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Developmental Status of 22 Children with Trisomy 18 and Eight Children with Trisomy 13: Implications and Recommendations

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Trisomy 18 and trisomy 13 are conditions often referred to as "incompatible with life" or "lethal anomalies." If there is long-term survival, the outlook is considered "grim." Developmental status is presumed to be minimal. Yet, Baty et al. [1994; 49:189–194] described a variety of developmental skills in their sample. An additional 22 children with trisomy 18 and eight with trisomy 13 are described here. A range of developmental skills is noted with strengths in the language and communication, gross and fine motor and social-emotional domains including indicating preferences, exploration of objects and a range of voluntary mobility. These results serve to expand the knowledge base on developmental status for these groups and advance the need to further explore developmental abilities rather than focus on deficits. Avenues for future research, implications, and recommendations are provided. © 2015 Wiley Periodicals, Inc.

Key words: trisomy 18; trisomy 13; developmental status; developmental skills

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INTRODUCTION

Trisomy 18 and trisomy 13 are conditions often referred to as "incompatible with life" or "lethal anomalies." Expectant parents and parents with newborns with a post-birth diagnosis are told to provide minimal medical interventions and, if the newborn survives, to expect a limited quality of life [Leuthner, 2004; Breeze et al., 2007; Catlin, 2010; Chervanek and McCullough, 2012; Merritt et al., 2012]. Some authors are more optimistic but caution about a brief and, potentially, grim outcomes for live born cases [Koogler et al., 2003; Wilkinson, 2010; Derrington and Dworetz, 2011; Lakovschek et al., 2011; Breeze and Lees, 2013]. Empirical studies verify these viewpoints with data from medical professionals such as neonatologists describing limited outcomes of medical interventions [McGraw and Perlman, 2008; Heuser et al., 2012]. In addition, the pessimistic outlook extends to developmental status with information provided that minimal to no discernable skills such as social smile and reaching for objects will be attained. For example, Jones et al. [2013] state "…mental deficiency is severe…" A number of authors offer a more hopeful perspective emphasizing the need for parents to make informed decisions based on the needs of their newborns and children and not place limits on their potential [Koogler et al., 2003; Carey, 2012; Lorenz and Hardart, 2014]. In addition, Fenton [2011] states "We perform ethical analyses assuming we truly understand how to apply beneficence or maleficence to a child with trisomy 18 or 13 or any other disorder in which there may be profound disability. As if the child can tell us what he or she is feeling. But we can do our best to assess and treat pain and discomfort. Smiles and laughter need no score pad. We know what they mean."

Few studies have investigated developmental status in cases with trisomy 18 and trisomy 13. Primary among them is Baty et al. [1994]. The authors reviewed data for 50 persons with trisomy 18 aged one to 232 months and 12 with trisomy 13 aged one to 130 months. Receptive language was identified as an area in which a variety of skills were demonstrated. Results indicated limited progress in expressive communication and motor development. Specifically, skills including the following were noted: understands words and phrases, follows simple commands, sits independently, uses a walker, recognizes, and interacts with others. The authors state "it became clear that many parents resented the early message that their child would never interact with his or her environment and family... Many families with surviving trisomy 18 or trisomy 13

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children think that the information they were given was more discouraging than necessary, and ignored the humanity of their child. It is important to parents that the accomplishments of their children are acknowledged by the medical community." It is also of note that the Baty sample was largely over one year of age (up to 130 months).

Additional studies include information about developmental status for cases with trisomy 18. Kosho et al. [2013] described 24 individuals. Most displayed social smiling, head control and reach and grasp skills. Though not focused on developmental outcomes, Kosho et al. [2006] included mention of several patients demonstrating head control, rolling over and smiling. As noted above in Baty et al. [1994] and by Barnes and Carey [2014], skill acquisition is present regardless of amount of delay in comparison to same age peers. Finally, Cereda and Carey [2012] explain, "Thus, children with trisomy 18, while showing marked developmental and cognitive disability have many more abilities than usually perceived in the stereotype and prior portrayals of the condition."

Recent investigations have focused on communication development. Braddock et al. [2012] and Liang et al. [2013] describe samples with mean ages of approximately 16 and 13 years, respectively. Parent report and observations of responses to communication temptation tasks were analyzed. Again, receptive language was more advanced than expressive language development. Based on 10 cases (n = 8 trisomy 18, n = 2 trisomy 13), the first study noted the use of eye gaze, hand gesture, open vowel sounds, and facial expressions to communicate. The second group (n = 32; 17 trisomy 18 and 15 trisomy 13) was reported to exhibit the following skills: attention-to-self, requesting an object, choice making and reject/protest. Over 60% were reported to produce at least one word, gesture or related communication form from an augmentative device. Conclusions highlighted the communication potential of children often judged to be incapable of developmental progress. Lastly, Janvier et al. [2012] describe developmental milestones in 64 individuals. Skills included laughing, pointing at objects, rolling, and sitting. Also, "…parents overwhelmingly described surviving children as happy and stated that they were able to communicate with them to understand their needs."

The purpose of the present study is to further describe surviving persons with trisomy 18 and trisomy 13 between 1 and 5 years of age. Specifically, data describing developmental status will be shared for samples diagnosed with the full type of these conditions in order to offer additional information on strengths and abilities for these children.

METHODS

The Tracking Rare Incidence Syndromes (TRIS) project collects data focusing on the prenatal and neonatal periods as well as perinatal and longitudinal medical needs and interventions. An additional component of the TRIS Survey is gathering developmental information at the time of baseline survey completion.

Instrumentation

The Full TRIS Survey is completed by parents with a child two months of age or older (child does not have to be living at time of survey completion; Modified TRIS Survey is for children living 60 days or less or stillborn). The survey was piloted in 2005–2006 and first available online in February 2007 at http://web3.coehs.siu.edu/TRIS/ Paper copies are provided by request. Completed surveys are returned by postal mail and manually entered into the project's database.

TRIS Full Survey is composed of three parts. Part I includes 15 items on labor and delivery and suspected and presenting medical conditions. An additional 10 items focus on the post-birth course and hospital discharge. There are also seven demographic items (e.g., marital status, education level). Part II examines family support (56 items). Part III collects data related to a range of health and medical interventions including surgeries and medications (61 items).

In addition, Part III includes the TRIS Developmental Matrix. The matrix lists developmental milestones from one month to five years of age across domains (cognitive, language and communication (expressive and receptive), fine motor, gross motor, social-emotional, and activities of daily living). Parents are requested to note the presence of each skill listed on the matrix along with the age at acquisition and additional comments as needed (e.g., use of a device or piece of equipment to demonstrate a skill).

The list of skills was informed by guidelines from the American Academy of Pediatrics (see http://www.healthychildren.org/English/ages-stages/Pages/default.aspx). From 1 to 24 months, skills are grouped in three to six month increments. Skills linked to age three, four and five are grouped by year. It should be noted that the number of skills varies by developmental domain such as existence of a greater number for language and communication compared with activities of daily living. This disparity is particularly evident in skills from birth to seven months. In addition, the skills included in the TRIS Developmental Matrix are those most likely to be acquired for cases with full trisomy 18 and trisomy 13. For example, few items were included in the gross motor domain beyond walking unassisted.

Procedure

Approval was received from the Human Subjects Committee at Southern Illinois University Carbondale prior to the start of data collection.

Participants were and continue to be recruited through announcements to rare trisomy-related websites, listservs and via articles in the Support Organization For Trisomy 18, 13 and related disorders (SOFT) newsletter. In addition, a Facebook page was launched in 2011 (https://www.facebook.com/TRIS.Trisomy.project). A series of entries about the project were posted to the Global Genes project's blog in 2013 (see http://globalgenes.org/raredaily/giving-a-face-to-trisomy-18-debbie-elaborates-on-this-rare-condition/ for an example). Contact among project participants is another avenue for recruitment (e.g., Facebook groups, attendance at SOFT Conference).

Participants complete a pre-enrollment form located at http://web3.coehs.siu.edu/tris/PreEnroll.php with name, phone number, state/province and country, e-mail address and child's name, date of birth (and death, if appropriate) and trisomy type. Within 24–48 hr, a unique login and password to access the appropriate survey

instrument is sent to the email address provided on the form. This log in and password is used for the duration of project participation. In addition, the server housing this data is updated and archived daily.

TRIS project staff is able to view participant progress since the survey can be accessed multiple times on the TRIS project website prior to completion (participants are minded to save additional data prior to leaving the survey). An initial email reminder is sent when a survey is not completed within one month of enrollment. Additional reminders are sent every two to three months. At survey completion, TRIS project staff receives a completion notice by e-mail.

Data Analysis

Upon receipt of a survey completion notice, demographic data are downloaded and entered into SPSS [2008].

The author visually inspected developmental status data. Each child's developmental range was determined by identifying a basal level (month level a child was reported to attain majority of skills) and a ceiling level (month level with attainment of two or less skills). The resulting values provide a developmental range such as 3–9 months. In addition, developmental skills were recognized even when an adaptation or modification was necessary. For example, expressive language included gestures, signs and/or use of an augmentative device as well as spoken language. Independent use of a gait trainer or walker was accepted for "Walks unassisted."

Participants

For the children with trisomy 18, between February 1, 2007 and March 7, 2012, 22 surveys were completed for cases with full t18 between 12 and 59 months of age (n = 225, 9.8%). At the time of survey completion, all were living. Mean age was 29.9 months (SD ± 17.41 months; range 13–58 months). Mean maternal age at the time of pregnancy was 31.6 years (SD ± 6.27 years; range 19–40 years). Mean paternal age at the time of pregnancy was 33.1 years (SD ± 6.53 years; range 19–45 years). The participant who did not provide this information adopted her child.

The majority of mothers were married (n. 17, 77.3%). Mother's education level varied from 7 years of formal schooling to more than 20 years. The majority of mothers resided in the United States (n = 18, 81.8%) and reported medium income level (n = 18, 81.8%).

Between February 2007 and August 2013, nine surveys were completed for children with full trisomy 13 between 12 and 59 months of age (n = 273, 3.3%). Mean age was 35 months (SD ± 10.70 months; range 15–53 months). Mean maternal age at the time of pregnancy was 32.5 years (SD ± 4.10 years; range 27–38 years). Mean paternal age at the time of pregnancy was 32.63 years (SD ± 4.63 years; range 26–39 years). As with the trisomy 18 sample, the majority of mothers were married (n = 7, 78%) and education level varied from seven years of formal schooling to more than 20 years. All participants resided in the United States at the time of survey completion.

The samples are further described in Bruns and Campbell [2014a, 2014b].

RESULTS

Developmental status for 22 children with trisomy 18 and eight with trisomy 13 (one participant did not complete the TRIS Developmental Matrix) are described. The descriptions below indicate skills demonstrated at the time of survey completion (refer to basal and ceiling levels described above). As such, previously acquired skills are not included. For example, rolling back to stomach or stomach to back (seven month skill) is identified as a case's most advanced skill. An earlier skills, kicking legs (three month skill), is also marked as acquired.

Trisomy 18 Sample

The sample with trisomy 18 included 22 children. The range in developmental status is primarily between 3–12 months for children aged 9–57 months at survey completion.

Approximately half were able to turn head toward voice and sounds, demonstrate preferences for toys/objects and look at pictures in books in the cognitive domain. Indicating needs by vocalizing was evident in 13 (59%) at survey completion with several children exhibiting more advanced language and communication skills including babbling consonant-vowel combinations and responds to "no."

Fine and gross motor skills were delayed but evident. Exploration of objects with hands and mouth was prevalent in this group. The ability to swipe at, grasp or shake objects was identified in six individuals (27%). Approximately a quarter of the group were noted to reach, grasp and transfer hand held items. Patients 5 and 21 displayed a pincer grasp to hold objects. Twelve (55%) were noted to kick legs and nine (41%) to roll back to stomach or stomach to back. Each child demonstrated at least one of the following social-emotional skills: coos or social smile to family member or caregiver, responds to social games, and engages in social play. Ten (45%) displayed purposeful actions relating to this developmental domain such as indicating preference for their main caregiver.

Activities of daily living such as attempting to assist or assisting with dressing were noted for Patients 1, 5, and 13. In addition, Patients 12 and 18 imitated household tasks such as sweeping with a broom and were reported to walk unassisted. Patient 12's parent shared that testing after completion of the survey indicated mosaicism. This may be the circumstance with Patient 18 but, to date, no testing has been completed.

Trisomy 13 Sample

The sample with trisomy 13 (n = 8; one parent did not complete the TRIS Developmental Matrix) provides some similarities to data described above for the t18 group. The range in developmental status is primarily between 3–9 months for children aged 15–53 months at survey completion.

All demonstrated exploration of objects and voluntary mobility including kicking legs, sitting without support and rolling. Demonstrating preferences was also identified in six individuals (75%). Children also showed evidence of imitation and production of open vowel sounds. Patients 7 and 9 offer representative examples

of these skills. Patient 1 demonstrated the highest level of skills (12–18 month range) including imitating simple gestures, scribbling and taking steps with assistance. Notably, Patient 8 demonstrated a pincer grasp and Patients 4 and 5 produced consonant-vowel combinations.

Activities of daily living such as attempting to assist with dressing and self-feeding were not exhibited.

See Tables I and II for detailed developmental status data for all trisomy 18 and trisomy 13 cases.

DISCUSSION

The data provided here offer a representation of developmental status in 22 children with trisomy 18 and eight with trisomy 13. The majority in both groups demonstrated skills in the 3–9 month range including indicates needs by vocalizing, rolls back to stomach or stomach to back, and engages in social play. Social-emotional development was a consistently identified developmental strength. Fine and gross motor skills were limited but a number of key skills were evident including exploration of objects with hands and sitting without support. The majority in both groups did not exhibit any skills in the domain of activities of daily living (e.g., self-feeding, assisting with dressing).

Individual variations were present in both groups with children only displaying skills below the three month level and a few with trisomy 18 with the most advanced developmental status with skills up to 48 months.

Confirming Previous Literature

Data confirm and extend the available literature but it is necessary to point out that the few existing studies describe older samples or focus on only one developmental area. For example, Baty et al. [1994] samples included 50 cases with trisomy 18 aged one to 232 months and 12 cases with trisomy 13 aged one to 130 months and Braddock et al. [2012] exclusively discusses communication development. The results described here are of younger children (below 60 months of age) and include all developmental areas.

Baty et al. [1994] reported a number of skills similar to the groups described here such as reaching for objects, rolling over, and sitting alone. The authors also note higher developmental age in social skills and activities of daily living. The latter was not found in the groups described here. It is unclear if this discrepancy is due to the younger chronological age of these cases and/or additional factors. Kosho et al. [2013] described similar skill development in 24 cases with trisomy 18 and commented "…continued to learn, although they show severe to profound developmental disability. Again, different from the individuals reported here, many were over 10 years of age. It is encouraging that some younger children with these conditions are gaining these developmental milestones before 60 months of age."

Communication in persons with trisomy 18 and trisomy 13 was the focus in Braddock et al. [2012] and Liang et al. [2013]. Once again, most were older and the Braddock et al. sample was small ($n = 10$; eight with trisomy 18 and two with trisomy 13). Liang et al. [2013] discussed 32 cases. Over half of parents reported their child (up to age 35) was able to produce one word, gesture or augmentative and alternative communication (AAC) form (device). The groups described here primarily produced open vowel and consonant-vowel combinations. Five children with trisomy 18 and two cases with t13 were noted to attempt to or were able to imitate simple gestures such as waving goodbye (12 month skills). AAC use was not included in the TRIS Developmental Matrix. A parent would need to provide that information in the comments section.

Limitations

A number of limitations potentially affected the results. First, data are collected through a parent-completed survey. Medical records (e.g., karyotype report) and other documentation such as results from developmental testing is not requested to be submitted for review. This was done to facilitate participation and, due to the international scope of the project, it would be necessary to locate volunteers or hire translators fluent in a parent's native language and knowledgeable of medical and developmental terminology in order to analyze the corresponding documentation. Accuracy of a parent-completed survey is also a concern whether parents are inflating or minimizing their child's developmental skills. Interestingly, the author has first-hand, anecdotal knowledge of this from conversations with parents. It is unclear if this is due to limited knowledge of development and/or the low expectations placed on their children.

Second, the TRIS Developmental Matrix requests the age at acquisition of each skill but this information was only provided by a few parents in each group and, thus, not included here. In addition, additional comments regarding use of a device or piece of equipment to demonstrate a skill, for example, were, for the most part, not supplied. A solution would be to contact parents after survey submission to complete these items. Unfortunately, due to staffing constraints, this is not feasible. The TRIS project staff is comprised of the author, one to two student assistants depending on university funding and two support staff to manage the database and project website.

The decision to not include all developmental skills to 60 months on the TRIS Developmental Matrix may have also impacted the results. Specifically, items were selected for likelihood of acquisition and brevity. Regarding the latter, the matrix is at the end of the TRIS Survey, which can take up to several hours to complete. To offset the possibility of parents not completing this last set of items, less were included. It was also taken into consideration that documenting their child's skills may cause distress. While all demonstrated skills should be cause for celebration, confirming their child's delays and deficits could be emotionally difficult.

The persons described here may not be representative of all children with these genetic conditions. Some of the advanced developmental skills described here may not reflect the majority of cases but can provide hope. This data also reinforces the need to examine each case as an individual rather than based on the diagnosis alone.

In addition, one child with trisomy 18 (Patient 12) was confirmed as having mosaicism and another suspected (Patient 18) after survey completion. Results indicated more advanced developmental status compared with the remaining 20 cases. As such, the developmental status of these two cases is skewed in comparison to the remaining 20 (12–48 months for both children).

TABLE I. Developmental Status of Trisomy 18 Cases (n = 22)

	Age at completion of baseline survey in months	Developmental status[a]	Cognitive development	Language and communication development
1	48	7–18 months	Identifies 3–5 body parts	Babbles consonant-vowel combinations
			Demonstrates preference for toy/object (e.g., reaches, vocalizes)[b]	Imitates simple gestures (e.g., waves bye, blows kiss)
2	56	3–9 months	Follows moving objects with eyes	Produces open vowel sounds (e.g., "ah", "ow")
			Turns head toward voice or sounds	Indicates needs by vocalizing
3	17 (age at passing)	3–12 months	Demonstrates preference for toy/object	Produces open vowel sounds
			Looks at pictures in books	Indicates needs by vocalizing
4	58	3–12 months	Responds to "no"	Babbles consonant-vowel combinations
			Demonstrates preference for toy/object	Indicates needs by vocalizing
5	57	7–12 months	Finds partially hidden toy	Indicates needs by vocalizing
			Demonstrates preference for toy/object	Imitates simple speech sounds or words (e.g., "mama")
6	16	3–12 months	Turns head toward voice or sounds	Produces open vowel sounds
			Looks at pictures in books	Indicates needs by vocalizing
7	9 (age at passing)	3–9 months	Turns head toward voice or sounds	Indicates needs by vocalizing
			Interacts with mirror image	
8	14 (age at passing)	1–3 months	Turns eyes toward face	n/a
			Watches face for at least 10 sec	
9	11 (age at passing)	1–3 months	Follows moving object with eyes	n/a
			Turns head toward voice or sounds	
10	20	3–12 months	Looks at pictures in books	Imitates simple speech sounds or words
			Demonstrates preference for toy/object	Attempts to imitate simple gestures, signs and/or words (e.g., waves goodbye, "mama")
11	13	3–12 months	Interacts with mirror image	Indicates needs by vocalizing
			Looks at pictures in books	
12	56[c]	12–48 months	Retells/signs nursery rhymes or children's songs	Follows two step directions
			Identifies primary colors	Says, signs or gestures "no" with intent
13	42	7–12 months	Looks at pictures in books	Babbles consonant-vowel combinations
			Demonstrates preference for toy/object	Attempts to imitate simple gestures, signs and/or words
14	13	3–12 months	Interacts with mirror image	Produces open vowel sounds
			Looks at pictures in books	Indicates needs by vocalizing
15	23	3–7 months	Follows moving object with eyes	Produces open vowel sounds
			Turns head toward voice or sounds	
16	14	3–7 months	Follows moving object with eyes	Produces open vowel sounds
			Turns head toward voice or sounds	Indicates needs by vocalizing
17	20	3–9 months	Turns head toward voice or sounds	Indicates needs by vocalizing
			Looks at pictures in books	
18	47[d]	12–48 months	Associates name with object (e.g., picture of ball with a ball)	Imitates simple gestures
			Identifies self in photo by pointing, saying name etc.	Follows two step directions
19	13	1–9 months	Turns head toward voice or sounds	Indicates needs by vocalizing
			Looks at pictures in books	
20	31	3–12 months	Demonstrates preference	Responds to "no"

(*Continued*)

TABLE I. (*Continued*)

	Age at completion of baseline survey in months	Developmental status[a]	Cognitive development	Language and communication development
			for toy/object	
			Associates name with object	Attempts to imitate simple gestures, signs and/or words
21	45	3–12 months	Turns head toward voice or sounds	Indicates needs by vocalizing
			Demonstrates preference for toy/object	
22	19	3–12 months	Turns head toward voice or sounds	Indicates needs by vocalizing
			Demonstrates preference for toy/object	Responds to "no"

	Fine motor development	Gross motor development	Social-emotional development
1	Uses hands to explore objects	Sits without support	Responds to social games
	Uses hands to build with blocks and other manipulatives	Stands for at least 5 sec (positioned against wall)	Prefers main caregiver
2	Reaches, grasps and transfers hand held items	Kicks legs	Engages in social play
	Uses raking motion on textured items (e.g., blankets, toys)	Rolls back to stomach or stomach to back	Shows preference for familiar adults
3	Reaches, grasps and transfers hand held items	Kicks legs	Responds to social games
	Uses hands to explore objects	Rolls back to stomach or stomach to back	Prefers main caregiver
4	Uses hands to explore objects	Rolls back to stomach or stomach to back	Responds to social games
	Reaches, grasps and transfers hand held items	Sits without support	Shows preference for familiar adults
5	Uses pincer grasp to hold objects	Crawls on stomach	Responds to social games
	Uses hands to explore objects	Stands for at least 5 sec (with adult support)	Prefers main caregiver
6	Swipes at, grasps or shakes small toy	Kicks legs	Engages in social play
	Explores objects with hands and mouth	Rolls back to stomach or stomach to back	Prefers main caregiver
7	Swipes at, grasps or shakes small toy	Kicks legs	Responds to social games
	Explores objects with hands and mouth	Rolls back to stomach or stomach to back	Shows preference for familiar adults
8	Brings one or both hands to mouth	Kicks legs	Coos or social smile to family member or caregiver
9	Explores objects with hands and mouth	Raises head and chest when lying on stomach	Coos or social smile to family member or caregiver
	Swipes, grasps or shakes small toy	Kicks legs	
10	Uses hands to explore objects	Kicks legs	Responds to social games
		Rolls back to stomach or stomach to back	Prefers main caregiver
11	Swipes at, grasps or shakes small toy	Kicks legs	Responds to social games
	Uses hands to explore objects	Rolls back to stomach or stomach to back	Prefers main caregiver
12	Builds a tower with blocks or similar construction toy	Walks unassisted	Prefers main caregiver
	Scribbles with pencil or crayon	Climbs (e.g., on and off couch)	
		Catches and throws a ball with two hands	
13	Reaches, grasps and transfers hand held items	Transfers to sitting position	Responds to social games
	Uses hands to explore objects	Takes steps with assistance	Shows preference for familiar adults

TABLE I. (*Continued*)

	Fine motor development	Gross motor development	Social-emotional development
14	Swipes at, grasps or shakes small toy	Kicks legs	Responds to social games
	Explores objects with hands and mouth	Sits without support	Prefers main caregiver
15	Brings one or both hands to mouth	Kicks legs	Coos or social smile to family member or caregiver
	Swipes at, grasps or shakes small toy	Rolls back to stomach or stomach to back	
16	Brings one or both hands to mouth	Kicks legs	Coos or social smile to family member or caregiver
	Swipes at, grasps or shakes small toy	Rolls back to stomach or stomach to back	
17	Swipes at, grasps or shakes small toy	Kicks legs	Coos or social smile to family member or caregiver
	Reaches, grasps and transfers hand held items		
18	Builds a tower with blocks or similar construction toy	Walks unassisted	Says, signs or gestures "no" to parent or caregiver (testing limits)
	Scribbles with pencil or crayon	Climbs	Plays cooperatively with siblings
		Catches and throws a ball using two hands	
19	Brings one or both hands to mouth	Kicks legs	Responds to social games
	Swipes at, grasps or shakes small toy		Shows preference for familiar adults
20	Uses pincer grasp to hold objects	Kicks legs	Responds to social games
	Uses hands to explore objects	Rolls back to stomach or stomach to back	Prefers main caregiver
21	Explores objects with hands and mouth	Stands for at least 5 sec	Engages in social play
	Uses pincer grasp to hold objects	Takes steps with assistance	
22	Reaches, grasps and transfers hand held items	Kicks legs	Engages in social play
	Uses hands to explore objects	Rolls back to stomach or stomach to back	Prefers main caregiver

[a]Represents scatter skills including multiple skills at lower and upper end of range.
[b]Examples provided.
[c]Confirmed mosaic trisomy 18 after completion of baseline survey.
[d]Mosaicism suspected.

Future Research

Efforts should be made to collect developmental data over time to create a fuller representation of developmental progress in cases with trisomy 18 and trisomy 13. The TRIS Follow-up Survey is completed once a year. A question is included about developmental progress but, again, project staff is unable to make the necessary email contacts and phone calls to collect this data. It is hoped that future funding will allow the addition of another research assistant to focus on this component of data collection and analysis. To provide perspective, the author and project research assistant completed two to four contacts with parents to complete each of 12 case studies during 2014 (see the completed case studies at http://www.coehs.siu.edu/tris/casestudies.html).

There is also a need to collect age of acquisition and adaptation data to confirm Baty et al. [1994] and provide additional information for medical professionals and parents about possible trajectories for development. Use of AAC devices (e.g., switches, iPad apps) and mobility equipment such as gait trainers should also be documented. Speech and language pathologists, occupational therapies and physical therapists providing services for cases with trisomy 18 and trisomy 13 can provide data to this end (see Braddock et al. [2012] and Liang et al. [2013]). Early intervention (birth to three) providers and school-based professionals can also support this effort through outreach and support from parents.

In addition to the data reported here, developmental status results have been reported for cases with trisomy 9 mosaic [Bruns,

TABLE II. Developmental status of trisomy 13 cases (n = 8[a])

	Age at completion of baseline survey in months	Developmental status[b]	Cognitive development	Language and communication development	Fine motor development	Gross motor development	Social-emotional development
1	37	12–18 months	Responds to "no"	Imitates simple speech sounds (e.g., mama)[c]	Uses hands to explore objects	Takes steps with assistance	Shows preference for familiar adults
			Demonstrates preference for toy/object	Imitates simple gestures (e.g., waves bye, blows kiss)	Scribbles	Attempts to throw a ball	Prefers main caregiver
2	42	7–12 months	Responds to "no"	Indicates needs by vocalizing	Explores objects with mouth	Cruises along furniture	Engages in social play
			Finds partially hidden toy	Imitates simple gestures	Uses hands to explore objects	Takes steps with assistance	Shows preference for familiar adults
4	33	3–9 months	Finds partially hidden toy	Produces open vowel sounds (e.g., "ah", "ow")	Swipes or shakes objects	Transfers to sitting position	Shows preference for familiar adults
			Demonstrates preference for toy/object	Babbles consonant-vowel combinations	Uses hands to explore objects	Commando crawls	Prefers main caregiver
5	24	7–9 months	Finds partially hidden toy	Babbles consonant-vowel combinations	Reaches, grasps and transfers hand held items	Sits without support	Shows preference for familiar adults
			Demonstrates preference for toy/object	Imitates simple speech sounds	Uses hands to explore objects	Commando crawls	Prefers main caregiver
6	36	3–9 months	Responds to "no"	Indicates needs by vocalizing	Reaches, grasps and transfers hand held items	Kicks legs	Responds to social games (e.g., peekaboo)
			Looks at pictures in books	Imitates simple speech sounds	Uses hands to explore objects	Rolls back to stomach or stomach to back	Prefers main caregiver
7	53	3–9 months	Watches face for at least 10 sec	Produces open vowel sounds	Reaches, grasps and transfers hand held items	Kicks legs	Social smile to family members
			Demonstrates preference for toy/object	Indicates needs by vocalizing	Uses hands to explore objects	Turns head toward voice or sounds	Prefers main caregiver
8	38 (age at passing)	3–12 months	Looks at pictures in books	Produces open vowel sounds	Uses pincer grasp to hold objects	Rolls back to stomach or stomach to back	Responds to social games
			Demonstrates preference for toy/object	Imitates simple gestures	Uses hands to explore objects	Takes steps with assistance	Prefers main caregiver
9	15	3–9 months	Watches face for at least 10 sec	Produces open vowel sounds	Uses raking motion on textured items (e.g., blanket, touch and feel book)	Kicks legs	Shows preference for familiar adults
			Demonstrates preference for toy/object	Indicates needs by vocalizing	Uses hands to explore objects	Turns head toward voice or sounds	Prefers main caregiver

[a]One parent did not provide developmental status data for her child (#3; 37 months of age at completion of baseline survey).
[b]Represents scatter skills including multiple skills at lower and upper end of range.
[c]Examples provided.

2011;]. Similar studies should be undertaken for cases with trisomy 18 mosaicism and trisomy 13 mosaicism.

Implications and Recommendations

A number of implications from the results presented here must be noted. First, documentation of developmental status can be utilized to contribute to decision making for interventions. The interventions may be medical such as surgery to assist with assisted or independent mobility or therapeutic such as initiation or continuation of speech therapy. All too often the "incompatible with life" label is used to make decisions rather than assessing medical needs and also examining developmental status.

The language and communication skills described here also highlight the importance of associated auditory acuity skills. In turn, medical professionals are encouraged to include checks of hearing to routine care and management. The same can be recommended for vision for acquisition of fine motor skills.

The children described here were below 60 months of age in contrast to the older samples described in Baty et al. [1994] and Liang et al. [2013]. While developmental status of older persons can inform decision-making, it is this author's proposal that information provided in Tables I and II should be shared with medical professionals, parents and caregivers in order to encourage discussion of developmental status as part of overall care and management. Further, acquisition of skills at younger ages is important to share with these audiences to promote a more favorable outlook in general and toward interventions.

CONCLUSION

As indicated by results here, a range of developmental skills are attainable for children with trisomy 18 and trisomy 13. This is in contrast to the "incompatible with life" and negative views on survivors. There is a need to further explore developmental abilities to inform both medical professionals and parents involved in their care.

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