Doença de Huntington

Musculatura oral

- Hipotonia e movimentos involuntários variam
- Não consegue manter língua fora da boca mais que uns segundos

FALA

Disartria na Doença de Huntington

- **Hipernasalidade**
- **Aspereza e soprosidade**
- **Dificuldade em controlar variação de intensidade**
- **Intervalos prolongados entre palavras**
- **Silêncios inapropriados**
- **Velocidade do discurso variável**
- **Inspiração ou expiração forçada em 20% dos indivíduos com coreia**
- **Alterações prosódia** (monointensidade, monofrequência, frases curtas, velocidade de fala diminuída, intervalos e silêncios inapropriados, prolongamento de fonemas, pausas articulatórias irregulares)

Características fala

- Articulação imprecisa
- Distorção vogais
- Intervalos prolongados
- Silêncios inapropriados - velocidade inapropriada
- Frases curtas
- Variações pitch, monopitch
- Qualidade vocal rouca e tensa-estrangulada
- Excesso na variação de loudness e redução ênfase
- Hipernasalidade
- Inspirações expiração súbitas

- PROSÓDIA - mais afetada

Disartria na Doença de Huntington

Intervenção nas alterações de fala na Doença de Huntington

Severidade da Disartria	Descrição	Intervenção
Ligeira	Alterações na fala são evidentes Movimentos coreicos sobrepõem-se aos movimentos de fala Inteligibilidade pouco afetada	Tarefas prosódicas Manutenção velocidade adequada Técnicas que reduzam quebras fonatórias
Moderada	Movimentos coreicos interferem com a inteligibilidade de fala	Técnicas comportamentais que promovam inteligibilidade da fala Sistemas aumentativos de comunicação
Severa	Fala não é funcional	Sistemas aumentativos de comunicação – sistema sim/não; calendários e ajudas de memória; quadros de alfabeto; uso de objetos reais/ miniaturas.

"…As the disease progresses the **effectiveness of communication** becomes increasingly compromised by a **combination of changes in motor function, diminishing cognitive linguistic abilities and neuropsychiatric changes**, such as depression and apathy…"

"…The European Huntington's Disease Network Standards of Care Speech and Language Therapy Working Group has brought together expert speech and language therapists from across Europe to **produce guidelines to improve the management of communication disorders for individuals with HD**. "

SPECIAL REPORT

For reprint orders, please contact: reprints@futuremedicine.com

Management of speech, language and communication difficulties in Huntington's disease

Alison Hamilton[1], Ulrika Ferm[2], Anne-Wil Heemskerk[3,4], Rita Twiston-Davies[5], Kirsty Y Matheson[6], Shiela A Simpson[6,7] & Daniela Rae[*6,7]; On behalf of the contributing members of the European Huntington's Disease Networks Standards of Care Speech and Language Therapist Group

Practice Points

- Speech and language therapy has an important role to play in the management of communication problems in Huntington's disease.
- As the disease progresses the effectiveness of communication becomes increasingly compromised by a combination of changes in motor function, diminishing cognitive linguistic skills and neuropsychiatric changes, such as depression and apathy.
- Signs and symptoms associated with Huntington's disease are distinctive but there is considerable variation between individuals on the extent, rate and natural course of disability within the disease. For these reasons assessment and review must be comprehensive and consideration should be given to a number of factors such as mood, motivation and behavior, which will be pertinent to performance.
- As a variety of symptoms can affect communication no single course of treatment will be effective throughout the disease. The consensus of opinion is that the therapy management will vary and that

"…SLP management of individuals with hyperkinetic dysarthria includes behavioral and compensatory strategies for addressing compromised speech and intelligibility…"

OPEN ACCESS Freely available online

Tremor and Other Hyperkinetic Movements

Reviews

Speech–Language Pathology Evaluation and Management of Hyperkinetic Disorders Affecting Speech and Swallowing Function

Julie M. Barkmeier-Kraemer[1*] & Heather M. Clark[2]

[1]Division of Otolaryngology, University of Utah, Salt Lake City, UT, USA, [2]Department of Neurology, Mayo Clinic, Rochester, MN, USA

Abstract

Background: Hyperkinetic dysarthria is characterized by abnormal involuntary movements affecting respiratory, phonatory, and articulatory structures impacting speech and deglutition. Speech–language pathologists (SLPs) play an important role in the evaluation and management of dysarthria and dysphagia. This review describes the standard clinical evaluation and treatment approaches by SLPs for addressing impaired speech and deglutition in specific hyperkinetic dysarthria populations.

Methods: A literature review was conducted using the data sources of PubMed, Cochrane Library, and Google Scholar. Search terms included 1) hyperkinetic dysarthria, essential voice tremor, voice tremor, vocal tremor, spasmodic dysphonia, spastic dysphonia, oromandibular dystonia, Meige syndrome, orofacial, cervical dystonia, dystonia, dyskinesia, chorea, Huntington's Disease, myoclonus; and evaluation/treatment terms: 2) Speech–Language Pathology, Speech Pathology, Evaluation, Assessment, Dysphagia, Swallowing, Treatment, Management, and diagnosis.

Results: The standard SLP clinical speech and swallowing evaluation of chorea/Huntington's disease, myoclonus, focal and segmental dystonia, and essential vocal tremor typically includes 1) case history; 2) examination of the tone, symmetry, and sensorimotor function of the speech structures during non-speech, speech and swallowing relevant activities (i.e., cranial nerve assessment); 3) evaluation of speech characteristics; and 4) patient self-report of the impact of their disorder on activities of daily living. SLP management of individuals with hyperkinetic dysarthria includes behavioral and compensatory strategies for addressing compromised speech and intelligibility. Swallowing disorders are managed based on individual symptoms and the underlying pathophysiology determined during evaluation.

Discussion: SLPs play an important role in contributing to the differential diagnosis and management of impaired speech and deglutition associated with hyperkinetic disorders.

Keywords: Hyperkinetic dysarthria, dystonia, dyskinesia, chorea, myoclonus, essential vocal tremor

Citation: Barkmeier-Kraemer JM, Clark HM. Speech–language pathology evaluation and management of hyperkinetic disorders affecting speech and swallowing function. Tremor Other Hyperkinet Mov. 2017; 7. doi: 10.7916/D8Z32B30

DEGLUTIÇÃO

Prevalência

- 100% dos doentes em algum momento da doença
- Envolvimento coreico do mecanismo respiratório em 40%
- Aspiração e aerofagia em 10%
- Erutação em 40%

Patofisiologia disfagia

- Hipercinéticos, hipocinéticos ou ambos
- Descoordenação podem influenciar de forma primária ou secundária as competências de comer e dos movimentos da deglutição
- Perturbações emocionais, como apatia e outras alterações cognitivas, como impulsividade
  - Dificuldades interpretar sinais, avaliação e tratamento da deglutição

Diagnóstico disfagia

- Taquifagia e erutação são os sinais que podem não ser observadas na avaliação instrumental → fisiologia da deglutição

- Avaliação instrumental
  - Paciência e posicionamento especial
  - Auto-alimentação - avaliar diferença face à alimentação por terceiros

- DESAFIO
  - Identificar foco da alteração
    - Respiratório
    - Oral
    - Faríngeo

Complicações disfagia

- Pneumonia de aspiração
- Desidratação
- Desnutrição
- Perda peso

- Medicação para corea → DETEORA DEGLUTIÇÃO

- Taquifagia, erutação e movimentos anormais
  - Isolamento social
  - Consequências psicosociais

- Incidência de obstrução da via aérea fatal → poderá ser elevada

Disfagia na Doença de Huntington

Caraterísticas	Descrição	Frequência	Possíveis mecanismos
Taquifagia	Deglutição rápida e descontrolada	Muito comum	Coreia bucolingual e demência afetam os estadios iniciais da deglutição resultando na transferência impulsiva e prematura dos alimentos.
Coreia respiratória	Movimentos involuntários no sistema respiratório	40% dos indivíduos	O descontrolo do ciclo inspiração-expiração provocado pela coreia aumento o risco de aspiração.
Erutação	Arrotos excessivos	Aproximada mente 40% dos pacientes hipercinéticos	Possivelmente relacionado com a presença de coreia na contração dos músculos expiratórios, deslocado pequenas quantidades de ar gástrico.

Disfagia na Doença de Huntington

Caraterísticas	Descrição	Frequência	Possíveis mecanismos
Aerofagia	Deglutição de ar	10% dos pacientes com DH com hipercinesia.	Coreia lingual e deglutições repetidas.
Aspiração	Penetração glótica de alimentos e líquidos	Menos de 10% em pacientes hipercinéticos; Mais frequente e com aspiração de maiores volumes no grupo com rigidez e bradicinesia.	Coreia laríngea pode provocar o rápido e forçoso encerramento das bandas ventriculares e das pregas vocais, promovendo penetração mas impedindo a aspiração. Deglutições múltiplas diminuem os resíduos acumulados.

"… Clinical assessment of the HD-h (hyperkinetic) cohort (30 patients) demonstrated rapid lingual chorea, swallow incoordination, repetitive swallows, prolonged laryngeal elevation, inability to stop respiration, and frequent eructations…"

Dysphagia in Huntington's Disease: A 16-Year Retrospective

Marion C. Kagel, M.A.[1] and Norman A. Leopold, D.O.[2]
[1]Department of Medicine, Divisions of Speech/Language Pathology, Crozer-Chester Medical Center, Upland, and
[2]Department of Neurology, Hahnemann University School of Medicine, Philadelphia, Pennsylvania, USA

Abstract. Degenerative diseases of the basal ganglia are commonly complicated by dysphagia. In 35 patients with Huntington's disease (HD), a hereditary neurodegenerative basal ganglia disease characterized by chorea, dementia, and emotional changes, an extensive battery of clinical and radiologic procedures helped to identify numerous abnormalities of deglutition. The results permitted the classification of our patients with HD into hyperkinetic (HD-h) or rigid–bradykinetic (HD-rb) groups. Although the two groups share multiple abnormalities, statistically significant intergroup differences were observed. Clinical assessment of the HD-h cohort (30 patients) demonstrated rapid lingual chorea, swallow incoordination, repetitive swallows, prolonged laryngeal elevation, inability to stop respiration, and frequent eructations. In the HD-rb group (five patients), frequently observed abnormalities included mandibular rigidity, slow lingual chorea, coughing on foods, and choking on liquids. Videofluoroscopic swallowing studies (VFSS) using a variety of barium-impregnated foods and liquids confirmed the abnormalities noted on the clinical assessment. Respiratory and laryngeal chorea, pharyngeal space retention, and aspiration were also identified. Numerous compensatory techniques introduced during videofluoroscopy benefited all patients.

Key words: Huntington's disease — Chorea — Rigidity — Respiration — Aspiration — Deglutition — Deglutition disorders.

Address offprint requests to: Dr. N.A. Leopold, Crozer-Chester Medical Center, POB #2, Suite 222, Chester, PA 19013, USA

Huntington's disease (HD) is a hereditary neurodegenerative disease of the central nervous system (CNS). Characterized by involuntary movements, dementia, and emotional impairment, HD appears most frequently during the fourth or fifth decades of life [1]. However, its phenotypy is variable, with a small percentage of patients presenting with parkinsonian features including bradykinesia or rigidity [2]. Extrapyramidal features change as HD progresses; many patients become more choreic and hypotonic, while others experience less chorea as muscle tone increases [3]. Chorea and varying degrees of bradykinesia and rigidity often coexist [4]. In some patients, parkinsonian signs are induced by phenothiazines administered for the treatment of chorea [4, 5]. Disease progression is slow and inexorable. Motor control of nearly all voluntary muscles is impaired, including those governing respiration, speech, and deglutition. Death usually occurs within 20 years, often from aspiration pneumonia [6].

Although multiple regions of the brain are affected in patients with HD, caudate and putaminal atrophy dominate the neuropathologic findings [1]. The result is a severe fall in CNS gamma-aminobutyric acid (GABA) levels [7]. Unfortunately, therapeutic strategies that elevate CNS GABA levels provide no substantive benefit. The current treatment is symptomatic and includes neuroleptics to reduce chorea and/or control schizophrenic-like behavior, anxiolytics, and antidepressants [5].

Dysphagia is a common, if not inevitable, complication of HD that usually presents as the disease advances [8]. In a preliminary study of dysphagia in patients with HD, numerous abnormalities occurred throughout ingestion, that is, during those

"… In the HD-rb group (rigid – bradykinetic) (five patients), frequently observed abnormalities included mandibular rigidity, slow lingual chorea, coughing on foods, and choking on liquids. …"

Dysphagia in Huntington's Disease: A 16-Year Retrospective

Marion C. Kagel, M.A.[1] and Norman A. Leopold, D.O.[2]
[1]Department of Medicine, Divisions of Speech/Language Pathology, Crozer-Chester Medical Center, Upland, and
[2]Department of Neurology, Hahnemann University School of Medicine, Philadelphia, Pennsylvania, USA

Abstract. Degenerative diseases of the basal ganglia are commonly complicated by dysphagia. In 35 patients with Huntington's disease (HD), a hereditary neurodegenerative basal ganglia disease characterized by chorea, dementia, and emotional changes, an extensive battery of clinical and radiologic procedures helped to identify numerous abnormalities of deglutition. The results permitted the classification of our patients with HD into hyperkinetic (HD-h) or rigid–bradykinetic (HD-rb) groups. Although the two groups share multiple abnormalities, statistically significant intergroup differences were observed. Clinical assessment of the HD-h cohort (30 patients) demonstrated rapid lingual chorea, swallow incoordination, repetitive swallows, prolonged laryngeal elevation, inability to stop respiration, and frequent eructations. In the HD-rb group (five patients), frequently observed abnormalities included mandibular rigidity, slow lingual chorea, coughing on foods, and choking on liquids. Videofluoroscopic swallowing studies (VFSS) using a variety of barium-impregnated foods and liquids confirmed the abnormalities noted on the clinical assessment. Respiratory and laryngeal chorea, pharyngeal space retention, and aspiration were also identified. Numerous compensatory techniques introduced during videofluoroscopy benefited all patients.

Key words: Huntington's disease — Chorea — Rigidity — Respiration — Aspiration — Deglutition — Deglutition disorders.

Address offprint requests to: Dr. N.A. Leopold, Crozer-Chester Medical Center, POB #2, Suite 222, Chester, PA 19013, USA

Huntington's disease (HD) is a hereditary neurodegenerative disease of the central nervous system (CNS). Characterized by involuntary movements, dementia, and emotional impairment, HD appears most frequently during the fourth or fifth decades of life [1]. However, its phenotypy is variable, with a small percentage of patients presenting with parkinsonian features including bradykinesia or rigidity [2]. Extrapyramidal features change as HD progresses; many patients become more choreic and hypotonic, while others experience less chorea as muscle tone increases [3]. Chorea and varying degrees of bradykinesia and rigidity often coexist [4]. In some patients, parkinsonian signs are induced by phenothiazines administered for the treatment of chorea [4, 5]. Disease progression is slow and inexorable. Motor control of nearly all voluntary muscles is impaired, including those governing respiration, speech, and deglutition. Death usually occurs within 20 years, often from aspiration pneumonia [6].

Although multiple regions of the brain are affected in patients with HD, caudate and putaminal atrophy dominate the neuropathologic findings [1]. The result is a severe fall in CNS gamma-aminobutyric acid (GABA) levels [7]. Unfortunately, therapeutic strategies that elevate CNS GABA levels provide no substantive benefit. The current treatment is symptomatic and includes neuroleptics to reduce chorea and/or control schizophrenic-like behavior, anxiolytics, and antidepressants [5].

Dysphagia is a common, if not inevitable, complication of HD that usually presents as the disease advances [8]. In a preliminary study of dysphagia in patients with HD, numerous abnormalities occurred throughout ingestion, that is, during those

Disfagia na Doença de Huntington

- Posicionamento e postura (reduzir hiperextensão de cabeça e tronco)
- Adaptação utensílios - colher, copo, prato...
- Modificação de consistências e volumes
- Supervisão nos momentos de alimentação
- Manobras posturais de proteção da via aérea
- Meios alternativos - SNG / PEG

Terapia

- Copos com doseador
- Copos de punho
- Pesos pernas

- Se utente tem forma mais rígida
  - Exercícios de fortalecimento poderão ser apropriados

- Alterações emocionais e comportamentais
  - dificultam tratamento
  - ponderar custo-benefício da intervenção

Terapia

- ★ Hipercinéticos
  - Taquifagia incontrolável, movimento lingual súbito e inibição respiratória comprometida na deglutição
- ★ Rigidez-bradicinesia
  - rigidez mandibular, mastigação ineficiente e trânsito oral prolongado

- ★ **Manobras compensatórias**

"…We found that the patient groups studied were heterogeneous and the methods used were highly variable, and no balanced advice for prevention and treatment was systematically proven…".

Dysphagia (2011) 26:62–66
DOI 10.1007/s00455-010-9302-4

REVIEW ARTICLE

Dysphagia in Huntington's Disease: A Review

Anne-Wil Heemskerk · Raymond A. C. Roos

Received: 9 December 2009 / Accepted: 14 August 2010 / Published online: 14 September 2010

Abstract Huntington's disease (HD) is a progressive neurodegenerative autosomal dominant disease characterized by disturbed movements and behavior and cognitive decline. The motor disturbances are both choreiform and hypokinetic. As a result of the combination of these signs, it is known that many patients with HD suffer from dysphagia. Little is known about the frequency and the characteristics of dysphagia in HD. Well-balanced strategies for treatment and prevention of dysphagia in HD are lacking. Therefore, we have performed a detailed survey of the literature. We found that the patient groups studied were heterogeneous and the methods used were highly variable, and no balanced advice for prevention and treatment was systematically proven.

Keywords Huntington's disease · Dysphagia · Deglutition · Deglutition disorders

HTT gene on the short arm of chromosome 4. The mutant protein huntingtin causes neurodegeneration in the brain, particularly in the caudate nucleus and putamen. Onset of HD on average occurs in the third or fourth decade of life and lasts about 15–20 years [1–8]. Death often results from aspiration pneumonia caused in turn by a progressive dysphagia [9–12]. Previous studies into dysphagia in HD investigated dysphagic features in the different phases of ingestion [13–17]. It is not known in what stage of HD the dysphagia becomes clinically apparent. The frequency of this incapacitating sign is not known. No well-proven strategies to prevent dysphagia are available. Therefore, we reviewed the literature on this clinically important sign in HD, looking for different methods and interventions to find a better strategy for treatment and prevention.

Methods

"…The current evidence is insufficient to make strong recommendations regarding the usefulness of physiotherapy, occupational therapy, or speech pathology for people with Huntington's disease…"

Effectiveness of Physiotherapy, Occupational Therapy, and Speech Pathology for People with Huntington's Disease: A Systematic Review

Belinda Bilney, Meg E. Morris, and Alison Perry

This review provides a summary of the current literature examining the outcomes of physiotherapy, occupational therapy, and speech pathology interventions for people with Huntington's disease. The literature was retrieved via a systematic search using a combination of key words that included Huntington's disease, physiotherapy, occupational therapy, and speech pathology. The electronic databases for Medline, Embase, CINAHL, Cochrane Controlled Trials Register, and PEDro were searched up to May 2002. Articles meeting the review criteria were graded for study type and rated for quality using checklists to assess study validity and methodology. The majority of articles that examined therapy outcomes for people with Huntington's disease were derived from observational studies of low methodological quality. A low level of evidence exists to support the use of physiotherapy for addressing impairments of balance, muscle strength, and flexibility. There was a small amount of evidence to support the use of speech pathology for the management of eating and swallowing disorders. The current evidence is insufficient to make strong recommendations regarding the usefulness of physiotherapy, occupational therapy, or speech pathology for people with Huntington's disease. There is further need for therapy outcomes research in Huntington's disease so that clinicians may use evidence-based practice to assist clinical decision making.

The aim of this article is to assist physiotherapists, occupational therapists, speech pathologists, and rehabilitation physicians to effectively treat people with Huntington's disease by providing a review and critical evaluation of the evidence on therapy outcomes. A second aim is to identify the types of interventions or treatments most frequently used by therapists in the management of people with this debilitating neurological condition. Recommendations for therapy are made based on the evidence presented.

BACKGROUND

Huntington's disease (HD) is an inherited neurodegenerative condition that occurs owing to a mutation of the gene (IT15) located on chromosome 4.[1,2] The condition is typified by progressive degeneration of the medium spiny neurons within the basal ganglia, primarily the caudate and puta-