Four-year Retrospective Speech/Language Report of a Child with Joubert Syndrome

Rachel Bissell, B. S. Joan Kwiatkowski, M. A.
Department of Communicative Disorders, University of Wisconsin – Madison

WAISMAN CENTER

Background

- Typical features of Joubert Syndrome [1] include
  - underdevelopment/complete lack of cerebellar vermis
  - cognitive disabilities varying in severity
  - difficulty coordinating voluntary muscle movements (ataxia)
  - decreased muscle tone (hypotonia) affecting gross and fine motor movements
  - individualized treatment based on symptomatology

- Other features, that may or may not be present [1] are
  - facial dysmorphologies
  - ocular impairments, not limited to oculomotor apraxia
  - respiratory issues
  - medical issues affecting various body systems
  - sensory-motor integration issues

- Limited findings for speech and language development [2] indicate
  - receptive and expressive language delays vary in severity
  - speech intelligibility ranges from good to poor
  - possible presence of apraxia of speech in combination with motor disorders in other systems such as ocular- and fine- motor

- The intent of the current report is to provide additional information on speech/language development and learning in children with Joubert Syndrome.

Methods

Participant

- R, a male, ranged in age between 6;10 and 10;10 during the period of this retrospective study. He received speech and language services in a university clinic prior to and during the study period.
- He was diagnosed with Joubert Syndrome at 2;0, with
  - a cleft through one area of the cerebellar vermis and underdevelopment of other areas
  - oculomotor apraxia
  - unspecified cognitive delays, warranting special education services
- He was diagnosed with apraxia of speech at 3;7.
- Due to delays in the development of speech and syntax, he has received speech/language services since 2;0.

Methods, continued

Procedure for Review of Clinic Files

- While R received speech and language services in the university clinic during fall, spring and summer semesters, because of the slow rate of change, only yearly progress reports between ages 6;10 and 10;10, were reviewed.

Summary of Status/Changes

- ***Speech Sound Production***
  - Vowels: delays in production of most non-rhotic vowels; most substitution errors resolved by 6;10; substitution of low-front vowel as in ‘sad’ for mid-front vowel as in ‘said’ persisted at 10;10; persistent distortion of rhotic vowels
  - Consonants: persistent delays in consonant production: lateral distortion of /s/ /z/, lateralized substitutions for other sibilants, substitution of voiceless cognate for final position /b/ /d/ /g/ and /v/, deletion of /l/ and /r/ in clusters, substitution of /w/ for /r/; by 10;10 was beginning to resolve substitution of ‘y’ for /l/
  - Intelligibility: persistent problems with intelligibility ranging from good to poor depending on content and context

- ***Prosody/Voice***
  - Phrasing: typically not linking words into phrase units; linking evident only in frequently used phrases (e.g., I don’t know); sometimes pauses between syllables in multi-syllabic words.
  - Rate: fluctuates from normal to too slow
  - Stress: often puts equal stress on each word; when pauses between syllables in multi-syllabic words, puts equal stress on each syllable
  - Loudness: ranges from normal to too loud; ability to voluntarily adjust loudness when asked suggests lack of self-monitoring
  - Pitch: appropriate
  - Voice: appropriate laryngeal quality and nasal resonance

- ***Expressive Language***.
  - Vocabulary: steady acquisition of vocabulary
  - Noun Phrase: correct use of pronouns; persistent delays = inconsistent use of articles, s-marked possessive and s-marked plural
  - Verb Phrase: persistent delays = inconsistent use of ed-marked and irregular past tense forms, s-marked 3rd person singular verb form, auxiliary ‘be’ forms, and copula; when inconsistently marks future tense uses “going to” rather than modal “will”
  - Negatives: improved from consistent *not/don’t* substitution at 6;10 to inconsistent use of *don’t* at 10;10; ability to self- correct identified errors for *don’t* at 10;10 suggests lack of self- monitoring; did not always use correct *do* + *not* form.

Summary of Status/Changes, continued

- ***Expressive Language, continued***
  - Questions: by 6;10 was inverting subject and verb; went from not including dummy-do at 7;10 to inconsistent use at 9;10, which persisted at 10;10
  - Complex/Compound Forms: While most utterances continued to be simple sentences, went from no complex/compound forms at 6;10, to inclusion of marked infinitive by 7;10, addition of conjunction *and* by 8;10, conjunction *if* and full propositional complements headed by *think* by 9;10, and conjunction *but* by10;10.

Observed Learning Patterns and Challenges

- Progress for the few resolved/resolving speech/language targets was slow. For example,
  - speech sound errors that resolved exceeded normative expectations; while most targeted non-rhotic vowels were learned by 6;10, the mid-front non-rhotic vowel and rhotic vowels were not correct at age 10;10
  - production of do + not forms in declarative statements was still inconsistent at 10;10
- Some errors never resolved. For example,
  - linking and stress issues, possibly because of difficulty coordinating respiratory and articulatory subsystems
  - lateral distortions and substitution errors for /r/, possibly because of learning challenges noted below
- Learning challenges were evident and needed to be addressed during shaping/evoking speech sounds and for generalization of speech sounds and syntactic forms.
  - shaping/evoking speech sounds was challenging seemingly because of apraxia of speech; the single effective strategy was to work from behaviors R could voluntarily produce, when these were available; e.g., shaping /l/ by sticking tongue out slightly between teeth and contacting the tongue to the lower surface of the top teeth
  - when a speech or syntactic target was being produced in structured teaching contexts, challenges to generalization appeared to be related to lack of self-monitoring, as R could readily correct errors when the clinician identified them; a promising combination of training strategies included auditory bombardment and evaluation of the clinician’s correct/incorrect productions to increase awareness of a target, production/self-evaluation during writing/reading exercises and production/self-evaluation when verbally communicating in meaningful contexts with the clinician and family members.
- The continuation of learning of speech sounds and syntactic forms well beyond the developmental period suggests that changes are occurring over time that create the potential for learning previously unsuccessful targets.
- The most productive strategy for identifying speech/language targets was to probe R’s potential for learning during a short diagnostic teaching period; if the target was too challenging, work was discontinued and the target was probed again during the next semester; e.g., /l/ was probed between 6;10 and near 10;10 when it was finally targeted.

While this report of R’s speech/language skills and learning challenges is more detailed than existing reports for children with Joubert Syndrome, it is likely not representative of all children with this syndrome. It can, however, serve as a guide to identifying skills and challenges in other children with Joubert Syndrome and developing a body of reference data.

References

1. Joubert Syndrome (Cerebellar vermis agenesis/hypoplasia). Joubert Syndrome Foundation & Related Cerebellar Disorders: www.jsfrc.org
2. Braddock, B.A., Farmer, J.E., Deidrick, K. M., Iverson, J. M., & Maria, B. L. (2006). Oromotor and communication findings in Joubert Syndrome: Further evidence of multisystem apraxia. *Journal of Child Neurology*, *2*, 160-163.

Acknowledgement : This project was supported by a grant from the National Institute on Deafness and Other Communication Disorders, NIDCD DC0049C and a core grant (HD00352) to the Waisman Center from the National Institute of Child Health and Development.