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Twenty-Two Survivors over the Age of 1 Year with Full Trisomy 18: Presenting and Current Medical Conditions

Deborah Bruns* and Emily Campbell

Southern Illinois University Carbondale, Carbondale, Illinois

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The purpose of the study is to provide data about 22 survivors over the age of 1 year with full trisomy 18 (12–59 months). Mothers completed the online, mixed method Tracking Rare Incidence Syndrome (TRIS) Survey provides data on birth information (e.g., gestational age, birth weight) and medical conditions identified at birth and at the time of survey completion. Data indicate similar birth characteristics to other studies and presence of syndrome related medical conditions including cardiac conditions, use of a variety of feeding methods, apnea, respiratory difficulties, and kidney issues. Associated interventions, sometimes considered "aggressive" or "intensive" treatments including cardiac surgeries were noted in the sample. Implications for treatment are provided and the need for additional research with this clinical subgroup is needed.

Key words: trisomy 18; cardiac conditions; feeding method; kidney issues; apnea; medical interventions

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INTRODUCTION

In recent years, there has been an increasing emphasis on palliative or comfort care, rather than aggressive or intensive treatment, for newborns with what medical professionals term "severe fetal anomalies" or "incompatible with life" who survive labor and delivery [Hentschel et al., 2006; Breeze et al., 2007; Romesberg, 2007; Everett and Albersheim, 2011; Merritt et al., 2012]. One of the groups discussed in this literature is those diagnosed with full trisomy 18 (Edward syndrome, t18) [Courtwright et al., 2011; Derrington and Dworetz, 2011; Yates et al., 2011]. The consensus leans toward non-treatment [e.g., Bos et al., 1992; Goc et al., 2006; McGraw and Perlman, 2008] with some opposing voices and encouraging data [Koogler et al., 2003; Bruns, 2010, 2013; Maeda et al., 2011].

Newborns with t18 frequently present with distinctive physical characteristics including low-set ears, clenched hands, and rocker bottom feet [Jones, 2006]. Common medical conditions include apnea (typically central rather than obstructive), cardiac anomalies such as ventricular septal defect (VSD), pulmonary hypertension, compromised respiratory functioning that may require mechanical support (e.g., oxygen delivered via canula, tracheostomy), kidney malformations and feeding difficulties. Cause of death is often associated with complications due to central apnea, cardiac and/or respiratory problems [Jones, 2006].

The majority of published reports point to a bleak prognosis for infants with t18. For example, a number of population-based studies stress early mortality and life threatening medical conditions [Rasmussen et al., 2003; Crider et al., 2008; Vendola et al., 2010]. Most research points to only 5–10% of infants reaching their first birthday [Rasmussen et al., 2003]. Yet, Rasmussen et al. do report the level of medical treatment recommended or received, only mortality rates. "Survivors" are not described in the literature much beyond case studies [see Kelly et al., 2002; Bhanumathi et al., 2006] with most focusing on children or adults with less severe forms of the syndrome [Tucker et al., 2007; Banka et al., 2013].

Descriptions of infants with t18 living for a short time often present a negative view emphasizing anomalies and defects [Shaw, 2008]. Long-term survivors demonstrate significant developmental delays and many experience chronic medical complications but throughout the literature, there is an added assumption of a limited quality of life used as a rationale for the denial of intensive treatment [Chervenak and McCullough, 2012; Merritt et al., 2012].

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*Correspondence to:
Deborah Bruns, Southern Illinois University Carbondale, MC-4618 Carbondale, IL 62901.
E-mail: dabruns@siu.edu

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A literature base describing survivors over the age 1 year is emerging with reports of children with t18 reaching their second, third, and beyond birthdays [Imataka et al., 2007; Bruns, 2010]. Recently, a number of studies reported from Japan highlight better outcomes from cardiac surgery and follow-up [Kaneko et al., 2008; Kobayashi et al., 2010; Maeda et al., 2011]. A study by Yates et al. [2011] found a willingness of cardiologists to recommend surgical intervention for cardiac defects. Most geneticists and neonatologists in the sample did not agree. Merritt et al. [2012] recommend that treatment decisions be tempered by the "best interest" of the infant. The authors use this point of reference to explain that some infants with t18 should not receive intensive treatment and the need to increase parents' knowledge of t18-related complications. Again, this draws on the common pessimistic view toward this population.

What is missing from the literature is detailed examination of survivors over the age of 1 year and empirical data specifically examining medical needs and longevity. Without this, the conventional view toward little to no intensive treatment at birth and in the immediate postnatal period will be maintained without consideration for other options.

The purpose of the present study is to examine medical conditions noted at birth and the immediate perinatal period, along with the presence or absence of the same conditions coupled with medical treatment and outcomes in a sample of long-term survivors with full t18 between the ages of 12–59 months. These data are offered to raise awareness of the survival of older children with t18 and to assist a shift in treatment decisions to ensure appropriate medical care for this population.

METHODS

The Tracking Rare Incidence Syndrome (TRIS) project began in 2007 with the intent to collect and analyzed parent provided data for an array of trisomy-related topics and disseminate the results to a variety of audiences including geneticists, nurses, and other medical professionals [Bruns, 2008].

Instrumentation

The TRIS Survey, for children living at least 2 months after birth (referred to here as "long-term survivors), 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). The TRIS project also collects data via the Modified TRIS Survey for infants in one of the following categories: (a) miscarried, (b) stillborn, or (c) lived 60 or less days. The data described here is from the TRIS Survey. 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. Finally, there are seven items in Section IV requesting demographic information (e.g., marital status education level). Part II examines sources of family support (eight sections with 56 items, see Bruns and Foerster, 2011 and Bruns and Schrey, 2012 for a description and results).

Part III includes items related to developmental, educational, therapeutic, related services, as well as past and current medical conditions including effectiveness of medications and surgical interventions (nine sections, 61 items). Part III also includes the TRIS Developmental Matrix for mapping the child's progress in the areas of cognitive, language, fine and gross motor, and social skills. The results described here are from Parts I and III.

Procedure

After receiving approval from Human Subjects Committee, parents were recruited through parent-to-parent contact, postings on trisomy-related listservs (tri-family, tri-med), and invitations posted to parent support organization websites such as Hope for Trisomy 18 and 13 and announcements in 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 hr. Participants are also given an option to receive a paper copy of the survey if they prefer. TRIS project staff enters the data upon receipt of the completed survey.

The TRIS project's Web site (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. The TRIS Research Coordinator prompts participants if the survey is not completed within 2 months.

A more detailed description of the TRIS Survey and procedures is available in previous publications [Bruns, 2008, 2010].

Participants

Between February 5, 2007 and March 7, 2012, 225 TRIS Surveys were completed for children and adults with t18, trisomy 13 and other rare trisomy conditions. Of the total (n = 225), 22 represented children with full t18 between 12 and 59 months of age (9.8%). At the time of survey completion, all children were living. Mean age was 29.9 months (SD ± 17.41 months; range 13–58 months). As of June 1, 2013, 15 children in the t18 subgroup were still living (68.2%). An additional 16 surveys were completed for children with t18 between 2 to 11 months of age and 19 represented survivors with t18 who were 60 months or older. (A manuscript is in preparation for the older group).

Information on maternal and paternal history was available for 21 (95%) participants. 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%). The remaining participants represented Australia, Canada, and parts of Western Europe. See Table I for additional information.

TABLE I. Participant Demographic Data at Time of TRIS Survey Completion (n = 22)

	Mean (±SD) Range
Child's age (n = 22)[a]	29.9 months (±17.41 months) Range: 13–58 months
Mother's age at birth (n = 21)[b]	31.6 years (±6.27 years) Range: 19–40 years
Father's age at birth (n = 21)[b]	33.1 years (±6.53 years) Range: 19–45 years
Marital status (n = 21)	n (%)
Single	3 (14.3%)
Married	17 (81%)
Separated	1 (4.5%)
Education level (n = 21)	
7–9 years	1 (4.8%)
10–12 years	3 (14.3%)
13–16 years	9 (42.9%)
17–20 years	7 (33.3%)
More than 20 years	1 (4.8%)
Income level (n = 21)[c]	
Low	3 (14.3)
Medium	17 (81%)
High	1 (4.8%)

[a]Total number of participants. Number of responses to individual items is noted by each characteristic.
[b]One participant adopted their child with trisomy 18; birth parent information is not available.
[c]Income level is not represented in dollar figures due to the international scope of the project (participants represented the US (n = 18), Australia (n = 1), Canada (n = 1), Germany (n = 1), and Sweden (n = 1), and their corresponding national currencies).

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 is then copied into a database in 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 Full Survey demographic data.

RESULTS

Data describing gestational age, birth weight, length, and hospital stay after birth are provided in addition to medical conditions and surgical procedures (see Tables II and III). Photos of three children from the sample are also provided to contextualize the data (Figs. 1–3).

Birth Information

Gestational age data was available for all infants. Mean age at birth was 39.1 weeks (SD ± 2.01) with a range of 36–44 weeks. Birth weight data indicated a mean weight of 2,166 g (SD ± 333.42). The range in weight was 1,730–3,260 g. Birth length data was available for 21 (95%) participants. Mean length at birth was 45.29 cm (SD ± 3.02) with a range of 41–53 cm. Five (24%) were male and 17 (77%) were female.

Length of hospital stay after birth varied. Of the 22 infants, 27% (six infants) were in the hospital for less than 7 days. Three infants were released from the hospital prior to 14 days after birth. Eleven (50%) infants stayed in the hospital for 2–4 weeks. The remaining two (9%) stayed in the hospital for 5–8 weeks. As the data indicates, the majority of infants remained in the hospital for less than four weeks after birth.

Of the 22 infants, eight (36%) had a suspected diagnosis of trisomy 18 prior to birth. Data indicate that the time of diagnosis was equally distributed between the second and third trimesters. Of these eight mothers, six (75%) chose to have further testing, and all received confirmation of diagnosis before birth. Of the infants diagnosed after birth (n = 15), 11 (73%) were diagnosed within 7 days. The remaining four (27%) were diagnosed within 2 weeks. (Data was unavailable for one participant).

Cardiac Conditions

Data indicated that 20 (90%) infants were diagnosed with a minimum of one cardiac condition prior to release from the hospital. Six (27%) infants were diagnosed with four cardiac conditions (heart murmur, an atrial septal defect (ASD), patent ductus arteriosis (PDA), and a VSD.

At the time of survey completion, one (5%) participant reported no existing cardiac condition. The child's conditions (ASD, PDA, and VSD) had resolved without medical intervention. Conversely, 11 children had surgery to correct cardiac conditions. Median age at time of surgery was 7 months with a range of 3–36 months. Repair of VSD was the most common. No cardiac banding was reported prior to complete repair. As indicated in Table III, survival after corrective cardiac surgery was largely effective with children living approximately 2 to 3 years post cardiac surgery. Kosho et al. [2013] also discuss positive outcomes of cardiac surgery related to resolution of defects and longevity. Other studies offer mixed outcomes [see Graham, 2004; Kaneko et al., 2008; Yates et al., 2011; Nelson et al., 2012].

Feeding Methods

Prior to release from the hospital, 21 infants required assistance with feeding (95%). Four (19%) participants reported their infants only received intravenous (IV) feeding. Five (24%) infants were fed exclusively with a gavage tube (oral through mouth or nasogastric, n-g, through nose). Thirteen infants required multiple forms of artificial nutrition prior to release from the hospital. Of the remaining infants, one (4%) required oral gavage as well as IV feedings and received a gastrostomy (g-tube) prior to discharge.

Data indicated five infants (23%) did not require use of a feeding tube at the time of survey completion and were able to receive all nutrition orally. Of the remaining 17, 12 (71%) required an NG tube, 13 (76%) required a gastrostomy (g-tube), and three (18%) used a jejunostomy tube (j-tube). One participant did not provide data on feeding methods at the time of survey completion.

TABLE II. Participant Birth Data and Medical Conditions at Birth

	Gender	Gestational age (weeks)	Birth weight (g)	Time in NICU	Apnea	Respiratory interventions	Cardiac conditions	Feeding methods at birth	Kidney issues	Surgery prior to discharge
1	F	41	2,296	7–13 days	No response	O	ASD, VSD	IV	KR	n/a
2	F	41	2,041	<7 days	CA	O	PDA, VSD	IV	n/a	n/a
3	M	40	2,300	2–4 weeks	No response	n/a	ASD, PDA, VSD	IV	n/a	CS
4	F	38	1,814	2–4 weeks	CA	n/a	ASD, PDA, VSD	H, NG, OG	HK	n/a
5	F	40	2,296	2–4 weeks	No response	O	n/a	IV, NG	HP	n/a
6	F	38	1,984	2–4 weeks	n/a	O, V, VA	PDA, VSD	IV, NG	n/a	GS
7	F	40	2,495	<7 days	n/a	O	PDA,VSD	H, NG, OG	n/a	n/a
8	F	42	2,523	2–4 weeks	n/a	n/a	ASD, PDA, VSD	G, NG, OG	n/a	CS, FS
9	F	44	3,260	<7 days	n/a	n/a	ASD	OF	n/a	n/a
10	F	39	2,296	7–13 days	CA	n/a	VSD	IV	n/a	CS
11	F	36	1,928	5–8 weeks	n/a	O	ASD, PDA, VSD	H, NG	n/a	n/a
12	F	40	2,155	2–4 weeks	n/a	O, V	PDA, VSD	IV, NG	n/a	n/a
13	M	40	1,758	2–4 weeks	CA, OA	O	VSD	H, NG, OG	KR	n/a
14	F	36	1,814	5–8 weeks	OA	O	ASD, PDA, VSD	OG	n/a	CS, FS, TEF
15	F	41	2,440	2–4 weeks	n/a	O, T	ASD, VSD	NG	n/a	CS, FS
16	F	38	2,155	<7 days	n/a	O, V, VA	ASD, PDA, VSD	IV, NG	n/a	n/a
17	F	37	1,729	2–4 weeks	CA, OA	O, V	ASD, VSD	IV, OG	n/a	n/a
18	F	38	1,956	<7 days	No response	n/a	PDA, VSD	NG	n/a	CS, FS
19	M	37	2,240	2–4 weeks	CA	O, V	ASD, PDA, VSD	G, IV, NG, OG	n/a	FS
20	M	38	2,211	7–13 days	CA	n/a	ASD, PDA, VSD	IV, OG	n/a	FS
21	F	41	2,126	<7 days	OA	n/a	n/a	NG	C	n/a
22	M	39	2,100	2–4 weeks	n/a	O	VSD	IV, NG	n/a	n/a

ASD, atrial septal defect; C, polycystic kidneys; CA, central apnea; CS, cardiac surgery; F, female; FE, feeding related surgery G, gastrostomy tube feeding; H, Haberman feeder; HK, horseshoe kidney; HP, hydronephrosis; IV, intravenous feeding; KR, reflux; M, male; NG, nasogastric tube feeding; O, supplemental oxygen; OA, obstructive apnea; OF, oral feedings; OG, oral gavage/mouth tube feeding; PDA, patent ductus arteriosis; VA, ventilator assistance; T, tracheostomy; TEF, tracheoesophageal fistula repair; V, ventilator; VSD, ventricular septal defect.

TABLE III. Participant Age, Status, and Medical Conditions at Survey Completion

	Age at survey completion (months)	Living on June 1, 2013	Apnea	Cardiac conditions	Feeding methods after discharge	Kidney issues	Other surgeries
1	48	Yes	No response	BAV, VSD	G	KR	UC, UR
2	56	No	n/a	ES, PDA, VSD	G	n/a	n/a
3	25	No	No response	TOF*	OF	n/a	CC
4	58	Yes	n/a	ASD, PDA	G, NG	HK	n/a
5	57	Yes	No response	n/a	OF	HP	n/a
6	16	Yes	n/a	ASD, PDA	G	n/a	O, AP
7	14	No	n/a	PDA*, VSD*	NG	n/a	TV
8	14	No	n/a	ASD*, PDA, VSD*	G, NG	n/a	n/a
9	13	No	n/a	ASD, VSD	OF	n/a	n/a
10	20	Yes	n/a	VSD*	no response	n/a	BMT
11	13	Yes	n/a	ASD, PDA*, VSD	NG	n/a	n/a
12	56	Yes	n/a	PDA, VSD	OF	n/a	A, BMT
13	42	Yes	n/a	BAV	G	KR	BIH
14	13	Yes	OA	ASD*, PDA*,VSD*	G, NG	n/a	EF, NF, PAPVR, RVH, SV
15	23	Yes	n/a	ASD*, VSD*	OF	n/a	n/a
16	14	Yes	n/a	ASD, PDA, VSD	G, NG	n/a	n/a
17	20	Yes	CA, OA*	ASD, PS, VSD	G, J, NG	n/a	T
18	47	Yes	No response	PS, TOF, VSD	G, J, NG	n/a	n/a
19	13	Yes	n/a	ASD, VSD	G, J, NG	n/a	NF, T, TT
20	31	Yes	n/a	ASD, VSD	G, NG	n/a	NF
21	45	Yes	OA	ASD, PS, VSD	G, NG	n/a	AN, NF
22	19	Yes	No response	VSD	NG	n/a	n/a

Note*, corrective surgery performed; A, adenoidectomy; AN, angioplasty; AP, appendectomy; ASD, atrial septal defect; BAV, bicuspid aortic valve; BIH, bilateral inguinal hernia repair; BMT, bilateral myringotomy and tubes; CA, central apnea; CC, cardiac catheterization; EF, esophageal fistula repair; ES, Eisenmenger's Syndrome; G, gastrostomy tube feeding; HK, horseshoe kidney; HP, hydronephrosis; J, jejunostomy tube feeding; KR, kidney reflux; NG, nasogastric tube feeding; NF, nissen fundoplication; O, omphalocele repair; OA, obstructive apnea; OF, oral feedings; PAPVR, partial anomalous pulmonary venous return repair; PDA, patent ductus arteriosis; PS, pulmonary stenosis; RVH, Right ventricular hypertrophy repair; SV, Sinus Venous repair; T, Tracheostomy; TOF, tetralogy of fallot; TT, tympanostomy tubes; TV, tricuspid valve repair; UC, unspecified cardiac surgery; UR, ureteral reimplantation; VSD, ventricular septal defect.

Apnea

Available data for 18 infants (n = 22, 82% of sample) indicated that nine (50%) experienced an apnea episode. Of the nine, five (55%) experienced episodes of central apnea (central nervous system problem resulting in limited or no muscle coordination for breathing). Of these five infants, two parents (40%) reported stimulation of their infant during apnea episodes. Two other infants of the nine (22%) experienced obstructive apnea (obstruction in airway passage). One of the two infants was fitted with a continuous positive airway pressure (CPAP) device. In addition to the infants described here, two others (22%) experienced episodes of central and obstructive apnea, and one (11%) of these two underwent a tracheostomy and was treated with caffeine.

At the time of survey completion, the two infants with obstructive apnea were still experiencing episodes at age 13 and 45 months. The use of CPAP was still necessary for one child at age 45 months. The 20-month-old infant who received a tracheostomy at 10 months of age continued to experience episodes of central and obstructive apnea.

Additional Respiratory Difficulties

Prior to first hospital discharge, 17 (78%) infants were identified with additional respiratory difficulties. Fourteen (63%) infants received supplemental oxygen and five required ventilator support. In addition, one infant was reported to undergo an adenoidectomy at 30 months without the presence of apnea. As noted previously, one infant underwent surgery for a tracheotomy before discharge but did not report need for assistance from a ventilator during the immediate postnatal period.

Kidney Issues

Of the 22 participants, five (23%) were diagnosed soon after birth with a kidney anomaly. Two participants reported their infant was diagnosed with kidney reflux, one participant reported primary vesicoureteral reflux, and one with a horseshoe shaped kidney. One infant had hydronephrosis and double ureters of the right kidney, while a second infant was diagnosed with kidney cysts. At the time of survey completion, kidney disorders persisted in four (80%) of the five infants. One infant with kidney reflux had ureteral reimplantation surgery at 24 months of age. At 48 months, the child still reported kidney reflux. The cysts that had been present in one child were resolved without surgery (no age specified).

DISCUSSION

The present study provides data on 22 survivors over the age of 1 year with t18. It is significant to note the resolution of some

FIG. 1. Patient 20: still living. Born April 2008; photo December 2011; 44 months.

FIG. 1. Patient 20: still living. Born December 2008; photo December 2011; 36 months.

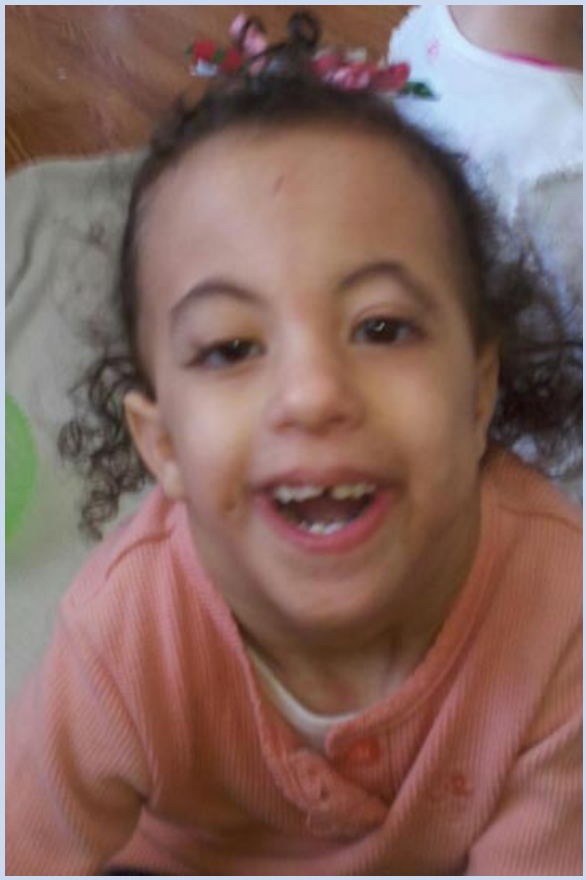

FIG. 2. Patient 6: still living. Born December 2008; photo February 2013; 50 months.

FIG. 2. Patient 6: still living. Born April 2008; photo February 2013; 58 months

common t18 medical issues with intensive treatment (e.g., cardiac surgery) in this sample.

Confirm and Disconfirm Previous Findings

This study reports on several commonly cited t18 issues with a sample of survivor over the age of 1 year. As such, medical conditions and their resolution in this group was markedly different compared with population-based literature [Rasmussen et al., 2003; Niedrist et al., 2006; Crider et al., 2008; Vendola et al., 2010]. The TRIS project largely represents surviving children in marked contrast to literature describing review of hospital registries, for example. Since data collection began in February 2007, 250 surveys have been completed on children with various trisomy conditions (t18, t13, t9) living at least 2 months. Further, data on presenting conditions at birth and their resolution are not found in many existent studies outside of the work underway in Japan [Kosho et al., 2006, 2013; Kaneko et al., 2008; Kobayashi et al., 2010].

The majority of existing studies do not include information about prenatal diagnosis. For example, Kosho et al. [2006] presents prenatal sonographic findings but does not indicate when the data were gathered. Method of prenatal diagnosis (amniocentesis or ultrasound) is offered by Kaneko et al. [2008] but not when

FIG. 3. Patient 1: still living. Born March 2005; photo March 2010; 60 months.

diagnosis was confirmed. Interestingly, Burke et al. [2012] focus on correlations with live birth for infants diagnosed before and after 20 weeks gestation. The authors conclude, "The findings from this study support the view that long-term survival for fetuses with prenatally diagnosed trisomy 18 appears to be much lower than when the condition is diagnosed postnatally, and is probably close to zero" [p. 2]. Better outcomes for infants without a prenatal diagnosis are described by Janvier et al. [2012]. A third of the current sample received a prenatal diagnosis during the second or the third trimester with the remainder receiving the t18 diagnosis within 2 weeks of birth. Importantly, mean age of the sample at the time of TRIS Survey completion was approximately 30 months.

Several current studies provide information about newborn characteristics. Results vary for full t18 newborns' gestational age, weight, and length at birth. For example, Kosho et al. [2006] found mean gestational age at 37 weeks, 5 days and another study at 39 weeks, 2 days by Lin et al. [2006]. Niedrist et al. [2006] point out longer gestation increasing survival as was evident in this sample with a mean of 39.1 weeks (range = 36–44 weeks). [Two studies [Baty et al., 1994; Lin et al., 2006] include birth weight (mean = 2195.39 g, mean = 1,977 g, respectively). In addition, Boghossian et al. [2012] report that infants with t18 were likely to have low birth weight and high mortality rate. Finally, Lin and colleagues report length (mean = 42.8 cm). The sample described here had comparable mean gestational age and longer length. Birth weight data indicated a mean weight of 2166.36 g (SD ± 333.42) with a range of 1,730–3,260 g. Interestingly, this result is between Baty et al. [1994] and Lin et al.'s [2006] finding but toward the higher mean weight. It is possible that longer gestation and higher birth weight contributed to survival in the immediate postnatal period and beyond. Data such as this along with recent findings by Kosho et al. [2013] may be utilized to differentiate survivors and non-survivors. Similar to data described here, Kosho et al. also found that diagnosis after birth and ability to feed orally without medical assistance as factors supporting survival. Of the sample described here, 23% of infants were able to feed orally at the time of survey completion. In addition, many of the infants did not require a prolonged hospital stay after birth, which may be an additional indicator of survival past 1 year for this population.

Data are available on cardiac surgery for infants with t18 in Japan. For example, Kobayashi et al. [2010] reported positive results for five infants with VSD repair with two still living over 2 years after surgery. Seven children post-VSD repair are described here along with surgeries to correct additional t18 cardiac anomalies such as ASD and PDA. TRIS project staff is further analyzing post-cardiac surgery survival data to determine longitudinal outcomes on this sample and a group of older survivors (over 60 months at the time of completion of baseline TRIS Survey). Other study samples also describe cardiac surgery with varying results [e.g., Graham, 2004; Nelson et al., 2012].

Feeding methods are another area not discussed in the present literature outside of Kosho et al. [2013] describing improved outcomes for oral feeders. The data here indicate a variety of feeding needs and their resolution. Bruns and Springer, [2013] report in more detail on feeding methods over time including introduction of enteral (tube) feeding and identification and treatment of reflux. This area requires further study to learn about the feeding needs in infancy and beyond.

Apnea is mentioned as a cause of death in studies on t18 [Embleton et al., 1996; Vendola et al., 2010]. Conversely, discussion of treatment of central and obstructive apnea is sparset. In our study, for some infants, apnea was a condition that changed over time and was treated accordingly. Importantly, while supplemental oxygen was needed during the postnatal period for a majority of the sample, the use of more invasive mechanical support was only noted for five infants. With the paucity of such data, it is unclear if this group is unique. Regardless, this finding warrants the need to include further respiratory-related data in subsequent investigations.

Jones [2006] describes a higher incidence of kidney issues in the t18 population than reported here (10–50% vs. 5–10%). It is unclear how this sample is different from the data set used by Jones and there is a dearth of studies for comparison. For this sample, it appears that kidney issues were not life threatening and necessary treatments were provided for their resolution. This is in contrast to the t18 profile described by Jones [2006] (Table IV).

Limitations

A diagnosis of full t18 and child aged 12–59 months at time of survey completion were criteria used to identify the sample described here. As such, the sample was small and purposive and reflected data already in the TRIS project database. As a result, this group is not representative of this condition. Results cannot be generalized.

Non-completion of survey items resulting in an incomplete data set for analysis (noted in the Results and accompanying Tables). The authors posit that non-completion may be due to limited access to medical documentation for the immediate postnatal period or possible unfamiliarity with medical terminology presented in the survey. In addition, it was not feasible to contact mothers up to 4 years after TRIS Survey completion to request data due to personnel limitations. Yet, it is important to note that mothers in this sample were able to recount detailed information highlighting an advantage of parent report for investigations such as this one compared to data that is population-based [Rasmussen et al., 2003; Niedrist et al., 2006; Vendola et al., 2010].

The data were collected from present reports and not from medical records. However in recent years this methodology is currently widely used and considered valid. Since data collection began, the first author has encountered additional information about common t18 medical conditions. Due to the nature of data collection, this information could not be added, as all participants must complete the same survey. Survey items cannot be changed or updated due to the continuing nature of the project.

Implications

Often, parents with a child with t18 are encouraged to forgo "heroic measures" due to the "lethal" diagnosis. This appears to be due to the overwhelming focus on mortality rather than addressing medical issues of survivors. For example, Courtwright et al. [2011] and

TABLE IV. Jones [2006] Profile Compared to the TRIS Project Sample (n = 22)

	Jones [2006]	TRIS project sample at birth (n = 22)	TRIS project sample at survey completion (n = 22)
Atrial septal defect	≥50%	55% (12/22)	36% (8/22)
Patent ductus arteriosis	≥50%	59% (13/22)	36% (4/22)
Ventricular septal defect	≥50%	86% (19/22)	55% (12/22)
Feeding difficulties	States likelihood of occurrence without a percentage	95% (21/22)	77% (17/22)
Apnea	States likelihood of occurrence without a percentage	50% (9/18)	11% (2/18)
Additional respiratory needs (ventilator support or tracheostomy)	States likelihood of occurrence without a percentage	29% (5/17)	9% (2/22)
Horseshoe kidney	10–50%	5% (1/22)	5% (1/22)
Double ureters	10–50%	5% (1/22)	5% (1/22)
Polycystic kidneys	10–50%	5% (1/22)	5% (1/22)
Hydronephrosis	10–50%	5% (1/22)	5% (1/22)

Goc et al. [2006] recommend against use of invasive medical interventions. Yet, there are authors who emphasize the need for medical professionals to take parent priorities and preferences into consideration in treatment decisions regardless of the t18 diagnosis [Yates et al., 2011]. Carey [2012] emphasizes the need to report both a balanced approach for parents so they can arrive at decisions on behalf of their infant. The data presented here are intended to empirically add to Carey's recommendations and be shared with parents prenatally so they can arrive at decision-making with knowledge of the range of possible outcomes rather than a sole emphasis on mortality. The new findings by Kosho et al. [2013] of greater longevity, successful medical interventions and positive family outcomes (e.g., child interacts with family members, parents' ability to adapt to their child's needs) should also be noted. Janvier et al. [2012] also emphasize positive parent appraisals related to their child with t18.

Treatment decisions need to be reached based on a combination of principles such as "best interest" of the infant [Merritt et al., 2012], parent preferences based on balanced information [Carey, 2012] and, importantly, on individual infant physical characteristics and medical condition. A recent study provides encouraging findings on positive outcomes for children with t18 and their families regardless of medical needs and longevity [Janvier et al., 2012]. In addition, in the first author's experience, mothers of young children with t18 report variations in their access to medical professionals who are knowledgeable about the condition. They also describe a range of positive and negative experiences with accessing services for their child.

Focusing on cardiac conditions, feeding methods, apnea, respiratory difficulties, and kidney issues also serves to highlight the expected medical complications for t18 survivors [Jones, 2006] along with consideration of treatment with an eye toward long-term survival. As the data indicate, surgeries were performed and treatment was provided to children deemed "incompatible with life." In addition, Lantos [in Boss et al., 2013] states "many clinicians object to life-sustaining treatment of infants with trisomy 13 and 18…These views are no longer tenable. Many infants with these trisomies survive for years." It is imperative to share this information to increase awareness of positive outcomes rather than solely emphasizing early mortality [e.g., Romesberg, 2007; Everett and Albersheim, 2011; Merritt et al., 2012]. There is no assurance that medical interventions will be successful but offering them is an initial step to changing views about this population.

McCaffrey [2011] also points out the need to view quality of a child's life rather than diagnostic label (t18) especially when providing treatment options to parents during the immediate postnatal period. Nelson et al. [2012] report the provision of medical interventions to children over 12 months of age. The authors also state, "to the degree that a uniform "non-intervention" paradigm ever existed in the past, though, the patterns of care over the past decade and a half suggest that such a paradigm is no longer universal."

In counterpoint to Merritt et al. [2012] and other authors who question providing interventions for this population, it is clear that this sample received intensive treatments and surgeries with positive outcomes. Nelson et al. [2012] and Graham [2004] also note the trend toward interventions. In addition, parent-reported data indicates quality of life (QoL) as largely positive with the children enjoying daily activities and routines and being a valued member of their family [Janvier et al., 2012].

Children with t18 deserve opportunities to grow and thrive. We recommend that their parents receive information about all treatment options with an emphasis toward positive outcomes than the futility of aggressive care [Bruns and Crosier, in preparation; Wilkinson et al., 2012]. Medical professionals commonly involved with these cases (e.g., neonatologist, clinical geneticist, pediatric cardiologist) must utilize the most current empirical research rather than reliance solely on mortality studies. These recommendations are coupled with Carey [2012] viewpoint "…recommend [ing] a balanced approach to counseling families of the newborn with trisomy 18 and 13 at the time of diagnosis and at decision points in management, that is, in the delivery room, newborn nursery, and clinic. The components of this counseling process should include presentation of accurate figures for survival that take

into consideration the individual clinical findings of the child, avoidance of language that assumes outcome…"

CONCLUSION

Results provide a new perspective on survivors over the age of 1 year with full t18 based on the success of intensive treatment of common medical conditions associated with the syndrome. Data also highlight the need for decision-making, taking into account individual characteristics rather than general, diagnosis-specific recommendations. In order to reach this outcome, additional research is needed to further examine this clinical subgroup.

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