FAMILIAL PERSISTENT DEVELOPMENTAL STUTTERING: DISFLUENCIES AND PREVALENCE

Gagueira desenvolvimental persistente familial: disfluências e prevalência

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ABSTRACT

Purpose: to characterize and to compare the frequency of speech disfluency in adults with familial persistent developmental stuttering in males and females, the stuttering severity and then to determinate the familial prevalence and the sex ratio of stuttering among the families members of probands. **Methods:** subjects were 30 adults who stutter (ages between 18 and 53 years old), divided in two groups: one with 20 males, and the other with 10 females. Data were gathered by clinical and familial history, fluency assessment and Stuttering Severity Instrument. **Results:** the percentages of stuttering-like disfluencies (SLD) (p=0.352), of other disfluencies (OD) (p=0.947) and of total disfluencies (TD) (p=0.522) were similar between males and females. A mean of 5.23% of SLD and 5.5% of OD were found among subjects. The mild subgroup was the prevalent one among the participants (83.3%). The members of male's families presented a greater risk to stutter when compared to females (p<0,001). From the 1002 members of the families analyzed, 85 presented stuttering, of which 53 were male and 32 female. **Conclusions**: there were no differences between males and females concerning the analyzed measures. Regarding the frequency of disfluencies, results around a half of the total disfluencies were characterized as SLD. The subgroup of familial persistent developmental stuttering was characterized mainly as mild. The risk among relatives of affected probands was 8.5%.The familial prevalence data showed that risk that a person has to manifest stuttering when there is some familial affected was 8.5%.The sex ratio of stuttering was 3.72 males to 1 female (3.72:1).

KEYWORDS: Speech Language and Hearing Sciences; Speech; Evaluation; Speech Disorders; Stuttering; Genetic

INTRODUCTION

Stuttering is a fluency disorder characterized by excessive atypical breaks during linguistic formulation[1-3], impairing smoothness[4] and time of speech[5]. Its main symptom is intermittent failure of the nervous system in generating appropriate command signals to the muscles to which the activity must be dynamically controlled so that fluent speech can be produced[6]. In this sense, objective assessments have an important role not only in the initial stage of diagnosis, but also in controlling the evolution of the disorder[7]. The measure of percentage of stuttering-like disfluencies is considered the "gold standard" of behavioral assessment of the disorder[8]. Also the classification of stuttering severity is indicated in the disorder diagnosis process [8-11].

When the disorder begins in childhood it is denominated as developmental stuttering. Historically, there is a variety of etiological explanations for stuttering [12] and although its origin is not yet well understood [13], there is a consensus that genetic factors act in approximately half the cases of persistent developmental stuttering [14]. In cases where there is familial recurrence, or in which two or more individuals of the same family are affected by

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the disorder, stuttering is denominated as familial[15]. This subgroup of stuttering was denominated as Familial Persistent Developmental Stuttering[14] and is the most prevalent.

Therefore, familial persistent developmental stuttering is considered a disorder with complex or multifactorial pattern [16]. The disorder is the result of complex interactions of predisposing factors such as genotype at one or more loci, in addition to various environmental components able to activate, accelerate or intensify the manifestation of stuttering.

Molecular genetic research has focused on investigating the susceptibility of genes that may contribute to the transmission of stuttering[12]. There is a diversity of research findings, suggesting that stuttering is probably a polygenic disorder with several genes that may increase the genetic predisposition [17,18].

Gene mapping studies, associated with varied and complex statistical analyzes, such as studies of genoma-wide linkage and association analysis, have been used extensively in the location and identification of specific loci and alleles involved, which provide definitive evidence of the genetic contribution to stuttering[19].

Professionals working in this area need to understand that there are subtypes of stuttering, and they should be characterized. Due to the complexity of the disorder, the characterization of clinical manifestations of stuttering is relevant and will assist the diagnostic and therapeutic processes.

This research, therefore, had the following objectives: to characterize and compare the frequency of disfluency speech of adults with familial persistent developmental stuttering, for males and females, severity of the disorder, and to determine the familial prevalence and gender ratio of stuttering among the relatives of the proband.

METHODS

This research was established as a cross-sectional, quantitative and qualitative study, conducted with adults who stutter, at the in the *Laboratório de Estudos da Fluência* [Fluency Study Laboratory] - LAEF of the *Centro de Estudos da Educação e da Saúde* [Education and Health Study Center] (CEES) of Universidade Estadual Paulista - FFC – Marilia.

This study was approved by the Ethics Committee of the Faculty of Philosophy and Sciences, Universidade Estadual Paulista - CEP / FFC / UNESP under protocol n ° 0724/2013.

A total of 30 adults participated in this study, presenting familial persistent developmental stuttering, 18-53 years (X = 31, SD = 8.9), with 20 males and 10 females. These adults are referred to as probands; this term is commonly used in genetic studies, and refers to the first member of the family affected, who seeks for treatment.

As inclusion criteria, all participants should present: age between 18 and 59 years (since one study showed that people over 60 years old had some differences in the profile of fluency) [20]; positive family history for the disorder; minimum duration of 12 months of disfluency; onset of stuttering must have occurred in childhood; mimimum of 3% of the stuttering like disfluencies [21,22] and presentt scores 18 or more points on the Stuttering Severity Instrument on the classification for the total score of the test and its severity (mild, moderate, severe or very severe) [11].

Adults who had other complaints, hearing, neurological, psychological and / or psychiatric disorders, were excluded.

The procedures were performed after signing the consent form in which the adults who stutter signed the agreement freely, authorizing the participation in this study.

For the selection of participants through the application of the criteria for inclusion and exclusion, adults who stutter were questioned orally on the identification data and family history. Pre-elaborated forms were used like the consente form of the participant, identification form, family history, fluency assessment protocol[21] and Stuttering Severity Instrument [11] protocol.

Data of the familial history to accomplish the heredogram were collected at the end of the clinical history. This procedure was done because it is known that the detailed distribution of stuttering between different classes of relatives allows estimating the disorder risk for each classes of relatives [23]. The probands were asked about the standard of fluency of their relatives and about the existence of someone in the family who presented stuttering, or had stuttered in childhood. To enable probands to answer these questions, the interviewer had a standard definition of stuttering offering examples that could illustrate it. Stuttering was defined as “interruption in the continuity of the flow of speech characterized as repetitions, prolongations, or blocks of sounds, syllables or short words” [16]. Examples of repetitions of sounds or syllables, repetitions of monosyllabic words, prolongations of sounds and blocks were offered.

The speech samples collected were transcribed in a total of 200 fluent syllables, considering the fluent and non-fluent syllables. Subsequently, the analysis of speech samples and characterized the types of disfluencies were performed according to the following description [21]:

- Other disfluencies: repetition of non monosyllabic word, phrase repetition, interjection, revision and incomplete sentence.
- Stuttering-like disfluencies: part os word repetitivo, monosyllabic word repetition, block and sound prolongation

For characterizing the frequency of disfluencies, the following measures were employed: percentage of total of disfluency, percentage of other disfluencies and percentage of stuttering-like disfluencies.

For the diagnosis of stuttering, it was adopted the criterion for presence of at least 3% of stuttering--like disfluencies, and stuttering disorder should be rated at least as mild in the Severity Sttutering Instrument[11]. Thus, cases of very mild stuttering, were excluded.

For statistical analysis the Mann-Whitney Test was employed, to verify possible differences between the considered variables between males and females. Another method of statistical analysis employed was the chi-square test for proportions, in order to verify possible differences between the prevalence of stuttering for relatives of probands from male and female genders. The significance level for statistical tests was 5% (0.050). Data analysis was performed using SPSS (Statistical Package for Social Sciences) program, version 22.0.

RESULTS

The results for the frequency of disfluency data, severity of the disorder and familial prevalence and sex ratio, were presented in tables and figure. Regarding frequency of disfluency is can be observed that there were no differences between males and females, regarding the values obtained on the stuttering-like disfluencies typical stuttering (SLD), the other disfluencies (OD) and total disfluencies (TD). For the three measures examined, the male group showed a trend toward greater variability in relation to female group, especially regarding stuttering-like disfluencies (SLD) and total disfluencies (TD) (Table 1).

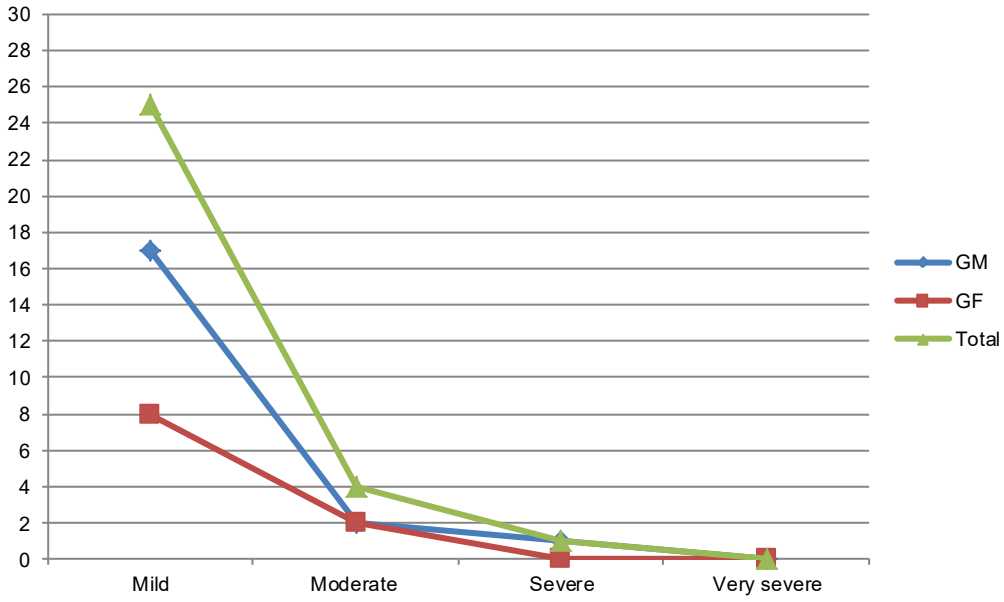
It is also worth noting that approximately half of the total disfluencies (TD) were characterized as stuttering-like disfluencies (SLD), and half as other disfluencies (OD) (Table 1).

Table 1 – Distribution of values of the percentage of the stuttering-like disfluencies, other disfluencies and of total disfluency of adult stutterers groups of males and females.

Frequency of disflluencies							
Variable	**Gender**	**N**	**Mean**	**Standard deviation**	**Minimum**	**Maximum**	**p-value**
%SLD	males	20	5.50	3.07	3.00	17.00	0.352
	females	10	4.70	1.97	3.00	8.50	
	total	30	5.23	2.75	3.00	17.00	
%OD	males	20	5.55	2.22	2.00	11.50	0.947
	females	10	5.40	1.78	3.50	8.50	
	total	30	5.50	2.06	2.00	11.50	
%TD	males	20	11.10	4.52	6.00	25.00	0.522
	females	10	10.10	3.43	7.00	17.00	
	total	30	10.77	4.16	6.00	25.00	

Legend: %SLD= Stuttering-like disfluencies; %OD= Other Disfluencies; %TD= Total Disfluencies; N: Number of participants.
P-value refers to Mann-Whitney test - significant values are in bold and with an asterisk.

The analysis of stuttering severity showed that the group of adults with familial developmental stuttering subtype showed higher prevalence of mild stuttering in relation to moderate, severe or very severe. This trend was also observed when the groups were separated into male and female. None of the participants expressed very severe stuttering (Figure 1).



Legend: GM = male group; GF = Female group.
Severity score: 10-17 = very mild; 18-24 = mild; 25-31 = moderate; 32-36 = severe; 37-46 = very severe (Stuttering Severity Instrument - SSI-3, Riley, 1994).

Figure 1 – Comparison of groups of males and females, and the total number of adults and the stuttering severity

The familial prevalence of stuttering in probands was investigated by counting the number of families of males and females who presented stuttering for each proband. It was observed that the prevalence of stuttering was statistically higher for relatives of males in relation to female family members, either for male probands and for the total of the probands. However, non statistically significant difference was observed, when it was analyzed the total of affected probands from the males compared to females, (p = 0.242) (Table 2).

A separate analysis by proband gender, showed that the class of greatest risk for the development of stuttering was the class of the relatives of male probands, descendant from female probands (0.164). The class that showed lower risk for the disorder was the class of female relatives of probands, descendant from female probands (0.032). The male/female ratio showed the highest prevalence of stuttering for males (Table 2)

Table 2 – Prevalence of familial stuttering in relatives of probands, represented by the number of stutterers males and females divided by the total number of relatives of male and female, and male / female (M / F) ratio.

	Male probands	Female probands	Total
Family male stutterers	40/372= 0.107	27/165 = 0.164	67/537 = 0.125
Family stutterers females	13/308 = 0.042	5/157 = 0.032	18/465 = **0.039***
p-value	**0.001***	**< 0.001***	**< 0.001***
Total of relatives stutterers (male and female)	53/680 = 0.078	32/322 = 0.099	85/1002 = 0.085
p-value	0.242		
M / F ratio	3.08	5.40	3.72

Legend: M / F = Male / female
P-value refers to the test of Chi-square - significant values are in bold and with an asterisk.

DISCUSSION

The contemporary literature has shown the importance of better understanding the stuttering subtypes. However, few studies have characterized the measures of disfluencies and the severity of the disorder in different subgroups, and described about the familial prevalence and gender ratio. Thus, this study characterized the frequency of speech disfluencies in adults with familial persistent developmental stuttering, the severity of the disorder as well as determined the familial prevalence and gender ratio of the relatives of probands with stuttering.

The data obtained from the spontaneous speech of adults with familial persistent developmental stuttering, has helped to confirm, that on average, the group showed 10.77% of ruptures, approximately half characterized as stuttering-like disfluencies (SLD) and the other half as other disfluencies (OD).This balance in the distribution of disruptions occurrence of stuttering adults between SLD and OD found in this study is similar to results of a survey comprising 40 children, who were stuttering speakers of Brazilian Portuguese [24].

However, the data on this similar distribution of disfluencies in the group of people who stutter, differ from the findings on the disfluency in fluent people, because adults who are fluent speakers of Brazilian Portuguese have shown that there is a greater quantity of other disfluencies, in relation to the stuttering-like disfluencies [20]. The data therefore suggest that the fluency profile of stutterers distinguishes from the profile of fluent people, on the distribution among other disfluencies and the stuttering-like disfluencies.

When the data were compared with fluent speakers of Brazilian Portuguese, it was observed that adults who stutter demonstrated increased value of frequency for total disfluencies, in relation to fluent (confidence interval in the studied age, ranged was from 8.32% to 9%) [20]. However, this number was relatively close (10.77%) in the results of this investigation. This finding was expected, since the main manifestation of stuttering is the increased frequency of stuttering-like disfluencies [1-3], and the sum of these disfluencies with other disfluencies has caused the increase on the total of disfluencies.

The results for the values concerning the frequency of disfluency corroborate to the previous description [25]. Quantitative values of disfluency of 10 stuttering adults of different severities, showed an average of 10.25% of total disfluencies, 5.25% from other disfluencies and 5.02% of the stuttering-like disfluencies[25].

It is worth noting that some stuttering definitions describe the presence of an increased amount of stuttering-like disfluencies[1-3]. The results of this study corroborate to these descriptions, as the average presented by the group was 5.23% of disfluencies, and the criteria used by the scientific community recognizes the minimum of 3% [22].

Specifically, on the frequency of stuttering-like disfluencies, the result obtained in this study (5.23%) was lower when compared to a survey conducted comprising 22 adults with persistent developmental stuttering, speakers of American English (7.1%) [26].

These findings are important for speech pathologists who realize the diagnosis of communication disorders, as they help to distinguish with greater safety, the fluency disorders, also contributing to the classification of the severity of the disorder, and hence the assessment of therapeutic efficacy.

However, it becomes important to highlight, that adults who stutter also express other disfluencies

which are part of the communication of any speaker[25]. These data suggest that this characteristic occurs for any age, considering that an investigation with children who stutter, also showed the presence of other disfluencies or also denominated as common disfluencies [24].

The findings concerning the severity of the disorder show no consensus in the literature. These results corroborate to a study of 10 adults who stutter, in which 50% were classified as mild stuttering, 10% moderate, 20% severe and 20% very severe[25], suggesting therefore that the mild stuttering is more prevalent. However, the results are not in accordance to the data for the probands from a study with 17 children who stutter, as 53% were classified as moderate stuttering, 29.4% mild, and 17.6% severe[27]. In the same publication, the author showed that 95% of the affected family members (total of 20 first-degree relatives of persons) showed mild stuttering, a result similar to our study.

A survey comprising 47 adult speakers of American English with familial stuttering, rated severity employing the Stuttering Severity Instrument[11], and found 19 with mild stuttering[28]. However, cases of stuttering classified as very mild were not considered in this study, so the comparative analysis was performed with a total of 28 adults. Thus, the results corroborate to those described above, since most of them presented mild stuttering (42.85%) [28].

Noteworthy is the fact that results on the severity and frequency of disfluency in familial persistent developmental subtype, were not found compiled in the bibliography.

With regard to the prevalence of stuttering, it was observed that the number of male relatives affected was higher than the number of female relatives, for total probands. This result confirms previous data that stuttering is a disorder that has a higher prevalence of male gender[7,15,24,29]. Therefore, relatives of male probands with stuttering had higher risk (12.5%) than female relatives (3.9%).

The prevalence of familial persistent stuttering was higher for relatives of male probands, than from females (16.4%) confirming the literature[30]. The risk of a male family member (12.5%) was 3.2 times greater than the risk of a female (3.9%). This finding corroborates to previous investigations that reported the highest prevalence of stuttering among males[16,24].

The data showed that, despite the risk of a family from a female proband (9.9%) have shown a tendency to be higher in relation to the risk of a family from a male proband (7.8%), this difference was not statistically significant (p= 0.242). Although the literature indicates that female probands have a higher prevalence of stuttering among their family [31], the results obtained in this investigation do not corroborate this description. The overall risk of a family member of a proband to present stuttering, regardless of gender, was 8.5%.

The results present restrictions, due to the number of participants, but they may contribute as basis for further research, and bring relevant clinical implications in the diagnosis and prognosis of the patient.

In this sense, the speech pathologist should include in his/her diagnostic routine, the procedure named as family history, in order to help determine the risk of the person for developing persistent stuttering, as well as to define the familial or isolated stuttering subgroup. In addition, this information will conduct the guidelines that the professional should provide to the family about the possible risks that a person may present to the onset of stuttering.

CONCLUSION

The results obtained in this study showed that, on average, the total disfluencies frequency of the group of adults with familial persistent developmental stuttering was 10.77%, of which approximately half were classified as other disfluencies, and the other half as stuttering-like disfluencies. Comparing the frequency of disfluency between male and female groups, there was non statistically significant differences. The subtype of familial persistent developmental stuttering was characterized mainly by a mild disorder, concerning severity. Although most of the disorder was classified as mild, stuttering can present an impact on the quality of life of the speaker and therefore needs to be valued.

The familial prevalence data showed that the risk for a person to present stuttering, when there is a stuttering affected relative was 8.5%, and this risk increases when the person is male. The ratio between genders showed that stuttering has affected more males, when compared to females, a ratio of 3.72:1.

The health professional must therefore enhance the knowledge about the different subtypes of stuttering, in an attempt to offer a qualified service in order to meet the real needs of different people who stutter.

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RE**SUMO**

Objetivo: caracterizar e comparar a frequência das disfluências da fala de adultos com gagueira desenvolvimental persistente familial do sexo masculino e feminino, a severidade do distúrbio e determinar a prevalência familial e a razão entre gêneros da gagueira nos familiares dos probandos. **Métodos:** participaram 30 adultos com gagueira (18 a 53 anos), divididos em dois grupos, sendo 20 do sexo masculino e 10 do sexo feminino. Os procedimentos realizados foram: história clínica e familial, avaliação da fluência e Instrumento de Severidade da Gagueira. **Resultados:** as porcentagens de disfluências típicas da gagueira (p=0,352), de outras disfluências (p=0,947) e do total das disfluências (p=0,522) foram semelhantes entre os grupos masculino e feminino. A média de disfluências típicas da gagueira foi 5,23% e de outras disfluências 5,50%. O subtipo leve foi manifestado pela maioria dos participantes (83,3%). Os familiares do gênero masculino apresentaram maior risco de apresentar gagueira (p<0,001). Do total de 1002 familiares, 85 apresentaram gagueira. No total de familiares afetados (n=85), 53 eram do sexo masculino e 32 do feminino. **Conclusão:** não houve diferenças entre os grupos masculino e feminino nas medidas analisadas. Quanto à frequência das disfluências, aproximadamente metade do total das disfluências foi caracterizada como disfluências típicas da gagueira. O subtipo de gagueira desenvolvimental persistente familial foi caracterizado principalmente por um distúrbio classificado quanto à severidade como leve. O risco dos familiares dos probandos afetados foi de 8,5%. A gagueira afetou mais pessoas do gênero masculino em relação ao feminino, numa proporção de 3,72:1.

DESCRITORES: Fonoaudiologia; Fala; Avaliação; Distúrbios da Fala, Gagueira; Genética

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