Year : 2016
Volume : 3
Issue Number : 1
Doi Number : 10.5455/JNBS.1444218339

Article history:
Received 17 December 2015
Received in revised form 02 February 2016
Accepted 22 February 2016

SWALLOW OUTCOME IN THREE FEMALE SIBLINGS WITH HUNTINGTON'S DISEASE AND CHOREA

HUNTINGTON VE KORA HASTALIĞI OLAN ÜÇ KIZ KARDEŞTEKI YUTKUNMA SONUÇLARI

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Abstract

The present study focuses on describing characteristics of swallow among Huntingtons Disease (HD) with Chorea before and after dysphagia therapy. Three female siblings of 21, 22 and 33 years having juvenile type onset of HD with chorea were included. The patients were evaluated comprehensively for swallowing using Manipal Manual for Swallowing Assessment. Each patient was subjected to ingestion of solid, thin liquid and thick liquid of 5ml and 10 ml quantified using a standard measurable cup. Descriptive statistics was administered on the data using statistical package SPSS (Version 17). On observation, all three patients presented with sensory and motor issues in addition to posture instability with abrupt body movements, food spillage, piece meal deglutition, intra bolus retention, wet voice and cough. Following which cognitive approach and behavioural approach based intervention was initiated. The symptoms of intra bolus retention and cough decreased post therapy with no change in sensory aspects. The present study evidences three female siblings with severe cognitive deficits and dysphagia secondary to HD. Despite rehabilitation being provided, they could not completely waiver off the symptoms. These evidences highlight the importance of identifying and addressing swallow based treatment outcomes in HD with chorea.

Keywords: Huntingtons disease, chorea, dysphagia, behavioural therapy

Özet

Bu çalışma, yutma zorluğu terapisinden önce ve sonra Koralı Huntingon Hastalığı(HD) arasındaki yutkunma özelliklerine odaklanmıştır. Çalışma, Koralı HD'nın ergenlik döneminde başlayan çeşidine sahip olan 21, 22 ve 33 yaşlarındaki üç kız kardeşten oluşmaktadır. Hastalar, Manipal Yutkunma Becerileri Kılavuzu kullanılarak yutkunmaları için kapsamlı bir şekilde değerlendirilmişlerdir. Her hasta, standart bir ölçme kabı kullanılarak katı, ince sıvı, 5ml ve 10ml'lik kalın sıvıları yutmaya maruz bırakılmışlardır. İstatistik programı olan SPSS (sürüm 17) kullanılarak betimleyici istatistikler elde edilmiştir. Gözlem sırasında, üç hasta da ani vücut hareketleriyle duruş dengesizliği, yiyecek dökme, kısmi yutma foksiyonu, intra kapsül retansiyonu, nemli ses ve öksürmenin yanı sıra duyusal ve motorsal bulgular ortaya koymuştur. Bilişsel ve davranışsal yaklaşıma dayanan müdahalelerin takibi başlatılmıştır. Öksürük ve intra kapsül retansiyonunun semptomları duyusal açılardan hiçbir değişlik olmadan tedavi sonrasında azalmıştır. Bu çalışma, yoğun bilişsel eksiklikleri ve Huntingon Hastalığı (HD)'na bağlı yutma zorluğu olan üç kız kardeşi inceler. Rehabilitasyon desteği almalarına rağmen, bu semptomlardan tam olarak kurtulamamışlardır. Bu bulgular, koralı Huntingon Hastalığı(HD)'nın yutkunmaya dayalı tedavi sonuçlarının tanımlanmasının ve gösterilmesinin önemini vurgulamaktadır.

Anahtar Kelimeler: *Huntingon Hastalığı, Kora, yutma zorluğu, davranışsal terapi*

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1. Introduction

HD is an autosomal dominant progressive neuro-degenetraive disorder, typically with an adult onset. Repetition of mutant protein HTT on the short arm Chromosome 4 leads to cerebellar atrophy, especially at the level of caudate nucleus and putamen. This in turn leads to progressive motor, emotional and cognitive decline with choreiform body movements (Rusz et al., 2013). In view of these facts, life expectancy of such individuals is significantly affected, with poor mortality and morbidity, due to untreated dysphagia (Sorenson & Fenger, 1992).

Swallowing issues begin right from the oral preparatory stages manifesting as postural instability, poor quantity of food intake, tachyphagia, in-complete mastication and reduced lingual control ending in premature swallow (Leopold & Kagel, 1985; Hunt & Walker, 1989; Hamakawa et al., 2004; Kagel & Leopold, 1992; Mochizuki et al., 1999). Oral stage has absent voluntary swallow, short oral transition time of 0.23 ms with greater quantity of intra bolus retention despite multiple swallows. These act as a barrier and impede safe pharyngeal stage swallow leading to irregular hyo-laryngeal movements, frequent coughing due to aspiration indicated in a wet voice quality. Despite these well defined HD characteristics till date there is no clinically apparent red flag that serves as a bio marker for early identification and prevention of dysphagia (Heemskerk & Ross, 2011).

Even though literature reports 30 years as the typical age of onset, the symptoms may be exhibited during early childhood and continue to late adulthood (Bates et al., 2002). Moreover, previous findings reported were questioned for lack of patients and poor method (Heemskerk & Ross, 2011). This calls in for more evidence in the era of evidence based practice for better understanding of the concept being investigated. Therefore, we hereby put forth swallowing related clinical findings of three female siblings diagnosed of having HD with chorea before and after dysphagia therapy.

2. Subjects and Methods

Three female siblings with complaint of involuntary body movements admitted for medical treatment in a multidisciplinary teaching hospital participated. The patients P1, P2 and P3 were of 21, 22 and 33 years of age (mean age of 25.33 years) respectively with history of juvenile onset of HD with chorea symptoms. The clinical diagnosis of HD with chorea was given based on previously established criteria of choreatic body movements, impaired motor control, social, behavioural and cognitive changes associated with positive family history (Huntingtons Study Group, 1994; Harper, 1991; Tabrizi et al., 2012). Following which details pertaining to onset of the problem were ascertained by asking the first onset of chorea, recurrent mood swings, poor social behaviour, memory issues, sleep disturbance, frequent falls. Based on this, age of onset was 17, 15 and 17 years of age (mean age of 16.33 years) respectively. Family history revealed non-consanguineous marriage with paternal history of similar symptoms in three members, who were deceased for same.

Cognitive status of the three siblings was determined using Mini Mental State Examination before initiating any further investigation (Folstein, Folstein, & McHugh, 1974). Manipal Manual for Swallowing Assessment was administered to assess the swallow abilities in the subjects (Balasubramanium & Bhat, 2012). This manual is proposed to comprehensively assess structure, function, phases and tolerance of swallowing across four sub-scales for Indian population.

Following the diagnostic evaluation, therapeutic regime was initiated keeping two domains into account, cognitive approach and behavioural approach (Nance, 2012). As all three patients presented with severe form of cognitive deficits, orientation therapy was initiated with focus on presentation of orientation information, like time, place and person oriented. We also counselled the mother with the aim to cut down her anxiety levels and understand the practicality of the condition, so as to provide better support services. In the behavioural approach, positional change, hydrating oral cavity, mixing sour liquids, consistency changes with quantity reduction, chin tuck manoeuvre, verbal prompt of /a:/ followed by spoon positioning was performed routinely. These measures taken were earlier reported as evidence for swallow rehabilitation in literature (Nance, 2012; Aubeeluck & Mokowitz, 2008). A Speech Language Pathologist provided therapy three sessions/day of 30 minutes each for five continuous days.

3. Results

Results of the swallow investigation revealed series of undesired ramified swallow outcomes across all three patients. We observed severe sensory-motor deficits associated with cognitive communication dysfunctions. Postural disturbance were also evidenced in the subjects, with P1 presenting with supine position while P2 and P3 sitting upright with back support. Scores of Mini Mental Status exhibited severe form of cognitive dysfunction with P1, P2 and P3 obtaining nil score. As a result, we placed them under the category of 24 hour compulsory assistance for everyday functioning. Results of the swallowing investigation carried out are represented in Table 1.

In assessment of structure, the first sub parameter of sensory aspect, none of the patients were able to

Table 1: : Scores of swallow assessment obtained pre and post-therapy across P1, P2 and P3.

	Assessment of Function				Phases of Swallowing		Total	
	Sensory		Motor					
	Pre	Post	Pre	Post	Pre	Post	Pre	Post
P1	48	48	58	50	23	17	129	115
P2	48	48	63	63	29	26	140	137
P3	48	48	57	51	14	10	119	109

identify light vs. deep pressure when stimulated with the tongue depressor. Moreover, only awareness of the stimuli was observed behaviourally. Apparently in the second sub parameter of assessment of structure, the motor aspect, the three patients presented with persisting open mouth, drooling, lingual chorea, decreased tongue range, strength and absent voluntary cough. It was also observed that P3 presented with posterior tongue position at rest, while P1 and P2 had neutral tongue placement during rest. Results of phases of swallow provided evidences of oral and pharyngeal stage dysphagia. Series of intra bolus retention symptoms were noted at the level of lateral buccal cavity and tongue blade across the three subjects. In addition to these, poor lip seal, delayed onset of voluntary swallow, prolonged hyo-laryngeal elevation, lingual chorea with piece meal deglutition and aspiration cough was noted for thin and thick liquids. Comparatively, better intake of solids was observed with decreased symptoms of multiple swallows, intra bolus retention and cough. Overall, bolus preparation time was increased for solids. No nasal regurgitation was observed. Lastly, subjects were tolerant to textures across oral and pharyngeal phase.

Issues pertaining to identification of sensory stimuli did not change even after post-therapy. All three patients persisted with just awareness of sensory stimuli to be present. After three sessions of therapy, we observed decreased drooling and better tongue range in P1 and P3 subjects. No clinical differences were notable in P2 subject. On the continuum, we observed better swallow performance in terms of reduced quantity of intra bolus retention for thick liquids. This however did not change for thin liquids post-therapy and the patient presented with intra bolus retention and aspiration cough. No changes were noted in cognitive aspect post-therapy. In general, post-therapy swallow investigation revealed that only the degree of the oral and pharyngeal dysphagia could be reduced with more sessions of safe swallow for nutrition intake.

4. Discussion

The present study was undertaken to profile swallow skills in individuals diagnosed of having HD with chorea. On comprehensive swallow evaluation, in all three female siblings we documented oral and pharyngeal stage of swallow problems. We attribute progressive sensory-motor degeneration in the cerebellum, reported to be more severe in juvenile onset type of HD, as the root cause for presentation of these symptoms. These neurological deficits incapacitate the ability to control respiratory and bucco-lingual muscles compulsorily required for safe swallow (Hamakawa et al., 2004). The findings of the present study are similar to the reports evidenced in literature which characterises sensory-motor deficits with oro-pharyngeal dysphagia (Leopold & Kagel, 1985; Hunt & Walker, 1989; Hamakawa et al., 2004; Kagel & Leopold, 1992).

Co-ordination between swallowing and breathing is an essential element in protecting the airway tract during swallowing. Absence of this bio-mechanical physiology hinders co-ordinated initiation and propulsion of bolus consequently leading to series of premature swallow events, thereby causing progressive dysphagia (Sorenson & Fenger, 1992). Adverse affects of dysphagia are the common cause for individuals with HD to have aspiration pneumonia, respiratory issues and malnourishment that ultimately leads to cessation of life (Sorenson & Fenger, 1992).

Motor disturbances, such as chorea, are also accounted for impeding safe swallow in our study. Presence of choreatic or dance like movement hinders normal rhythmic, repetitive sequences of single motor movements (Willingham & Koroshetz, 1993). Evidences of lingual chorea in our patients must have dictated in-coordinated swallow, piece meal deglutition and intra bolus retention characteristics.

Functioning of tongue by means of rotation, thrust movements, bolus positioning and touch to palate are crucial aspects of normal swallow. Hence, impairment in the buccal-lingual muscles can affect individual's ability to pool out bolus trapped in the buccal cavity, specifically lateral sulci. Further on, complicating these motor movements is the lack of maintenance of posture (Reilman et al., 2012). In our all three patients we observed posture instability, one among various reasons that facilitates poor bolus preparation and delayed lingual movements in oral phase subsequently terminating as a pre-mature swallow.

In addition to these, sensory deficits are also a contributing factor. We attribute lack of sensory awareness as the possible reasons in our patients who presented with poor bolus preparation, bolus positioning and failure in pooling out intra-bolus residue. As per the swallowing manual, sensory deficits were assessed with two types of differential pressure being applied and the patient responding. Now due to poor cognition and lack of language skill we cannot accurately pin point degree of sensory deficits. Supportive of declining cognitive profile, evidences of language impairment have also been reported in HD patients (Azambuja et al., 2012).

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Strong evidences have been documented in literature which roots that brainstem regulates the central pattern generator for swallowing (Jean, 1990). Despite these viewpoints, several researchers have reported activation of cortical regions during voluntary swallow, in which few are suggestive of bilateral representations and some unilateral (Robbins et al., 1993; Smithard et al., 1997). Hence, intact cortical functions i.e., cognition is one of the basic pre-requisites for performing daily activities. This happens to be affected in HD with chorea, both initial and progressive stages (Cleret de Langavant, 2013). Studies draw focus upon impaired attention, short term memory deficits and executive function issues indicating a degenerative cognitive profile (Naarding, Kremer, & Zitman, 2001). Although we did not administer a detailed cognitive test battery, results of Mini Mental Status Examination helped us arrive at the conclusion of severe cognitive impairment in our patients. Literature reports Mini Mental Status Examination to be superior in sensing cognitive decline than other test battery like Montreal Cognitive Assessment thereby justifying adoption of present method (Gluhm et al., 2013)

Studies have suggested that manifestation of psychiatry issue in addition mood disorders hinders prognosis, while adding burden on the parents (Rickards et al., 2011). In all our patients, presence of suicidal tendencies, lack of sleep and depression was reported. These symptoms highlight what Paulson and colleagues report of stage II type of HD, which characterizes loss of independent functioning (Paulson et al., 2005). Severe cognitive impairment associated with stage II type of HD puts such individuals at greater risk of poor social life, dignity, safety, nutrition, bowel movements and functional competence. Presentation of these complex, multifaceted symptoms makes it very difficult for the family in providing care, as noted in the present study.

Having known the fatal consequence of HD with chorea it is alarming that very few clinical setups have focussed upon the principle 'there is never anything we can do for HD' (Nance, 2012). Despite these remarks there have been few initiatives, like palliative and hospice care, that addresses challenging deficits observed in later stages of HD (Dellefield & Ferroni, 2011). We state that, from the current case series observed, availing palliative and hospice care could provide them better quality of life in addition to receiving rehabilitation services. Apparently, initiating palliative and hospice care in terminal stages of progressive neurologic disease is mostly in developed countries, and in India it is still in primitive stages and lacks awareness for initiating implementation. This calls in for a more constructive approach in setting up institutes that primarily addresses and delivers ideologies of palliative and hospice care.

5. Conclusion

The present study was undertaken to profile swallow skills before and after intervention in individuals diagnosed of having HD with chorea. Results of swallow investigations revealed sensory-motor issues with oro-paryngeal dysphagia and cognitive communicative deficits. Post-therapy we observed lesser degree of dysphagia characteristics but with no improvement in cognitive skills. Presentation of these clinical case scenarios cues for accommodating compensatory strategies. Prognosis of HD in later stages is not so bright. However, therapeutic services must focus to facilitate quality of life by adopting palliative and hospice care based rehabilitation. Lastly, the present study calls in for more clinical evidence in early identification and rehabilitation of HD with chorea.

References

Aubeeluck, A., & Moskowitz, C. B. (2008). Huntington's disease Part 3: Family aspects. Brazil Journal of Nursing, 17(5), 328-331.

Balasubramanium, R. K., & Bhat, J. S. (2012). Manipal manual for swallowing assessment. Manipal: Manipal University Press.

Bates, G., Harper, P. S., & Jones, L. (2002). Huntington's disease. (3rd ed.). New York: Oxford University Press.

Dellefield, M. E., & Ferrini, R. (2011). Promoting excellence in end-of-life care: Lessons learned from a cohort of nursing home residents with advanced Huntington disease. Journal of Neuroscience Nursing, 43(4), 186-192.

Folstein, M. F., Folstein, S. E., & McHugh, P. R. (1975). Mini-mental state: A practical method for grading the cognitive state of patients for the clinician.Journal Psychiatry Research, 12(3), 189-198.

Hamakawa, S., Koda, C., Umeno, H., Yoshida, Y., Nakashima, T., Asaoka, K., & Shoji, H. (2004). Oropharyngeal dysphagia in a case of Huntington's disease. Auris Nasus Larynx, 31(2), 171-176.

Harper, P. S. (1991). Huntington's Disease. W.B. Saunders: London.

Heemskerk, A. W, & Roos, R. A. (2011). Dysphagia in Huntington's disease: A review. Dysphagia, 26(11), 62-66.

Hunt, V. P., & Walker, P. O. (1989). Dysphagia in Huntington's disease. Journal of Neuroscience Nursing, 21(2), 92-95

Huntington Study Group. (1996). Unified Huntington's disease rating scale: Reliability and consistency. Journal of Movement Disorder, 11(2), 136-142.

Kagel, M. C., & Leopold, N. A. (1992). Dysphagia in Huntington's disease: A 16-year retrospective. Dysphagia, 7(2), 106-114.

Leopold, N. A., & Kagel, M. C. (1985). Dysphagia in Huntingtons disease. Archives of Neurology, 42(1), 57-60.

Mochizuki, H., Kamakura, K., Kumada, M., Goto, J., Kanazawa, I., & Motoyoshi, K. (1999). A patient with Huntington's disease presenting with laryngeal chorea. European Neurology, 41(2), 119-120.

Nance, M. A. (2012). Therapy in Huntington's Disease: Where Are We?. Current Opinion in Neurology and Neuroscience Reports, 12(4), 359-366.

Paulsen, J. S., Nehl, C., Hoth, K. F., Kanz, J. E., Benjamin, M., Conybeare, R., McDowell, B., & Turner, B. (2005). Depression and Stages of Huntington's Disease. Journal of Neuropsychiatry and Clinical Neurosciences, 17(4), 496-502.

Rickards, H., De Souza, J., van Walsem, M., van Duijn, E.,

Simpson, S. A., Squitieri, F., & Landwehrmeyer, B. (2011). Factor analysis of behavioural symptoms in Huntington's disease. Journal of Neurology, Neurosurgery and Psychiatry, 82(4), 411-412.

Rusz, J., Klempir, J., Baborova, E., Tykalova, T., Majerova, V., Cmejla, R., Ruzicka, E., & Roth, J. (2013). Objective acoustic quantification of phonatory dysfunction in huntington's disease. PLoS One, 8(6), 1-7.

Sorensen, S. A., & Fenger, K. (1992). Causes of death in patients with Huntington's disease and in unaffected first degree relatives. Journal of Medical Genetics, 29(12), 911-914.

Tabrizi, S. J., Reilmann, R., Roos, R. A., Durr, A., Leavitt, B., Owen, G., et al. (2012). Potential endpoints for clinical trials in premanifest and early Huntington's disease in the TRACK-HD study: Analysis of 24-month observational data. Lancet Neurology, 11(1), 42-53.

Willingham, D. B., & Koroshetz, W. J. (1993). Evidence for dissociable motor skills in Huntington's disease patients. Psychobiology, 21, 173-182.

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