

Desempenho em consciência fonológica, memória operacional, leitura e escrita na dislexia familiar*****

Phonological awareness, working memory, reading and writing performances in familial dyslexia

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Abstract

Background: familial dyslexia. **Aim:** to characterize and compare the phonological awareness, working memory, reading and writing abilities of individuals whose family members are also affected. **Method:** in this study 10 familial nuclei of natural family relationship of individuals with dyslexia were analyzed. Families of natural individuals living in the west region of the state of São Paulo were selected. **Inclusion criteria** were: to be a native speaker of the Brazilian Portuguese language, to have 8 years of age or more, to present positive familial history for learning disabilities, that is, to present at least one relative with difficulties in learning. **Exclusion criteria** were: to present any neurological disorder genetically caused or not, in any of the family members, such as dystonia, extra pyramidal diseases, mental disorder, epilepsy, attention deficit and hyperactivity disorder (ADHA); psychiatric symptoms or conditions; or any other pertinent conditions that could cause errors in the diagnosis. As for the diagnosis of developmental dyslexia, information about the familial history of the adolescents and children was gathered with the parents, so that a detailed pedigree could be delineated. Neurological, psychological, speech-language, and school performance evaluations were made with the individuals and their families. **Results:** the results of this study suggest that the dyslexic individuals and their respective relatives, also with dyslexia, presented lower performances than the control group in terms of rapid automatic naming, reading, writing and phonological awareness. **Conclusion:** deficits in phonological awareness, working memory, reading and writing seem to have genetic susceptibility that possibly determine, when in interaction with the environment, the manifestation of dyslexia.

Key Words: Dyslexia; Learning; Reading.

Resumo

Tema: dislexia familiar. **Objetivo:** caracterizar o desempenho em consciência fonológica, memória operacional, leitura e escrita do probando com dislexia e de seus familiares afetados. **Método:** participaram deste estudo 10 núcleos familiares de parentesco natural de indivíduos com queixa específica de problemas de leitura e compreensão. Foram selecionadas famílias de probandos naturais e residentes na região do oeste do estado de São Paulo. Os requisitos de inclusão dos probandos foram: ser falante nativo do Português Brasileiro, ter idade acima de oito anos, apresentar histórico familiar positivo para os problemas de aprendizagem, ou seja, apresentar no mínimo um outro parente com dificuldade para aprender em três gerações. Os critérios de exclusão para o grupo de probandos foram: apresentar qualquer distúrbio neurológico-genético tais como distonia, doenças extras piramidais, deficiência mental, epilepsia, transtorno do déficit de atenção e hiperatividade (TDAH); sintomas ou condições psiquiátricas; ou outras condições pertinentes que poderiam gerar erros no diagnóstico. Para o diagnóstico de dislexia do desenvolvimento foram coletados dados de antecedente familiar na histórica clínica com os pais das crianças e adolescentes para realização do heredograma. Foram realizadas avaliações neurológica, fonoaudiológica, psicológica e de desempenho escolar nos probandos e em seus parentes. **Resultados:** os resultados deste estudo sugeriram que os probandos e seus familiares com dislexia apresentaram desempenho inferior ao grupo controle quanto à nomeação rápida, leitura, escrita e consciência fonológica. **Conclusão:** alterações em consciência fonológica, memória de trabalho, leitura e escrita tem susceptibilidade genética que possivelmente em interação com o meio ambiente determinam o quadro de dislexia.

Palavras-Chave: Dislexia; Aprendizagem; Leitura.

Introduction

Dyslexia is both familial and heritable. Family history is one of the most important risk factors, being that from 23% to as much as 65% of children who have a parent with dyslexia are reported to have the disorder as well^{1,2}. The rate among siblings of affected persons is approximately 40% and among parents it ranges from 27% to 49%.

Despite the strong genetic involvement, dyslexia does not commonly segregate in families in a simple mendelian fashion^{3,4}. Rather, ability to do reading related to cognitive tasks tends to decrease as a function of increasing genetic relatedness of relatives to the dyslexic probands. This finding suggests that several or many genetic factors determine reading ability, and that some or all of these factors might interact with one another to bring about particular influences on the reading ability.

Linkage analysis studies showed various locus chromosomes that can have candidate genes to dyslexia. This locus include the chromosomes 1p5, 2p6, 6p7, 15q8, and 18p9. Some genes have been associated to dyslexia such as: KIAA031910 and DCDC211 in chromosome 6p and EKN1 in chromosome 15q12.

Identification of these genetic variants may mean that a child's particular risk of developing certain types of reading problems could be estimated before severe problems develop.

Based on what was exposed above, this article will present a study made with families of children with developmental dyslexia which aims at:

- . Investigating familial prevalence and gender reason of learning disabilities among the families of individuals with dyslexia,
- . Characterizing phonological, working memory, reading and writing abilities of dyslexic individuals and their families who are also affected.

Method

This study has been previously approved by the Committee of Ethics in Research of the Science and Philosophy Faculty of the State University of São Paulo - UNESP/Marília - São Paulo - Brazil, under the protocol number 2231/2006.

In this study 10 familial nuclei of natural family relationship of individuals with dyslexia were analyzed. The requisites of inclusion of the dyslexics were: being a native speaker of Brazilian Portuguese, being 8 years old or more, presenting positive

familial history of learning disabilities, that is, presenting at least another relative with difficulties for learning in 3 generations. The criteria of exclusion for the group of dyslexics were: presenting any neurological disorder, genetically caused or not, in family members, such as dystonia, extra pyramidal diseases, mental retardation, epilepsy, attention-deficit/ hyperactivity disorder (ADHD); psychiatric symptoms or conditions; or other pertinent conditions which could cause errors in diagnostics.

The clinical history was made with the parents of the dyslexics with the purpose of verifying the presence of any problem, such as neurological, hearing, cognition and/or visual deficits and complaint of speech-language disorders.

Specific data about the natural history of dyslexia, time of beginning and specific disabilities related to reading and writing, was checked with the parents.

The data of the familial antecedents for the detailed pedigree were collected in the end of the clinical history. In order to obtain precise information, the relatives were encouraged to check the information referred to the reading problems with their own parents or other relatives of first degree. The information of the detailed pedigree, for this study, was used to determine the familial prevalence and the sexual ratio of dyslexia in the first degree relatives.

The dyslexics and their parents were evaluated in the Ambulatory of Child Neurology of the Clinical Hospital of State University of São Paulo - UNESP (UNESP-Botucatu - São Paulo - Brazil) and in the Centre of Study of Education and Health (CEES/ UNESP- Marília - São Paulo - Brazil). Only the relatives of the dyslexics that had complaints of reading comprehension difficulties were evaluated.

The subjects were considered dyslexic when presented the following criteria in interdisciplinary assessment: static balance; appendicular coordination; motor persistence, dynamic balance, torso-limb coordination and sensitivity alteration in the neurological evaluation, discrepancy between verbal intellectual quotient and performance intellectual quotient in the psychological assessment, memory, reading and writing alteration in the neuropsychological battery, phonemic, syllabic, rhyme and alliteration alteration in the phonological awareness test, reading level and reading speed lower to age and school level, phonological disorder in the phonological assessment, single word and non-word reading and writing, thematic essay, and partial comprehension

of text reading.

The interdisciplinary assessment of learning disabilities was considered when the subjects or their relatives presented, besides the criteria above, syntactic-semantic structure alteration in both oral language and the thematic essay, besides difficulties in arithmetic calculus, both in resolving single calculus and resolving calculus that depended on the reading comprehension of a text.

With the purpose of obtaining adequate reliability of the data in this study, data collection was realized with a control group. Thus, the groups in this study were distributed as:

. Group 1 (G1): formed by 10 dyslexic, between 8 and 15 years old, 8 (80%) males and 2 (20%) females, studying from 2nd to 6th grades.

. Group 2 (G2): formed by 10 students without learning difficulties paired according to gender, age and school level with the G1.

. Group 3 (G3): formed by 22 relatives of the dyslexic, aged from 8 to 70 years old, 6 (27,2%) males and 16 (72,8%) females, studying from 2nd to 5th grades.

. Group 4 (G4): formed by 22 subjects without learning difficulties paired according to gender, age and school level with the GIII.

The procedures of assessment utilized in this study, after the signing of a Term of Post-Informed Consent (according to resolution of the National Health Council - CNS 196/96), were the following:

. traditional neurological evaluation: this evaluation is composed of a neurological examination that evaluates: Perimeter and Cranial form, Conscious state, spontaneous, passive, automatic, reflex and involuntary movement, static and dynamic balance, coordination, sensitivity, cranial nerves and meningoradicular signs, in accordance to standards used in the Medical College of UNESP -Botucatu-São Paulo-Brazil. It was conducted by a child neurologist at the Clinical Hospital- HC/FM/UNESP-Botucatu-São Paulo - Brazil - to exclude neurological disorders.

. intelligence scale for children - WISC-III-R13: it was used in the subjects aged from 6 to 16 years old, aiming at quantitatively measuring their general cognitive level, through mental operations such as associations, deductions, types of reasoning, among others.

. adult intelligence scale -WAIS-R14: it was used in the subjects from 17 years of age and older, aiming at quantitatively measuring their general cognitive level, through mental operations such as

associations, deductions, types of reasoning, among others.

The use of the WISC and WAIS was conducted by psychologists at the Clinical Hospital- HC/FM/UNESP-Botucatu-São Paulo - Brazil:

. school performance test¹⁵: the subjects were submitted to the application of an assessment the fundamental capacities in school performance, such as writing, arithmetic and reading.

. rapid automatized naming - RAN¹⁶: the subjects were submitted to the Rapid Automatized Naming test, computer version, composed of subtests of color, digits, letters and objects naming. The subjects were told about that the time would be measured from the initial moment of the test.

. phonological awareness test¹⁷: the procedure used was the Phonological Awareness Test. The test consists of ten subtests, each one consisting of four items to verify synthesis, segmentation, manipulation and transposition syllabic and phonemic, rhyme and alliteration. The findings of the test are presented by score.

The results were analyzed statistically. We have adopted the significance level of 5% (*= 0,0050), for the application of the statistic test in this study. The SPSS program was used (Statistical Package for Social Sciences), in its 13.0 version, for obtaining the results. The statistically significant results are marked by an asterisk (*).

Results

Sexual prevalence and sexual ratio

In the results presented here about the familial prevalence and the sexual ratio for dyslexia, we considered all affected relatives (n=38) of the dyslexics, however only 22 were evaluated. The other 16 relatives were not evaluated for some reasons such as mental retardation, encephalic trauma, death or living far away from the data collection place.

The familial prevalence of dyslexia was investigated by counting the number of relatives with dyslexia for each subject. The prevalence of dyslexia in male relatives of all subjects was considered smaller (0,185) than in females relatives (0,312). The difference of the dyslexia prevalence among the two genders was significant (0,001) according to Table 1.

From the 22 relatives evaluated, 6 were males and 16 were females. Added to the dyslexics this

resulted in a total of 32 affected individuals (14 males and 18 females). The sexual prevalence of all affected individuals evaluated was 0,77 male individuals to 1,00 female. Among the relatives of dyslexics there were 12 individuals with dyslexia diagnosis and 10 with learning disabilities diagnosis. The data about the sexual ratio in the dyslexia showed a higher prevalence in males than in females (1,75).

Characterization of phonological, working memory, reading and writing findings

Following, we present the results of the cognitive and linguistic performance in the tests used in this study of the 10 familial nuclei.

When the Mann-Whitney Test and the Friedman Test were used for comparing the average score in the WISC in Group 1 and among the average score of WAIS in Group 3, significant statistic difference was observed in the performances of the dyslexics and performances of the relatives of dyslexics from VIQ and PIQ (table 2). In the same table it is possible to see the comparison between the performance of Group 1 and the Group 2 in the School Performance Test, using the T of Student Test controlled for Levene Test, statistically significant difference between the obtained score and expected score.

In table 3, significant statistic difference was verified in the performances of the dyslexics and the relatives of the dyslexics in the digit span, picture completion and coding subtests when compared to the performances of Group 1 and Group 3 with their respective control groups, Group 2 and Group 4.

The findings showed statistically significant difference in the subtests of syllabic synthesis and transposition and all the phonemic subtests when the T of Student Test controlled by Levene Test was used. By comparing the findings of Group 3 and Group 4, statistically significant difference was not verified only in the subtests of syllabic manipulation (SMan) when applied the T of Student Test controlled by Levene Test.

By comparing the performances of Group I and Group II in the Rapid Automatized Naming using the Friedman Test and T of Student Test controlled by Levene Test, significant statistic difference in the subtests of letters, number and objects naming was observed. By comparing the performance of Group 3 and Group 4, statistically significant difference was observed in all the subtests.

TABLE 1. Familial prevalence of developmental dyslexia in the relatives of dyslexics, represented in number of individuals with developmental dyslexia of both male and female genders, divided by the total number of male and female relatives and sexual ratio (M/F).

	Male Gender	Female gender	Total
<i>Relatives with the learning difficulties of male gender</i>	12/67 = 0,179	1/3 = 0,333	13/70 = 0,185
<i>Relatives with learning difficulties of female gender</i>	23/73 = 0,315	2/7 = 0,285	25/80 = 0,312
Ratio M/F	0,52	0,5	0,52

TABLE 2. Distribution of Average (A), Standard deviation (Sd) and value of p (P) to comparison of performances of the dyslexics (G1) and the dyslexic's relatives (G3) in relation to verbal, performance and total intelligence quotients of the WISC, WAIS and School Performance Test.

<i>Skills</i>		<i>G1</i>	<i>G3</i>
Verbal Intelligence Quotient (VIQ)	A	74,20	84,36
	Sd	13,51	6,37
Performance Intelligence Quotient (PIQ)	A	87,90	89,00
	Sd	19,59	11,23
	P	0,031*	0,012*
Verbal Intelligence Quotient (VIQ)	A	74,20	84,36
	Sd	13,51	6,37
Total Intelligence Quotient (TIQ)	A	81,20	86,36
	Sd	16,33	12,46
	P	0,109	0,278
Performance Intelligence Quotient (PIQ)	A	87,90	89,00
	Sd	19,59	11,23
Total Intelligence Quotient (TIQ)	A	81,20	86,36
	Sd	16,33	12,46
	P	0,072	0,053
Writing obtained score	A	12,50	22,73
	Sd	7,69	5,01
Writing - age expected score	A	30,70	32,45
	Sd	4,32	0,86
	P	< 0,001*	< 0,001*
Arithmetic obtained score	A	13,30	17,18
	Sd	4,11	5,48
Arithmetic - age expected score	A	22,40	24,41
	Sd	5,83	3,81
	P	0,001*	< 0,001*
Reading obtained score	A	28,80	32,64
	Sd	9,59	10,95
Reading - age expected score	A	68,40	68,73
	Sd	1,90	1,28
	P	< 0,001*	< 0,001*

TABLE 3. Distribution of Average, Standard deviation and value of p to the comparison of performance from G1, G2, G3 and G4 in digit span, picture completion and coding subtests of WISC and WAIS, syllabics, phonemics and supra-phonemics subtests of Phonological Awareness Test and color, letters, digit and objects subtests of RAN.

	Skills	Groups	Average	Standard Deviation	Value of P	
WISC/WAIS	Digit Span	1	7,50	0,71	< 0,001*	
		2	9,80	0,42		
		3	7,55	0,96	< 0,001*	
		4	9,55	0,51		
	Coding	1	7,40	0,97	< 0,001*	
		2	9,60	0,52		
		3	7,18	1,44	< 0,001*	
		4	9,59	0,50		
	Picture Completion	1	7,40	0,97	< 0,001*	
		2	9,60	0,52		
		3	7,36	1,33	< 0,001*	
		4	9,68	0,48		
Phonological Awareness	Syllabic Synthesis	1	1,00	1,05	0,008*	
		2	0,00	0,00		
		3	0,82	1,01		
		4	0,00	0,00	0,001*	
	Phonemic Synthesis	1	3,80	0,63	< 0,001*	
		2	1,20	0,79		
		3	3,91	0,43	< 0,001*	
		4	1,09	0,68		
	Syllabic Segmentation	1	0,40	0,84	0,168	
		2	0,00	0,00		
		3	0,55	0,91		
		4	0,00	0,00	0,011*	
	Phonemic Segmentation	1	4,00	0,00	< 0,001*	
		2	0,40	0,52		
		3	4,00	0,00	< 0,001*	
		4	0,50	0,51		
	Rhyme	1	0,40	0,84	0,168	
		2	0,00	0,00		
		3	0,45	0,86		
		4	0,00	0,00	0,021*	
	Alliteration	1	0,40	0,84	0,151	
		2	0,00	0,00		
		3	0,45	0,86		
		4	0,00	0,00	0,021*	
	Syllabic Manipulation	1	0,40	0,84	0,151	
		2	0,00	0,00		
		3	0,27	0,70		
		4	0,00	0,00	0,083	
	Phonemic Manipulation	1	3,80	0,63	0,000*	
		2	0,70	0,67		
		3	3,82	0,59	< 0,001*	
		4	0,82	0,73		
	Syllabic Transposition	1	2,00	0,00	< 0,001*	
		2	0,00	0,00		
		3	2,00	0,00	< 0,001*	
		4	0,00	0,00		
	Phonemic Transposition	1	3,80	0,63	< 0,001*	
		2	0,40	0,52		
		3	3,64	0,79	< 0,001*	
		4	0,41	0,50		
	Score	1	20,00	1,63	< 0,001*	
		2	2,70	1,06		
		3	19,91	1,69	< 0,001*	
		4	2,82	1,05		
	RAN	Collor	1	34,92	29,17	0,434
			2	42,50	2,84	
			3	26,65	28,75	0,049*
			4	39,55	4,23	
Letters		1	52,80	7,94	< 0,001*	
		2	28,50	2,42		
		3	51,09	7,07	< 0,001*	
		4	30,68	4,08		
Digit		1	45,60	5,40	< 0,001*	
		2	27,00	2,62		
		3	47,23	6,32	< 0,001*	
		4	28,32	4,34		
Objects		1	18,23	27,68	0,008*	
		2	47,90	2,08		
		3	8,85	20,13	< 0,001*	
		4	42,59	6,36		

Discussion

The present study indicates that from the 10 families who participated, 12 relatives of the subjects presented dyslexia, and 10 presented learning disabilities, corroborating previous findings¹⁰ regarding the evidence that in families that present one member with dyslexia, at least one more member also presents similar difficulties.

The sexual ratio found in this study for the dyslexic was 4:1, showing higher prevalence for males (M/F ratio = 0,52). Although this study was not conclusive because of the small sample, it points out important aspects to be considered such as: smaller prevalence of dyslexia among the males relatives of all dyslexic in comparison to females, and higher frequency of male subjects suggesting that dyslexia is a disorder that can be related to chromosome X. Segregation studies indicate that the disorder is usually inherited as autosomal dominant⁴. Recently, a study developed in the UK evidenced that single word reading disorder is related to Xq269 and Xq2718. A recessive allele of an X linked gene increasing the risk for dyslexia could explain why males are more commonly or more severely affected than females.

The findings of this study in relation to cognitive level corroborate previous findings^{19,20}, which were evidenced in studies with dyslexics with VIQ inferior to PIQ. This discrepancy has been noticed in the dyslexics as well as in their relatives, not only related with the verbal and execution quotient, but also between the quotients that indicate information processing speed and working memory, demonstrating that this would be a profile of a dyslexic person and their affected relatives.

Another characteristic of these students with specific reading disabilities noticed in this study was their difficulty in relation to arithmetic calculus.

According to the literature²¹ subjects with specific reading disabilities present disorders in information processing, and as this processing is based on cognitive and linguistic skills, the comprehension problems and arithmetic calculus, which need lexicon-mental correspondence and number representation, impair the performance of arithmetic.

There is a consensus that phonological awareness plays a critical role in reading acquisition. The phonological deficit hypothesis has been supported by a number of studies that have specifically identified delays in sensitivity to rhyme,

alliteration, and phonemic segmentation as precursors to the development of reading²⁴. In Brazil, these studies with dyslexia have specifically identified delays in alliteration, manipulation and phonemic segmentation¹⁹⁻²².

These finding about phonological deficit hypothesis have also been extended to adult populations. A recent study²⁵ described that phonological deficits have also been found in adults with dyslexia.

In this study, the dyslexics and their relatives with dyslexia presented lower performance than the control group in the RAN, and the same happened with the reading, writing, arithmetic and phonological tests, what indicates that there are difficulties in the phonological processing, recognition, and speed of information processing, in addition to attention difficulties, phonological skills, rapid naming and working memory^{26,27}.

The findings of this study showed the relationship between RAN, phonological processing, reading and writing, because the subjects with dyslexia and their parents presented naming speed, working memory and, phonological skills alteration, what was directly reflected in their inferior performance in reading, writing and arithmetic calculus.

In cases of dyslexia there are usually phonological deficits. These deficits are related to a difficulty in accessing and retaining phonological information, which is necessary to perform reading and writing tasks. However, we must consider that these deficits may have family origin, according to the literature^{21,23,24,28}, corroborating the etiological genetic of the phonological deficit hypothesis in these dyslexics and their relatives in this study.

Conclusion

The sexual ratio found in this study for the dyslexic showed higher prevalence for the males. There was smaller prevalence of dyslexia among the male relatives of all subjects in comparison to females, and high frequency of male subjects with dyslexia.

Subjects with dyslexia presented disorders in phonological accessing of information, temporal processing speed, working memory, phonological storage, what is directly related to poor associations and memorizing, resulting in alterations in writing and calculations.

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