencies in fragile X syndrome were really stuttering or some other type of nonfluency. Clearly, a significant number of individuals with fragile X syndrome show symptoms that are indicative of developmental stuttering. It has been contended that the speech of persons with fragile X syndrome is very specific and recognizable, even to the extent that it would sometimes be possible to diagnose the disorder solely on basis of an individual’s speech. Nonfluent speech is among the 13 items of the checklist for screening fragile X developed by Hagerman (1987). Nonetheless, a diversity of labels have been used to characterize the speech of individuals with fragile X. One finds such various labels as a perseverative speech pattern, a repetitive speech pattern, dysfluent speech, stuttering, cluttering, fast speech, dysrhythmia and also verbal apraxia and developmental apraxia. Symptoms mentioned are repetitions, prolongations and interjections of sounds, repetitions of words and phrases, blocks, reformulations with false starts and self-corrections, and also palilalia (Belser & Sudhalter, 2001; Hagerman & McBogg, 1983; Jacobs, Glover, & Mayer, 1980; McLaughlin & Kriegsman, 1984; Rhoads, 1984; Vilkman, Niemi, & Ikonen, 1988; Wolf-Schein et al., 1987). These patterns of speech are indeed consistent with developmental stuttering, but are also consistent with other types of nonfluencies that are not stuttering. Both stuttering and other speech disfluencies are significantly noted in individuals diagnosed with fragile X syndrome. Unfortunately, consistent patterns and descriptions of the fluency breakdowns of speakers with fragile X syndrome have not been shown to be a differentially diagnostic pattern of the speech of individuals with fragile X syndrome at this time. However, the prevalence of stuttering in fragile X syndrome appears to be somewhat lower than the prevalence of stuttering in Down syndrome, but slightly higher than in autism or non-specific forms of mental retardation.

The distribution of the dysfluencies in individuals with fragile X syndrome appears to be similar to that seen in developmental stuttering, i.e., repetition of initial parts of sentences (Lubs, Travers, Lujan, & Carroll, 1984), repetition and prolongation of initial sounds of words, and blocks on initial plosives (Paul & Leckman, 1984; Vilkman et al., 1988). Yet, Rhoads (1984), in a group of 15 affected males in Hawaii, found that repetition of words or phrases occurred particularly at the ends of sentences. This unusual pattern of fluency breakdown may be useful in differential diagnoses in the future, however, the patterns of sentence ending word and phrase repetitions has not been verified in other studies of speech characteristics of individuals with fragile X syndrome. As mentioned earlier, fragile X syndrome usually affects females less severely than males. This also seems to hold for the occurrence of fluency disorders (Jacobs, Mayer, Matsuura, Rhoads, & Yee, 1983; Madison, George, & Moeschler, 1986). This does not mean, however, that there is never nonfluent speech in females (Grigsby, Kemper, Hagerman, & Myers, 1990). These studies do not offer the same detail of the fluency breakdowns as the studies on males, but it is clear that females diagnosed with fragile X syndrome are also susceptible to significant fluency breakdowns.

In summary, when specific types of nonfluencies are considered, individuals with fragile X syndrome appear to have a higher prevalence of stuttering than non-specific forms of mental retardation and autism, but a lower prevalence than individuals diagnosed with Down syndrome.
Prader-Willi syndrome is a multiple anomaly disorder with an incidence estimated at 1/5000 to 1/25000 (Akefeldt, Gillberg, & Larson, 1991; Burd, Vesely, Matsof, & Kerbeshian, 1990; Greenswag & Alexander, 1995). The syndrome results from the loss of imprinted genomic material within the paternal 15q11.2-13 locus. Deletion of the same part of the chromosome in the mother leads to Angelman syndrome. Typical clinical features of Prader-Willi syndrome include hypotonia at birth, hypogonadism (absent or decreased function of the sex glands), hypopigmentation (whitening of the skin), mild to moderate mental retardation, hyperphagia (excessive eating) with childhood-onset obesity, small hands and feet, and craniofacial abnormalities such as a small forehead, a small mouth, and almond-shaped eyes (Holm, Sulzbacher, & Pipes, 1981).

There is evidence that speech and language development is frequently problematic in Prader-Willi syndrome (Akefeldt, Akefeldt, & Gilberg, 1997; Branson, 1981; Downey & Knutson, 1995; Dyson & Lombardino, 1989; Edmonston, 1982; Hall & Smith, 1972; Kleppe, Katayama, Shipley, & Foushee, 1990; Munson-Davis, 1988; Zellweger, 1988) and a number of authors make note of fluency disorders.

Kleppe et al. (1990) investigated fluency in 18 Prader-Willi children and found percentages of nonfluency ranging from 1 to 34%. Interjections and revisions were frequently observed, accounting for 36 and 26% of all nonfluencies observed. Word repetitions, part-word repetitions, phrase repetitions, and incomplete phrases occurred to a lesser degree (16, 10, 9, and 3%, respectively, of the total number of nonfluencies exhibited). Sound repetitions and prolongations, some of the hallmarks of stuttering, did not occur. Kleppe et al. (1990) concluded that many of their subjects were disfluent, but as a group the Prader-Willi children did not present the clinical picture of stuttering. Only 1 child was considered by them to have the symptoms of stuttering. It is not entirely clear how these researchers came to this conclusion in light of the part-word (sound) repetitions noted in their reports.

Branson (1981) also reported the presence of fluency breakdowns in only 1 of 21 subjects. She described the repetitions, additions, and circumlocutions found in this subject’s spontaneous speech and attributed many of them to the marked word recall problems that the youngster showed. These symptoms are indicative of both stuttering (through the use of repetitions) and disfluent speech (as marked by the use of circumlocutions and additions).

In a more comprehensive study Defloor, Van Borsel, and Curfs (2000) investigated speech fluency in 15 individuals with Prader-Willi syndrome. Their subjects ranged in chronological age from 9.9 to 20.0, and total IQ (WISC-R) from 40 to 94. Speech samples, collected in four different speech modalities (spontaneous speech, repetition, monologue, and automatic series), were analysed for frequency, type and distribution of nonfluencies. All subjects showed patterns of nonfluency with the most fluency breakdowns occurring during spontaneous speech samples (average dysfluency index: 13.03%). Somewhat fewer nonfluencies were seen during monologue (average dysfluency index: 10.22%). During repetition and automatic series, nonfluencies occurred but with relatively low frequency (average dysfluency index: 1.14 and 0.54%, respectively). All subjects produced interjections, revisions, part-word repetitions, whole-word repetitions, and phrase repetitions, indicating patterns of both stuttering and disfluency. Interjections were the primary type of nonfluencies and accounted on average of 52.58% of the total number of nonfluencies. Revisions, part-word repetitions, whole word-repetitions and phrase repetitions accounted for 11.10, 10.84, 9.85 and 5.34%, respectively. On the other hand, incomplete phrases, prolongations, blocks, and broken words occurred in 14, 13, 10 and 5 subjects, respectively. The distribution of the SLD in the speech sequence was largely similar to that of developmental stuttering. The SLD occurred more
frequently on the first words of a sentence than on words in other positions. They also occurred predominantly in the initial position of words rather than on other locations within a word and were more prevalent in lexical words than in function words. Unlike individuals that stutter, however, the participants of this study did not show any secondary characteristics of stuttering. In addition, disfluencies outnumbered SLD in their speech. On average, more than half of all the subjects’ nonfluencies (52.58%) consisted of interjections and revisions (i.e., disfluencies). Whole-word repetitions were among the most frequently observed types of SLD (respectively, 11.10 and 9.85% of the total number of nonfluencies exhibited). The presence of some instances of word-final nonfluencies was noted in two subjects. This presence of word-final nonfluencies and a finding that monosyllabic words were more often affected than bisyllabic or polysyllabic words are not characteristic of developmental stuttering (Brown, 1945; Van Borsel, Van Coster, & Van Lierde, 1996; Van Borsel, Van Lierde, & Van Cauwenberge, 1997; Wingate, 1967). These symptoms may help in the differential diagnosis of stuttering associated with Prader-Willi syndrome.

In summary, simple counts of all nonfluencies, or SLD do not distinguish stuttering in Prader-Willi syndrome from developmental stuttering. However, an in-depth analysis of the distribution of stuttering moments has been shown to differentiate the stuttering associated with Prader-Willi syndrome from developmental stuttering. Specifically, the lack of secondary behaviors in the presence of SLD, and some notation of word-final disfluencies may help to differentially diagnose the stuttering associated with Prader-Willi syndrome.

5. Tourette syndrome

A syndrome that is often discussed in relation to stuttering is Tourette syndrome. Tourette syndrome, named after the French physician, Georges Gilles de la Tourette, is characterized by the presence of multiple motor tics and one or more vocal tics that cause marked distress or significant impairment in social, occupational, or other important areas of functioning. It has its onset before the age of 18 and is not due to the direct physiological effects of a substance or a general medical condition. It is a relatively frequent condition with an estimated prevalence ranging between 0.03 and 1% (Cardoso, Veado, & Teotonio de Oliveira, 1996; Van de Wetering, Cath, & Buitelaar, 1996). Males are three to four times more likely to be affected than females. Most likely, Tourette syndrome is genetically determined. An autosomal dominant inheritance with sex-specific penetrance and variable expression has been suggested but its exact etiology and pathogenesis are still unclear (American Psychiatric Association, 1987; Kurlan, 1993).

It is often stated that stuttering is quite common in individuals with Tourette syndrome. For example, in the Diagnostic and Statistical Manual of Mental Disorders (DSM IV) (American Psychiatric Association, 1987), stuttering is listed as one of the initial symptoms of the Tourette’s complex. This definition may lack the specificity that we are attempting to use as markers for differential diagnosis of fluency disorders. The estimated incidence of “stuttering” in individuals with Tourette syndrome ranges from 15.3 to 31.3% (Comings & Comings, 1993; Pauls, Leckman, & Cohen, 1993). The fluency failures described range from “repetitions, hesitations, and false starts” (O’Quinn & Thompson, 1980), “interjections, whole-word repetitions, prolongations, phrase and part-word repetitions, revisions” (Rassas Cohn, Shames, McWilliams, & Ferketic, 1983), “interjections, phrase repetitions, revisions, part-word repetitions, whole-word repetitions (Van Borsel & Vanryckeghem, 2000), to “stutter like behavior” (Singer, Pepple, Ramage, & Butler, 1978), and “stutter-like repetitions of sounds” (Aronson, 1980). By definition, the fluency failures listed as hesitations and false starts (O’Quinn and Thompson), and interjections, phrase repetitions, and revisions (Rassas Cohn) are not stuttering, but are more indicative of speech disfluency.
and Donaher (2003) found the presence of an “unusual type and distribution of disfluencies, that included many word and sentence medial disfluencies, and many word and sentence final disfluencies” in two case studies of Tourette syndrome. Also, the presence of palilalia has been noted in individuals with Tourette syndrome. Palilalia is a disorder of speech characterized by compulsive repetition of a phrase or word, often at the end of a sentence, that the patient sometimes utters with increasing rapidity and with decreasing voice volume (Brain, 1961; Critchley, 1970). Once again, this is one of the commonly described fluency failures in Tourette syndrome (Cardoso et al., 1996; Jankovic, 1997; Ludlow, Polinsky, Caine, Bassich, & Ebert, 1982). In a Brazilian cohort of 32 patients with Tourette syndrome, Cardoso et al. (1996) observed palilalia in 9% of cases (3/32). It is apparent that a careful description of nonfluency behaviors can help differentially diagnose stuttering associated with Tourette syndrome from that of developmental stuttering.

Recently, the relationship between Tourette syndrome and stuttering was also studied from another perspective. Abwender et al. (1998) examined 22 developmental stutterers for the presence of neuropsychiatric features commonly seen in Tourette syndrome, including tics, obsessive compulsive behaviors, and attention deficit disorders. Eleven individuals displayed motor tics and symptoms of obsessive compulsive behaviors at rates similar to those seen in persons with Tourette syndrome. According to Abwender et al. (1998), their findings are consistent with models suggesting extrapyramidal involvement in stuttering and raise the possibility that developmental stuttering and Tourette syndrome are pathogenetically related.

The suggestion of a possible relationship between developmental stuttering and Tourette syndrome and perhaps a shared underlying pathogenesis is most interesting. However, such a line of thinking may, at the present moment be premature. While it is often assumed that stuttering is fairly common among those with Tourette syndrome, especially in the medical literature, data that support this idea are lacking. Few studies have investigated the nature of the fluency failures in Tourette syndrome and the results of these studies suggest that the disfluencies displayed do not conform to the classic pattern of stuttering.

Rassas Cohn et al. (1983), who studied the disfluencies in a 25-year-old male with Tourette syndrome, reported that in spontaneous speech only one fifth of their subject’s fluency failures were SLD. Similar findings were arrived at by Van Borsel and Vannryckeghem (2000) in an 18-year-old male with Tourette syndrome. During an assessment that included a variety of speech tasks, only 15% of the nonfluencies were of the kind that most researchers would consider characteristic of stuttering. Overall, the subject’s speech pattern seemed to bear little resemblance to typical developmental stuttering.

A recent study in three individuals (Van Borsel, Goethals, & Vannryckeghem, 2004) confirms the above findings. The study investigated the speech of two boys (9.11 and 12.7 years) and a girl (12.2 years) with Tourette syndrome in different speech modalities (automized speech, word and sentence repetition, reading aloud, conversational speech, monologue, semantic fluency (retrieval of names of animals), singing, and repeating the final word of sentences (for instance: of the sentence “wij worden door hem afgehaald” repeat the last word; correct answer: “afgehaald”). All three individuals demonstrated nonfluencies, but the pattern was unlike that typically seen in developmental stuttering. The vast majority of their nonfluencies (respectively, 71.5 and 69.7% in the two boys and 67.5% in the girl) were disfluencies rather than stuttering behaviors. Moreover, the few nonfluencies characteristic of stuttering did not always follow the distributional tendencies typically seen in stuttering (i.e., more stuttering on longer words than on shorter words, more on lexical words than on function words, and predominantly at the beginning of a sentence rather than at the end). The occurrence of a substantial number of interjections, phrase repetitions, and whole-word repetitions and also a certain percentage of incomplete phrase and revisions might
be considered suggestive of disorganized wording; not stuttering. However, two of the three subjects obtained scores on the Communication Attitude Test (Brutten & Vanryckeghem, 2003) that reflected a negative speech-associated attitude as typically seen in children who stutter. Two of the three subjects showed examples of phrase and/or word repetition at the end of clauses as typically seen in palilalia. However, in contradistinction to patients with palilalia (Lebrun, 1997), the subjects did not show a definite tendency to repeat words and phrases rather than part words, they did not repeat units several or many times in succession, and they did not have any problems in the task that required them to repeat only the last word of a sentence (Van Borsel, Schelpe, Santens, De Vos, & De Vos, 2001). In conclusion, the subjects’ fluency failures were not consistent with genuine stuttering, nor were they consistent with patterns of genuine cluttering, nor were they consistent with patterns of genuine palilalia. These patterns have the potential for differential diagnosis of a subtype of fluency breakdown that is exclusive to fluency disorders in genetic syndromes.

The study by Tetnowski and Donaher (2003) also noted unusual patterns of fluency as well as unexpected patterns to fluency-induction tasks. Both subjects displayed patterns of fluency breakdown not typically observed in developmental stuttering (word-medial and word-final repetitions). Additionally, one of the subjects showed no response to delayed auditory feedback (DAF) as seen in developmental stuttering. Fluency breakdowns showed no change from non-DAF to DAF conditions across various tasks including monologue, dialogue and reading. In this case, not only were the nonfluencies of the subjects qualitatively different from developmental stuttering, but the response to fluency-inducing tasks was different than expected when compared to developmental stuttering.

De Nil, Sasisekaran, Van Lieshout, and Sandor (2005) found that there is not unequivocal proof indicating a higher prevalence of stuttering in individuals with Tourette syndrome. This study investigated the speech of 69 children diagnosed with Tourette syndrome (mean age 12.49) and a group of 27 control participants (mean age 10.9) during conversation and reading. During conversation no significant differences were found for SLD between the individuals with Tourette syndrome and the control participants. However, the Tourette syndrome individuals showed a statistically significant higher frequency of non-stuttering-like disfluencies (word repetitions, filled and unfilled pauses, hesitations, and interjections). During reading, the older children showed a higher percentage SLD compared with the control group, while the opposite was true for younger children. The percentage of non-stuttering-like disfluencies of both groups during reading was comparable.

In summary, the nonfluencies of individuals with Tourette syndrome are qualitatively different from the nonfluencies of the typical developmental stutterer. Specifically, word-medial and word-final nonfluencies seem to be characteristic of the nonfluency patterns in Tourette syndrome. In addition, the reaction to fluency-inducing techniques (unfortunately not attempted in all studies) may also add clues to a differential diagnosis to fluency disorders in genetic syndromes.

6. Neurofibromatosis type I

Neurofibromatosis type I, formerly called Von Recklinghausen disease, is a progressive autosomal dominant disorder with an estimated prevalence of two to three cases per 10,000 population (Friedman, 1999). The condition is characterized by the presence of neurofibromas and abnormal cutaneous pigmentation including café-au-lait spots and axillary freckling. The responsible gene is situated on the long arm of chromosome 17 (Barker et al., 1987).

A review of the literature (Bekaert, Van Borsel, & Mortier, 2003) shows that individuals with neurofibromatosis have a high risk of developing speech and language problems. Specific
research of these problems is scanty, however. A number of authors report the presence of fluency problems. These have been characterized as disorders in speech rate (Lorch, Ferner, Golding, & Whurr, 1999; Riccardi, 1981; Solot et al., 1990), speech rhythm (Solot et al., 1990; White, Smith, Brooke, Bigler, & Schauer, 1986) and stuttering (Köhler, 1990; Rondal, 2001). None of these studies, however, provides further details of the nature of the disorder or information about the pathogenesis. In a recent survey by Cosyns, Vandeweghe, Mortier, Janssens, and Van Borsel (submitted for publication), 8 out of 57 participants (i.e., 14%) reportedly stuttered. Seven of them provided more information on the disfluencies that characterized their stuttering: all of them mention the occurrence of blockages. This description of nonfluencies is more typical of developmental stuttering. However, we are hesitant to make this distinction at this point until in-depth analyses of speech nonfluencies are conducted. The loci of these nonfluencies may give us an indication of whether the nonfluencies are more “stuttering-like”, or whether they are more “abnormal” as seen in other genetic syndromes.

7. Turner syndrome

Turner syndrome is a syndrome that occurs only in females with an estimated incidence of 1/2500 (Hook & Warburton, 1983). It is known to result from a missing X-chromosome with its main features being a short stature, ovarian dysgenesis (defective development of the ovaries), neck webbing, congenital peripheral lymphedema (swelling of the lymph passages), coarctation of the aorta, cubitus valgus (deformity of the elbow in which the elbow deviates from the midline of the bone when extended), dysplastic (abnormally developed) nails, and pigmented nevi (a benign, coloured growth on the skin) (Gorlin, Cohen, & Levin, 1990).

It has been suggested that most individuals with Turner syndrome show some form of speech and language disability, but that the defects may be subtle and subject to environmental influences (Jung, 1989). According to Sparks (1984) females with Turner syndrome are less likely to need speech and language intervention than those with other sex-linked chromosome abnormalities. A survey (Van Borsel, Dhooge, Verhoye, Derde, & Curfs, 1999) in 128 females with Turner syndrome ranging in age from 2.4 to 58.8 years nonetheless showed that almost one quarter of the subjects (31 or 24%) were receiving or had received treatment for speech or language problems. Stuttering was mentioned in four cases (3.1%). It is not clear if this figure is typical for the population in Turner syndrome in general. As far as we are aware, there are not other data available on the prevalence and nature of fluency disorders in Turner syndrome. Once again, future research is the key to distinguishing whether the nonfluencies related to Turner syndrome are different from those of developmental stuttering. Only an in-depth analysis of fluency type, loci, and nonfluency description will answer this debate.

8. Discussion and conclusion

It is clear that certain syndromes may be associated with a fluency disorder but also that knowledge is presently still very limited. This review exemplifies this shortcoming. For a number of syndromes, it is clear that they are closely linked with a fluency disorder and research on the prevalence and nature of the speech fluency disorder has already been conducted. This appears to be the case for Down syndrome, fragile X syndrome, Prader-Willi syndrome and Tourette syndrome. For other syndromes knowledge is only anecdotal and fragmentary. A fluency disorder may be present but little data is available about the frequency of occurrence and the precise nature of the associated fluency disorders. This is the case with both neurofibromatosis type I and Turner
syndrome. For still other syndromes it is not known if they may be associated with a fluency failure. Information on the synopsis of these genetic syndromes is provided in Table 1.

The fact that stuttering is only rarely mentioned in a list like that of Shprintzen’s (1997) is in itself quite remarkable. A substantial number of the syndromes included in this list (almost two thirds, see Van Borsel, 2004) are associated with mental retardation. Since the prevalence of stuttering in the mentally retarded is generally assumed to be higher than the normal 1% (Bloodstein, 1995) one would expect that stuttering is reported more often. Perhaps the severity of the cognitive deficit could explain this discrepancy. In some cases of genetically determined mental retardation, the retardation can be so severe that these individuals will not likely develop complex speech patterns. Others syndromes are possibly associated with very severe speech and language disorders that are much more prominent and overshadow a fluency breakdowns.

In summary, little information is available about fluency disorders in comparison to other communication disorders in genetic syndromes. Even in those syndromes that have been researched for the prevalence and nature of fluency disorders, the picture is still far from clear. Regarding prevalence, for example, data are often based on small samples and in several instances information is lacking on how the data were obtained. It is often unclear which speech tasks were studied, how speech was analysed, or which criteria were used for diagnosing stuttering or another fluency disorder. The delineation between stuttering-like disfluencies and other forms of fluency breakdown is not consistent and/or well defined. The situation is similar for the nature of the fluency disorders. In Down syndrome, for example, there is a question as to whether the fluency breakdowns represent stuttering, cluttering, or perhaps both. The nonfluent speech of individuals with fragile X syndrome has been characterized as stuttering, as cluttering, and also as verbal apraxia. Careful analysis of the specific speech characteristics associated with fragile X has given us some information related to prevalence of stuttering (see Table 1). In Prader-Willi syndrome and Tourette syndrome, the same careful analysis of speech behaviors indicates that their nonfluencies consist predominantly of nonfluencies that would not be considered to be fluency failures typical of stuttering (see Table 1). The main conclusion so far is that none of the syndromes reviewed in this study show a specific pattern or prevalence of nonfluency identical to that of developmental stuttering. Although subtypes of stuttering have been described in the literature, such as stuttering associated with acquired neurological disorders (Helm-Estabrooks, 1999) and acquired psychogenic stuttering (Baumgartner, 1999), the stuttering associated with genetic syndromes does not mimic these patterns either. Another subtype may be needed to describe stuttering associated with genetic syndromes.

Another question is whether syndrome-specific patterns of dysfluency exist. There certainly is a possibility that some syndromes are associated with a typical, highly recognizable pattern of dysfluency. The pattern of dysfluency displayed in Tourette syndrome, for instance, is definitely quite different from that in Down syndrome. For fragile X syndrome it has even been suggested that the speech pattern is so distinct that a diagnosis can be arrived at purely on the basis of speech. However, the evidence gathered in this review does not indicate that the speech fluency patterns for fragile X syndrome are exclusive but fine-grained analyses of fluency behaviors have yet to be completed.

It is also possible that consistent nonfluency behaviors have yet to be defined within a genetic syndrome due to the subject pools that were used in these studies. The possible variations within a syndrome may be due to variables such as age, gender, degree of mental retardation, and the presence of other speech and language disorders. As far as age is concerned, it is quite likely that for individuals with a genetic syndrome, the clinical picture changes over the years with various degrees of growth, environmental experiences, opportunity, and multiple other variables. This is widely accepted in the case of developmental stuttering, and for all communication development
Table 1
Descriptions and differences of nonfluent speech behaviors in genetic syndromes

<table>
<thead>
<tr>
<th>Genetic syndrome</th>
<th>Type of nonfluency noted</th>
<th>Notes on prevalence</th>
<th>Secondary behaviors</th>
<th>Differential diagnostic patterns</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>SLD</td>
<td>Disfluency</td>
<td>Notes on prevalence</td>
<td>Not studied</td>
</tr>
<tr>
<td>Down syndrome</td>
<td>Yes</td>
<td>Yes</td>
<td>Higher than general population</td>
<td>Not studied</td>
</tr>
<tr>
<td>Fragile X syndrome</td>
<td>Yes</td>
<td>Yes</td>
<td>Lower percentage of SLD than Down syndrome; higher percentage of SLD than non-specific mental retardation or autism</td>
<td>Not studied</td>
</tr>
<tr>
<td>Prader-Willi syndrome</td>
<td>Yes</td>
<td>High percentage of disfluencies</td>
<td>Unclear</td>
<td>None noted</td>
</tr>
<tr>
<td>Tourette syndrome</td>
<td>Yes</td>
<td>Yes</td>
<td>Higher than general population</td>
<td>Not studied</td>
</tr>
<tr>
<td>Neurofibromatosis type I</td>
<td>Yes</td>
<td>Unclear</td>
<td>Higher than general population</td>
<td>Not studied</td>
</tr>
<tr>
<td>Turner syndrome</td>
<td>Yes</td>
<td>Unclear</td>
<td>Slightly higher than general population</td>
<td>Not studied</td>
</tr>
</tbody>
</table>
in general. Regarding gender, one should not only take into account the possibility of a different prevalence in males and females but also of a possible gender difference in severity of expression of the symptoms (as for instance in fragile X syndrome). The degree of mental retardation too may be assumed to cause variation. It is evident, for instance, that the risk of developing secondary symptoms is greater in individuals with a milder retardation who are well aware of the disorder. According to Boberg, Ewart, Masson, Lindsay, and Wynn (1978), diagnosing a disorder becomes increasingly difficult with increasing severity of the retardation, perhaps because other and/or more severe speech and language disorders are more likely to occur which blur and complicate the picture. In more severe forms of mental retardation the nonfluent speech patterns may be more a reflection of the retardation, than being specific to the syndrome. In addition, it is likely that more than one type of fluency disorder may be associated with a particular syndrome. The occurrence of both stuttering and cluttering in Down syndrome is a case in point.

In conclusion, a great deal more research is necessary before definite statements about the occurrence of fluency disorders in genetic syndromes, and the possibility of differentiable, diagnostic nonfluencies in different syndromes can be formulated. Differential diagnosis of nonfluency in genetic syndrome requires more than a surface analysis of speech. Qualitative descriptions of fluency types and loci may be the key to successful diagnosis and treatment. Since “stuttering” is listed as a symptom of many of these genetic syndromes, it is possible that clinicians may try to intervene with programs established for developmental stuttering. No data exist on whether programs that have high success rates with developmental stuttering (e.g. Lidcombe Program, Onslow, Packman, & Harrison, 2003) will have the same success rates with stuttering associated with genetic syndromes. Research is necessary that compares the speech of individuals with a particular genetic syndrome with that of normal speakers, with that of developmental stutters, and with that of individuals with other genetic syndromes. Variables such as age, gender, degree of mental retardation, and the presence of other speech and language disorders should be taken into account. Preferably, analyses should include a variety of speech samples collected in different speech tasks, as well as fluency-induction strategies. This research should also use a clear and well-defined classification of speech nonfluencies. The results of such analysis may ultimately serve as a starting point for treatments that address the specific and perhaps unique problems of each syndrome.

CONTINUING EDUCATION

Fluency disorders in genetic syndromes

QUESTIONS

1. Mental retardation is typically not associated with which of the following genetic disorders:
   a. Down syndrome
   b. fragile X syndrome
   c. Prader-Willi syndrome
   d. Tourette syndrome
2. Both stuttering-like disfluencies, and non-stuttering disfluencies are associated with:
   a. Down syndrome
   b. fragile X syndrome
   c. Tourette syndrome
   d. all of the above
3. Stuttering, with the absence of secondary behaviors has been noted in:
   a. Prader-Willi syndrome
   b. Tourette syndrome
   c. Neurofibromatosis type I
   d. Turner syndrome

4. The percentage of stuttering noted in fragile X syndrome is:
   a. lower than the percentage of stuttering in Down syndrome and autism
   b. higher than the percentage of stuttering in Down syndrome and autism
   c. higher than the percentage of stuttering in Down syndrome but lower than the percentage of stuttering in autism
   d. lower than the percentage of stuttering in Down syndrome but higher than the percentage of stuttering in autism

5. What statement best summarizes the findings of this study:
   a. a high percentage of stuttering is clearly present in all genetic syndromes
   b. a high percentage of disfluency is clearly present in all genetic syndromes
   c. the relationship of fluency disorders to genetic syndromes is unclear at this time
   d. neither stuttering nor disfluency is present in all genetic syndromes

References


