Brief Communication

Oromotor and Communication Findings in Joubert Syndrome: Further Evidence of Multisystem Apraxia

ABSTRACT

This study provides descriptive information in the areas of oromotor abilities and communication to better understand the spectrum of disability in individuals with Joubert syndrome. Participants included 21 individuals with the diagnosis of Joubert syndrome (mean age 10.45 years). Participants completed oromotor and receptive language measures. In addition, all of the participants' speech and gesture communication from a narrative task was coded and analyzed from videotape. Caregivers reported the participants' level of fine and gross motor function. The results show that individuals with Joubert syndrome exhibit a distinct oromotor pattern consistent with verbal and lingual apraxias. Despite significant motor skills deficits and oculomotor apraxia, persons with Joubert syndrome produced gestures when communicating, and those whose speech was less intelligible used a higher rate of gesture compared with those with greater verbal output. These findings suggest a new form of apraxia not previously described in the condition and are consistent with previous research that suggests that persons with Joubert syndrome typically do not exhibit classic symptoms of autism spectrum disorder. (*J Child Neurol* 2006;21:000–000).

Joubert syndrome is a rare neurogenetic disorder characterized by midbrain-hindbrain malformations, including agenesis of the cerebellar vermis, dysplasia of the dentate and olivary nuclei, and nondecussation of major brainstem tracts.[1,2] Clinical characteristics of Joubert syndrome can include oculomotor apraxia, nystagmus, gaze palsy, reduced visual acuity, motor ataxia, hypotonia, poor coordination, motor speech dysarthria, and developmental delay.[1,3,4]

In a report describing the original proband, Andermann et al highlighted the prominence of the motor speech impairment found to be characteristic of Joubert syndrome both at the time of diagnosis and on reexamination 30 years later.[3] The original proband was able to protrude his tongue incompletely but had no lateral tongue movement (lingual apraxia). His speech had features of pseudobulbar dysarthria and was judged to be similar to the speech pattern of another group of patients with bilateral perisylvian polymicrogyria. Despite the presence of pseudobulbar speech qualities, the authors interpreted cerebellar and brainstem dysfunction as the cause of the dysarthria. Although oromotor dysfunction is generally cited as a clinical marker of Joubert syndrome, little additional research is available that systematically examines the nature of this disability.

Other research reported in the literature provides some information about the quality of communication skills among individuals with Joubert syndrome, but detailed analyses are lacking. For example, Gitten et al examined parent reports of developmental delays in 32 children with Joubert syndrome.[5] At a mean chronologic age of 68.7 months, this group displayed skills at the 19-month developmental level in both expressive and receptive language. In a subsequent series of studies, Fennell et al identified deficits in verbal fluency, verbal memory, and picture recognition vocabulary on standardized tests, but the sample sizes were quite small (n = 2–6 children).[6] Finally, in a study of autistic behavior in Joubert syndrome, Ozonoff and colleagues reported that only 4 of 11 (36%) children were verbal and only 3 of 10 (30%) used adequate nonverbal behaviors to regulate social interaction.[7] This study suggested that autism spectrum disorders and associated communication impairments might be common co-occurring conditions in Joubert syndrome.

To our knowledge, no study has conducted an in-depth examination of oromotor and communication skills in larger samples of individuals with Joubert syndrome. The goal of the present study was to provide more descriptive information in these areas. Three research questions were addressed: (1) Is a distinct pattern of oromotor functioning evident in Joubert syndrome? (2) What is the average communicative functioning of persons with Joubert syndrome? and (3) To what extent is the rate of gesture related to the severity of the language and/or motor disorder?

Methods

Participants included 21 of 47 (45%) persons with Joubert syndrome and their caregivers who attended a biannual international conference on Joubert syndrome. Participants ranged in age from 32 months to 19 years 9 months (mean age = 10.45 years, SD = 5.50 years). Fifty-two percent of the participants were male. With the exception of one African-American participant, the participants were Caucasian. Demographic information was available for a subset of 17 mothers and 17 fathers. The mean age of the mothers was 39.42 years (SD = 4.8 years) and of the fathers was 40.67 years (SD = 5.8 years). Slightly less than half of the mothers (47%) and fathers (47%) achieved a bachelor's or graduate degree. All participants were from homes in which English was the primary language. Five participants were excluded from the study (four were unable to complete the gesture-speech observation owing to severe verbal and nonverbal deficits, and one was non–English speaking).

A certified speech-language pathologist and child neuropsychologists conducted the assessment in accordance with a research protocol approved by the Institutional Review Board. It consisted of measures of oromotor functioning, speech and gesture use, receptive vocabulary, and motor functioning.

Oromotor Functioning

The examiner instructed persons with Joubert syndrome to complete tasks designed to assess the range of excursion, speed, and accuracy of oromotor movement.[8,9] First, participants were asked to sustain phonation over time following a practice trial (eg, "Say 'ah' as long as you can while I time you"). The examiner then recorded the length of the vowel prolongation in seconds. Next, participants were instructed to retract and protrude their lips as in a smile and pucker on command or to imitation. Participants were also instructed to stick their tongue out past their lips and move their tongue side to side and up and down on command or to imitation. Finally, participants were asked to complete alternating movements of the lips, tongue, and velum (eg, "Say 'pataka' as fast as you can and as clearly as you can until I tell you to stop"; time-by-count diadochokinetic rate).[9] Participants were allowed three practice trials of about 3 seconds with the examiner. The examiner recorded whether participants were able to complete 10 repetitions over time with speed, accuracy, and periodicity.

Gesture-Speech Observation

Each individual with Joubert syndrome and their primary caregivers were videotaped as they engaged in a verbal narrative regarding a cartoon script or a picture book.[10,11] For younger participants or those participants with difficulty viewing the cartoon drawings owing to visual impairments, the large picture book was used. Caregivers were instructed to talk about the drawings or book with the participants, and participants were then asked

to provide a verbal narration regarding the story sequence. Caregivers were allowed to provide visual and verbal prompts as needed to elicit conversation from the participants and were allowed as much time as necessary to complete the task.

All of the participants' speech and gestures were coded from the videotaped cartoon or book narrations. Speech was transcribed verbatim and coded using a standard procedure for analyzing free-speech samples.[12] Measures of verbal communication included (1) the number of different words (calculated as the number of different root words produced in the course of the observation); (2) the mean length of utterance in morphemes (calculated as the number of morphemes divided by the total number of intelligible utterances); and (3) intelligibility rating (calculated as the number of completely intelligible utterances divided by the total number of intelligible and unintelligible utterances).

Gesture productions were coded using rating criteria established by Butcher and Goldin-Meadow[13]: (1) the gesture must not itself be a direct manipulation of some relevant person or object. To be conservative, all acts that were performed on objects were excluded with one exception: if a participant held up an object to bring it to another's attention, an act that appeared to serve the same social-communicative function as the pointing gesture, it was counted as a gesture; (2) the gesture must not be part of a ritual act (eg, blowing kisses to someone); and (3) the gesture must not be in direct imitation of the caregiver's preceding gesture. Lexicalized gestures were also scored as gesture occurrences (eg, Amerind, American Sign Language). To assess intercoder reliability, 2 of 21 speech-gesture observation segments were independently transcribed and coded by a second trained coder. Mean percent agreement was 87% for gesture occurrence (n = 97). Gesture rate was calculated as the number of gestures divided by the total number of communications produced by each participant over the course of the gesture-speech observation.

Receptive Vocabulary

An examiner also asked participants to complete the Peabody Picture Vocabulary Test-III (PPVT-III)[14] using pictured stimuli enlarged by 30%. The Peabody Picture Vocabulary Test-III is a standardized measure of one-word receptive vocabulary skill (mean = 100, SD = 15).

Motor Functioning

As part of a larger assessment of adaptive behaviors,[15] caregivers of persons with Joubert syndrome were asked to complete the Scales of Independent Behavior-Revised (SIB-R)[16] to describe participants' fine and gross motor skills (mean = 100, SD = 15).

Results

Table 1 presents findings from the oromotor measurement. Nearly all participants were able to protrude their lips as in a pucker, retract their lips as in a smile, and stick out their tongues past their lips. In contrast, only about one third to half of the participants were able to move their tongue side to side, upward, and downward. Although participants clearly understood commands for tongue movement, some were observed using head thrusts to comply with lateral movement in a fashion similar to what they use for lateralizing eye movements (oculomotor apraxia). No vertical head thrusts were noted. The most notable deficits were in the production of alternating movements of the lip, tongue, and velum (ie, "pataka") and in sustained phonation over a 5-second duration because fewer than one third were able to complete these tasks. All participants maintained their nutrition through oral feedings, although a few required modifications to the texture of their diet to promote safe swallowing.

Persons with Joubert syndrome displayed significant impairments in both expressive and receptive language. As shown in Table 1, the mean length of utterance was less than two words, a finding consistent with the level of language complexity found in children at approximately 26 months of age. The mean number of different root words produced over the course of the gesture-speech observation was approximately 30, and the mean percentage of intelligible utterances was 61%. Receptive vocabulary was in the mildly deficient range, on average, compared with same-age peers.

Despite very significant impairments in motor functioning (motor standard score = 40), participants made relatively frequent use of gesture (mean gestures per communications = 0.44, SD = 0.21). The gesture rate for each participant was ranked from low to high, and a median split was performed between 0.42 and 0.43. This enabled us to group participants into two subgroups: a low-gesture subgroup (n = 10) and a high-gesture subgroup (n = 11). These two groups did not differ significantly in age (low-gesture subgroup, mean = 12.41 years, SD = 4.91; high-gesture subgroup, mean = 8.66 years, SD = 5.61; U = not significant).

Table 1 provides descriptive data in the areas of receptive and expressive language and motor skills for the low-gesture and high-gesture subgroups. Relative to low-gesture participants, high-gesture participants produced a significantly lower number of different words (U = 18.5, P < .01) and a significantly lower mean length of utterance (U = 15.5, P < .01). They tended to exhibit more unintelligible utterances (U = 30, P < .08). No differences were found between the low-gesture and high-gesture subgroups in one-word receptive language ability (Peabody Picture Vocabulary Test-III; low-gesture subgroup, mean = 65.72, SD = 12.98; high-gesture subgroup, mean = 65.5, SD = 14.33, U = not significant).

Relationships between gesture rate and expressive language measures were also examined using partial correlations controlling for participant's chronologic age. Strong negative associations were found between the gesture rate and the mean length of utterance (r = –.72, P < .002), the number of different words (r = –.76, P < .001), and speech intelligibility (r = –.55, P < .03). Finally, relationships between gesture rate and receptive language standard score (Peabody Picture Vocabulary Test-III) and motor standard score (Scales of Independent Behavior-Revised) were not statistically significant.

Discussion

This is the first known study to provide an in-depth examination of communicative functioning in persons with Joubert syndrome. In the area of oromotor function, participants exhibited a distinct pattern that might distinguish them from other groups with neurologic impairment. They showed marked difficulties in cranial nerve XII function for bilateral tongue lateralization, elevation, and depression yet relatively spared abilities to protrude the tongue and to retract and protrude the lips bilaterally. Although delays in the initiation of tongue movement were not quantified as previously described for oculomotor function in individuals with Joubert syndrome, the observed horizontal head thrusts for tongue lateralization are similar to those in ocular lateralization.[17] These findings suggest that individuals with Joubert syndrome have oculomotor and lingual apraxias. Alternating movements of the articulators were especially difficult for individuals with Joubert syndrome, which might be due to verbal apraxia given that cerebellar input is required for finely tuned speech output. In addition, the participants displayed poor abilities to sustain phonation for short durations. This might indicate that individuals with Joubert syndrome have poor coordination between the respiratory and phonatory systems of speech or suspected deficits within cranial nerve IX and/or X transmissions. Despite the degree of oromotor involvement and speech dysarthria, nearly all participants were able to eat orally without signs or symptoms of oral or pharyngeal dysphagia or aspiration.

Specifically, examination of cranial nerve XII in persons with Joubert syndrome indicates that the impairment is not consistent with unilateral hypoglossal involvement as sometimes seen in other groups of persons with related brain disorders and resultant speech dysarthria (eg, the tongue deviates to the side of neurologic impairment). On the other hand, persons with Joubert syndrome do not show complete bilateral hypoglossal involvement. In cases of complete hypoglossal involvement, one would expect inability to lateralize the tongue side to side and inability to protrude the tongue fully past the central incisors. We find it clinically significant that nearly all participants were able to eat orally despite the described oromotor deficits and concerns with phonatory sufficiency. It might be that persons with Joubert syndrome have functional pharyngeal plexus innervation (eg, cranial nerves IX, X, XI) for use in safe swallowing but have difficulty in coordinating the phonatory and respiratory systems during speech because of abnormal corticobulbar connectivity. On the basis of the observations in our study, we hypothesize that lingual apraxia with sparing of

Table 1. Speech-Language and Motor Functioning for the Total Sample and for High- and Low-Gesture Subgroups

Assessments	*Total Sample* % (n)	*High-Gesture Subgroup* % (n)	*Low-Gesture Subgroup* % (n)
Oromotor			
Labial			
Retract lips	100 (21/21)	100 (11/11)	100 (10/10)
Protrude lips	95 (21/21)	91 (10/11)	100 (10/10)
Lingual			
Side to side	43 (9/21)	27 (3/11)	60 (6/10)
Upward	35 (7/20)	10 (1/10)	60 (6/10)
Downward	50 (10/20)	20 (2 /10)	80 (8/10)
Protrude tongue	86 (18/21)	82 (9/11)	90 (9/10)
Alternating movement	30 (6/20)	10 (1/10)	50 (5/10)
Sustained phonation	33 (6/18)	11 (1/9)	56 (5/9)
Oral feeding			
Oral nutrition	100 (21/21)	100 (11/11)	100 (10/10)
With modifications	14 (3/21)	27 (3/11)	0 (0/10)
	Mean (SD)	*Mean (SD)*	*Mean (SD)*
Expressive language	*n* = 21	*n* = 11	*n* = 10
Number of words	30.19 (23.59)	17.45 (19.81)	44.20 (19.59)
Mean length of utterance	1.85 (0.96)	1.30 (0.68)	2.47 (0.86)
Intelligibility rating	61% (30)	49% (33)	74% (22)
Receptive vocabulary*	65.59 (13.37) (*n* = 17)	65.50 (14.33) (*n* = 10)	65.71 (12.98) (*n* = 7)
Motor standard score†	40.17 (28.18) (*n* = 18)	45.20 (30.93) (*n* = 10)	33.88 (24.85) (*n* = 8)

*Peabody Picture Vocabulary Test-III (mean = 100, SD = 15).
†Scales of Independent Behavior-Revised (mean = 100, SD = 15).

facial nerve and pharyngeal plexus function for safe swallowing is a unique feature of individuals with Joubert syndrome and related disorders of hindbrain malformation.

Consistent with previous studies, persons with Joubert syndrome demonstrated very significant deficits in language functioning and motor ability.[5] The present research extends this literature by examining the relationship between gesture and verbal modalities during a direct observation of spontaneous communication. Participants with the most difficulty in expressive language produced gesture at a higher rate. These findings suggest that persons with Joubert syndrome are not passive communicators. Instead, they attempt to communicate with caregivers through the use of available words, naturalistic gesture, and lexicalized sign. These results stand in sharp contrast to individuals with autism spectrum disorders, who tend to use few gestures that involve sharing reference and directing social-communicative interactions.[18–21] This supports a previous finding that individuals with Joubert syndrome typically do not exhibit the classic characteristics of autism.[22]

Diagnostic criteria for Joubert syndrome include the presence of a characteristic "molar tooth" sign on magnetic resonance imaging of the brainstem isthmus.[2] As of this publication, four loci and two genes have been implicated in disorders that share the molar tooth sign malformation.[23] The findings from the present study provide information about oromotor and communication profiles that might shed light on the disorder and provide a more complete understanding of malformed neuromotor circuits in Joubert syndrome.

In future research, these fine-grained profiles will need to be examined in various hindbrain malformation disorders to uncover molecular and structural central nervous system etiologies. Further, it is necessary to determine if the degree of apraxia in speech is related to a broader motor disturbance in Joubert syndrome, encompassing other systems, such as oculomotor and fine motor abilities. In addition, analyses of the interplay among gesture, language, cognition, and motor ability are crucial to understanding the neurobehavioral profile of Joubert syndrome.

Barbara A. Braddock, MHS
Department of Psychological Sciences
University of Missouri-Columbia
Columbia, Missouri

Janet E. Farmer, PhD
Kathleen M. Deidrick, PhD
Department of Health Psychology
University of Missouri-Columbia
Columbia, Missouri
Jana M. Iverson, PhD
Department of Psychology
University of Pittsburgh
Pittsburgh, Pennsylvania
Bernard L. Maria, MD, MBA
Department of Pediatrics
Medical University of South Carolina
Charleston, South Carolina

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Address correspondence to Dr Janet Farmer, Department of Health Psychology, University of Missouri-Columbia, One Hospital Drive, DC116.88, Columbia, MO 65212. Tel: 573-882-8847; fax: 573-884-4540; e-mail: farmerje@health.missouri.edu.

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