

TRIS project: A year in review (2016-2017)

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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.

TRIS project components

- TRIS Modified Survey for infants living up to 60 days.
- TRIS Full Survey (baseline survey) collects information on birth history, medical issues, developmental progress and support needs
- TRIS Follow-up Survey collects annual updates on key items from the TRIS Survey
- Dissemination of survey results through presentations and publications
- Outreach for raising awareness and recruitment

A year in review: 2016-17

- Update on survey completions
- Recruitment of new enrollees
- Parent requests
- New publications
- In process data analyses and manuscripts
- Upcoming activities

TRIS Survey completions

Completions as of July 15, 2017:

Modified TRIS Survey = 221/257; 85.9%

Full TRIS Survey = 473/814; 58.2%

TRIS Follow-up Survey

Year 1 = 288

Year 6 = 45

Year 2 = 204

Year 7 = 32

Year 3 = 145

Year 8 = 17

Year 4 = 102

Year 9 = 5

Year 5 = 62

Year 10 = 2

TRIS Survey completions continued

Completions for t9m = 81

Completions for t8m = 29

In process to complete a third manuscript for t9m and plan to work on initial data analyses and write up for t8m; will be, to date, largest sample of condition

Recruitment of new enrollees

- Outreach via project's Facebook page
 - Post updates with project news, during Trisomy Awareness month etc.
 - Respond to messages/follow-up
- Posting on and responses to posts and comments on various trisomy related Facebook pages
- Google alerts for "Trisomy in the News"
- Parent to parent contact
- Responding to emails requesting information

Parent requests

Topics including:

- Intestinal issues and resolution
- Seizure activity and treatment
- Success of medical interventions including cardiac surgery
- Trisomy type of long-term survivors (Follow-up survey Years 7-9)
- Resources to share with medical team such as case studies and outcome data

New publication

Donovan, J. H., Krigbaum, G. & Bruns, D. A. (2016). Medical interventions and survival by gender of children with trisomy 18. *American Journal of Medical Genetics Part C*, 172(3), 272-278. (Special issue)

Research has typically shown limited aggressive medical interventions and low survival rates for children with full trisomy 18. Recent studies provide more positive results. ***This study examined 82 children with full trisomy 18 drawn from the Tracking Rare Incidence Syndromes (TRIS) project database. Children were classified into three groups according to the highest intervention received: “hospice or no intervention” (n.5, 6.1%), “necessary interventions (enteral feeding, ventilator use)” (n.46, 56.1%), and “aggressive interventions (surgery)” (n.31, 37.8%).*** Seven of 14 male children (50%) and 52 of 68 female children (76.5%) were living at the time of survey completion. Additionally, information about any interventions used during the care of these children was also provided. It was found that three males (37.5%) and 28 females (48.3%) had used hospice care at some point; 12 males (85.7%) and 61 females (89.7%) received enteral feeding at some point; 7 males (58.3%) and 25 females (38.5%) had ventilator; and 7 males (50%) and 33 females (48.5%) underwent some form of surgery. ***These results suggest improved outcomes when given necessary and aggressive medical interventions.*** Implications and recommendations for further research are provided.

In process data analyses and manuscripts

- Data trends over time in prenatal diagnosis in children with trisomy 18 and after birth care
- Apnea and its treatment in children with trisomy 18
- Cardiac surgery in children with full trisomy 13
- Data from Modified survey examining cardiac conditions

In process data analyses and manuscripts continued

- **Trisomy 8 mosaic** sample (n=29; will be largest group described in the literature)
- Auto-inflammatory conditions in children with **trisomy 8 mosaicism**; recruiting for researcher at the National Institutes of Health (NIH); recruitment is ongoing
- Phenotypic, medical and developmental data in sample with **trisomy 9 mosaicism** (n=31; in addition to published articles with n=14 and n=25, respectively)

Online modules (spring 2017)

Preparation of online modules for parents and medical professionals describing prenatal markers, phenotype, medical and health needs and maintenance, and developmental strengths of children diagnosed with trisomy 18, trisomy 13 and trisomy 9 mosaicism. Additional module will highlight trisomy 8 mosaicism. Inclusion of photos in all modules.

Each module includes a PDF of a Power Point presentation, resource list and glossary of medical terms.

Requests for feedback from parents and professionals.

Online modules

Modules are available in the following ways:

- TRIS project website at <http://tris.siu.edu/modules/index.php>
- by request via TRIS project Facebook page
- Link posted to SOFT website at <http://trisomy.org>
- Link posted to International Trisomy Alliance website at <http://www.internationaltrisomyalliance.com>
- as requested, by postal mail

Screen shot of Online modules introduction page

The screenshot shows a web browser window displaying the TRIS project online modules introduction page. The browser's address bar shows the URL tris.siu.edu. The page features a navigation menu on the left with links for Home, Flyer, Survey, Informational Modules, Case Studies, TRIS Project Update, Publications and Presentations, Give to TRIS, Photo Gallery, and Staff. Below the navigation menu is a 'Contact Us' section for the TRIS Project, listing Dr. Deborah A. Bruns as the contact person, along with her title, phone number (618-453-2311), email (dabrun@siu.edu), and physical address (Wham Building, Room 223, MC 4616, Carbondale, IL 62901). Social media icons for Facebook, Twitter, and Instagram are also present.

The main content area is titled "TRIS project online modules for families and medical professionals". It contains the following text:

To date, over 1000 families from around the world have enrolled in the TRIS project. Data from submitted surveys have led to 15 publications and over 20 presentations (national and international meetings and conferences). In addition, 12 case studies were developed in 2014 and are available for download at <http://tris.siu.edu/case-studies/index.php>.

Yet, a recurring need has been availability of concise and accessible information to showcase the abilities and needs of children diagnosed with low incidence trisomy conditions. To this end, online modules have been developed providing historical background, relevant literature, overview of daily and medical care, and recommendations for monitoring key medical needs based on research and anecdotal reports. Additional family support information has been included highlighting positive aspects of the experience along with unmet needs. Finally, quotes from newspaper articles describing raising a child with a low incidence trisomy condition are also shared to draw attention to several common as well as unique issues families face.

It is hoped these modules are shared with expectant parents, families with a child diagnosed with full trisomy 18, full trisomy 13 or trisomy 9 mosaicism and also with medical professionals and others caring for the child including therapists and teachers. It is especially important to disseminate to those on a child's medical and developmental/educational team so that the focus is on strengths within the context of addressing needs and facilitating continued growth.

Questions and comments are welcomed, please send to tris@siu.edu (project email address) and/or dabrun@siu.edu (Principal Investigator's email address).

Below the text are three tabs labeled "T18 MODULE", "T13 MODULE", and "T9M MODULE". The "T18 MODULE" tab is selected, and the content below it includes:

- TRIS Project Full T18 Online Module
- Resource List for Trisomy 18
- Glossary of Medical Terms

On the right side of the page, there is a vertical banner with two photographs of children. The top photo shows a young girl with blonde hair, smiling, wearing a blue patterned shirt. Below the photo is the caption "Nora, 4 years old, Trisomy 13". The bottom photo shows a young girl with dark hair, wearing a pink shirt, holding a doll. Below the photo is the caption "Twila, 5 years old, Trisomy 18". Above the banner is a small graphic with the text "Trisomy TRIS ... OSIS" and a "Like" button. At the bottom of the page, there is a "MISSION STATEMENT" link.

Online modules continued


Sample slide from t18 module

Phenotype

Facial features including:

- Prominent occipital bone
- Short palpebral fissures
- Hypoplasia of orbital ridges
- Low-set ears
- Micrognathia
- Cleft lip and/or palate (*low incidence*)

Karson
DOB 07/23/13



Jones KL, Jones MC, & Del Campo M. (2013).
Smith's recognizable patterns of human malformation. Elsevier Health Sciences.

MacBook Pro

Online modules continued

Sample slide from t13 module

Phenotype

Facial features including:

- Microphthalmia
- Colobama of iris
- Cleft lip (60-80%)
- Cleft palate
- Low-set ears
- Shallow supraorbital ridges
- Upslanting palprebral fissures
- Absent philtrum



Jones KL, Jones MC, & Del Campo M. (2013).
Smith's recognizable patterns of human malformation. Elsevier Health Sciences.

Zion
5/2/01-2/16/10

Online modules continued

Sample slide from t9m module


Phenotype

Facial features including

- Microphthalmia
- Broad, bulbous nose
- High arched or cleft palate
- Low-set ears
- Hypertelorism
- Short palpebral fissures

Jones KL, Jones MC, & Del Campo M. (2013).
Smith's recognizable patterns of human malformation.
Elsevier Health Sciences.

UNIQUE:
<http://www.rarechromo.org/information/Chromosome%20%209/Trisomy%209%20mosaicism%20FTNP.pdf>



Alana
DOB 12/9/09

MacBook Pro

Upcoming activities

- New TRIS project logo – thanks to Kathy (mom to Mackenzie, t9m)
- Case studies highlighting unique medical needs and interventions
- Key interventions (e.g., cardiac surgery, tracheotomy) and positive outcomes across and within rare trisomy conditions (t18, t13, t9, t8)
- Analyze and write up family support data

Sample mock up of logo



Additional ideas for the next year

- 1.
- 2.
- 3.
- 4.
- 5.

For additional information

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TRIS project website links:

<http://tris.siu.edu> (homepage)

<http://web3.coehs.siu.edu/tris/PreEnroll.php>

<http://www.coehs.siu.edu/tris/casestudies.html>

TRIS project Facebook page: over 1500 Likes

<https://www.facebook.com/TRIS.Trisomy.project>