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ORIGINAL ARTICLE

Reported communication ability of persons with trisomy 18 and trisomy 13

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Abstract

Objective: The aim of this study was to describe the communication ability of individuals with trisomy 18 and trisomy 13 syndromes.
Methods: Parents reported on children's potential communication acts, words, spontaneous gesture, and augmentative and alternative communication (AAC) using a parent report inventory ($n = 32$; age range 3–35 years). Potential communicative acts are defined as behaviors produced by an individual that may be interpreted by others to serve communicative functions.
Results: Potential communicative acts categorized as body movement displayed the highest median rank for reported occurrence followed by vocalization and facial expression. Although symbolic forms were ranked lower, more than half of the parents (66%) reported that their children produced at least one word, gesture or AAC form. Challenging behaviors or stereotypic movement displayed lowest median ranks.
Conclusions: Results are discussed in terms of communication potential and the need to address AAC in trisomy 18 and 13.

Keywords

Augmentative and alternative communication, gestures, potential communicative acts, trisomy 13, trisomy 18

History

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Introduction

Two common chromosome syndromes, trisomy 18 and trisomy 13, are distinct conditions; yet share some common characteristics [1]; namely, in both conditions, trisomy refers to a range of genetic disorders resulting due to the presence of three copies of a chromosome, rather than two. Although severe to profound neurodevelopmental disorders are well documented in persons with trisomy 18 and trisomy 13, little is known about their communication potential given that only 5–8% of persons born with the disorder survive the first year of life [1]. Since these genetic disorders are not as universally lethal as once presented, more information is needed into the communication potential of those who survive the neonatal period and are ultimately discharged home with their families.

The trisomy 18 and 13 syndromes, also known as Edwards syndrome and Patau syndrome, respectively, are the second and third most common trisomy syndromes after trisomy 21 (Down syndrome) [2]. Most infants are prenatally diagnosed by screening based upon maternal age, maternal serum marker or detection of sonographic abnormalities. Trisomy 18 is characterized by low birth weight, prominence of the posterior portion of the cranium, clenched hands, heart malformations, kidney defects and intellectual disability. Trisomy 13 is characterized by low birth weight, polydactyly, scalp defects, orofacial clefts or cleft lip and palate, eye malformations, decreased muscle tone and intellectual disabilities. Major causes of death include central apnea, cardiac failure and respiratory insufficiency. These syndromes affect over 2000 families in the United States annually [2].

Research into the communication potential of surviving persons with trisomy 18 and 13 may aid others as they seek to recognize the developmental trajectory of this understudied group of learners. The aim of this study was to describe potential communicative acts in a larger sample of survivors with trisomy 18 or trisomy 13 using a parent report instrument, known as the Inventory of Potential Communicative Acts [3]. A potential communicative act is described in the literature as any behavior produced by an individual that may be interpreted by others to serve a communicative purpose [3, 4]. Two specific research questions were addressed: (1) What is the pattern of reported communication behavior in a group of individuals with trisomy 18 and 13? and (2) What differences in reported communication behavior are apparent between subgroups of individuals with trisomy 18 vs. trisomy 13?

Only a few studies have examined the communicative abilities in persons with trisomy 18 and trisomy 13. In one such study, investigators interacted with 10 individuals with trisomy 18 or 13 at a mean age of 15.96 years during a communication temptation task using bubbles and toy play [5]. Communicative behaviors were then coded from

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videotape. Relative to behaviors classified as hand gestures or words, participants produced higher proportions of behaviors involving visual-eye movement, facial expression, and vocalizations in interactions with the examiner. In addition, auditory comprehension was reported higher than verbal expression, as no participant was observed to produce intelligible spoken words. Intentional communication, such as pushing away or throwing an object to protest an activity, was observed and reported in more than half of the participants [5].

Braddock and others observational findings were consistent with a larger study of 50 individuals with trisomy 18 or 13. In this retrospective review of participants' developmental records, relative to motor and verbal communication skills, participants demonstrated strengths in receptive language and daily living skills [1]. Functionally, older individuals understood words and phrases, produced a few words and/or manual signs, recognized and interacted with others, and often played independently [1].

The current study extends the present body of literature in two important ways. First, it is the first known prospective study of communication ability in a larger group of individuals ascertained with trisomy 18 ($n = 17$) and trisomy 13 ($n = 15$). Second, the current study makes use of a parent report tool designed to examine participants' communicative abilities over a wide range of functions. In this way, parents are able to report on their children's communicative abilities within everyday contexts. Parent-report tools may offer an improved means by which representative data can be gathered on the complete communication profile of persons with a severe communication disability in terms of type and context for which communications routinely occur.

It is expected that parents of individuals with trisomy 18 or trisomy 13 will report that their children produce both idiosyncratic and conventional forms of communication for varied communicative functions. In line with the past studies of communication abilities in trisomy 18 and 13, it is predicted that participants will produce high levels of potential communicative acts involving eye movement, facial expression, and vocalizations. It is also predicted that participants will produce some gestures and manual signs for communication.

Results in regard to the communicative potential of persons surviving with trisomy 18 and 13 may assist families and medical practitioners as they move to make medical and ethical decisions in the individualized care plan of infants with new diagnoses [6]. The medical–ethical dilemma involves the decision to offer aggressive resuscitative measures for infants diagnosed with trisomy 18 and 13, who have an uncertain prognosis and limited life expectancy. Therefore, research into the communicative potential of persons surviving with trisomy 18 or 13 is needed as families and medical practitioners engage in active treatment planning in the early newborn period.

Methods

Procedure

Interested study participants were recruited through the Support Organization for Trisomy 18, 13 and Related Disorders (SOFT). Investigators contacted parents of persons with trisomy 18 or 13 via email or telephone. Following informed consent, parents of persons with Trisomy 18 or 13 were asked to complete two questionnaires: (1) Case history form detailing participants' general health and developmental history, and (2) the Inventory of Potential Communication Acts (IPCA) parent report measure [3, 4]. Depending on individual preferences, investigators emailed study questionnaires and asked parents to complete each response item and return by email or scheduled a convenient time with the parents to obtain and record their responses to each item by telephone.

The IPCA is an interview protocol designed for use with parents, teachers, or therapists of children who have severe communication disability. The IPCA was used to obtain information about the participants' behaviors that might be interpreted by parents of persons with trisomy 18 and 13 as forms of purposeful communication. The interview protocol included a series of 54 questions addressing 10 major communicative functions: (a) social convention; (b) attention-to-self; (c) reject/protest; (d) requesting an object; (e) requesting an action; (f) requesting information; (g) comment; (h) choice making; (i) answer; and (j) imitation.

Participants

Thirty-two individuals between the ages of 3 and 35 years participated in the study (*mean age* = 12.3 years, $SD = 8.88$; male = 10, female = 22). Within diagnostic subgroups, the study included 17 individuals with trisomy 18 (*mean age* = 13 years, $SD = 9.71$; male = 2, female = 15) and 15 individuals with trisomy 13 (*mean age* = 11.5, $SD = 8.10$; male = 8, female = 7). To be eligible for participation, parents were asked to confirm their children's medical diagnosis of trisomy 18 or trisomy 13 prior to the completion of the study questionnaires. All participants were from English-speaking background.

As part of a case history form, parents reported on each participant's hearing sensitivity, visual acuity, and oral feeding status. Reductions in hearing sensitivity were reported in 17 of 32 (53.1%) participants. Visual impairment was reported in 28 of 32 (87.5%) participants. Six of 32 (18.8%) individuals with trisomy 18 and 13 tolerated oral intake by mouth, while the remaining 26 used a percutaneous endoscopic gastrostomy (PEG) or nasogastric (NG) tube to maintain nutrition. All had positive reports of medical concerns and lengthened hospital stays following birth.

Coding

Data taken from each participant's IPCA interview protocol were summarized to generate categories of potential communicative acts interpreted by parents as serving a communicative function. The following categories of potential communicative acts were adapted from previous research in individuals with severe communication disability [3, 4]. Reported potential communicative act were classified into one of the eight mutually exclusive categories by type: (a) facial expression, or those reported behaviors such as smile or frown; (b) eye movement or instances in which the child would make eye contact or look away or toward;

(c) vocalization such as crying, whining or laughing; (d) challenging behaviors, or behaviors such as biting, hitting, destruction of items; (e) body movement, reports of full body movement, arm/hand movement, moving toward someone or an object; (f) stereotypic movement, or those acts that involved motor repetition such as arm flapping, body rocking and spinning; and (g) symbolic forms such as spontaneous productions of spoken words, gestures, augmentative and alternative communication (AAC) modes to include unaided manual signs, unaided eye pointing, aided speech generating device, and (h) imitative acts, or any reported potential communicative act produced in direct imitation of another's behavior. See Table I for an expanded listing of potential communicative acts taken directly from Sigafoos et al. [3].

Spontaneous gesture (classified under symbolic forms) was further classified by the type based on past gesture research [7, 8]. Three gesture types were classified: (a) deictic gesture, or an action used to signal out a referent from other possible ones as in pointing or showing, or in the production of a ''give-me'' gesture (i.e., empty-handed gesture with palm up with fingers curling back and forth or still), (b) conventional gesture, or emblematic gesture learned as part of one's culture and often understood by others without accompanying speech, such as waving good-bye or shaking head no, and (c) iconic gesture, or an empty-handed gesture used to depict an action or attribute of the intended referent, such as in waving arms to indicate bird in flight.

Reliability

Inter-rater agreement for potential communicative acts was assessed by having two coders independently code 15% (5 of 32 randomly selected IPCA interview protocols). Mean percent inter-rater agreement between the two trained coders was high for classifying reported potential communicative acts by category type: 96% for facial expression ($n = 12$), 95% for eye movement ($n = 10$), 93% for vocalizations ($n = 19$), 96% for body movement ($n = 112$), 80% for stereotypic movement ($n = 2$), 93% for symbolic forms ($n = 14$), 98% for imitative acts ($n = 26$). Challenging behaviors were coded at a low occurrence in the randomly selected sampling; therefore, it was not included in reliability calculations ($n = 1$).

Results

Potential communicative acts

As the measure of central tendency, the median was reported and considered in the following analyses to provide a more representative picture of the communicative abilities of persons with trisomy 18 and 13. To control for variation in parent reporting of potential communicative acts, numbers of different potential communicative acts by category were ranked from high to low for all 32 participants. Categorical ranks ranged from one (i.e., high) to eight (i.e., low). This range was used because it corresponded to eight possible IPCA categories of coded behavior. From this, a median rank value was assigned for each IPCA category of behavior. Median rank for body movement was calculated at 1.0, followed by vocalization ranked at 2.0, and facial expression ranked at 3.0. Symbolic forms, eye movement, and imitative acts IPCA categories were equally rank ordered at 4.0. Finally, stereotypic movement and challenging behaviors IPCA categories were equally ranked ordered at 5.0. See Table II for median ranks of coded IPCA categories of behavior.

Difference in symbolic forms between trisomy 18 and 13 subgroups

To examine the difference in the number of reported symbolic forms between subgroups of persons with trisomy 18 and those with trisomy 13, a Mann–Whitney non-parametric statistical test was used. Results indicated no statistically significant difference in the reported number of different symbolic forms between subgroups of persons with trisomy 18 and trisomy 13 ($U = 121.5$, $p = $ ns). Median frequency of reported symbolic forms was identical for persons with trisomy 18 ($median = 1.0$, $AD = 1.40$) and for persons with trisomy 13 ($median = 1.0$, $AD = 2.58$).

Table I. Examples of potential communicative acts taken directly from Sigafoos et al. [4].

Vocalization	Body Movement	Face/eye Movement	Breathing	Challenging Behaviors	Stereotypic Movement	Symbolic Forms
Sound/noise	Moves closer	Purses lips	Rapid	Aggression	Arms flapping	Speech
Yell/scream	Moves away	Stares	Slow	Tantrum	Hand wringing	Manual signs
Grunt	Tenses	Opens eyes	Hold	Self-injury	Body rocking	Gestures
Cry/whine	Wiggles	Closes eyes	Swallow	Destruction of items	Head weaving	Head nod
Laugh	Repositions body	Shifts eye	Sigh			Eye point
	Reaches/touches	Gazes away	Blow			Picture board
	Pushes/pulls	Gazes toward				
	Points					

Table II. Median rank order for 8 IPCA categories of potential communicative acts.

Category	Body Movement	Vocalization	Facial Expression	Symbolic Forms	Eye Movement	Imitative Acts	Stereotypic Movement	Challenging Behaviors
Median rank	1	2	3	4	4	4	5	5
Average deviation	0.53	0.73	0.83	1.09	0.92	0.91	1.03	1.18

Table III. Percent of open ended questions completed by parents for 10 IPCA categories of communicative function.

Communicative function	Percent completion of open-ended questions
Comment	0.85
Social convention	0.67
Attention-to-Self	0.62
Reject/Protest	0.62
Choice making	0.55
Answer	0.53
Requesting an object	0.41
Imitation	0.28
Requesting an action	0.28
Requesting information	0.05

IPCA percent completion by communicative function

To understand parents' patterns of reporting for IPCA communicative functions, percent of completion scores were calculated for each participant. To calculate percent completion scores, the number of completed responses was divided by the total number of IPCA open-ended questions for each of the ten communicative functions. Based on parents' responses on the IPCA interview protocol, commenting was the communicative function most frequently reported (*mean* = 85%, *range* = 67–100%), followed by social convention (*mean* = 67%, *range* = 50–100%), then by attention-to-self and reject/protest (*mean* = 62%, *range* = 25–100%, *range* = 33–100%, respectively), choice making (*mean* = 55%, *range* = 20–100%), answer (*mean* = 53%, *range* = 25–100%), and requesting an object (*mean* = 41%, *range* = 17–80%). Relative to other response categories, requesting information had the lowest mean value for category of communicative function (*mean* = 5%, *range* = 0–5%). Table III summarizes percent of open ended questions completed by parents for 10 IPCA categories of communicative function.

Symbolic forms

All symbolic forms were further reviewed given that their reported occurrence provided for a measure of participants' ability to use symbols for referential communication. Examination of symbolic forms was completed through visual inspection of IPCA parent responses and associated codes. All items coded as a symbolic form and its corresponding IPCA communicative function were listed and counted for each participant. Twenty-one of 32 parents (66%) reported that their children produced at least one symbolic form. Three broad types of symbols were reported to include, spontaneous gesture (conventional and deictic), AAC use (unaided and aided), and spoken one-word productions.

For this subgroup of individuals who were reported to use symbols ($n = 21$), the most frequently reported symbolic form was a spontaneous gesture for a head shake used to represent ''no'' (85.7%). This conventional gesture was used for the communicative functions of answering, protesting/rejecting, or choice making. Parents also reported that nine children produced a head nod gesture to represent ''yes'' for the communicative function of answering questions (43.9%). Further, about a quarter of parents reported that their children produced a clapping gesture to represent ''look at me'', ''well done'', ''yes or no'' or ''this one'' for the communicative functions of attention-to-self, commenting, answering, or requesting an object (28.6%).

Four individuals were reported to spontaneously produce deictic gesture types. For example, one individual held up her feeding tube to show that she wanted a feeding, while another put her fingers inside her mouth as if to point out ''I want food.'' Another individual patted her thigh to show that she was in need of a diaper change (i.e., requesting an action). In another example, an individual was reported to spontaneously produce a ''give-me gesture type'' with hands together palm up in front of self as if to beg for varied communicative functions (i.e., attention-to-self, comment, requesting an object, and requesting an action).

Eight of 21 (38%) ''symbol users'' were reported to use at least one manual sign for choice making, requesting an object, social convention, requesting an action, and attention-to-self. For example, participant 18-3 had learned to tap his chin to communicate ''mom'' and tap his head to communicate ''dad'' for the purpose of social convention. Only three parents reported that their children verbally produced an intelligible and consistent word form in communication (i.e., ''mama'', ''hi'', ''mom''). All words forms were reported to be produced for the communicative function of social convention. Of note, participant 13-5 was viewed as an outlier in the data set because he was reported to produce 11 different symbolic forms relative to the group mean of 2.0 different symbolic forms. Table IV provides a listing of all reported symbolic forms by type and corresponding communicative functions for reported ''symbol users'' ($n = 21$). In addition, Table IV provides examples of children's mean age in months for emergence of communicative symbolic forms based on research in typical development (TD) [9, 10]. Item-by-item comparisons may serve as a rough estimate when approximating communication developmental level (in months) for persons with trisomy 18 and 13.

Conclusions

This study described potential communicative acts in a sample of 32 individuals with trisomy 18 and trisomy 13. This is the largest prospective study describing communication ability in individuals with trisomy 18 and 13. Given the large sample size, this study is of importance because it provides families and medical practitioners with a more representative picture of the communicative abilities of persons with Trisomy 18 and 13 surviving upwards to 35 years of age. From parents' report, a recognizable pattern of communicative behavior was apparent in persons with trisomy 18 and 13 as parents were able to infer meaning from their children's largely unconventional communicative acts across varied communicative functions. Parents most consistently reported that their children with trisomy 18 or 13 made use of potential communication acts for the functions of comment, social convention, attention-to-self, and reject/protest.

Consistent with the authors' initial prediction, data showed highest median ranks for IPCA categories of behaviors classified as body movement, vocalization, and facial expression. Although motor impairment has been documented in the

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Table IV. Symbolic forms by type, communication function, and percentage of reports for all ''symbol users'' ($N = 21$).

Symbolic Forms	Type	Communicative Functions	Percent of all Symbol Users	Trisomy18-Participant # and Trisomy13-Participant #	Estimated mean age in months for Symbol Emergence (by form/type) in Typical Development (TD)
Head Shake ''No''	Spontaneous Gesture: Conventional	Answer Reject/Protest Choice Making	18/21, 85.7%	18-1, 3, 4, 6, 7, 9, 10, 12, 13, 15 13-2, 5, 6, 7, 10, 11, 13, 14	Behavior Regulation: about 13.33 months [10]
Head Nod ''Yes''	Spontaneous Gesture: Conventional	Answer	9/21, 43.9%	18-2, 3, 4, 9, 13, 15 13-2, 5, 7	Social Interaction: at about 15.96 months [10]
Clap	Spontaneous Gesture: Conventional	Attention to Self, Comment, Answer, Request Object	6/21, 28.6%	18-2, 4, 7, 8, 13, 15 13-5	Social Interaction: (excitement) 13.2 months [10]
Wave Goodbye	Spontaneous Gesture: Conventional	Social Convention	4/21, 19.0%	18-7 13-5, 7, 14	Social Interaction: (in context) 8.42 months [10]
Flip Hands	Spontaneous Gesture: Conventional	Social Convention (farewell)	1/21, 4.76%	18-9	Social Interaction: (in context) 8.42 months [10]
Hold Hands Up For ''Stop''	Spontaneous Gesture: Conventional	Choice Making	1/21, 4.76%	18-15	
Puts Fingers in Mouth (Contact point to mouth or showing feeding tube)	Spontaneous Gesture: Deictic	Request Object (something to eat or to be fed)	3/21, 14.3%	18-12 13-2, 11	Showing, giving or pointing out about 10 months [9]
Pats or shows thigh for diaper change	Spontaneous Gesture: Deictic	Request Action	1/21, 4.76%	13-5	Showing, giving or pointing out about 10 months [9]
Puts Hands Together in Front of Self As if to Beg (give-me gesture)	Spontaneous Gesture: Deictic	Attention to Self, Comment, Request Object, Request Action	1/21, 4.76	13-11	Behavioral Regulation: (reaching with open and closed hands) about 9.5 months [10]
Eye Point	AAC	Choice Making (between 2 objects)	2/21, 9.52%	18-5, 9	
Signs ''More''	AAC: Manual Sign	Request Object	2/21, 9.52%	13-2, 14	
Signs ''Food''	AAC: Manual Sign	Request Object	2/21, 9.52%	13-5, 11	
Taps Chin for Mom	AAC: Manual Sign	Social Convention	1/21, 4.76%	18-3	
Taps Head for Dad	AAC: Manual Sign	Social Convention	1/21, 4.76%	18-3	
Brings Hands Together	AAC: Manual Sign	Request Object (more)	1/21, 4.76%	13-2	
Brings Hand to Waist	AAC: Manual Sign	Request Action	1/21, 4.76%	13-2	
Signs ''Want''	AAC: Manual Sign	Request Object	1/21, 4.76%	13-5	
Signs to Change Pants	AAC: Manual Sign	Request Action	1/21, 4.76%	13-5	
Signs to Take off Shoes	AAC: Manual Sign	Request Action	1/21, 4.76%	13-5	
Signs to Get Up	AAC: Manual Sign	Attention to Self	1/21, 4.76%	13-5	
Signs ''Drink''	AAC: Manual Sign	Request Object	1/21, 4.76%	13-5	
Signs by Patting Mouth	AAC: Manual Sign	Request Object	1/21, 4.76%	18-15	
Signs with Palm to Mouth	AAC: Manual Sign	Request Object (something to eat)	1/21, 4.76%	13-14	
Signs with Back of Hand to Mouth	AAC: Manual Sign	Request Object (something to drink)	1/21, 4.76%	13-14	
Uses Tech Talk Device	AAC: Aided	Request Object, Request Action, Choice Making	1/21, 4.76%	13-14	
Says ''Mama'' or ''Mom''	Verbal: Word Production	Social Convention	2/21, 9.52%	18-3, 13-14	Joint Attention: word or word approximate to comment about 11.42 months [10]
Says ''Hi''	Verbal: Word Production	Social Convention	1/21, 4.76%	13-7	

developmental histories of individuals with trisomy 18 and 13 [1], parents appeared to pay attention and ultimately reinforced children's body movements for communicative function.

Body movement

Parents likely reported higher numbers of different body movements because they could make sense of their children's directed and/or consistent patterns of movement. For example, most parents reported that their children directly reached out to a familiar person or object for a particular communicative function. In reporting, parents appeared to pay attention to the direction of their children's movement (such as in referencing movement away from or towards a point of reference). In other reporting, parents appeared skilled in making inferences about their children's mental state based on a consistent body movement. For example, one parent reported that her child shook her leg to comment that she was ''happy.'' In cases such as this, parents appeared to make inferences about their children's mental state based on the presence of highly consistent body movements. To be meaningful, body movements were likely produced by persons with trisomy 18 and 13 in a consistent manner and observed by parents over many trials.

Further, reports of high numbers of different body movements indicated that parents were highly responsive to their children's often unconventional and/or idiosyncratic forms of communication. Within practiced and familiar routines, children's communicative acts were likely modified and shaped through parent–child interactions over time. Aside from paying attention to and reinforcing children's body movement, parents also reported that they trained children's motor hand movements in the form of manual signs or natural hand gestures.

Eye movement

Relative to body movement, vocalization, and facial expression IPCA categories of behavior, the IPCA category for eye movement was ranked slightly lower. It may be that parents reported less reliance on eye movement because fewer variations of movement can be established with the eyes alone (i.e., look toward, look up, look down, look left, look right, close/open eyes). In addition, a high percentage of children with trisomy 18 and 13 had some degree of reported visual impairment, which may have impacted upon their functional use of eye movement for potential communicative function.

Challenging behaviors

Parents rarely reported challenging behaviors as potential communicative acts despite an array of challenging behaviors that may exist in persons with severe communication disability (such as tantrums, self-injury, and aggression). Specifically, only 6 of the 32 (18.8%) participants were reported to produce challenging behaviors as a potential communicative act. Further, when parents' word choices were reviewed from the IPCA, most parents used few negative terms or referenced specific challenging behaviors in their children. Rather parents referenced more positive or neutral behaviors as judged in written IPCA responses. One parent commented when reporting vocalization as a potential communicative act, ''He makes a cute sweet sound that just melts your heart so you just come close to him naturally.'' Another parent commented on her child's positive affect, ''She is such an easy going child, I really can't say she's been angry.''

Symbolic forms

When subgroups were examined based on number of different symbolic forms, no statistically significant difference was found between trisomy 18 and trisomy 13 subgroups. This result indicated that subgroups of individuals with trisomy 18 and 13 looked more similar than dissimilar in the amount (and possible type) of symbolic forms produced.

For example, when the larger group was examined, 21 of 32 (66%) participants were reported to produce at least one type of symbolic form. When types of symbolic forms were examined (i.e., spontaneous gesture, AAC, spoken words), these data clearly show that individuals with trisomy 18 and 13 made progress towards communicative goals in the area of augmentative and alternative communication (AAC). Eight participants were reported to produce at least one manual sign. One participant made use of an AAC speech-generating device, while another two used eye pointing to select between two objects.

Augmentative and alternative communication

Based on the reported pattern of results, AAC should be targeted early in development and refined over time with the goal of maximizing the functional language and communication abilities of learners with trisomy 18 and 13. Parents should be informed that AAC is often used to supplement natural speech, and should not be viewed as an intervention which hinders the development of spoken words [11, 12].

In both naturalistic and direct teaching, parents and others should pair AAC, such as gestures and manual signs, picture boards or speech-generating devices, with spoken words to enhance children's auditory comprehension abilities. The pairing of the spoken word with AAC is of upmost importance for three reasons. First, the pairing of words and AAC will serve to enhance auditory comprehension abilities which have been found to be strength in persons with trisomy 18 and 13. Second, the pairing of words and AAC will promote verbal language skills if and when speech skills should develop in persons with trisomy 18 and 13. Third, the pairing of words and AAC will provide for a more conventional form of communication by which other caregivers (aside from parents) may communicate best with persons with trisomy 18 and 13. These data as well as past studies show support for AAC benefits in language learning [8].

Spontaneous gesture

Spontaneous gesture provided a robust measure by which to examine communication ability in persons with trisomy 18 or 13. For example, deictic showing, giving, and pointing gestures emerge in a predictable sequence starting at about

10 months of age in young children with typical development [9]. In typical development, body movement such as reaching or moving the body in a particular way to signal a communicative act decrease in frequency while deictic showing, giving and pointing gestures increase in frequency [13]. Only four individuals with trisomy 18 or 13 were reported to make use of deictic type gestures for showing, give-me gesture, and contact pointing. No participant was reported to make use of a distal pointing gesture (i.e., no reports of gesturing with index finger extended towards an out of reach referent).

No participant made use of empty-handed iconic gestures to display a pictorial quality of the intended referent. This is not surprising because iconic gesture productions are found to be tied to longer spoken utterances in children developing language typically [8]. However, conventional gesture types were observed in trisomy 18 and 13, as seen in clapping, waving good-bye, shaking head no, and nodding head yes. Some earlier developing conventional gestures may be evident in young children's communicative repertories around the onset of first words. For example, on average, young children wave in context for social interaction around 8 months, and shake their head ''no'' to protest around 13 months [10].

Spoken words

It is important to note that only three individuals with trisomy 18 or 13 were reported to produce spoken words for ''mama'', ''mom'' and ''hi.'' This finding is consistent with past studies documenting few spoken words in persons with trisomy 18 and 13 [1, 5]. One-word approximations or first words typically emerge in children at about 12 months of age [10].

Clinical implications

Information about the communication ability of persons with trisomy 18 and 13 may assist in medical decision-making. According to the study by McGraw and Perlmann, there is a shift in the treatment approach for children with trisomy 18 [6]. Neonatologists have become more likely to consider resuscitation owing to their desire to honor parents' wishes. Literature on the natural history and development of children with trisomy 18 and 13 who have survived past the first year of life and beyond may be helpful to parents experiencing a new diagnosis. This study may also assist parents as they plan for future educational and therapeutic needs of their children. This study also documented the need to consider the possibility of reduced hearing sensitivity and visual acuity when structuring communication goals for persons with trisomy 18 and 13.

Although the present study adds new information to the literature, it is limited in several ways. First, the study relies on parent report and no direct observational measures were used to validate parent reporting. Second, given the open-ended questioning style of the IPCA, coded responses were dependent on parents' vocabulary choices and descriptive abilities used to summarize their children's communicative behaviors. Third, because parents required differing levels of support to complete the IPCA, investigators offered parents two methods by which to submit their IPCA responses. It is unclear if parents' responses would vary given IPCA completion using a supported phone interview with the investigator in comparison to a self-completed email format.

In sum, parent report indicated that persons with trisomy 18 and 13 produced highest median ranks for body movement, vocalization, and facial expression. Symbolic forms followed in rank as spontaneous gestures and manual signs were reported in the communication repertoires of more than half of the participants. Only three participants were reported to produce spoken words. The described pattern of results provides a strong rationale for AAC intervention. More information about the developmental progression of individuals with trisomy 18 and 13 over their lifetime is needed. Future research is planned in conjunction with families who care for individuals with trisomy 18 and 13 and their support organizations.

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Declaration of interest

The authors report no conflicts of interest. The authors alone are responsible for the content and writing of this paper.

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References

1. Baty BJ, Jorde LB, Blackburn BL, Carey JC. Natural history of trisomy 18 and trisomy 13: II. Psychomotor development. American Journal of Medical Genetics 1994;49:189–184.
2. Carey JC. Perspectives on the care and management of infants with trisomy 18 and 13: Striving for balance. Current Opinion in Pediatrics 2012;24:672–678.
3. Sigafoos J, Arthur-Kelly M, Butterfield N. Inventory of potential communicative acts. Enhancing everyday communications for children with disabilities. Baltimore, MD: Paul H Brookes Publishing Co; 2006.
4. Sigafoos J, Woodyatt G, Deen D, Tait K, Tucker M, Roberts-Pennell D, Pittendreigh N. Identifying potential communicative acts in children with developmental and physical disabilities. Communication Disorders Quarterly 2000;21:77–86.
5. Braddock B, McDaniel J, Spragge S, Loncke F, Braddock SR, Carey JC. Communication ability in persons with trisomy 18 and trisomy 13. Augmentative Alternative Communication 2012;28: 266–277.
6. McGraw MP, Perlman JM. Attitudes of neonatologists toward delivery room management of confirmed trisomy 18: Potential factors influencing a changing dynamic. Pediatrics 2008;121: 1106–1110.
7. McNeill D. Hand and mind. Chicago: University of Chicago Press; 1999.
8. Nicoladis E, Mayberry RI, Genesee F. Gesture and early bilingual development. Developmental Psychology 1999;35: 514–526.
9. Bates E, Camaioni L, Volterra V. The acquisition of performatives prior to speech. Merrill-Palmer Quarterly 1975;21:205–226.

10. Crais E, Douglas DD, Campbell CC. The intersection of the development of gestures and intentionality. Journal of Speech, Language and Hearing Research 2004;47:678–694.
11. Lloyd LL, Fuller DR, Arvidson HH. Augmentative and alternative communication intervention on speech production of individuals with developmental disabilities: A research review. Journal of Speech, Language, and Hearing Research 1997;49:248–264.
12. Romski MA, Sevcik RA. Augmentative communication and early intervention: Myths and realities. Infants and Young Children 2005; 18:74–185.
13. Blake J, Dolgoy SJ. Gestural development and its relation to cognition during the transition to language. Journal of Nonverbal Behavior 1993;17:87F–102F.