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Speech in spinocerebellar ataxia

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ABSTRACT

Spinocerebellar ataxias (SCAs) are a heterogeneous group of autosomal dominant cerebellar ataxias clinically characterized by progressive ataxia, dysarthria and a range of other concomitant neurological symptoms. Only a few studies include detailed characterization of speech symptoms in SCA. Speech symptoms in SCA resemble ataxic dysarthria but symptoms related to phonation may be more prominent. One study to date has shown an association between differences in speech and voice symptoms related to genotype. More studies of speech and voice phenotypes are motivated, to possibly aid in clinical diagnosis. In addition, instrumental speech analysis has been demonstrated to be a reliable measure that may be used to monitor disease progression or therapy outcomes in possible future pharmacological treatments.

Intervention by speech and language pathologists should go beyond assessment. Clinical guidelines for management of speech, communication and swallowing need to be developed for individuals with progressive cerebellar ataxia.

1. Spinocerebellar ataxia

Spinocerebellar ataxias, SCAs, are a heterogeneous group of hereditary, neurodegenerative, progressive disorders affecting the cerebellum and its efferent and afferent pathways. Age of onset is often in the third or fourth decade of life (Dürr, 2010). Clinical phenotype is dominated by progressive gait and limb ataxia, but in addition there is generally a range of concomitant neurological symptoms, e.g. oculomotor disturbance, retinopathy, spasticity, extrapyramidal movement disorders, peripheral neuropathy, sphincter disturbances. Dysarthria and cognitive impairment is also present in many cases.

The first gene causing autosomal dominant hereditary ataxia (spinocerebellar ataxia, type 1) was identified in 1993 (Orr et al., 1993). Since then there has been a rapid development of molecular genetic diagnostics, and to date up to 30 genetic loci and 20 genes have been identified (Perlman, 2011). Autosomal dominantly inherited ataxias were initially considered to be caused by expansions of coding CAG repeats within the coding region of the gene, as in SCA1, SCA2, SCA3, SCA6, SCA7, SCA17, and DRPLA (dentatorubro-pallidoluysian atrophy), subtypes of SCAs that were identified early (often referred to as polyglutamine expansion SCAs). The CAG repeats lead to a toxic process resulting in neurodegeneration and neuronal cell death. In addition, expansions in non-coding regions of genes have also been discovered as in SCA10, SCA12, and SCA31, referred to as non-coding-expansion SCAs. SCA4, SCA5, SCA11, SCA13, SCA14 and SCA27 result from point mutations, SCA15 and 16 with deletions and SCA6 is a channelopathy (Bird, 2013; Dürr, 2010; Perlman, 2011).

Anticipation is seen in many SCAs; i.e. increasing severity and earlier onset of disease in subsequent generations, related to increased repeat expansion from one generation to another (Mariotti & Di Donato, 2001). Prevalence of SCA is uncertain but has been suggested to be between 3–8/100,000 individuals (Craig, Keers, Archibald, Curstin, & Chinnery, 2004; Koht & Tallaksen, 2007; Van De Warrenburg, Sinke, Verschuuren-Bemelmans, Scheffer et al., 2002). Thus, SCA is a rare disorder, compared to e.g. Parkinson's disease with a prevalence in Europe of around 200/100,000 individuals over the age of 65 (Campenhausen et al., 2005; de Rijk et al., 2000) but comparable to e.g. Huntington's disease which has a prevalence of up to 10/100,000 (Hoppit, Calvert, Pall, Rickards, & Sackley, 2010; Morrison, 2010). Regional differences in prevalence of SCA exist, and the most common subtypes have been suggested to be SCA1, SCA2, SCA3, SCA6 and SCA7, accounting for about 70% of dominant SCA cases (Margolis, 2002).

2. Ataxic dysarthria

Ataxic dysarthria was characterized based on perceptual ratings of speech samples from individuals with cerebellar pathology by Brown, Darley, and Aronson (1970). Symptoms were found in the

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three clusters associated with cerebellar pathology defined by Darley, Aronson, and Brown (1969); i.e. articulatory inaccuracy, prosodic excess and phonatory–prosodic insufficiency. Most prominent speech dimensions related to articulation were imprecise consonants, irregular articulatory breakdown and distorted vowels. Prominent speech dimensions related to prosodic excess were excess and equal stress, prolonged phonemes and intervals. Dimensions related to phonatory–prosodic insufficiency were harsh voice, monopitch and slow speech rate. Some of the speech dimensions are less frequent in less severe impairment, e.g. distorted vowels, harsh voice, phonemes prolonged and monotony. Later descriptions of ataxic dysarthria, particularly based on acoustic methods, have included reduced speech rate in a range of different speech tasks, alterations in speech rhythm and lengthening of segments as well as increased duration and variability of inter-stress intervals (Hartelius, Runmarker, Andersen, & Nord, 2000; Kent et al., 2000). Articulatory imprecision has also been confirmed in subsequent studies of ataxic dysarthria, and has been associated with reduced control of range, velocity, force and timing of the articulators (e.g. Kent, Netsell, & Abbs 1979).

In studies using more recently available methodology articulatory kinematics in ataxic dysathria has been studied. Using electromagnetic articulography (EMA), slower articulatory durations were found in subjects with ataxic dysarthria secondary to Friedreich's ataxia. In addition, it was demonstrated that longer consonant phase durations were associated with greater articulatory distances and not only with slowed movement execution (Folker et al., 2011).

Acoustic studies of phonation in ataxic dysarthria have shown both increased jitter and shimmer (cycle-to-cycle variations in frequency and amplitude) which relate to the perceptual dimension "harsh voice" as well as long-term phonatory instability which relates perceptually to "vocal instability" or "voice tremor" (Kent et al., 2000).

In summary, the overall impression of ataxic speech is that it is slow, imprecise and with a characteristically changed prosody. Common patient complaints include a sense of "drunk" or intoxicated speech, stumbling over words, speech deterioration with alcohol and poor coordination of breathing with speech (Duffy, 2005). Patients have often noted that speech difficulties are reduced when speech is slowed down.

3. Dysarthria in spinocerebellar ataxia, SCA

Based on neuropathological considerations, dysarthria in SCA would be expected to resemble previous descriptions of ataxic dysarthria. Dysarthria is a feature in most clinical characterizations of subtypes of SCA, but there are very few more detailed descriptions of type and degree of speech symptoms to date.

Several speech tasks were studied with acoustic instrumentation in two subjects with SCA7 and one subject with SCA2 by Schalling and Hartelius (2004). Reduced speech rate, increased pause duration, increased and more variable durations of alternating motion rate (AMR), sequential motion rate (SMR) syllables and inter-stress intervals (ISI) in addition to vocal instability was demonstrated in the three subjects. Schalling, Hammarberg, and Hartelius (2007) studied 21 individuals with hereditary, progressive ataxia and 21 matched control subjects, with perceptual and acoustic methods. Twelve of the subjects had a diagnosis of SCA2, 3, 7 or 8, based on molecular testing, and nine were diagnosed clinically by a neurologist with cerebellar ataxia (of unknown etiology). Perceptual analysis was performed by four experienced speech–language pathologists. The most prominent speech symptoms were equalized stress, imprecise consonants, vocal instability, monotony and reduced speech rate. A factor analysis showed that perceptual speech symptoms were associated primarily with two major factors. The first factor was called *speech-timing and articulation* (including characteristics such as imprecise consonants, prolonged intervals, imprecise vowels and equalized stress, all speech characteristics related to difficulties with coordination and timing). It was hypothesized that this factor mainly reflected common underlying speech motor programming difficulties related to articulatory disturbance. The second factor was called *voice quality* (including characteristics such as harsh voice, strained–strangled voice and glottal fry, all related to vocal hyper-function). This factor may reflect other aspects of underlying neurophysiological pathology, associated with phonatory aspects. The third factor only had one speech characteristic with a high factor loading and was therefore not considered important.

Acoustic findings in Schalling et al. (2007) confirmed perceptual impressions and included reduced speech rate, increased variability of segment durations as well as increased vocal instability shown as significantly higher coefficient of variation of F0 compared to control subjects.

Another term for the prominent and frequently found perceptual symptom equalized stress is "scanning speech". In a study by Ikui et al. (2012) of 20 Japanese-speaking subjects with spinocerebellar degeneration of different etiologies, speech was investigated particularly with reference to this concept. Potential differences in the manifestations of ataxic speech between a syllable-timed language like Japanese and stress-timed languages like the Germanic languages were explored. In a syllable-timed language the syllables durations are of equal length, whereas in stress-timed languages, stress is placed at equal intervals in the phrase, thus inter-stress intervals are isochronous but syllable durations are more variable. Ikui and colleagues found that speech rate was reduced in ataxic subjects compared to control subjects. In addition, duration of vowel segments (morae) was both longer and more variable in speech produced by ataxic subjects. In addition, long Japanese vowels were also produced with more variable duration in ataxic subjects compared to controls and the distinction between long and corresponding ordinary vowel became unclear. Thus, scanning speech in Japanese is caused by a breakdown of isochrony, i.e. a difficulty in maintaining invariable vowel length in speech. In addition, a tendency towards shortening of long vowels also contribute to the impression of "scanning" and also contribute to unclear distinction between long and ordinary vowel in Japanese. The findings by Ikui et al. differ from previous studies of scanning speech in stress-timed languages, e.g. by Hartelius et al. (2000) or by Ackerman and Hertrich (1994), who found more isochronous syllables in ataxic subjects compared to healthy controls, and may reflect differences in the nature of syllable-timed and stress-timed languages.

In an effort to look for differences in perceptual characteristics across SCA subtypes and also differences in speech signs across speech tasks, Sidtis, Ahn, Gomez, and Sidtis (2011) examined speech in 26 subjects with SCA1, 5 or 6. Perceptual ratings were performed by speech–language pathology master students. Speech samples were rated from 1 to 5 on primary and secondary dimensions. Primary dimensions included articulation, rate, rhythm and prosody (rhythm referring to pitch accents and prosody referring to melodic contour of the intonational entity). Samples rated as abnormal on the primary dimensions were also rated on 11 secondary dimensions derived from the Mayo Clinic protocol in order to further characterize the speech abnormality (Duffy, 2005). The secondary dimensions were irregular articulatory breakdown, imprecise consonants, distorted vowels, prolonged phonemes, excess and equal stress, excess loudness variation, hypernasality, voice tremor, harsh voice, breathy voice, strained–strangled voice.

Some differences between SCA-subtypes were identified. A global measure of impairment indicated that the group with SCA6 was