Dysarthria in rapid-onset dystonia-parkinsonism

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Introduction

Rapid-onset dystonia parkinsonism (RDP) is a rare movement disorder characterized by sudden onset of dystonia and parkinsonism with infrequent symptom change after this period [1, 2, 3]. Nonmotor symptoms may include cognitive changes and psychiatric difficulties. It occurs due to ATP1A3 mutations with de novo and familial variants[4].

Communication changes with RDP, while important to differential diagnosis, have been insufficiently characterized, with descriptions limited to hypophonia and dysarthria. The onset, progression, and profile of the dysarthria is not known.

We analyzed the speech of two 18 year old identical twin sisters with RDP:
RQ1. What is the dysarthria profile?
RQ2. What is the pattern of onset and progression of dysarthria?

Measures

History
- Twin A: sudden onset of motor symptoms at age 15 following a minor traffic collision
- Twin B: sudden onset of motor symptoms at age 18 with no associated trigger

RQ1: a standard motor speech evaluation and connected speech sample were analyzed via perceptual evaluation (3 expert SLPs)

RQ2: Speech samples were obtained from social media video blogs
- 48 sampling dates were selected over a period of 2 years
- Each sample was divided into phrases, and the 10 longest phrases (ms) were analyzed

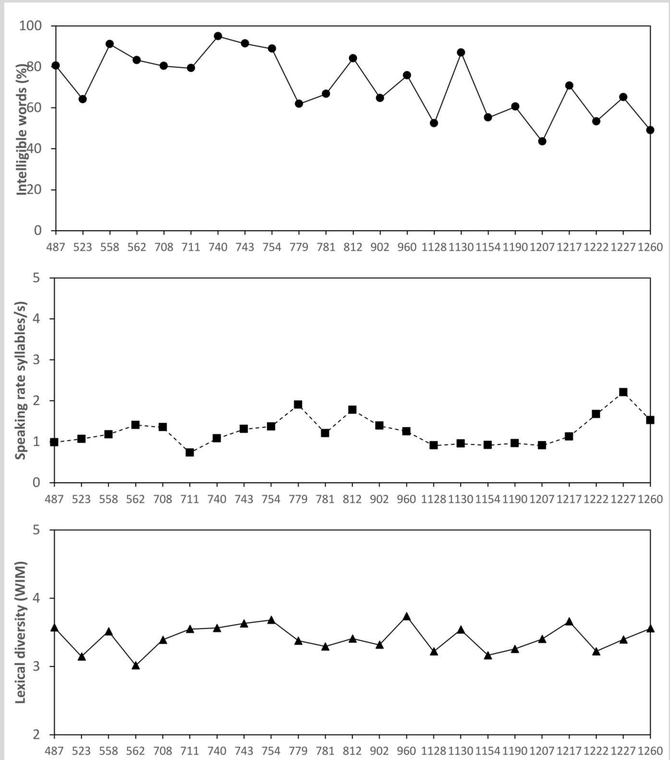
  - *Intelligibility*: % words understood for phrases presented in random order averaged across two raters
  - *Speech rate* (syllables/second)
  - An index of lexical diversity, the *Word Information Measure (WIM)*[5]

Results

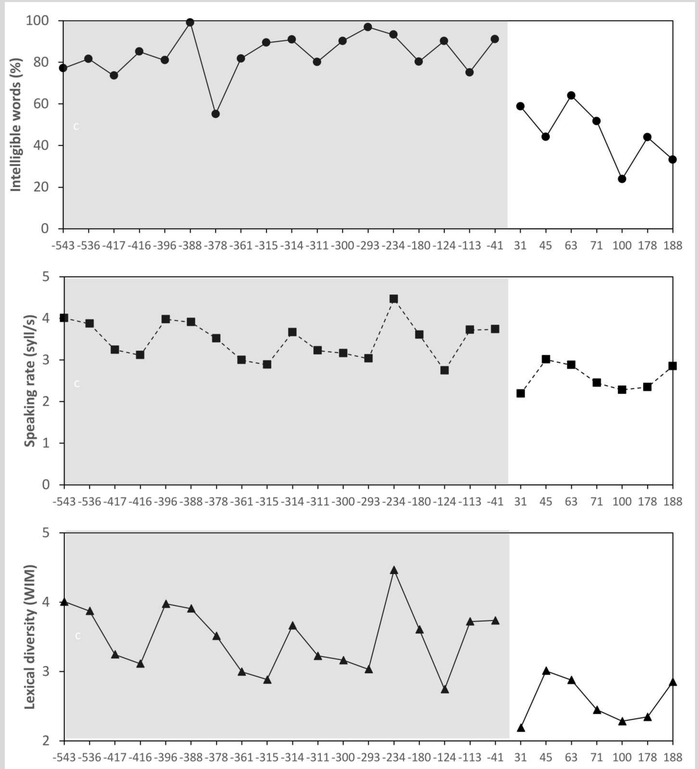
RQ1. What is the dysarthria profile?

Expert perceptual analysis indicated the same profile for both twins: mixed hyperkinetic-hypokinetic dysarthria
- Hyperkinetic elements: prominent mandibular dystonia primarily affecting bilabial consonant articulation
- Hypokinetic elements: hypophonia, breathy vocal quality, short rushes of speech, consonant imprecision

Twin A: First sample 487 days after primary onset



Twin B: First sample 583 days before primary onset



RQ2. What is the pattern of onset and progression?
- A linear mixed effects model of intelligibility over time predicted declining intelligibility ($p<0.001$), with a decrease of -0.04% intelligibility per day
- For Twin A, intelligibility appeared to demonstrate a gradual decrease during this period >487 days after symptom onset
- For Twin B, abruptly decreased intelligibility, lexical diversity, and rate were associated with the self-reported time of onset

Discussion

1. The motor speech profile was consistent with the limb and gait symptoms of parkinsonism and dystonia
2. Onset of motor speech deficits appeared to share the dramatic pattern of limb symptoms
3. Intelligibility appeared to continue to decline in the months and years post onset though individuals with RDP generally report symptom stability[2]

Future Directions

1. Is there progressive decline in motor speech function *post* primary symptom onset?
2. Do speech and language changes occur *prior* to primary motor onset for mutation-positive individuals in the asymptomatic phase?
3. Do communication measures predict psychiatric[6] and cognitive changes[7]?
4. Does dysarthria in RDP respond to behavioral therapy?

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PDF of poster and code for data collection and analysis available at github.com/ktcslp/motorspeech2020