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Nine Children Over the Age of One Year with Full Trisomy 13: A Case Series Describing Medical Conditions†

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Trisomy 13 (Patau syndrome), identified by Patau and colleagues [1960; Lancet 1: 790–793] is the third most common autosomal condition. Population studies indicate less than one in 10 children reaches their first birthday. In the face of mixed findings and recommendations for treatment, additional research is needed to further determine what contributes to longevity and implications for treatment for presenting medical conditions. The purpose of the present study is to report on presenting medical conditions and the presence or absence of the specific conditions (age at survey completion). Data on nine survivors (seven female, two male) with trisomy 13 indicated mean gestational age of approximately 36 weeks, birth weight ranging from 1100 to 3290 g and mean length of 45.3 cm. Length of hospital stay after birth varied. The majority of infants presented with well-known physical characteristics. Medical conditions and their treatment varied at birth and at survey completion. Notably, several infants' cardiac anomalies resolved without surgical intervention. Surgeries were provided for a range of conditions including gastrostomy tube placement to address feeding issues and removal of intestinal blockage. There were no reports of holoprosencephaly. Implications and recommendations are provided.

Key words: trisomy 13; medical intervention; cardiac conditions; surgery; Patau syndrome

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INTRODUCTION

Trisomy 13 (Patau syndrome), identified by Patau et al. [1960] is the third most common autosomal condition and occurs in one in 6,000–10,000 live births. In addition, Crider et al. [2008] and Irving et al. [2011] describe a rise in pregnancies with trisomy 13 coupled with lower prevalence rate due to prenatal diagnosis and terminations. For live births, survival is often presented in days or week increments [Brewer et al., 2002; Rasmussen et al., 2003; Lin et al., 2007; Crider et al., 2008; Vendola et al., 2010]. The population indicates less than one in 10 children reach their first birthday [Irving et al., 2011; Lakovschek et al., 2011; Sibidue et al., 2011; Wu et al., 2013].

Common physical characteristics include cleft lip and/or palate, microphthalmia, microcephaly, and postaxial polydactyly. [Lin et al., 2007; Petry et al., 2013]. Many infants also present with cardiac anomalies including patent ductus arteriosus (PDA) or ventricular septal defect (VSD), respiratory difficulties including central apnea and upper airway problems and feeding difficulties [Rios et al., 2004; Pont et al., 2006; Hsu and Hou, 2007; Jones et al., 2013]. In addition, Lakovschek et al. [2011] and Morris and Savva [2008] discuss the high incidence of fetal loss after prenatal diagnosis of trisomy 13.

Tsukada et al. [2012] offer improved outcome with intervention. Median survival for 16 patients was 733 days with 31% (n = 5) living more than three years. A majority received mechanical ventilation (n = 9) and six had surgical intervention. Graham et al. [2004] also report on successful cardiac surgery in their sample, which included 11 children with trisomy 13. Additional reports of better outcome are available [Hsu and Hou, 2007; Kaneko et al., 2008; Maeda et al., 2011]. There are also recent United States data that point to the increased delivery of surgical interventions and other hospital-based care for infants, toddlers, and older children with trisomy 13 [Nelson et al., 2012].

Peroos et al. [2012] provide an account of an 8-year-old with trisomy 13. The child presented with the physical character-

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istics described as well as seizures, scoliosis, and failure to thrive. Notably, the child was not diagnosed with any cardiac anomalies. Conversely, researchers in Japan describe a 9-year-old admitted with heart failure and sepsis [Yukifumi et al., 2011]. Older research discusses survivors as well, including a 19-year-old female [Redheendran et al., 1981]. Baty et al. [1994] present data on 31 children (some with the mosaic form of trisomy13) including surgical interventions. The authors note the average length of survival of their sample was 12.8 months. Recently, Bruns [2011] reported on 30 long-term survivors (mean = 48.4 months). The group exhibited the characteristic physical attributes and medical conditions such as VSD and feeding difficulties. Mean gestational age was 38.1 weeks and birth weight 2789 g. The majority of infants were released from the hospital within 4 weeks (n = 28, 93.3%).

In the face of mixed findings and recommendations for treatment, additional research is needed to further determine what contributes to longevity and implications for treatment for presenting medical conditions. Additional data can also serve to highlight the need for consideration of individual survival trajectories rather than decision-making solely based on pre- or postnatal diagnosis [Carey, 2012].

The purpose of the present study is to report on presenting medical conditions and the presence or absence of the same conditions at a later point in time (age at survey completion). The focus is on children with full as opposed to mosaic, partial or with a Robertsonian translocation, trisomy 13 between 12 and 59 months.

METHOD

The Tracking Rare Incidence Syndrome (TRIS) project began in 2007 with the intent to gather and analyze parent provided data for their child with conditions including trisomy 13 and dissemination of resulting data to interested audiences.

Instrumentation

The TRIS Survey was developed from three sources: (a) medical literature from 1990 to 2005; (b) rare trisomy specific parent listservs; and (c) printed materials from the Support Organization for Trisomy 18, 13 and related disorders (SOFT). A brief description follows.

Part I of the TRIS Survey includes four sections with a total of 43 items. Section I examines mothers' pregnancy history (11 items). Section II covers the newborn's birth and presenting medical conditions (15 items). Section III has 10 items examining the newborn's neonatal care and hospital discharge. Items in Section IV (n = 7) request demographic information (e.g., marital status education level). Part II examines sources of family support (eight sections with 56 items [Bruns and Foerster, 2011; Bruns and Schrey, 2012]. Part III of the TRIS Survey is comprised of nine sections, (61 items total). Sections include developmental data, educational and therapeutic services, as well as past and current medical conditions and treatment (e.g., medications, surgical interventions). The results described here are from Parts I and III.

Procedure

Parents were recruited through several methods including parent-to-parent contact, messages to trisomy-related listservs, and invitations posted to parent support organization websites such as Hope for Trisomy 18 and 13 and announcements and TRIS project updates submitted to the SOFT newsletter.

To enroll, participants provide their name, phone number, state/province and country, e-mail address along with child's name, date of birth and death (if appropriate) and trisomy type. The TRIS Research Coordinator then sends each participant a login and password via email to access the survey within 48–72 hr. Participants also have the option to receive a paper copy of the survey.

The TRIS project's Website (http://web.coehs.siu.edu/Grants/TRIS/) includes an introductory page outlining the sections of the TRIS Survey. A link from this page directs participants to a secure server with detailed information about the survey and a consent form. Participants can then access the survey and can save the data they enter and return to it as necessary for completion. TRIS project staff enters the data upon receipt of a completion email message sent from the TRIS project server. The TRIS Research Coordinator prompts participants if the survey is not completed within two months.

A more detailed description of the TRIS Survey and procedures is available in Bruns [2010; 2011] and Bruns and Campbell [2014].

Participants

Between February 2007 and August 2013, 225 TRIS Surveys were completed for children and adults with trisomy 18, trisomy 13, and other rare trisomy conditions. Of the total, nine represented children with full trisomy 13 between 12 and 59 months of age (4%). Mean age was 35 months (SD ± 10.70 months; range 15–53 months). As of September 1, 2013, seven children in the sample were living (78%).

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). The participant who did not provide this information adopted her child at the age of two months. The majority of mothers were married (n = 7, 78%). Mother's 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. See Table I for additional information.

Data Analysis

Survey data and participant demographics are linked to each participant's unique project ID number. All data are downloaded to a spreadsheet in Microsoft Excel for visual inspection. The data are then copied into a database in SPSS 16 [SPSS, 2008]. Due to the number of surveys available for analysis, frequencies, percentages, means, and standard deviations were computed as appropriate. Non-parametric, descriptive statistical analyses were also used with TRIS Survey demographic data. In addition, refer to Bruns and Campbell [2014].

RESULTS

Data describing gestational age, birth weight, length, and hospital stay after birth is provided in addition to phenotypic characteristics, medical conditions, and surgical procedures (see Tables II and III). Results are also presented in comparison to Jones et al. [2013] (see Tables IV and V).

TABLE I. Demographic Data at Time of TRIS Survey Completion (n = 9)[a]

	Mean (±SD) Range
Child's age in months (n = 9)	35 (±10.70) Range: 15–53
Mother's age in years at birth (n = 8)[b]	32.5 (±4.10) Range: 27–38
Father's age in years at birth (n = 8)[b]	32.63 (±4.63) Range: 26–39
Marital status	n (%)
Single	1 (11)
Married	7 (78)
Divorced	1 (11)
Education level	
7–9 years	1 (11)
10–12 years	2 (22)
13–16 years	2 (22)
More than 20 years	4 (44)
Income level[c]	
Medium	8 (89)
High	1 (11)

[a]Total number of participants. Number of responses to individual items is noted by each characteristic.
[b]One participant adopted their child with trisomy 13; birth parent information is not available.
[c]Income level is collected by level not dollar amounts due to the international scope of the project; all participants in sample represented in Table I identified as living in the United States.

Birth Information

Gestational age data was available for all infants. Mean age at birth was 36.44 weeks (SD ± 3.21) with a range of 33–40 weeks. Birth weight and length data were only available for seven (78%) infants. Birth weight data indicated a mean weight of 2336.81 g (SD ± 752.52 g). Range was 1105.63–3288.54 g. Mean length at birth was 45.29 cm (SD ± 3.86 cm) with a range of 38–48 cm. Two (22%) were male (Participants 1 and 6) and seven (78%) were female (Participants 2, 3, 4, 5, 7, 8, and 9).

Length of hospital stay after birth varied. Of the nine infants, one (11%) was in the hospital for less than seven days. Two infants were released from the hospital in less than two weeks. Three (33%) infants stayed in the hospital for 2–4 weeks. The remaining three (33%) remained in the hospital for 5–8 weeks.

Of the nine infants, one (11%) had a suspected diagnosis of trisomy 13 prior to birth. This mother did not choose to have further testing. Seven infants (78%) were diagnosed within seven days after birth. The remaining infant was diagnosed at 6 weeks.

Phenotypic Characteristics

Seven (78%) infants were identified at birth with polydactyly of the hands, while two also presented with polydactyly of the feet; two infants (Participants 2 and 5) had their extra digits removed before hospital discharge. Seven (78%) infants we identified with low-set ears. Participants 1, 4, and 7 (n = 3; 33%) displayed micrognathia and Participants 1 and 6 (n = 2; 22%) microcephaly. One (11%) infant was diagnosed with a cleft lip and palate, while one infant was diagnosed with cleft palate. Five infants presented with rocker bottom feet.

In addition, Participant 1 presented with small sacral defect (size and type unknown; information not provided) and a missing rib. Participants 3 and 9 were born with umbilical hernias that were repaired prior to discharge. Participant 6 was born with an encephalocele that was also repaired prior to hospital discharge. Participant 9 presented with microphthalmia. Finally, Participants 1, 2, and 9 showed evidence of a coloboma.

Cardiac Conditions

Data indicated that seven (78%) infants were diagnosed with a minimum of one cardiac condition prior to release from the hospital after birth. Participants 3 and 7 were diagnosed with a trial septal defect (ASD). Participant 3 was additionally diagnosed

TABLE II. Participant Birth Data and Presenting Medical Conditions (n = 9)

	Gender	Gestational age (weeks)	Birth weight (grams)	Time in NICU	Apnea	Respiratory interventions	Cardiac conditions	Feeding methods	Kidney issues	Surgeries prior to discharge
1	M	38	3288	2–4 weeks	NP	NP	NP	OF	NP	NP
2	F	35	1814	7–13 days	NP	NP	NP	NG	NP	RD
3	F	40	2948	<7 days	OA	O	ASD, DC	IV	NP	UR
4	F	39	2778	7–13 days	OA	O	ASD, PDA, VSD	IV, OG	NP	NP
5	F	37	NR	2–4 weeks	NP	CP, O, R	ASD, PDA	H, NG	NP	PS
6	M	38	2438	2–4 weeks	OA	CP, O	ASD, VSD	IV, G, NG	NP	EC, GS,
7*	F	38	NR	5–8 weeks	NP	NR	ASD	IV, G	DU, UTI	GS
8	F	35	1984	5–8 weeks	CA, OA	O	PDA	IV, OG	NP	NP
9	F	33	1105	5–8 weeks	OA	O, R	ASD, PDA	IV, NG	NP	LP, NF, UR

Notes. AP = appendectomy; ASD = atrial septal defect; CA = central apnea; CP = continuous positive airway pressure; CS = cardiac surgery; DC = dextrocardia situs inversus; DU = double ureters; EC = encephalocele closure; G = gastrostomy tube feeding; GS = gastrostomy tube placement surgery surgery; H = Haberman feeder; IV = intravenous feeding; LP = Ladd's procedure for bowel malrotation; NF = Nissen fundoplication; NG = nasogastric tube feeding; NP = condition not present; NR = no response; O = supplemental oxygen; OA = obstructive apnea; OF = oral feedings; OG = oral gavage feeding; PDA = patent ductus arteriosis; PS = polydactyly removal surgery; R = respirator; UR = umbilical hernia repair; UTI = urinary tract infections; VSD = ventricular septal defect.
*Participant was adopted at 2 months of age.

TABLE III. Participant Birth Data and Presenting Medical Conditions (n = 9)

	Age at survey completion (months)	Living on 9/1/2013	Apnea	Cardiac conditions	Feeding methods	Kidney issues	Surgeries
1	37	No	NP	NP	OF	PC	BIH, GS, VNS
2	42	Yes	NP	NP	OF	NP	TT
3	37	Yes	NP	ASD	G	NP	TR
4	33	Yes	OA	ASD, PDA, VSD	G, J, NG	NP	A, AP, LP, NF CS, T
5	24	Yes	NP	ASD	NG	NP	NP
6	36	No	NP	ASD, VSD	G	NP	NP
7*	53	Yes	NP	PM	G	NP	IB, VNS
8	38	No	CA, OA	NP	G, J, NG	NP	NP
9	15	Yes	NP	ASD	G, J, NG	NP	NP

Notes. A = adenoidectomy; AP = appendectomy; ASD = atrial septal defect; BIH = bilateral inguinal hernia repair; CA = central apnea; CS = craniosynostosis repair; G = gastrostomy tube feeding; GS = gastrostomy tube placement surgery; IB = intestinal blockage removal and repair; J = jejunostomy tube feeding;; NF = nissen fundoplication; NG = nasogastric tube feeding; NP = condition not present; OA = obstructive apnea; OF = oral feedings; PC = polycystic kidneys; PDA = patent ductus arteriosis;; PM = prolapsed microvalve; T = tonsillectomy; TR = tumor removal from anal region; TT = tympanostomy tubes; VNS = vagus nerve stimulator placement; VSD = ventricular septal defect
*Participant was adopted at 2 months of age.

with dextrocardia and situs inversus. Participant 8 was diagnosed with patent ductus arteriosis (PDA). Participants 5 and 9 were diagnosed with ASD and PDA. Participant 6 was diagnosed with ASD and ventricular septal defect (VSD). One (11%) infant, Participant 4, was diagnosed with three cardiac conditions (ASD, PDA, and VSD).

At the time of survey completion, three (33%) participants (5, 8, and 9) reported resolution of PDAs without medical intervention. Participant 7 was also diagnosed with a prolapsed mitral valve. No cardiac surgeries were reported.

Feeding Methods

Prior to release from the hospital, eight infants required assistance with feeding (89%). One infant (11%) (Participant 3) only received intravenous (IV) feeding. One (11%) infant was fed exclusively with a nasogastric tube. Six (67%) infants required multiple forms of artificial nutrition prior to release from the hospital. Of those infants, two (22%) (Participants 6 and 7) received a gastrostomy tube (g-tube) prior to discharge.

Survey completion data indicated two infants (22%) did not require use of a feeding tube and received all nutrition orally after hospital discharge. Of the remaining seven, four (44%) required an n-g tube (Participants 4, 5, 8, and 9), six (67%) required a g-tube (Participants 3, 4, 6, 7, 8, and 9) and three (33%) used a jejunostomy tube (j-tube) (Participants 4, 8, and 9) for nutrition between the time of hospital discharge after birth and survey completion. It is important to note that most participants received multiple feeding methods at birth and time of survey completion including several instances of oral feeding.

TABLE IV. Jones [2013] Profile of Physical Characteristics Compared with Sample (n = 9)

	Sample (n = 9) % (n)	Jones [2013] %
Low-set ears	78 (7)	Not described
Micrognathia	33 (3)	≤ 50
Cleft lip	11 (1)	60-80
Cleft palate	22 (2)	≥ 50
Microcephaly	22 (2)	≥ 50
Polydactyly	78 (7)	≥ 50
Rocker bottom feet	56 (5)	Not described
Microphthalmia	11 (1)	≥ 50
Coloboma	33 (3)	≥ 50

Source: Jones KL, Jones MC, Del Campo, M. 2013. Smith's recognizable patterns of human malformation (7th edition) (pp.20–23). Philadelphia, PA: Elsevier Saunders.

Apnea

Data indicated that five infants (56%) experienced an obstructive apnea episode after birth (obstruction in airway passage). Of the five, one (20%) (Participant 8) also experienced episodes of central apnea (central nervous system problem resulting in limited or no muscle coordination for breathing).

At the time of survey completion, two (n = 5, 40%) infants with obstructive apnea were still experiencing episodes at age 33 and 38 months, respectively (Participants 4 and 8). One of the two infants also continued to experience episodes of central apnea at 38 months. The infant with obstructive apnea was treated with glandular Botox and 3–4 l of oxygen while sleeping. The infant with obstructive and central apnea was repositioned during apnea episodes to assist with breathing.

Additional Respiratory Difficulties

Prior to first hospital discharge, six (67%) infants were identified with additional respiratory difficulties. Six infants received supplemental oxygen and two (33%) of which required ventilator support. In addition, three of the six infants (Participants 5, 6, and 9) were reported to receive continuous positive airway pressure (CPAP).

TABLE V. Jones [2013] Profile of Medical Conditions Compared with Sample (n = 9)

	Sample at birth (n = 9)	Sample at survey completion (n = 9)	Jones [2013]
Atrial septal defect	67% (6/9)	56% (5/9)	80%
Patent ductus arteriosis	44% (4/9)	11% (1/9)	80%
Ventricular septal defect	22% (2/9)	22% (1/9)	80%
Feeding support	89% (8/9)	78% (7/9)	Not described
Apnea	56% (5/9)	22% (2/9)	≥50% in early infancy
Additional respiratory difficulties	22% (2/9)	0% (0/9)	Not described
Double ureters	11% (1/9)	11% (1/9)	≤50%
Polycystic kidneys	0% (0/9)	11% (1/9)	31%
Holoprosencephaly	0% (0/9)	0% (0/9)	≥50%
Seizures	78% (7/9)	33% (3/9)	≥50%
Umbilical hernia	22% (2/9)	0% (0/9)	≥50%
Intestinal malrotation	22% (2/9)	0% (0/9)	≤50%

Source: Jones KL, Jones MC, Del Campo, M. 2013. Smith's recognizable patterns of human malformation (7th edition) (pp.20–23). Philadelphia, PA: Elsevier Saunders.

Kidney Issues

Of the nine participants, one (11%) was diagnosed soon after birth with double ureters (affected kidney is unknown – not provided). At the time of survey completion, one additional child was identified with a polycystic kidney (affected kidney was not specified).

Seizures

Seven (78%) infants were identified with seizures at birth or the immediate postnatal period. Participant 1 was identified as having generalized myoclonic seizures. Participant 3 was diagnosed with tonic-clonic seizures. Participant 4 experienced petit mal seizures. Participants 6 and 7 were identified with tonic-clonic, petit mal, grand mal, and mixed type seizures. Participant 8 tonic-clonic seizures and infantile spasms and Participant 9 tonic-clonic and grand mal seizures.

At the time of survey completion, three (33%) participants were still experiencing seizures. Participant 3 continued to have tonic-clonic seizures. Participants 7 and 9 continued to have multiple types of seizures (tonic-clonic, petit mal, and grand mal seizures; tonic-clonic, grand mal seizures, and mixed type seizures, respectively). Participants 1 and 7 had vagus nerve stimulation (VNS) implants to treat seizure activity at the ages of 36 and 42 months. Additionally, Participant 1 was on the ketogenic diet from 20 to 36 months but it proved ineffective for controlling seizures.

Refer to Tables II,III and V for data described above.

Photos of two participants from the sample are provided to further contextualize the data (Figs. 1 and 2) and emphasize the similarities and variations in phenotype and medical conditions across this sample.

DISCUSSION

Data on nine children with trisomy 13 indicated mean gestational age of approximately 36 weeks, birth weight ranging from 1100 to 3290 g and mean length of 45.29 cm. Length of hospital stay after birth varied. One infant was discharged in less than seven days, and three remained in the hospital for up to eight weeks. The majority of infants presented with polydactyly, low-set ears and rocker bottom feet. Micrognathia, cleft lip, and cleft palate were only present in approximately one-third of the sample. Interestingly, only one infant had a suspected diagnosis of trisomy 13 prior to birth. All remaining infants received their diagnosis by 6 weeks of age.

Medical conditions and their treatment also varied. Notably, several infants' cardiac anomalies resolved without surgical intervention. Most received one or more types of tube feeding. Apnea continued for several participants even with interventions. Kidney issues were largely absent in this sample. Surgeries were required for a range of conditions including gastrostomy tube placement to address feeding issues, removal of intestinal blockage, craniosynostosis, and placement of tympanostomy tubes. One infant received an encephalocele repair soon after birth. Importantly, there were no reports of holoprosencephaly.

Confirm and Disconfirm Previous Findings

The results presented here for nine children with full trisomy 13 offer a number of different findings from previous studies. For example, Baty et al. [1994] describes 31 children with trisomy 13.

FIG. 1. Patient 8 born September 2009; died November 2012.

FIG. 2. Patient 3 Sofia born January 2004; living.

Mean birth weight was 2676 g while the mean for this sample was approximately 10% lower (2336.81 g). In addition, seven infants were born between 38 and 42 weeks with a mean of 36.44 weeks (gestational age not provided for two infants) and required less than two weeks of hospitalization. In a recent report, Houlihan and O'Donoghue [2013] report mean gestation at 38 weeks (range 35–40 weeks; n = 21). The authors note preterm delivery for fetal distress or growth restriction with limited longevity after birth. Future studies should examine the contribution of gestational age as a determining factor in long-term survival.

Baty et al. [1994] also discussed that approximately one-third of their sample was living at the age of one year and 13% at the age of five. Other studies offer lower survival rates [e.g., Lin et al., 2007; Crider et al., 2008; Petry et al., 2013]. Mean age when TRIS Survey was completed was 35 months and most of the sample (78%) was still living several years later.

The majority of infants presented with common physical characteristics including polydactyly, low-set ears and rocker bottom feet while one participant presented with a cleft palate and another with both cleft lip and cleft palate. Hsu and Hou [2007] reported all infants with rocker bottom feet (n = 13) and over 75% had cleft lip and/or palate. In Peroos et al. [2012] case study of an 8-year-old female with trisomy 13, polydactyly, low-set ears and cleft palate were noted. Importantly, in Hsu and Hou's sample, there was only one occurrence of holoprosencephaly and none in the TRIS project sample. Tsukada et al. [2012] identified this condition in four cases (n = 16) with longest survival of 50 days. The absence of this brain anomaly may be an additional factor in long-term survival.

Petry et al. [2013] noted a high incidence of cardiac defects in 24 infants with trisomy 13 (additional six were mosaic). Close to 40% of infants in Pont et al. [2006] were diagnosed with a condition such as ASD or VSD. ASD was the most prevalent in the TRIS project sample (66% at birth and 55% at survey completion). Tsukada et al. [2012] report on 16 patients. Approximately half of the sample was diagnosed with ASD, PDA, and/or VSD. The authors also include "respiratory failure" as a complication. Data from the present study specified five instances of obstructive apnea and one of central apnea at birth and only two children experiencing this condition at survey completion. It appears that data regarding apnea is not typically reported in investigations of this clinical subgroup.

The group described here also underwent a variety of surgical procedures shortly after birth as well as in the months and years prior to survey completion including closure of encephaolocele, placement of gastrostomy tube, treatment of bowel malrotation and placement of a vagus nerve stimulator. Interestingly, no cardiac surgeries were reported. Similarly, only 3% of Nelson et al. [2012] sample underwent cardiac procedures (n = 1075). The most common surgeries were related to gastrointestinal conditions including placement of gastrostomy tube (n = 13). Several studies in Japan describe positive outcomes for treatment of cardiac defects including medications and/or surgical interventions [Kaneko et al., 2008; Maeda et al., 2011].

Existing studies do not report on feeding as described here and in Bruns and Campbell [2014]. Cleft lip and/or cleft palate are noted [e.g., Pont et al., 2006; Lin et al., 2007] but not feeding method. Further, the data shared here includes frequency of oral and eternal feeding at more than one point in time (neonatal period and at survey completion). Tsukada et al. [2012] reports on a sample of 16 with 10 diagnosed with cleft lip and cleft palate and only one child receiving corrective surgery. The child was one of four still living at the time of data analysis.

The results presented from this study and previous investigations involving children over the age of one year with full t13 provide an overview of presenting conditions and interventions. They also offer a view of changes over time in relation to care such as the provision of cardiac surgery. Case study and series studies further articulate common medical needs and their resolution along with, largely, highlight positive outcomes [e.g., Baty et al., 1994; Hsu & Hou, 2007]. Long-term survival is possible when neonates gestate for over 37 weeks, present without key medical conditions, such as holoprosencephaly, and are accorded necessary interventions perinatally and through the early childhood years.

Limitations

Study criteria consisted of a diagnosis of full trisomy 13 and between the ages of 12 and 59 months at time of survey completion. As such, participants who passed away within this time frame were not eligible and their data was not compared to the nine children described here. The resulting sample was small and purposive. Other trisomy 13 studies report on data from larger samples [Lin et al., 2007, n = 28; Kaneko et al., 2008, n = 31; Tsukada et al., 2012, n = 16]. These investigations either collect data over a longer time span and/or compile data from a specialized hospital or national hospital system. Participation in the TRIS project is voluntary and relies on outreach for recruitment including a Facebook page (https://www.facebook.com/TRIS.Trisomy.project), TRIS project brochures shared with parents and medical professionals, and conference presentations. In addition, although the scope of the project is worldwide, this subgroup of participants was all identified as living in the United States at the time of survey completion. Geographic location could affect availability of specialized medical care and interventions. As such, the results described cannot be generalized.

Since initial development of the TRIS Survey in 2005–06 and beginning data collection in 2007, additional trisomy 13-related medical conditions have been identified including brain anomalies, such as holoprosencephaly, and ocular conditions such as colobomata. Survey items cannot be revised or updated due to the continuing nature of the project. It should be noted that space is provided for additional responses ("Other: please specify"). This was the means to collect this additional data from parents with children with trisomy 13. Yet, efforts are underway to analyze additional data with an eye toward further examining presenting conditions and corresponding longitudinal interventions [Bruns and Springer, in preparation]. Development of an additional, more in-depth survey for this clinical subgroup is also being considered.

Data reported here are exclusively from parent report. Inaccuracies and omissions are possible. The TRIS project is unique in that data are collected directly from parents rather than reviewed from a registry or similar database [refer to Vendola et al., 2010; Wu et al., 2013]. The intent is to collect information at a greater breadth and depth as well as longitudinally. The TRIS Follow-up Survey is completed each year the child is living after the initial TRIS Survey, which is described here. At the present time, two children with full trisomy 13 have seven years of data in the TRIS database (initial survey in 2007 and six follow-up surveys; additional six have other rare trisomy conditions including full trisomy 18 and partial trisomy 6p).

Implications

No participants in this sample were diagnosed prenatally. One parent reported suspicion of a genetic condition during pregnancy but tests were not run prior to birth to confirm it. This is important to note as provision of medical interventions may have been positively affected as a result. In contrast, Bell et al. [2014] describes differences in delivery room care based on prenatal or perinatal diagnosis of trisomy 13. This was coupled with most surviving newborns receiving comfort care or withdrawal of care ($n = 38$; 11% surviving to hospital discharge). Also, as pointed out previously, no children in this sample were diagnosed with holoprosencephaly. This, too, could contribute to a willingness to offer medical interventions. The literature discusses recommendations of withholding medical procedures due to prenatal diagnosis of trisomy 13 [Courtwright et al., 2011; Merritt et al., 2012]. This, again, points to decision-making based on diagnosis rather than individual needs. Bruns [2013] posits than an infant or child's response to treatment should factor more into the provision of care than the largely negative view found in the existing literature. In addition, since this sample may have been unique, there is a need for additional investigations to further delineate the constellation of present and absent medical issues contributing to longevity.

Carey [2012] advocates for a balanced approach to care, one that encompasses medical literature and parent perspectives such as those presented in Janvier et al. [2012]. Carey also emphasizes care decisions made on an individual child's needs rather than diagnosis so that care and management recommendations then become personalized rather than defined by much of the current literature, which focuses on lethality [e.g., Merritt et al., 2012]. In addition Janvier et al. [2012] discuss the happiness their child brings or brought to their lives. The authors also noted "most parents reported that their family was trisomy 13strengthened since the birth, and often the death, of a child with t13 or 18. Parental decision-making was not homogeneous and reported outcomes were diverse…" (p. 297).

McCaffrey [2011] and Wilkinson et al. [2012] call for a shift from a lethal perspective to accepting ambiguity and treating each child on an individual basis. The parental perspective is shared in Locock [2005]. Parents describe their decision to continue their pregnancy after a trisomy 13 diagnosis at 20 weeks gestation. They emphasized the value of their son's brief life (born at 35 weeks and lived 3 min). Readers are also directed to http://www.livingwithtri13.org for descriptions of decision-making on behalf of infants and children with trisomy 13. The website also includes an array of photos of living children (see http://www.livingwithtri13.org/trisomy-13.htm).

Yates et al. [2011] discuss results of a survey examining the provision of cardiac interventions including surgeries to children with trisomy 13. Cardiologists were more likely to recommend this type of intervention than neonatologists or geneticists. The authors invite further discussion on the topic, which needs to occur. Interestingly, in the sample described here, there were no cardiac surgeries but a variety of other surgical interventions were completed (see Tables II and III). A willingness among medical professionals to perform these procedures becomes paramount.

Framing the results provided here within the available literature offers conflicting viewpoints. The answer may lie in the underlying thesis of the study. Low survival rates found in large scale studies (population-based, hospital registries) may reflect a tendency toward lack of medical interventions based on perceptions of a limited quality of life [Brewer et al., 2002; Irving et al., 2011]. This orientation increases mortality of infants and children with trisomy 13. It is evident that some infants are born with life threatening conditions that do not respond to medical intervention (e.g., holoprosencephaly). Yet, infants and children with trisomy 13, such as those described here, can be provided with procedures and surgeries to correct or reverse common trisomy 13 conditions and live and thrive for years. It becomes clear that both types of outcomes (early mortality with little to no medical interventions, survivors given varying amounts and types of medical interventions) must be studied. Additional research is needed to better determine treatment options for children with this genetic condition.

It is becoming clear that outside of life-threatening conditions, demonstration of the same phenotypic characteristics and medical issues does not result in uniform outcomes. Recommendations and follow through for each affected infant and child must be decided on an individual basis with data from multiple and up-to-date sources [Bruns, 2013; Janvier and Watkins, 2013]. This, in turn, provides parents with balanced information for decision-making on their child's behalf in place of dire predications and withholding or refusing medical care [Thiele et al., 2013].

CONCLUSION

Similar to Bruns [2011], the data here illustrate a more positive outlook on children with full trisomy 13. Results highlight birth

information, phenotypic characteristics, medical conditions and related interventions for survivors with trisomy 13 over the age of one year. This is a needed counterpoint to most of the available literature. It is hoped that sharing these findings contributes to further study of this group and a corresponding change in perception from a "lethal anomaly" to valuing infants and children diagnosed with trisomy 13.

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