Research Article

Characteristics of Fluency and Speech in Two Families With High Incidences of Stuttering

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Purpose: This study presents data from 2 families with high incidence of stuttering, comparing methods of phenotype assignment and exploring the presence of other fluency disorders and corresponding speech characteristics.

Method: Three methods for assigning phenotype of stuttering were used: self-identification, family identification, and expert identification. Agreement on which individuals were assigned by each of these methods was studied. Multiple measures of fluency and speech production were obtained.

Results: Self-reports and descriptions of blocking rather than self-identification as a person who stutters demonstrated the best agreement with expert identification of stuttering. Family identification showed poor agreement with both

expert and self-identification of stuttering. Using binary categories of fluent or stuttering, 90% of individuals in 1 family were classified by expert consensus. Only 70% of the other family could be similarly categorized. Experts required 2 other categories, cluttering and other fluency disorders, to fully characterize dysfluency within this family. These 2 families also demonstrated differences in speech production.

Conclusion: Some families with high incidence of stuttering may also have high incidence of other fluency disorders and other speech-production difficulties. This finding may have ramifications for genetic studies, including criteria for defining phenotype and collapsing data across multiple families.

Nineteen years ago, two reviews of genetic studies of stuttering (Felsenfeld, 1996; Yairi, Ambrose, & Cox, 1996) concluded that the lack of consistent phenotype definitions and diagnostic protocols had confounded early genetic work (Kraft & Yairi, 2011). Over the intervening years, studies using new genetic approaches and technologies have implicated a wide range of loci with poor interstudy agreement (see Table 3 in Kraft & Yairi, 2011). After reviewing these studies, Kraft and Yairi (2011) reiterated the 1996 conclusion and called for researchers to adopt “identical diagnostic procedures and measurement, and matching definitions of phenotypes” (p. 44). They proposed standardization as a basis for collaborative data sharing to increase the data pool and achieve greater statistical power.

Of course, the adoption of standardized approaches will only advance our knowledge if the adopted methods,

measures, and definitions are appropriate, valid, and reliable. Of particular relevance in this regard is determining who is affected with stuttering. In this context, the presence of other fluency disorders within families may need consideration—that is, the appropriate phenotype may be specific to stuttering or may be related more generally to dysfluency. Such alternatives could include either the presence or absence of other fluency disorders (categorical) or degree of dysfluency (continuous). However, information on the presence of other fluency disorders has not routinely been included in previous genetic studies of stuttering. The purpose of this report is to share findings that bear on this issue.

Three methods have been used for assigning affected versus unaffected with stuttering in adults: (a) self-identification (asking a person if he or she stutters; e.g., Kidd, Heimbuch, & Records, 1981; Suresh et al., 2006), (b) family identification (asking family members to identify those who stutter in their families; e.g., Felsenfeld, van Beijsterveldt, & Boomsma, 2010; Kidd et al., 1981; MacFarlane, Hanson, Walton, & Mellon, 1992; Viswanath, Lee, & Chakraborty, 2004; Wittke-Thompson et al., 2007), and (c) expert identification (classified by one or more qualified professionals; e.g., Canhetti-Oliveira & Richieri-Costa, 2006; Kang et al., 2010; Levis, Ricci, Lukong, & Drayna,

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Editor: Jody Kreiman

Associate Editor: Hans-Georg Bosshardt

Received March 10, 2014

Revision received August 19, 2014

Accepted June 24, 2015

DOI: 10.1044/2015_JSLHR-S-14-0080

Disclosure: The authors have declared that no competing interests existed at the time of publication.

2004; Raza, Riazuddin & Drayna, 2010; Riaz et al., 2005; Shugart et al., 2004). The assumption has been made that individuals who identify themselves or other family members as persons who stutter (PWS) are providing reliable and valid information (e.g., Wittke-Thompson et al., 2007). However, we found no reports describing congruity of results (reliability) from these differing methods (i.e., were the same individuals identified by different approaches?).

At this time, it is not clear whether the trait being transmitted is specific to stuttering or may relate more broadly to speech dysfluency. Some have posited that there is a continuum of fluency disorders with severe stuttering at one end of the continuum and normal fluency at the other end (Adams & Runyan, 1981). Others have posited several discrete fluency disorders, with stuttering being the best known. At least two other fluency disorders are recognized: cluttering (St. Louis, Raphael, Myers, & Bakker, 2003; van Zaalen-Op't Hof, Wijnen, & De Jonckere, 2009) and highly nonfluent (Felsenfeld et al., 2010; van Beijsterveldt, Felsenfeld, & Boomsma, 2010). Children have primarily been given this latter classification. In adults, *nonfluent* is usually applied to patients with a specific type of aphasia. However, fluency textbooks do use the term *other fluency disorders* for adults (e.g., Silverman, 2004), which may be the adult corollary of highly nonfluent.

A small body of literature suggests that both cluttering and high nonfluency in children may have genetic underpinnings. One study has documented a high incidence of cluttering in one family with a dominant inheritance (Op't Hof & Uys, 1974). Twin studies have provided evidence that high nonfluency may share some genetic contributions with stuttering (van Beijsterveldt et al., 2010). Based on answers from parents about the presence of specific fluency symptoms, dizygotic (DZ) and monozygotic (MZ) twins were assigned the phenotypes of stuttering or not and highly nonfluent or not. Bivariate genetic modeling using cross-twin–cross-trait correlations (i.e., nonfluency in one twin is correlated with stuttering in the other twin) demonstrated higher correlations between MZ twin pairs (0.56) than DZ twin pairs (0.38), suggesting contributions from both genetic (cross-correlation is higher in MZ twins) and environmental (MZ cross-correlation is smaller than twice the DZ cross-correlation) factors (van Beijsterveldt et al., 2010). On the basis of these findings, it is quite possible that families previously studied because of their high incidence of stuttering may also include family members who clutter and/or are highly nonfluent.

This last point may affect the advisability of pooling data from all families with high incidences of stuttering (a recommendation made by Kraft & Yairi, 2011). It may be that families with high incidence of stuttering differ with respect to the presence of fluency disorders occurring in conjunction with stuttering. Should data be collapsed from families with a high incidence of both stuttering and other fluency disorders with data from families with a high incidence of stuttering only? Would collapsing data from families who differ in this respect confound the search for genetic underpinnings?

In this study, our objectives were twofold: (a) to compare self- and family identification of PWS with expert identification in two families with high incidences of stuttering and (b) to determine whether other fluency disorders and/or related speech production problems were present in these same two families.

Methods

Our research team identified two families for whom the incidence of stuttering was reported to be higher than in the general population. A subset of the larger family had been previously reported (MacFarlane et al., 1992). We included all of the family members who could be located, who agreed to participate, and who were physically and mentally capable of participation. This process yielded 100 subjects (75 from the larger family and 25 from the smaller). The first author conducted extensive interviews with each subject, typically in their homes.¹ As described in the following sections, specific portions of these interviews were video recorded for later review and analyses.

When possible, each individual was placed in a fluency category. The category of fluent (FL) was applied to family members by self-identification and expert identification. The category of stuttering (ST) was applied to family members by self-identification, family identification, and expert (panel) identification. The categories of cluttering (CL) or other fluency disorders (OFD) were added following the panel's review and discussion.

Self-Identification

Each family member was interviewed while being video recorded. The interview commenced with a series of questions. The first few questions were direct inquiries about whether the individual stuttered, either currently or as a child. No definition of stuttering was offered, allowing each subject to use his or her own criteria for stuttering. Because Sheehan and Martyn (1966) and Finn (1996) demonstrated the merits of asking about stuttering symptoms rather than simply asking "Do you stutter?"—especially for issues of persistence and recovery of stuttering—the next questions were framed to assess the presence or absence of specific stuttering symptoms. These questions probed blocks, repetitions, prolongations, hesitations, and struggle/tension. In probing specific symptoms, a standard description of each symptom was given in order to be certain the interviewee understood the question (see Appendix A for questions). In response to all questions, subjects were encouraged to explain, elaborate, describe, and discuss. We also asked questions related to speech variables often associated with fluency disorders (Daly & Burnett, 1996; Daly & Cantrell, 2006; St. Louis, 1996). On the basis of studies of cluttering,

¹Pedigrees were also constructed; samples for genetic analyses were obtained; and information relating to educational history and attitudes toward communication, health, and language were collected. These are outside the scope of this report and are not included herein.

primarily, these included articulation errors, difficulties producing multisyllable words, fast talking, and indistinct speech or mumbling. Questions were designed for individuals who are naïve to professional terminology in speech-language pathology. Again, subjects explained, described, and discussed the bases for their responses (see Appendix A).

Expert Identification²

An expert panel composed of seven certified speech-language pathologists was asked to assign each family member to a fluency category. Panel members were Edward Conture, Barry Guitar, Tom Gurrister, Peter Ramig, Ben Watson, and the first two authors.³ Panel members are all clinicians with experience treating stuttering; most have published articles in peer-reviewed journals and/or written books on stuttering. Some panel members have extensive experience in treating children, and others specialize in adults, but all have worked with both. Further, although they specialize in fluency disorders, all were competent and experienced in the diagnosis and management of other speech, voice, and language disorders.

Panel members were provided with video recordings, created from the original recordings to achieve a randomized sequence of 100 subjects identified only by number. Each video interview included the following:

1. The interview questions described in the self-identification section
2. A 2-min monologue on a topic chosen by the subject
3. The “Washington” passage from the *Stuttering Severity Instrument* (SSI; Riley, 1994) read aloud at a normal rate
4. The “Plumber” passage from the *Stuttering Severity Instrument* (Riley, 1994) read aloud as quickly as possible

Information not provided to the panel included the following:

1. Family membership (which family an individual belonged to or which individuals were genetically related and which were spouses)
2. The first author’s impressions and categorizations (on the basis of the complete face-to-face interviews)
3. Family identification data (those individuals identified by other family members)

Using a standardized form, panel members were asked to indicate the appropriate fluency category for each

individual. Because of the possibility that fluency disorders other than stuttering could be present and to help validate their existence, the panel members were asked to indicate the presence of specific types of dysfluencies (such as blocks, repetitions, hesitations, prolongations) and speech variables frequently associated with cluttering (e.g., articulation errors, difficulties producing multisyllabic words, fast talking, indistinct speech or mumbling; Daly & Burnett, 1996; Daly & Cantrell, 2006; St. Louis, 1996).

No directions were given regarding how the panel members should make their decisions. They could view the recordings as many times as needed; they were free to count dysfluencies or apply other standardized diagnostic techniques. They were told that although ST, CL, OFD, and FL were possible categories, there existed no a priori assumptions as to how many (if any) subjects would be assigned to any particular category. No definitions or descriptions were given for any category. Panel members were only asked to apply their clinical knowledge and experience to the decision-making process and to complete one form per subject.

After each expert had independently reviewed the recordings and completed the forms, the panel met together for a face-to-face consensus conference. Consensus for categorization was defined as agreement by four of seven judges. When differences of opinion existed, video recordings were reviewed, and panel members discussed the bases for their categorizations and/or the reasons for their inability to assign a category. Discussions continued until consensus was reached (i.e., agreement by at least four of seven panel members) or until it was agreed that consensus could not be reached.

Family Identification

As part of the interview, each family member was asked to identify relatives who stutter. ST was the only fluency or speech category assessed by family identification. The number of family members identified by one family member was compared with the number identified by two or more family members.

Profiling Fluency and Related Speech Characteristics

Multiple measures were used to further describe the speech of family members and to validate the panel’s fluency categories. These included quantitative measures of dysfluency, quantitative measures of speech rate, and occurrence of specific speech characteristics.

Type of Dysfluency

Type of dysfluency was based on the time-honored concept of dysfluencies typically associated with stuttering (e.g., blocks, prolongations, and repetitions) versus those that typically arise in normal (fluent) speakers (e.g., hesitations, interjections, revisions, and pauses; Wingate, 1964). Panel members reported which types of dysfluencies were

²Our use of a full panel should not be taken to imply that this approach is recommended or even proposed for wider use. We were probing the difficulty of the task and seeking optimal results for a limited sample.

³With respect to cluttering, two additional experts (Klaas Bakker and Florence Myers) were consulted. They reviewed video recordings and provided insights but regrettably did not participate in the consensus conference.

present for each subject, and the number of panel members reporting each type was tabulated for each subject. When a particular type of dysfluency was reported by at least four of seven panel members, it was considered characteristic of that subject. The number of individuals in each fluency category (i.e., FL, ST, CL, or OFD) characterized by each dysfluency type was tabulated and percentage computed.

Analyses

Separate chi-squared analyses⁴ were used to determine whether significant differences existed between observed versus expected numbers of individuals who demonstrated each type of dysfluency (block, repetition, prolongation, and hesitations) between fluency categories (FL, ST, CL, and/or OFD), and whether more than two fluency categories were present in a family between the fluency-disordered categories (ST, CL, and/or OFD). The null hypothesis prediction was that the distributions of each type of dysfluency would be the same for each fluency category (Conover, 1980) and for each of the fluency-disordered categories. For these analyses, each individual was included in one, and only one, fluency category.

Dysfluency Measures

Three direct dysfluency measures (percentage of dysfluent time during monologue, percentage of dysfluent time during reading at regular rate, and dysfluencies per minute during the monologue) were used as well as two rate measures (word rate on the basis of reading without instruction as to speed and fast rate on the basis of reading with instruction to read as quickly as possible). These five measures were all made by the first author from the videotaped samples. Descriptions and the reliability of each of these measures are included.

Percentage of dysfluent time. This measure reflects both frequency and duration but does not discriminate between the two. Percentage of dysfluent time was calculated for two speaking tasks, monologue and reading, because they differ in their communication demands. The duration of dysfluent speech and the total duration of each speech task were measured using a computer program that started and stopped a timer with a press of the space bar. For total duration of a speech task, the timer was started at the beginning of the speech task and stopped at the end of the task, and this process was repeated until two duration measures were within 3 s of one another. Those two duration measures were considered to be reliable. Then, they were averaged.

For duration of dysfluent speech, the timer was started and stopped at the beginning and end of each dysfluency throughout the entire speech task. This produced a sequential list of duration times on a spreadsheet. The number of entries on that list was the total number of dysfluencies for the speech task. This process was repeated until the agreement on total number of dysfluencies from two lists

was within three. Those two total numbers of dysfluencies were considered to be reliable. Then, the actual durations of all dysfluencies on each list were added to determine the total duration of dysfluent speech per list, and then durations from the two lists were averaged to arrive at total dysfluent time. Total dysfluent time was divided by total duration and multiplied by 100 to yield a percentage (Rosenburger, Wheelden, & Kalotkin, 1976).

Dysfluencies per minute. This measure provides a count of every dysfluent event but does not identify type of dysfluency or duration. This measure was computed for the monologue task by tallying the number of dysfluencies (an average of the total number of dysfluent events on the spreadsheets for each of two lists; see above) and dividing by the total duration of the sample in seconds (the averaged time from start of speech to end of speech), then multiplying by 60 (Rosenburger et al., 1976).

Word rate. This reflects aspects of frequency and duration of dysfluencies as well as global differences in speed of speech production (Perkins, Bell, Johnson, & Stock, 1979). It was calculated for the reading task by subtracting dysfluent time (reliability discussed above) from total time (reliability discussed above) and dividing by the number of fluent syllables.

Fast rate. This focuses on the speaker's ability produce faster speech and was calculated for the fast reading task by dividing total time (reliability discussed above) by number of syllables. Measurement of dysfluent time was not feasible for the fast reading task.

Analyses

For each of the five dysfluency measures described above, *t* tests were used to assess differences between the mean for all fluency-disordered (i.e., ST, CL, and/or OFD) family members and the mean of all fluent (FL) family members, using the panel's categorizations. If more than two fluency categories were present in a family, single-factor analyses of variance (ANOVAs) were then used for each of the five dysfluency measures to test for differences between the means of each of the fluency-disordered categories (ST, CL, and/or OFD).

Related Speech Variables

The panel noted occurrences of articulation errors, pronunciation errors, slurred or indistinct speech, unintelligibility at fast rates, and fast talking. For reliability, at least four of seven of the panel members had to agree on the presence of a specific speech problem. The number of individuals in each fluency category (FL, ST, CL, and OFD) demonstrating each of these characteristics was counted.

Analysis

Separate chi-squared analyses were computed to determine if significant differences existed between the observed versus expected number of individuals demonstrating these speech characteristics (articulation errors, pronunciation errors, slurred or indistinct speech, fast talking, and

⁴Note: Chi-square analysis cannot be used when a variable has 0% or 100% occurrences. Therefore, the chi-square analysis was only used when there was at least one occurrence.

unintelligibility with fast talking) between fluency categories (FL, ST, CL, and/or OFD) and between the fluency-disordered categories (ST, CL, and/or OFD). The null hypothesis prediction was that the distributions of each of the speech characteristics would be the same for each fluency category and for the three fluency-disordered categories (Conover, 1980). Each individual was included in one, and only one, fluency category.

Results

Description of Families and Subjects

Subjects

Subjects were 81 genetically related individuals from two unrelated families, plus 19 spouses of family members (see Table 1). Results for spouses (who could be considered a control group) were generally unremarkable (as would be anticipated for a control group) and are not included in this report.

Sixty genetically related subjects were from the larger family, designated herein as the NORTH family, and 21 were from the smaller family, designated as the SOUTH family. In the NORTH family ($N = 60$), 30 male and 30 female family members ranged in age from 7 to 80 years. Ten percent (6/60) were under the age of 18. In the SOUTH family ($N = 21$), 11 men and 10 women ranged in age from 19 to 80 years (see Table 1). All subjects were informed of risks and signed consent or assent (the six under the age of 18) forms approved by the Institutional Review Board from the National Institute on Deafness and Other Communication Disorders.

Organization of Results

At first, we collapsed data across all individuals in each fluency category and conducted analyses without referring to family membership. Results derived from these analyses were unsatisfactory because they were inconsistent and lacked discernible patterns. Reviewing the data led us to believe that marked differences between the two families precluded meaningful use of this approach. Therefore, we analyzed data from each family separately. The results are presented in the following order: (a) comparison of approaches for identifying those who stutter, (b) presence of fluency disorders other than stuttering, and (c) comparison of speech and fluency between the fluency categories and between the two families.

Comparison of Approaches for Identifying PWS

Identification of PWS in the SOUTH Family

Self-identification. Of the 21 members of the SOUTH family, nine (42%) identified themselves as PWS. Three of the nine reported they stuttered as children but did not believe they still stuttered. One family member denied stuttering but reported stammering. This individual maintained a distinction between stuttering and stammering and cited specific examples of behavioral differences. When asked about stuttering behaviors, all nine reported experiencing blocking and explicitly described associated sensations of tension and struggle. One additional family member did not self-identify as a PWS but reported blocking and described sensations including tension and struggle.

Panel (expert) identification. Ten (47%) were identified as stuttering by consensus of the panel. All nine of those who self-identified were included among these 10. The 10th was the family member who denied currently stuttering but, when questioned about blocking, immediately responded to the descriptions of internal perceptions of blocking with tension and struggle. This subject, like several others in the SOUTH family, described strategies successfully used for avoiding, controlling, or disguising blocks. Because these strategies prevented overt repetitions or prolongations, the subject denied currently stuttering.

Two of the three who believed their childhood stuttering had been resolved were judged by the panel to still be stuttering. The third was judged to be recovered but was included in the ST group. The panel was unable to agree (i.e., at least four of seven) on whether the self-identified "stammerer" belonged to the ST, OFD, or FL category.

Family identification. Although this method identified more PWS than either of the other two, there was close agreement between those identified by only one family member and those identified by two or more: 14 versus 13. These included the nine who self-identified as stuttering and all 10 identified by the panel. The remaining three were identified as normal speakers by the panel and did not self-report stuttering. The fourth (identified by only one family member) was the individual who self-identified as stammering.

One family member was identified as stuttering by 100% of the other family members and a second by 54%. Although neither was severe, these two demonstrated a full range of overt stuttering symptoms. Family identification appeared to overidentify individuals as PWS compared with self- or panel identification but included those individuals identified by other approaches.

Table 1. Number of family and nonfamily members in each of the two families.

Family	Family members			Spouses			Totals
	Male	Female	Total	Male	Female	Total	
SOUTH family	11	10	21	2	2	4	25
NORTH family	30	30	60	7	8	15	75
Totals	41	40	81	9	10	19	100

Identification of Stuttering in the NORTH Family

Self-identification. Fourteen members of the NORTH family identified themselves as PWS. Seven (half) of these reported they stuttered as children but did not believe they currently stuttered. Twelve of the 14 reported experiencing blocking and described sensations of tension and struggle associated with their blocks.

Panel (expert) identification. The panel identified 11 family members as PWS. Among those 11, only seven self-identified as PWS, and four denied stuttering. Among these 11, 10 responded affirmatively to questions about blocking and elaborated on the standard description with personal descriptions of internal sensations of blocking with accompanying tension and struggle. The other individual was observed by the panel to have blocks but did not self-identify as experiencing blocks.

Seven individuals who identified themselves as PWS were not so identified by the panel. The interviews suggested that these individuals either recognized that they had speech difficulties or had been told they were not normally fluent and had assumed the label of stuttering. The panel decided that the speech problems evidenced by these subjects were not, and probably never had been, stuttering. In contrast to self-reports of recovery, the panel did not believe any member of the NORTH family had recovered from childhood stuttering.

Family identification. Nineteen members (32%) of the NORTH family were identified as stuttering by at least one other family member, although only eight (13%) were identified by two or more family members. Of the 19, only six were among those identified by the panel. Of the eight identified by multiple family members, only four were identified by the other methods. Of the remaining 13 (including four with multiple identifications), all denied stuttering and blocking, and the panel concluded they did not stutter.

One family member was identified as stuttering by 81% of the other family members. This individual demonstrated a full range of overt stuttering symptoms. No other family member was identified by more than 4%. Family identification appeared to overestimate the number of PWS compared with self- or panel identification and did not necessarily include those identified by other approaches.

Presence of Other Fluency Disorders

For the remainder of the Results section, the fluency categorizations are all based on consensus by the panel (i.e., agreement by at least four of seven). Those that are not categorized as FL are considered to have a fluency disorder.

Presence of Fluency Disorders Other Than Stuttering in the SOUTH Family

Two categories, ST and FL, were sufficient to describe 19 of 21 (90%) members of the SOUTH family. The panel unanimously agreed that one of the remaining two did not stutter or clutter but was not a normally fluent speaker,

leading to categorization as OFD. The panel was unable to reach consensus on the family member who self-identified as a stammerer. They all agreed that the individual was not FL, but fewer than four of the panel members believed the individual should be categorized as ST. Table 2 summarizes the number of individuals in each category.

Presence of Fluency Disorders Other Than Stuttering in the NORTH Family

Only 70% of the NORTH family was encompassed when binary categories (ST and FL) were used. Thus, the panel used two additional fluency-disordered categories, CL and OFD, for the remaining 18 nonstuttering but dysfluent members of the NORTH family.

In spite of their best efforts, the panel was unable to reach a consensus for 10 (17%) members of the NORTH family. In the majority of cases, the issue creating lack of consensus was determining whether the individual should be categorized as CL. Although all panel members were familiar with the characteristics of cluttering, they could not agree as to which symptoms or symptom clusters were necessary and/or sufficient for categorization. Therefore, after the consensus meeting, we sought advice from two cluttering experts, Klaas Bakker and Florence Myers. They reviewed a subset of video interviews, which included the nonconsensus subjects as well as a sample of other family members. With their assistance, issues creating nonconsensus were resolved, and six of the 10 nonconsensus subjects were categorized.

The expanded panel identified seven members (12%) of the NORTH family as CL. There was an overlap between those categorized as CL and those categorized as ST. Three individuals stuttered and cluttered. To clarify differences between stuttering and cluttering, these three individuals were categorized under the designation of stuttering plus cluttering (ST + CL). Now, instead of two fluency-disordered categories (ST and CL), there were three (ST, ST + CL, CL).

The panel agreed that another seven (12%) members of the family neither stuttered nor cluttered but were equally certain that these seven were not normally fluent speakers. These seven constituted the fourth fluency-disordered category, designated by default as OFD. The four subjects for whom consensus was not reached were not included in any further analyses (see Table 2).

Table 2. Number of family members within each fluency category assignment by the expert panel for the NORTH and SOUTH families.

Family	ST	ST + CL	CL	OFD	FL	Nonconsensus	Total
SOUTH	10			1	9	1	21
NORTH ^a	8	3	4	7	34	4	60

Note. ST = stuttering; ST + CL = stuttering and cluttering; CL = cluttering; OFD = other fluency disorders; FL = fluent.

^aTotal who stutter = 11; total who clutter = 7.

Comparison of Fluency and Related Speech Characteristics Between Families and Between Fluency Categories

The fluency categories are all based on consensus by the panel (i.e., agreement by four of seven). Those not categorized as FL are considered to have a fluency disorder.

Profiling Fluency and Related Speech Characteristics for the SOUTH Family

Decisions by the panel placed 19 of 21 family members in one of two categories, either FL (nine) or ST (10). Therefore, these two categories were used for profiling fluency and related speech characteristics in the SOUTH family.

Types of dysfluency. Figure 1 illustrates the percentage of individuals who demonstrated blocks, prolongations, hesitations, and repetitions in the ST and FL categories for the SOUTH family. Ninety percent (nine of 10) of individuals in the ST category demonstrated blocks, because one individual (10%) was deemed to have recovered on the basis of a history of experiencing blocks but no current evidence of blocks. No blocks or repetitions were present for those in the FL category, so chi-squared testing was not completed for these two dysfluency types. The null hypothesis was accepted for both dysfluency types (prolongations and hesitations), meaning that the distribution of individuals demonstrating prolongations and the distribution of individuals demonstrating hesitations was similar for the ST and FL groups.

Dysfluency measures. The means for the three dysfluency measures for FL and ST categories in the SOUTH family are summarized in Table 3. The only significant difference was percentage of dysfluent time in the reading task, ANOVA: $df = (1, 17)$, $F = 7.98$, $p = .01$. From these findings, it is clear that, as a group, the ST members of the SOUTH family produced short, infrequent dysfluencies. Even though their dysfluencies were short and infrequent,

they were recognized as blocks by experienced clinicians because they were characterized by perceptible struggle and/or tension.

Rate measures. The mean articulation and fast rates for FL and ST categories in the SOUTH family are also reported in Table 3. No significant differences were found between categories for either rate measure.

Related speech variables. The FL and ST categories in the SOUTH family did not differ with respect to related speech measures in terms of the number of individuals presenting with articulation errors, pronunciation errors, slurred or indistinct speech, fast talking, or unintelligibility at fast rates. A few family members self-reported having some of these speech patterns, but not enough of the panel members observed them to reach our criterion for agreeing that a characteristic was present (i.e., reported by at least four of seven).

Conclusions and Implications for Findings in the SOUTH Family

The prevalence of stuttering in the SOUTH family was about 50% as contrasted with less than 0.5% in the general adult population, supporting the designation of familial stuttering. Of the 10 who stuttered, seven were men and three were women, yielding a 2.3:1 male-to-female ratio, which is slightly higher than ratios reported by Drayna, Kilshaw, and Kelly (1999; i.e., 1.57:1) and MacFarlane et al. (1992; i.e., 1.6:1).

The overt behavioral symptomatology demonstrated by the stuttering members of the SOUTH family was mild. One was recovered, and seven of the remaining 10 were able to avoid, control, manage, or disguise their moments of stuttering in ways that masked the existence of their problem. These seven can be described as demonstrating covert or interiorized stuttering. The nature of their problem was more apparent in their descriptions and awareness of speech difficulties (and in the strategies they used to avoid or prevent stuttering) than in the observable behavioral manifestations. The remaining two exhibited moderate overt stuttering symptoms.

Of the quantitative measures, only one—percentage of dysfluent time in reading—provided discriminating potential. With the exception of stuttering, the speech of the SOUTH family was well within normal limits. This family can be described as high incidence of stuttering, very low incidence of other fluency disorders, and very low incidence of co-occurring speech characteristics.

Profiling Fluency and Related Speech Characteristics for the NORTH Family

Types of dysfluency. Figure 2 illustrates the percentage of NORTH family members in each of the five categories demonstrating each type of dysfluency. Blocks were present in ST, ST + CL, and OFD categories but not in the FL or CL categories. Prolongations, hesitations, and repetitions, however, were present in the FL, ST, ST + CL, and OFD categories. Only prolongations and hesitations were present in the CL category. The null hypotheses of the distributions of prolongations being the same for all five

Figure 1. The percentage of individuals as assigned by panel consensus (at least 4 of 7 panel members) from the two categories of the SOUTH family that demonstrated blocks, prolongations, repetitions, and hesitations. Fluent ($n = 9$); ST = stuttering ($n = 10$).

Table 3. Mean percentage fluency, mean number of dysfluencies per minute, and mean rate measures for the two fluency categories assigned by consensus of the expert panel in the SOUTH family.

Category	<i>n</i>	Dysfluent time monologue	Dysfluent time reading	# dysfluencies per minute monologue	Articulation rate, syllables per second	Fast rate, syllables per second
ST	10	9%	3%	7.65	4.47	5.52
FL	9	9%	0%	8.22	4.67	5.74

Note. Significant differences are bolded. ST = stuttering; FL = fluent.

categories and the distribution of hesitations being the same for all five categories was rejected (hesitations: $df = 4$; $p = .01$; prolongations: $df = 4$; $p = .02$). The null hypotheses of the distributions of hesitations being the same for all four fluency-disordered categories and the distribution of prolongations being the same for all four fluency-disordered categories were accepted. The percentage of occurrence of dysfluencies for the ST + CL category was observed to be more similar to the ST category than the CL category (see Figure 2).

Dysfluency measures. Table 4 summarizes the means for the three dysfluency measures for each of the five categories for the NORTH family. Comparisons of all fluent to all fluency-disordered individuals revealed significant differences between percentage of dysfluent time in the monologue task (t test: $df = 54$, $t = 5.089$, $p = .000004$) and in number of dysfluent events (t test: $df = 54$, $t = -5.165$, $p = .000005$). Comparisons of the four fluency-disordered categories revealed significant differences between percentage of dysfluent time in the monologue task, ANOVA: $df = (3, 18)$, $F = 3.704$, $p = .03$, and in the number of dysfluent events, ANOVA: $df = (3, 18)$, $F = 4.5427$, $p = .015$.

The most intriguing element in these results was the pattern of differences between the reading and monologue

speech tasks. The mean percentage of dysfluent time for FL speakers was almost identical for both tasks. The mean percentage of dysfluent time for ST, ST + CL, and OFD speakers was greater for the monologue task compared with the reading task. In contrast, the mean percentage of fluent time for CL speakers was greater for the reading task compared with the monologue task.

Rate measurements. The means for the two rate measures for each of the five categories for the NORTH family are also presented in Table 4. No significant differences were found when comparing all fluent to all fluency-disordered individuals for either rate measure. No significant differences were found among the four fluency-disordered categories for either rate measure. The ST and ST + CL categories had the slowest mean rates, validating the observation that stuttering is associated with slower speech rates.

Related speech variables. The percentage of individuals in each of the five categories demonstrating the presence of articulation errors, pronunciation errors, slurred speech, fast talking, and unintelligibility at fast rates for the NORTH family is illustrated in Figure 3. Chi-squared analyses were completed comparing the presence or absence of speech production variables in the fluent and the four fluency-disordered categories. The null hypothesis that the distributions of slurred speech and unintelligibility at fast rates and fast talking were similar for all fluency categories was rejected (slurred speech: $df = 4$, $p = .0035$; unintelligibility at fast rates: $df = 4$, $p = .0035$; fast talking: $df = 4$, $p = .0005$). The null hypothesis that the distributions of articulation errors and pronunciation errors were similar between all fluency categories was accepted. The null hypothesis that the distributions of unintelligibility at fast rates as well as fast talking were similar between the four fluency-disordered categories was rejected (unintelligibility at fast rates: $df = 3$, $p = .01$; fast talking: $df = 3$, $p = .06$). The null hypothesis that the distributions of articulation errors, pronunciation errors, and slurred speech were similar between the four fluency-disordered categories was accepted. The percentage of occurrence of speech characteristics for the ST + CL category appeared to be more similar to the CL category than the ST category for the characteristics of fast talking and unintelligibility at fast rates and more similar to the ST category for articulation and pronunciation errors (see Figure 3). It was observed that there was a greater percentage of occurrence of individuals with speech production errors than with types of dysfluency in the CL category (see Figures 2 and 3).

Figure 2. The percentage of individuals as assigned by panel consensus (at least 4 of 7 panel members) from the five fluency categories of the NORTH family that demonstrated blocks, prolongations, repetitions, and hesitations. Fluent ($n = 34$); ST = stuttering ($n = 8$); ST + CL = stuttering plus cluttering ($n = 3$); CL = cluttering ($n = 4$); OFD = other fluency disorders ($n = 7$).

Table 4. Mean percentage fluency, mean number of dysfluencies per minute, and mean rate measures for each of the fluency categories assigned by consensus of the expert panel in the NORTH family.

Category	<i>n</i>	Dysfluent time monologue	Dysfluent time reading	# dysfluencies per minute monologue	Articulation rate, syllables per second	Fast rate, syllables per second
ST	8	11%	16%	12.51	3.48	3.86
ST + CL	3	15%	5%	11.48	3.38	3.98
CL	4	5%	13%	3.66	3.85	4.43
OFD	7	13%	3%	11.41	3.8	4.39
FL	34	5%	6%	5.27	3.68	4.5

Note. Those that are bolded are significantly different between fluent and dysfluent (ST, ST + CL, CL, and OFD) speakers. Those that are in italics are significantly different between the “dysfluent” fluency categories. ST = stuttering; ST + CL = stuttering and cluttering; CL = cluttering; OFD = other fluency disorders; FL = fluent.

Conclusions and Implications of the Findings in the NORTH Family

The prevalence of stuttering in the NORTH family was about 13% as contrasted with less than 0.5% in the general adult population, supporting the designation of familial stuttering. Of the eight who stuttered, seven were male and one was female, yielding a 7:1 male-to-female ratio, which is much higher than ratios reported by Drayna et al. (1999; i.e., 1.57:1) and MacFarlane et al. (1992; i.e., 1.6:1).

The panel resolved the spectrum of symptoms observed in this family into four fluency-disordered categories, ST, ST + CL, CL, and OFD. The 22 dysfluent subjects differed significantly from the 34 fluent subjects on measures of dysfluencies per minute and percentage of dysfluent time during monologue and the number of individuals demonstrating slurred speech, fast talking, and unintelligibility at fast rates.

Each of the four fluency-disordered categories differed from one another on at least one variable. Any individual in the categories that included stuttering (ST, ST + CL) reported blocking, either currently or in the past.

Figure 3. The percentage of other speech measures (articulation errors, pronunciation errors, slurred speech, fast talking, and unintelligibility at fast rates) present as assigned by panel consensus (at least 4 of 7 panel members) from the five categories of the NORTH family. Fluent (*n* = 34); ST = stuttering (*n* = 8); ST + CL = stuttering plus cluttering (*n* = 3); CL = cluttering (*n* = 4); OFD = other fluency disorders (*n* = 7).

Individuals in the categories that included cluttering (CL, ST + CL) had significantly higher incidences of related speech characteristics, including fast talking and unintelligibility at fast rates, than other fluency-disordered groups. Individuals in the category of OFD had a high percentage of hesitations compared with other type of dysfluencies. The NORTH family can be described as high incidence of stuttering, high incidence of other fluency disorders, and high incidence of co-occurring speech production issues.

Discussion

Clearly, the results from one study of only two families cannot be generalized to all families with high incidence of stuttering. However, our findings strongly suggest that careful studies of fluency and speech in families with increased incidence of stuttering should be included in the design of future genetic studies of stuttering.

The first objective of this study was based on the differing methods that previous genetic studies have used to identify family members as stuttering versus fluent. Our results suggest that family identification of stuttering was not reliable.

We hoped that the combined clinical acumen of our expert panel might lead to agreement on the minimum characteristics needed to categorize an individual as a PWS. Using their categorization as the gold standard, we could then examine agreement with self-identifications and family identifications.

The most helpful finding from this study was the post hoc analysis of the symptom(s) that the expert panel apparently relied on to make the diagnosis of stuttering. Blocking appeared to be the differentiating symptom. A combination of observing blocking behaviors and an individual's agreement with and elaboration of descriptions of internal perceptions of blocking with struggle or tension proved to be the necessary conditions for diagnosing stuttering. One subject was not observed blocking, but the panel assigned that individual to the stuttering category on the basis of descriptions of childhood blocking. The concept of blocking as the salient characteristic of stuttering is not original. It has lurked in the literature for many years, but was most clearly and explicitly advocated by Perkins (1990, 1996).

The use of blocking (with associated tension and struggle) as the salient characteristic of stuttering was important in panel decisions of recovery and persistence of stuttering, an important consideration as both run in families (Ambrose, Cox, & Yairi, 1997). For example, in the SOUTH family, one subject who self-reported as recovered was accepted as recovered by the panel. This subject reported experiencing blocking in childhood, but panel members observed no current blocking behaviors. At least two other members of the SOUTH family were unsure whether they had recovered. They reported blocking with overt stuttering in childhood but learned to disguise or avoid overt stuttering. They described internalized blocks that still occurred in their speech and the methods they used to prevent overt stuttering. The panel was able to detect some of their blocks, so they were not considered recovered.

In contrast, several members of the NORTH family believed they had recovered from childhood stuttering. However, when their current speech was analyzed and their reports of childhood symptoms were considered, the panel decided that these speakers had not recovered from stuttering. Some still had observable blocks, suggesting persistence, and others were categorized as CL or OFD and probably had never stuttered. Thus, the use of blocking rather than the label of stuttering made distinguishing between persistence and recovery easier. This result has also been reported in two studies designed to establish techniques to validate recovery from stuttering (Finn, 1996; Sheehan & Martyn, 1966). Both of these studies found that recovered PWS reported having experienced characteristic features of stuttering in the past. Finn (1996) reported that 100% of recovered PWS reported having repetitions, prolongations, or blocks, with half reporting associated struggle or tension. Sheehan and Martyn (1966) also reported discrepancies between those who labeled themselves as PWS and those who demonstrated stuttering behaviors.

The use of symptom description rather than category label reflects a larger semantic issue, involving how individuals conceptualize the term *stuttering*—that is, some individuals thought they stuttered because others in the family stuttered, and their own speech was not normal, so they labeled themselves as PWS. In some cases, others had labeled them as PWS, and they accepted that label. In reality, they demonstrated other speech or fluency disorder(s). This is a false-positive identification and occurred more frequently in the NORTH family.

In direct contrast, some individuals reported they did not stutter because they did not exhibit the overt symptoms characteristic of the more severely affected members of their family. These individuals were observed as having blocks but denied ever stuttering or considered themselves recovered. This is a false-negative identification and occurred more frequently in the SOUTH family.

The semantic issue also appeared to have ramifications for how family members identified stuttering in other family members. Family identification demonstrated poor agreement with expert and self-identification. In the SOUTH family, more individuals were identified by family members

than by either expert or self-identification. However, those who were identified by expert or self-identification were included in those identified by family members. In contrast, in the NORTH family, only half of those identified by expert or self-identification were also identified by family members. Both of these findings emphasize our critical need for a standard operational definition of stuttering.⁵

Our second objective was to explore the existence of other fluency disorders in families with a high incidence of stuttering. If other fluency disorders were not found, then the binary categorization of family members as fluent versus stuttering would be adequate. Of our two families, the SOUTH family can best fit this type of categorization (i.e., 90%). However, when other fluency disorders are found, as in the NORTH family, then this categorization strategy may need to be revised. Instead, a binary categorization of fluent versus dysfluent may be preferable. Alternatively, categorization may not be the optimal approach, and a continuum of dysfluency may prove more viable.

Further, these dramatic differences between two families with high incidence of stuttering may have implications with respect to analyses of data across families. Should data be collapsed across multiple families without respect to critical differences between families? Collapsing data across families has been recommended as a strategy for increasing statistical significance (Kraft & Yairi, 2011). Findings from this study suggest caution and the need for more exploratory research.

As with stuttering, we need standard operational definitions for cluttering and other fluency disorders. In this study, individuals categorized by consensus of our panel members as cluttering differed on speech measures rather than on fluency measures, suggesting the importance of these variables in such a definition. This finding is in line with other researchers who have reported that persons who clutter are not significantly different in number of dysfluencies (Bakker, Myers, Raphael, & St. Louis, 2011; Myers, Bakker, St. Louis, & Raphael, 2012). Instead, those categorized as cluttering evidenced a range of speech symptoms, including pronunciation errors, slurred speech, unintelligibility of speech at faster rates, and fast talking. These were all included as cluttering symptoms by St. Louis (1996) and on a clinical checklist for diagnosing cluttering by Daly and Burnett (1996), subsequently revised (Daly & Cantrell, 2006). Also in line with previous research (van Zaalen-Op't Hof et al., 2009), those in the cluttering category did not have increased articulation rates compared with other fluency-disordered categories in spite of being perceived as fast talkers. It is possible that listeners perceive fast talking when a speaker's rate degrades articulatory accuracy or intelligibility regardless of the absolute syllable per second speech rate (van Zaalen-Op't Hof et al., 2009).

Individuals categorized by the expert panel as OFD revealed frequent hesitations (without tension or struggle).

⁵Although a widely accepted theoretical definition is desirable, an operational definition is essential.

This category of fluency disorder was present in both families, although to a much greater degree in the NORTH family (seven individuals) than the SOUTH family (one individual). This fluency-disordered group requires further study to help clarify the distinction between a continuum of fluency, as proposed by Adams and Runyan (1981), and distinct fluency-disordered groups.

This study did not investigate and does not speak to all of the potentially important variables, including language (such as word-finding difficulties) and/or environmental factors. It does offer some specific guidance in terms of future directions. It does suggest a possible operational definition of stuttering that would be worth exploring in other families with a high incidence of stuttering. This work also demonstrates that families with a high incidence of stuttering differ with respect to the presence of other types of fluency disorders. Thus, a more complete assessment of fluency and speech production abnormalities is warranted in these families. Results identify three factors that may need to be considered as part of phenotype assignment: presence of stuttering, presence of other fluency disorders, and presence of speech production abnormalities. What we cannot say is whether these are dichotomous or continuous traits.

Acknowledgments

This research was funded by the Intramural Division of the National Institute on Deafness and Other Communications, Protocol #97-DC-0002, titled "Genetic Linkage Analysis in Developmental Stuttering: Gene Mapping in Extended Kindreds and Candidate Gene Analyses" (Principal Investigator: Sheila Stager).

We would like to acknowledge the contribution of those who joined the authors in serving as experts on our panel: Edward Conture, Barry Guitar, Tom Gurrister, Peter Ramig, and Ben Watson. They gave significantly of their time in reviewing the patient tapes, filling out the forms, and coming to Bethesda for the consensus conference. The analyses could not have been completed without their clinical acumen. We would also like to acknowledge Klaas Bakker and Florence Myers, who reviewed a subset of the subjects following the consensus conference. Again, they gave significantly of their time in reviewing patient tapes and their efforts helped to clarify the classification of those individuals for whom the panel could not achieve consensus. We would also like to acknowledge the willingness, time, and patience of the family members who participated in this study.

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Appendix A

Interview Questions

Have you ever considered yourself to be a person who stutters?

Is it typical in your speech to repeat the first sound in a word, like *b-b-b-ball*?

Is it typical in your speech to prolong the first sound in a word, like *sssssssssam*?

Do you find that your mouth is in a position to say a word, but the word just does not come out immediately?

If yes, is it because you know exactly what you want to say and there is something blocking your ability to say it, or are you unsure of what you want to say and are planning it?

Do you feel any tension or struggle in the muscles that you use to speak when you are speaking?

Do you have a lot of hesitations, *uhms*, *ers*, in your speech?

Is it typical for you to look ahead at what you are going to say and change a word because you think you are going to have difficulty saying that word?

If yes, what kind of words will you change?

Are there any movements that you do with your hands, arms, feet, or head to get speech started?

Has anyone reported that your voice sounds too low in pitch or too high in pitch, either one?

Has anyone reported that you speak too loudly or too softly, either one?

If yes, are you aware when it happens, or does someone have to tell you?

Has anyone reported that you speak too quickly or too slowly, either one?

If yes, are you aware when it happens, or does someone have to tell you?

Have you ever lost your voice for a period of time not associated with a cold or flu?

Has anyone suggested that your speech is slurred or indistinct to them?

As a child, did you ever pronounce certain sounds incorrectly, like always saying /w/ for /r/ or lisping?

Do you consider your vocabulary to be above average, average, or below average that of your peers?

Do you have any difficulty pronouncing words with many syllables in them, such that you drop syllables or mix up the sounds?

Have you ever been seen by a speech-language pathologist for any reason?

If yes, why and when?

Has your hearing ever been tested? When? Was it within normal limits? Do you have any reason to believe that your hearing is not within normal limits now?