OROFACIAL MYOFUNCTIONAL EVALUATION IN INDIVIDUALS WITH NEUROFIBROMATOSIS TYPE 1

Avaliação da motricidade orofacial em indivíduos com neurofibromatose tipo 1

Carla Menezes da Silva [1], Cristiane Aparecida dos Santos [1], Nilton Alves de Rezende [1]

ABSTRACT

Purpose: to describe and characterize alterations in orofacial individuals with neurofibromatosis type 1 and to correlate these changes with clinical manifestations of neurofibromatosis type 1 and the muscular strength. **Methods:** the study included 24 individuals with neurofibromatosis type 1, 12 males and 12 females, aged between 14 and 50 years, matched by sex and age, with volunteers not affected by the disease. All subjects were evaluated for maximal handgrip force ($F_{máx}$) and evaluated for orofacial motricity using the MBGR protocol. **Results:** the main findings of this study demonstrated that individuals with neurofibromatosis type 1 had higher prevalence of abnormal structures phonoarticulatory and significant differences ($p < 0.05$), when compared to the control group in relation to body posture and speech organs, mobility, muscle tone, pain on palpation and stomatognathic functions. The total score of the test also showed Spearman correlation coefficient significant at 5% over $F_{máx}$. **Conclusion:** patients with neurofibromatosis type 1 had a higher prevalence of phonoarticulatory changes with significant impacts on the functions of breathing, chewing, swallowing and speaking. These alterations were correlated with the reduction of systemic muscular strength.

KEYWORDS: Neurofibromatosis 1; Speech, Language and Hearing Sciences; Miofunctional Therapy; Muscle Strength.

INTRODUCTION

Neurofibromatosis (NF) is a group of diseases caused by genetic mutations classified as neurofibromatosis type 1, neurofibromatosis type 2, and schwannomatosis[1]. Neurofibromatosis type 1 (NF1) is one of the most frequent genetic disorders in humans[2]. NF1 is characterized by neurocutaneous lesions such as café-au-lait spots, axillary, inguinal, and perioral ephelides, cutaneous neurofibromas, Lisch nodules, and bone dysplasia[3]. In most cases, the clinical manifestations have been deemed discrete benign but with potential involvement of additional cognitive and behavioral changes that can interfere with learning and psychosocial development[3].

In addition to these anomalies, a preliminary study conducted at the Reference Center for Neurofibromatosis of Minas Gerais (CRNF-MG) observed phonoaudiological alterations in 30 to 40% of individuals with NF1, including dysphonia, articulatory imprecision, and hypernasality[4]. Other studies carried out with NF1 individuals of different age groups reported similar results with regard to communication impairments involving the voice and orofacial movement[5-9].

The clinical evaluation of orofacial movement (OM), or orofacial myofunctional evaluation, is fundamental to the process of phonoaudiological diagnosis in this area, because it gives clinicians an understanding of the anatomical and functional status of the stomatognathic system (SS). In addition, it aids in therapeutic planning and provides data on prognosis[10].

The CRNF-MG results stating that individuals with NF1 exhibit muscle hypotonia due to decreased overall muscle strength[11] led us to investigate the possible correlation between hypotonia and changes in orofacial mobility. The aim of this study was to

[1] Universidade Federal de Minas Gerais, MG, Brasil.

Conflict of interest: non-existent

describe and characterize orofacial movement changes in individuals with NF1 who were matched for sex and age with healthy controls, and to correlate the observed changes with clinical manifestations of NF1, in particular, with the reduction in overall muscle strength.

METHODS

The study was approved by the Research Ethics Committee of the Federal University of Minas Gerais (UFMG) (ETIC 0307.0.203.000-09) and all volunteers, individuals with NF1, and healthy individuals or guardians gave informed consent in writing. All adult subjects participating in the study as well as parents and/or guardians of adolescent subjects read and signed the informed consent form.

This was a case-control study with a non-probabilistic sample based on typicality. The sample consisted of 24 individuals with NF1 who were matched for sex and age with 24 individuals without the disease. The individuals with NF1, confirmed by clinical diagnosis, were recruited from the CRNF-MG, which operates in the Department of Dermatology of the Hospital das Clínicas of the UFMG—outpatient clinic professor: João Gontijo. The control subjects were selected from among volunteers at the research institution.

The inclusion criterion for the NF1 group was the presence of at least three of the following: 1) six or more café-au-lait spots with a diameter of 1.5 cm or greater after puberty, or 0.5 cm or greater before puberty; 2) two or more neurofibromas of any type or one or more plexiform neurofibromas; 3) ephelides in skin fold areas (especially in the axillary or inguinal areas); 4) optic glioma; 5) two or more Lisch nodules; 6) sphenoid dysplasia or dysplasia/thinning of the long-bone cortex; and/or 7) a first-degree relative with NF1.

Exclusion criteria included presence of neurofibromatosis type 2 or schwannomatosis, NF1 suspected but not confirmed, or plexiform neurofibromas of the oral cavity or neck.

The Orofacial Myofunctional Evaluation, MBGR Protocol[11], was performed by a single speech therapist who also recorded the time of the examination. The duration of the assessment was approximately 45 min. The clinical examination protocol included observation of body posture, extra and intraoral morphological analysis, and evaluation of orofacial mobility, tonus, and sensitivity, as well as assessment of breathing, chewing, swallowing, and speech functions. Chewing and swallowing of solids were evaluated using food (cream-cracker biscuit) and liquid (water).

All individuals with NF1 and volunteers without NF1 were evaluated with regard to maximum handgrip strength (S_{max}) via three standardized measurements of maximum strength, using a manual dynamometer (0-100 kg Kratos®, Brasil). The muscle cross-sectional area was calculated based on the forearm circumference (cm), and muscle strength per unit of area (S_{area}) was calculated (kg/cm^2). The length of the forearm (from the wrist fold to the cubital fossa) on each side and the circumference of the forearm two-thirds of the way from the wrist fold were measured. The skin fold was measured (mm) at the same point where the circumference was measured, on the flexor side, using a clinical compass. The cross-section area was calculated based on the forearm circumference (cm) minus the skin fold of the forearm.

The results of this study were statistically analyzed using the Statistical Package for the Social Sciences software (SPSS version 15.0). The Mann-Whitney test was used to compare quantitative variables between the NF1 group and the control group. Correlation analysis was performed using Spearman's correlation coefficient. The level of significance was set at 0.05 in all tests.

RESULTS

In this study, the data did not follow a normal distribution. Table 1 shows the results obtained in the evaluation of OM. The individuals without NF1 exhibited lower means in all parameters of the protocol, with the exception of pain on palpation; lower means indicate less impairment of the structures and functions of the SS.

Figure 1 shows the postural changes that were observed in the group of individuals with NF1; shoulder posture distortions were the most prevalent. There were no postural changes in the group of individuals without NF1. This parameter was significantly different between the groups.

Table 1 – Average scores on individual items of the MBGR protocol, in subjects with neurofibromatosis Type 1 and control group

MBGR variables	Average Score		p-Value
	NF1 (n = 24)	Control (n = 24)	
Body posture			
Head	0.62	0.0	0.0002*
Shoulders	1.0	0.04	0.0000*
Extraoral examination			
Frontal view	1.8	0.8	0.0001*
Frontal numerical view	1.5	0.2	0.0000*
Masseter	0.1	0.04	0.1607
Jaw	0.04	0.0	0.3173
Lips	0.7	0.1	0.1274
Lateral view	0.7	0.5	0.0159*
Intraoral examination			
Lips	0.0	0.0	-
Cheeks	1.6	1.3	0.2075
Tongue	2.3	0.2	0.0000*
Palate	1.3	0.2	0.0292*
Palatine tonsils	0.04	0.1	0.5390
Teeth and occlusion	3.5	0.8	0.0017*
Mobility			
Lips	3.8	0.2	0.0000*
Tongue	2.9	0.2	0.0000*
Cheeks	1.1	0.04	0.0042*
Soft palate	0,8	0.4	0.0824
Jaw	1.4	0.2	0.0000*
Tonus	3.5	0.08	0.0000*
Sensitivity			
Pain on palpation	0.0	0.0	-
Functions			
Breathing	1.0	0.3	0.0113*
Chewing	3.3	0.5	0.0000*
Swallowing 1st test	0.9	0.0	0.0048*
Swallowing 2nd test	0.8	0.0	0.0048*
Swallowing 3rd test	2.2	0.2	0.0000*
Speech 1st test	0.2	0.0	0.0768
Speech 2nd test	0.2	0.0	0.0768
Speech 3rd test	0.08	0.0	0.9762
Speech 4th test	3.5	0.2	0.0000*
Total score	42.1	7.2	0.0000*

(*) Mann-Whitney test.
Legend: NF1 = Neurofibromatosis Type 1.

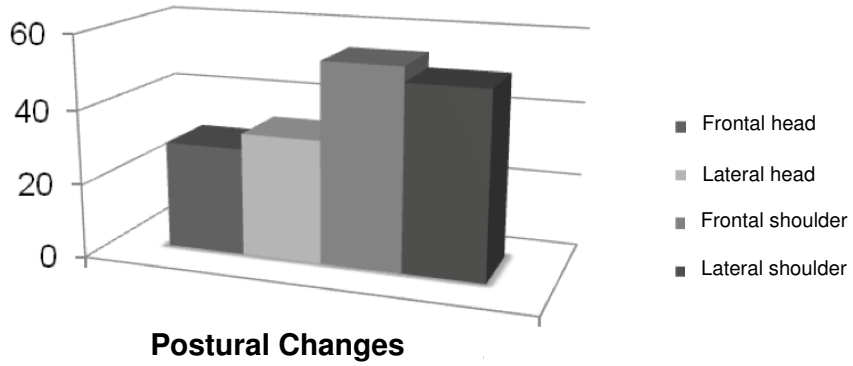


Figure 1 – Prevalence of postural changes in the neurofibromatosis Type 1 group

Table 2 shows that tonus change was more prevalent in individuals with NF1 than in those without NF1. Women with NF1 exhibited a higher prevalence of changes in tonus than did men with NF1.

Table 2 – Mean score on tonus item and prevalence of abnormal tonus, comparison between the neurofibromatosis Type 1 group and the control group, stratified for sex

Tonus	Women		Men	
	NF1	Control	NF1	Control
Mean	3.8	0.1	4.0	0.0
Adequate %	16.7	83.4	8.4	100
Abnormal %	83.3	16.6	91.6	0.0

Legend: NF1= Neurofibromatosis Type 1.

Figure 2 shows that in the NF1 group, the structures that exhibited highest prevalence of tonus reduction were the upper lip, the tongue, and the cheeks. Other structures such as the lower lip, chin, nasolabial fold, and mouth floor also exhibited reduced tonus, although with lower prevalence.

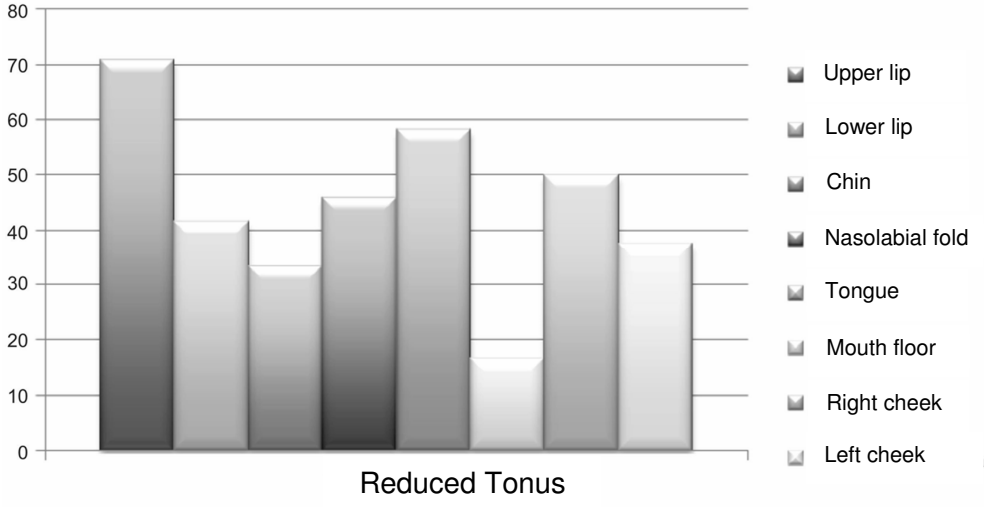
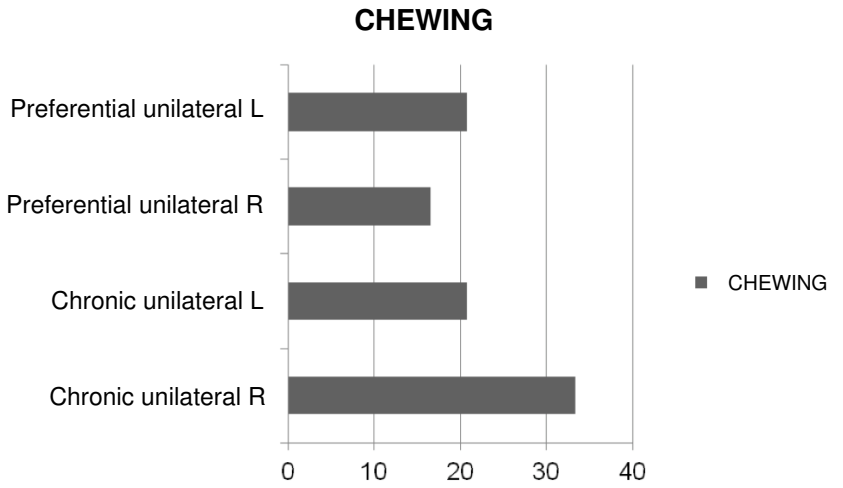


Figure 2 – Prevalence of tonus reduction in the neurofibromatosis Type 1 group in the various structures evaluated

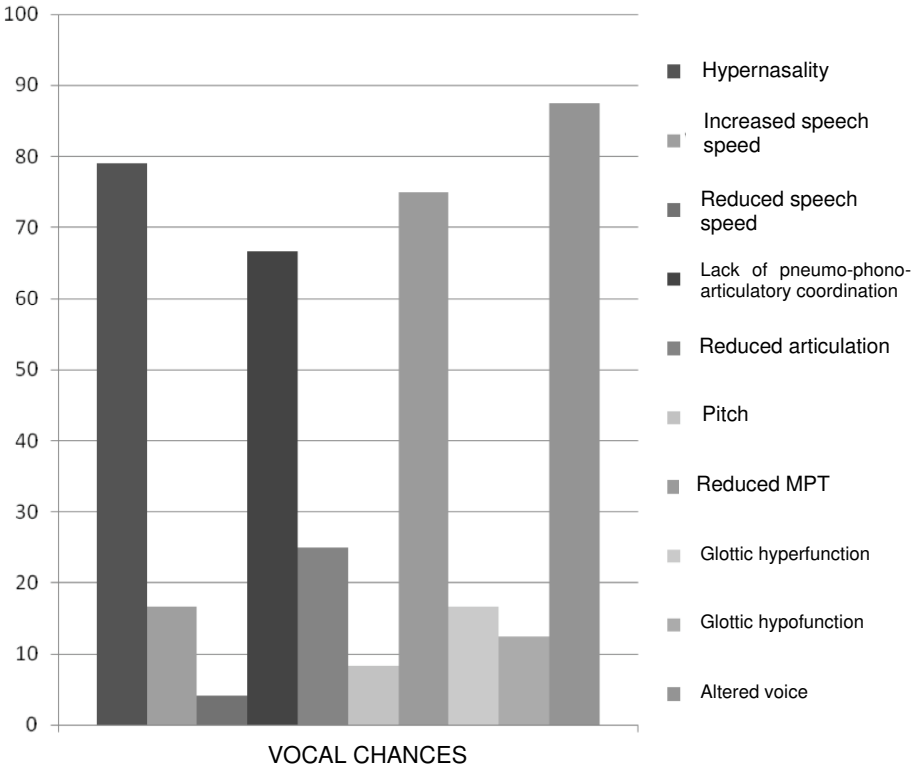
The chewing pattern of individuals in the NF1 group was altered to mainly chronic unilateral (right side). Other altered chewing patterns were preferential unilateral and chronic unilateral (left side) (Figure 3).

The speech of individuals with NF1 was characterized by imprecise articulation, hypernasality, and reduced intensity, as summarized in Figure 4. Other changes associated with voice, pitch, and a lack of pneumo-phono-articulatory coordination were also observed.



Legend: Preferential unilateral E = Preferential unilateral (left); Preferential unilateral D = Preferential unilateral (right); Chronic unilateral E = Chronic unilateral (left); Chronic unilateral D = Chronic unilateral (right)

Figure 3 – Prevalence of chewing patterns in the neurofibromatosis Type 1 group



Legend: MPT = maximum phonation time.

Figure 4 – Prevalence of vocal abnormalities in the neurofibromatosis Type 1 group

Changes in the structural and functional features of the SS were observed in the NF1 group. Structural changes included facial asymmetry, and functional changes comprised mobility disturbances, tongue tremor, and speech changes (Table 3).

As shown in Table 4, the prevalence of voice abnormalities and the maximum phonation time were higher in the NF1 group than in the control group.

Table 3 – Prevalence of structural and functional changes in the stomatognathic system in the neurofibromatosis Type 1 group and in the control group

Changes in the stomatognathic system	NF1		Control	
	N	%	N	%
Structural				
Facial asymmetry	4	16.6	0	0.0
Long facial type	23	95.8	15	62.5
Lower 1/3 larger than the median 1/3	6	41.6	4	16.6
Asymmetry between the cheeks	8	33.3	1	4.1
Masseters with different volumes	4	16.6	1	4.1
Tooth marks on the cheeks	20	83.3	15	62.5
Functional				
Lip mobility	21	87.5	6	41.6
Tongue mobility	23	95.8	7	29.1
Cheek mobility	8	33.3	0	0.0
Soft-palate mobility	10	41.6	4	16.6
Tongue tremor	17	70.8	1	4.1
Speech alteration	3	12.5	0	0.0
Articulatory imprecision	7	25	0	0.0

Legend: NF1 = Neurofibromatosis Type 1.

Table 4 – Vocal abnormality prevalences and mean maximum phonation times: comparison between the neurofibromatosis Type 1 group and the control group, stratified for sex (perceptive-auditory analysis item of the MBGR protocol)

	Women		Men	
	NF1	Control	NF1	Control
	%	%	%	%
Type of voice				
Adequate	25	91.7	0	83.4
Altered	75	8.3	100	16.6
Altered s/z ratio	41.6	0.0	33.3	0.0
Maximum phonation time				
/a/	11.2 s	15.4 s	12.3 s	22.7 s
/s/	10.8 s	16.0 s	10.5 s	19.2 s
/z/	10.2 s	15.5 s	8.5 s	21.1 s
Quantitative s/z ratio	0.9 s	0.9 s	1.2 s	0.9 s

Legend: NF1 = Neurofibromatosis Type 1.
Note: The results of the maximum phonation time are expressed in (s) seconds.

Table 5 shows that S_{max} was higher in healthy women than in women with NF1; however, no statistically significant difference was observed. A similar trend in S_{max} was observed in healthy and NF1 men, but the difference was significant (*p* value = 0.035).

Table 6 shows the Spearman's correlation coefficients between s/z ratio (a subtest of speech quality of the MBGR Protocol), and total score on the protocol, and hand grip S_{max}. The coefficients were significant despite their small absolute values.

Table 5 – Maximum handgrip strengths stratified for sex, in individuals with neurofibromatosis Type 1 and in control individuals, averaged across individuals

Sex	Maximum handgrip strength (N/cm²)		p-Value
	NF1	Controls	
Women	4.068	4.519	0.204
Men	4.837	6.789	0.043*
Groups	4.452	5.654	0.035*

(*) Mann-Whitney test.
Legend: NF1= Neurofibromatosis Type 1.

Table 6 – Spearman's correlation coefficient between maximum handgrip strength and total score in the MBGR protocol

		Spearman's correlation coefficient	p-Value
Muscle strength	s/z ratio	-0.290	0.045*
	Total score	-0.394	0.006*

(*) Spearman's correlation coefficient significant at 5%.

DISCUSSION

This study showed a higher prevalence of structural and functional changes in the SS of individuals with NF1 than in healthy individuals. These findings have not been reported previously and are important for the phonoaudiological diagnosis of the population under study.

Head and shoulder posture changes were not observed in the group of healthy volunteers, whereas 58.3% of individuals in the NF1 group exhibited these distortions. The most prominent characteristics in individuals with NF1 were upper torso posture abnormalities such as kyphosis, internal rotation, shoulder elevation, and asymmetry in static posture. These data suggest that muscle hypotonia and muscle strength reduction, previously described in patients with NF1[12] and confirmed in the present study, are generalized, affecting not only the major groups of skeletal muscles but also minor groups of muscles such as the head and neck muscles.

The respiratory system comprises a set of structures that are essential for effective breathing. On clinical examination, most individuals with NF1 exhibited middle-upper or thoracic respiration and nasal resonance. This type of breathing requires more involvement of the ribs and is often associated with shoulder elevation and shorter inhalations and exhalations. Costal-diaphragmatic or middle-lower respiration is the ideal type of breathing for speech because it produces a better flow of exhaled air, promoting good glottic coaptation necessary for adequate phonation. This type of breathing is directly associated with the activation of torso and abdominal muscles and is antagonized by important body changes seen in our NF1 group. These changes include deformed thoracic cage, flaccid and distended abdominal muscles, incorrect position of the head relative to the neck that causes changes in the spine to compensate for this bad posture, and forward rotation of the shoulders that causes compression of the thorax.

Reduction in respiratory muscle strength in individuals with NF1 has also been demonstrated in a recent study, although respiratory function as assessed by spirometry was found to be normal[12].

The results describe some typical structural and positional characteristics of the phono-articulatory organs (PAO) in this population, organs such as lips, tongue, teeth, cheeks, hard palate, and soft palate. The long facial type was predominant in both groups; however, in the NF1 group the lower third was longer than the middle third, whereas in the control group, the two thirds tended to be similar. Although the data were not based on a cephalometric assessment of individuals under study, in particular of the individuals with NF1, it is possible to infer that these changes are associated with hypotonia, especially hypotonia of the masseter muscle, one of the main muscles involved in jaw elevation. In individuals with shorter faces (i.e., predominantly horizontal growth), chewing was more vigorous. The individuals with longer faces were found to be less vigorous chewers and to have lower bite force[13].

Intraoral examination showed that 83% of individuals with NF1 exhibited tooth marks in both cheeks whereas in individuals without NF1 this percentage was 62.5%. Tooth marks on the cheeks may be associated with a lack of intraoral space and/or excessive tension applied to the dental arches. With regard to the tongue, 70.8% of individuals with NF1 had tongue tremor on protrusion and increased tongue width.

Reduced tonus of the tongue, lips, and cheeks was observed. For a less subjective clinical OM evaluation, the therapist must keep in mind that poor use of these structures and abnormal muscle form, function, and/or posture may indicate presence of abnormal muscle tonus, i.e., hypertonia (rigid muscle mass) or hypotonia (flaccid muscle mass)[14]. In addition, it is very important to distinguish postural tonus from action tonus.

The results of this study enabled us to characterize the functioning of the SS in the group of individuals with NF1, who exhibited poor coordination of the PAOs and difficulties in executing oral movements in response to a verbal order from the evaluator. This indicates that the subject lacks a model of the correct movement of the assessed structure, a problem that may be associated with auditory processing disorders that have been described in this population[15] or with oral dyspraxia.

The results showed a predominance of excessive nasal resonance in the group of individuals with NF1; however, adequate mobility of the soft palate with regard to symmetry and extension was observed in most individuals. Effective occlusion of the velopharyngeal sphincter depends not only on adequate mobility of the soft palate, but also on the medial approximation of the lateral walls of the pharynx; therefore, those individuals who did not exhibit abnormal soft palate mobility may instead have functionally compromised pharyngeal constrictor muscles[16,17].

The chewing pattern observed in the NF1 population was chronic unilateral, whereas the control group exhibited a preferential unilateral pattern. In the NF1 population, food oral transit was increased; this result was expected because the consistency of the food used in the evaluation did not allow for the formation of a cohesive mass during mastication.

No dependences on the consistency of foods (i.e., solid vs. liquid) were observed during deglutition. The liquid swallowing test performed on some individuals with NF1 showed accentuated contraction of the perioral muscles. This excessive contraction manifested as atypical deglutition. Atypical deglutition corresponds to inadequate movement of the tongue and/or other structures that are involved in the oral phase of deglutition, without changes in the form of the oral cavity. Because the individuals with NF1 who participated in this study did not exhibit any abnormalities in bone or dental form, the results are in line with the different deglutition patterns proposed in previous studies with different populations[14].

Only 12.5% of individuals in the NF1 group showed abnormalities in speech production, namely phonetic distortion, which had the same form in all such cases. These findings have been previously described in children and adults with NF1[5]. With regard to resonance, hypernasality was highly prevalent in this subgroup (79.1%), as was lack of pneumo-phono-articulatory coordination (66.6%). Some cases of articulatory imprecision (25%) were found. These changes were not observed in the control group. Reduction in muscle tonus can be associated with alterations in the SS, which directly influence speech production and quality, as well as with lack of coordination of muscle activation necessary for speech articulation.

Muscle hypotonia had been observed clinically in 32% of individuals with NF1 who were examined at the CRNF-MG; sample enlargement led to the observation that 67% of NF1 individuals exhibited lower muscle strength than healthy individuals matched for sex and age[12].

These findings led to complementary studies on muscle strength, including the use of an enlarged sample of individuals with NF1 of different age groups, and the evaluation of other muscle groups involved in stomatognathic functions, such as facial muscles.

We hypothesize that the finding of muscle weakness is related to the genetic cause of NF1, which directly results in a neurofibromin deficiency, thence motor coordination dysfunction (central origin

hypothesis of the reduction of S_{max} in NF1). Motor disorders of neurological origin are well described in NF1; muscle and skeletal changes observed in NF1 could be the result of neurological developmental impairments that lead to reduced muscle strength[12].

Furthermore, the sedentarism seen in individuals with NF1 is a potential contributor to reduced muscle strength. This behavior compromises the entire bone and muscle structure of the body, directly affecting posture and the functioning of orofacial structures.

Uncoordinated movements and inadequate posture can affect the normal movement of muscles and their performance.

In the phonoaudiological clinic, it is assumed that adequate performance of orofacial functions, including breathing, chewing, swallowing, and speech, is closely associated with a balanced relationship between static/passive structures and dynamic/active structures, the latter controlled by the central nervous system (CNS) and responsible for the harmonious functioning of the face[13].

Static or passive structures consist of the dental arches, maxilla, and jaw, which are connected by the temporomandibular joint. These structures also include other cranial bones and the hyoid bone. Dynamic or active structures consist of the neuromuscular units that mobilize the static parts. These interconnected structures form a system with specific features that perform shared functions[18]. The clinical studies cited here help increase and consolidate our knowledge of the muscular phenotype of NF1; however, the cause underlying the findings reported in them remain unknown. The etiology of reduction in muscle strength in individuals with NF1 has not been clarified and is probably multifactorial, either a result of neurofibromin deficiency directly affecting the skeletal muscles (muscular dysplasia), or a result of indirect action of various neurological changes (CNS dysplasia) that are potentially correlated with muscle phenotype[12]. Nevertheless, it is possible to infer that changes in overall muscle strength can affect facial muscles, leading to significant orofacial dysfunction in individuals with NF1.

CONCLUSION

This study demonstrates that SS functions are compromised in individuals with NF1, leading to interference with the quality and effectiveness of communication. However, aspects of the problem investigated in this study require complementary studies with a more homogeneous sample of individuals with NF1 and the use of adequate equipment to quantify the performance of muscles involved in breathing, chewing, swallowing, and speech. Such studies would support a phonoaudiological clinical intervention aimed at improving orofacial muscle strength and stomatognathic functions in patients with NF1 who exhibit abnormalities of orofacial movement.

RESUMO

Objetivo: descrever e caracterizar as alterações da motricidade orofacial de indivíduos com neurofibromatose tipo 1 (NF1) e correlacionar as alterações encontradas com manifestações clínicas da neurofibromatose tipo 1, mais especificamente com a redução da força muscular. **Métodos:** participaram deste estudo, 24 indivíduos com neurofibromatose tipo 1, sendo 12 do sexo masculino e 12 do sexo feminino, com idades entre 14 e 50 anos, pareados por sexo e idade, com indivíduos voluntários não acometidos pela doença. Todos os indivíduos foram avaliados quanto à força máxima de preensão manual ($F_{máx}$) e submetidos à avaliação da motricidade orofacial utilizando o protocolo MBGR. **Resultados:** os principais achados deste estudo demonstraram que os indivíduos com neurofibromatose tipo 1 apresentaram maior prevalência de alterações das estruturas fonoarticulatórias e diferenças significantes ($p < 0,05$) quando comparados ao grupo controle em relação à postura corporal e de órgãos fonoarticulatórios, mobilidade, tônus, dor à palpação e funções estomatognáticas. O escore total do teste também apresentou coeficiente de correlação de Spearman significantes com nível de significância menos que 5% em relação a $F_{máx}$. **Conclusão:** os indivíduos com NF1 apresentaram maior prevalência das alterações fonoarticulatórias com repercussões significantes sobre as funções de respiração, mastigação, deglutição e fala. Estas alterações estiveram relacionadas com a redução global da $F_{máx}$.

DESCRITORES: Neurofibromatose 1; Fonoaudiologia; Terapia Miofuncional; Força Muscular

REFERENCES

1. Ferner RE, Huson SM, Thomas N, Moss C, Willshaw H, Evans DG et al. Guidelines for the diagnosis and management of individuals with neurofibromatosis 1. J Med Genet. 2007;44(2):81-8.

2. Korf, BR, Rubestein AE. Neurofibromatosis: a handbook for patients, families, and health care professionals. 2nd ed. New York: Thieme; 2005.

3. Barker D, Wright E, Nguyen K, Cannon L, Fain P, Goldgar D et al. Gene for von Recklinghausen neurofibromatosis is in the pericentromeric region of chromossome 17. Sciene. 1987;236:1100-2.

4. Silva CM, Valentin HO, Rodrigues LOC, Rezende NA. High incidence of voice and motor control disturbances in NF1: a preliminary report. In: Models, mechanisms and therapeutic targets. NF Conference 2007: Proceedings of the Childrens Tumor Foundation Conference; 2007 Jun 10-12. Park City, USA; 2007. p. 52-3.

5. Cosyns M, Mortier G, Corthals P, Janssens S, Saharan N, Stevens E et al. Speech fluency in neurofibromatosis type 1. J Fluency Disord. 2010;35(1):59-69.

6. Cosyns M, Mortier G, Corthals P, Janssens S, Saharan N, Bogaert F et al. Articulation in schoolchildren and adults with neurofibromatosis type 1. J Commun Disord. 2012;45(2):111-20.

7. Cosyns M, Mortier G, Corthals P, Janssens S, Claes K, Borse JV. Objective assessment of nasality in Flemish adults with neurofibromatosis type 1. Am J Med Genet A. 2011;155A(12):2974-81.

8. Zhang I, Husein M, Dworschak-Stokan A, Jung J, Matic DB, Siu V et al. Neurofibromatosis and velopharyngeal insufficiency: is there an association? J Otolaryngol Head Neck Surg. 2012;41(1):58-64.

9. Cosyns M, Mortier G, Corthals P, Janssens S, Borsel J. Voice characteristics in adults with neurofibromatosis type 1. J Voice. 2011 Nov;25(6):759-64.

10. Genaro KF, Berretin-Felix G, Rehder MIBC, Marchesan IQ. Avaliação Miofuncional Orofacial – protocolo MBGR. Rev CEFAC. 2009;11(2):237-55.

11. Souza JF. A aptidão física está comprometida na neurofibromatose tipo1, mesmo nas formas de menor gravidade. [Dissertação]. Belo Horizonte (MG): Universidade Federal de Minas Gerais; 2013.

12. Souza JF, Passos RLF, Guedes ACM, Rezende NA, Rodrigues LOC. Muscular force is reduced in neurofibromatosis type 1. Rev Assoc Med Bras. 2009;55(4):394-9.

13. Bianchini EMG. Mastigação a ATM: avaliação e terapia. In: Marchesan IQ. Fundamentos em Fonoaudiologia: aspectos clínicos da motricidade oral. Rio de Janeiro: Guanabara Koogan; 1998. p. 37-49.

14. Marchesan IQ. Deglutição atípica ou adaptada: como considerar os problemas da deglutição? In: Junqueira P, Dauden ATB. Aspectos atuais em terapia fonoaudiológica. São Paulo: Pancast; 1996.

15. Batista PB, Silva CM, Valentim HO, Rodrigues LOC, Rezende NA. Avaliação do processamento auditivo na neurofibromatose tipo 1. Rev Soc Bras Fonoaudiol. 2010;15(4):604-8.

16. Penido FA, Noronha RMS, Caetano KI, Jesus MSV, Di Ninno CQMS, Britto ATBO. Correlação entre os achados do teste de emissão de ar nasal e da nasofaringoscopia em pacientes com fissura labiopalatina operada. Rev Soc Bras Fonoaudiol. 2007;12(2):126-34.

17. Mourão D, Souza GS, Torres LV, Vaz RN, Prado SG. Estudo sobre desenvolvimento fonológico em fissurados: implicações na fala e na linguagem. Estudos. 2006;33(5/6):425-41.

18. Douglas CR. Fisiologia da mastigação. In: Douglas, CR. Fisiologia Aplicada à Fonoaudiologia. São Paulo: Pancast; 2002. p. 325-50.

Received on: August 26, 2013
Accepted on: March 28, 2014

Mailing address:
Carla Menezes da Silva:
Rua Cordisburgo 90/401, Santa Inês
Belo Horizonte – MG – Brasil
CEP: 31080-060
E-mail: carlamenezesfono@gmail.com