SIU Southern Illinois University
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TRIS project findings: A resource for parents and professionals

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TRIS
Tracking Rare Incidence Syndromes

The Tracking Rare Incidence Syndromes (TRIS) project seeks to increase awareness and knowledge for families and professionals touched by rare trisomy conditions and aims to facilitate improved decision making for optimal services and supports for affected children and their families.

This session will provide an overview of research findings of the Tracking Rare Incidence Syndromes (TRIS) project highlighting responses to parent requests for data on medical conditions and developmental information.
Data has also been shared with Facebook groups and medical professionals with the intent of raising awareness and knowledge of needs and outcomes for children with these conditions.

Annabel, 9 years

TRIS project components

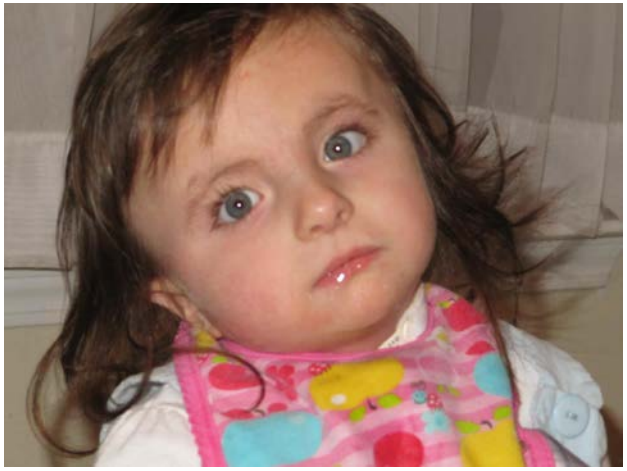
- TRIS Survey (baseline survey) collects information on birth history, medical issues, developmental progress and family support needs. In addition, TRIS Follow-up Survey collects annual updates on key items from the TRIS Survey (e.g., surgeries). TRIS Modified Survey for infants living up to 60 days.
- Dissemination of results through presentations and publications
- Collaboration with families and experts in field
- Outreach for raising awareness and recruitment

Types of inquiries

- Parents seeking assurance
  - Incidence questions regarding specific conditions
  - Treatment questions
  - Outcomes
- Trisomy syndrome specific questions
  - Trisomy 18 mosaic
  - Trisomy 9p
  - Partial trisomy 6p

Trisomy 18 parent inquiries

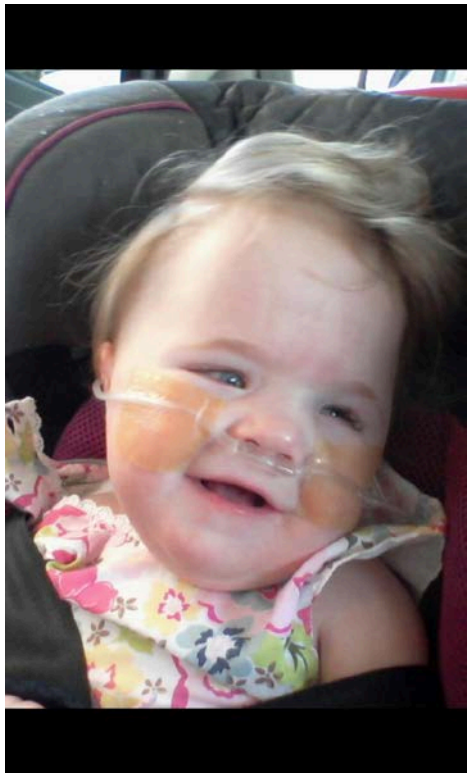
- Birth plans (expectant father with son with prenatal diagnosis of trisomy 18; passed in utero)
- Reflux in children with t18 (mom with daughter with t18 via Facebook; culminated in a publication)



- Tracheostomies and respiratory interventions in children with full trisomy 18 (moms on Trisomy 18 Mommies Facebook group)
- Incidence of microcephaly and hypothyroidism in children with trisomy 18 mosaic (mom with son with trisomy 18 mosaic via Facebook)

Trisomy 13 parent inquires

- Incidence of outcomes of hypoplastic left heart syndrome (pregnant mom with daughter with prenatal diagnosis of trisomy 13)
- Developmental milestones in trisomy 13 (dad with infant with t13)
- Oral health in children with trisomy 13 (mom with daughter with trisomy 13 mosaic via Facebook; preparing a manuscript including trisomy 18 as well)



Trisomy 9 parent inquiries

- Communication modalities in children with trisomy 9 variants (several parents on Trisomy 9 Mosaic Families on Facebook)
- Incidence of seizures and management (dad with son with trisomy 9 mosaic)
- At 2013 trisomy 9 gathering:
  - Incidence of apnea (mother of infant with trisomy 9 mosaic)
  - Speech therapy (several parents asked)
- Request to gather data on children with trisomy 9p (mother with adult son with t9p via Facebook)

Designing a birth plan

- Expectant dad with son with prenatal diagnosis of trisomy 18 (passed in utero)
- Discussed options regarding:
  - Monitoring during labor
  - Oxygen, ventilation and other types of respiratory support
  - Feeding options
  - Parameters of Do Not Resuscitate order
- Interest in collecting and analyzing birth plans and disseminating findings to obstetricians and neonatologists

Reflux in children with trisomy 18

Mom with daughter with trisomy 18 via Facebook; culminated in a publication:

Bruns, D. A. & Springer, S. A. (2013). Feeding changes in children with trisomy 18: Longitudinal data on primary feeding method and reflux identification and treatment. *Topics in Clinical Nutrition, 28*(4), 324-334.

Research indicates that approximately 40% to 70% of children with disabilities and identified and treated for feeding difficulties such as reflux. The available literature on children with trisomy18 does not describe feeding needs or treatment. The results described here address primary feeding method along with identification and longitudinal treatment of gastroesophageal reflux in 10 children with trisomy 18. Data indicate videofluoroscopic swallowing study as the primary diagnostic procedure and treatment with medication as largely effective. The majority of the sample also moved to tube feedings to address feeding difficulties. It is hoped that this information is an initial step to further understanding the nutritional needs of this unique population. Implications for practice and a need for further research are recommended.

Tracheostomies and respiratory interventions in children with full trisomy 18 (n=22)

- Of 22 children between the ages of 12 and 59 months with full trisomy 18, three (14%) required a tracheostomy. At the time of survey completion only two (66%) continued to use one.
- Sixteen (72%) participants reported their infant had respiratory difficulties while in the NICU. One infant received a tracheostomy prior to being discharged from the NICU; the infant was intubated and required a CPAP regulator before being released. This infant has since had their tracheostomy removed before 23 months of age.
- The two remaining infants had a tracheostomy procedure at 4 and 10 months respectively. Both infants required a ventilator while in the NICU. It was reported by the parent of the 10 month old, the child was given a tracheostomy as a means of treating central and obstructive apnea. The second parent indicated that her child had central apnea before the tracheostomy was placed.

Tracheostomies and respiratory interventions in children with full trisomy 18 (n=22) continued

- At the time of survey, 33% (n=7) of parents felt that breathing problems are their child's most critical medical need. Two of the seven (29%) children are those who currently have a tracheostomy, and use a ventilator for breathing assistance. Both parents have indicated that they are able to meet their child's need in that area.
- Of the remaining parents (n=5, 71%) only one (20%) parent does not feel that they are able to meet the needs of their child's problems with obstructive apnea and reflux. This child currently requires a CPAP machine as a means of addressing apnea.

Two inquiries about trisomy 18 mosaic

- Mom with son with trisomy 18 mosaic via Facebook:
- Incidence of microcephaly
  - Perinatal diagnosis
  - Diagnosed after birth
  - Comorbid brain anomalies
- Incidence of hypothyroidism
  - Related trisomy diagnosis
  - Management and treatment effectiveness
  - Age of diagnosis

Incidence and outcomes in infants with trisomy 13 diagnosed with hypoplastic left heart syndrome

- Pregnant mom with daughter with prenatal diagnosis of trisomy 13; mom also asked about “trisomy 13 friendly” neonatal intensive care units in her area

- Identified three children in TRIS project database (two infants and one five year old female child); infants passed away without intervention. Remaining child received interventions for respiratory and feeding needs, not for cardiac condition. Child lived in the same state; reached out to mom and two moms were in contact.

Milestones reached by children with trisomy 13 by 15 months

- Question was asked by father with a child with full trisomy 13. He was interested in how his daughter's development compared with children of the same age with full trisomy 13. Child was 15 months of age and functioning at a 3-9 month level.

- The comparison sample was comprised of four children between 12-18 months with full trisomy 13 at completion of Developmental Matrix included in TRIS baseline survey.

1-3 month milestones reached by young children with full trisomy 13

1-3 Month Milestones	Yes	No	% Achieved
Thrusts arms or legs	3	1	60%
Coordinates breathing sucking swallowing	4	0	100%
Turns eyes towards voice	4	0	100%
Follows faces with eyes	4	0	100%
Molds when held	3	1	60%
Raises head and chest when lying on stomach	3	1	60%
Opens and closes hands	4	0	100%
Swipes, grasps, and shakes small toy	4	0	100%
Coos or social smile	4	0	100%
Produces open vowel sound(s)	3	1	60%

7-9 month milestones reached by young children with full trisomy 13

7-9 Month Milestones	Yes	No	% Achieved
Rolls from back to stomach	2	2	50%
Transfers to sitting position	2	2	50%
Sits without support	2	2	50%
Explores objects with hand and mouth	4	0	100%
Engages in social play (peekaboo)	3	1	60%
Show preference for familiar adults	2	2	50%
Demonstrates preference for objects	1	3	20%
Explores with hands for more than 2 minutes	2	2	50%

Trisomy 13 and trisomy 18 oral health data

- Mom with daughter with trisomy 13 mosaic via Facebook
- Analyzed data for children with full, mosaic, partial and other types of trisomy 13 (translocations) in the following areas:
  - Frequency of dental visits
  - Excessive plaque
  - Enamel hypoplasia (soft/absent enamel)
  - Tooth decay
  - Periodontal disease
- Preparing a manuscript including trisomy 18 types as well; available literature was sparse

Language development in children and adults with trisomy 9 mosaic

- Data based on 26 participants aged 4-289 months with trisomy 9 mosaic completing the TRIS Full Survey. Twenty-one (81%) listed speech therapy as a critical learning need. 25 (95%) total participants had been enrolled in speech therapy prior to taking the survey.
- At the time of survey completion, 18 (69%) were still receiving speech/ language therapy an average of 60 minutes per week. Six (23%) participants received services at school, seven (27%) at their home, seven (27%) at a center, and four (15%) at a clinic. Also, at the time of survey completion, 19 (73%) participants were fed orally. One (3%) participant received feeding specific therapy from an SLP.
- Augmentative and assistive communication (AAC) devices: Use of devices in at least one or more settings including home, school and community. Several children were noted to use sign language as their primary method of communication.

Trisomy 9 AAC use (n=36)

Age	At Home in the Past	At Home Now	Outside Home in the Past	Outside Home Now
1-3 (n=14)	0	0	0	0
3-5 (n=7)	1 (14%)	0	1 (14%)	1 (14%)
6-8 (n=4)	2 (50%)	3 (75%)	3 (75%)	3 (75%)
9-11 (n=2)	2 (100%)	2 (100%)	2 (100%)	2 (100%)
12-14 (n=2)	2 (100%)	1 (50%)	2 (100%)	1 (50%)
15-20 (n=2)	2 (100%)	1 (50%)	1 (50%)	1 (50%)
21+ (n=5)	2 (40%)	0	2 (40%)	1 (20%)
Total= 36	**11 (31%)**	**7 (19%)**	**11 (31%)**	**9 (25%)**

Prevalence of seizures in trisomy 9 mosaic and partial trisomy 9

- Dad with son with trisomy 9 mosaic asked about incidence of seizures and management

- Based on June 2013 data, of 54 children and adults with trisomy 9 types, seven (13%) reported seizures prior to survey completion.
  At the time of survey completion, no additional participants reported seizures and only three (43%) participants continued to report seizure activity. Seizure management data was also collected.

Seizure management in trisomy 9

Participant	Diagnosis	Types of Seizures before Survey	Seizures at Survey Completion	Medication(s) prescribed	Most effective medication
1	t9 mosaic	UT	None	None	n/a
2	t9 mosaic	PM	PM	Epilim	Epilim
3	t9 mosaic	GM, TC	TC, GM	Depakote, Klonapin	no answer
4	Partial t9	UT	None	Phenobarbital	no answer
5	Partial t9	GM	None	None	n/a
6	Partial t9	TC	None	Dilantin	Dilantin
7	Partial t9	GM, M, PM, TC	GM, M, PM, TC	Clonazepam, Diastat,Keppra, Tegretol	None

Notes: GM= Grand mal; M= Mixed type PM= Petit mal; TC= Tonic clonic; UT= Unknown type

Prevalence of apnea in children with trisomy 9 mosaic

- Asked by the mother of an 18 month old child with trisomy 9 mosaic at the 2013 trisomy 9 gathering. Child had not been released from hospital due to complications with apnea.

- Of 24 children and adults with trisomy 9 mosaic and partial trisomy 9. two (9%) children were diagnosed with central apnea and six (25%) with obstructive apnea. Two (33%) children had an adenoidectomy and tonsillectomy. One (17%) child with obstructive apnea had an adenoidectomy and one (17%) child had a tonsillectomy. One (17%) child with obstructive apnea had an adenoidectomy, tonsillectomy, and received a tracheostomy.

For additional information

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TRIS project website:
http://web.coehs.siu.edu/Grants/TRIS/
http://web3.coehs.siu.edu/tris/PreEnroll.php
http://www.coehs.siu.edu/tris/casestudies.html

TRIS project Facebook page:
https://www.facebook.com/TRIS.Trisomy.project