SIU Southern Illinois University
CARBONDALE

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 2 = 204
  - Year 3 = 145
  - Year 4 = 102
  - Year 5 = 62
  - Year 6 = 45
  - Year 7 = 32
  - Year 8 = 17
  - Year 9 = 5
  - 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.

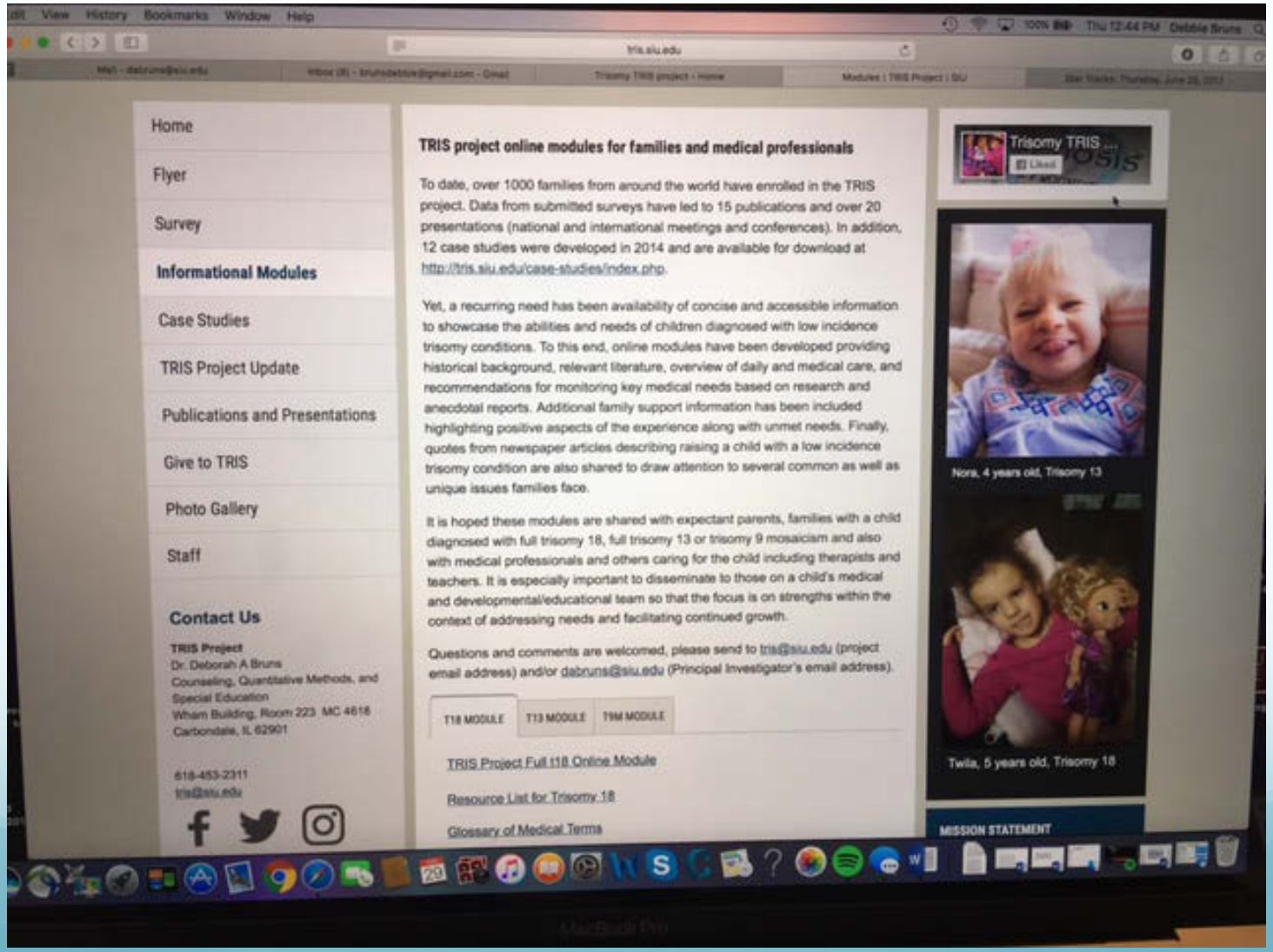
Requests for feedback from parents and professionals.

Online modules

Modules are available in the following ways:

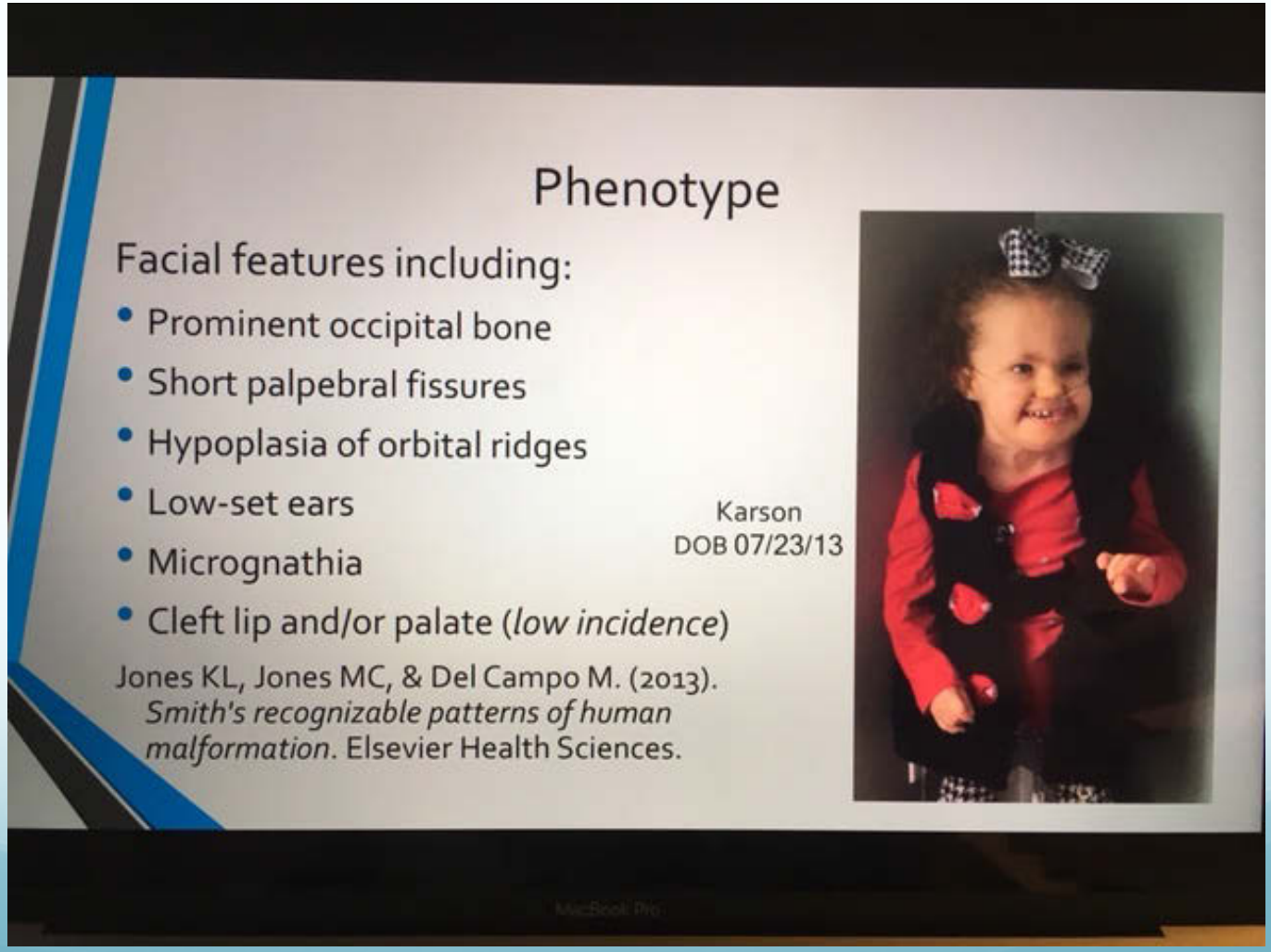
- TRIS project website at http://tris.siu.edu/modules/index.php
- by request via TRIS project Facebook page
- Link posted to SOFT website at http://trisomy.org
- Link posted to International Trisomy Alliance website at http://www.internationaltrisomyalliance.com
- as requested, by postal mail

Screen shot of Online modules introduction page



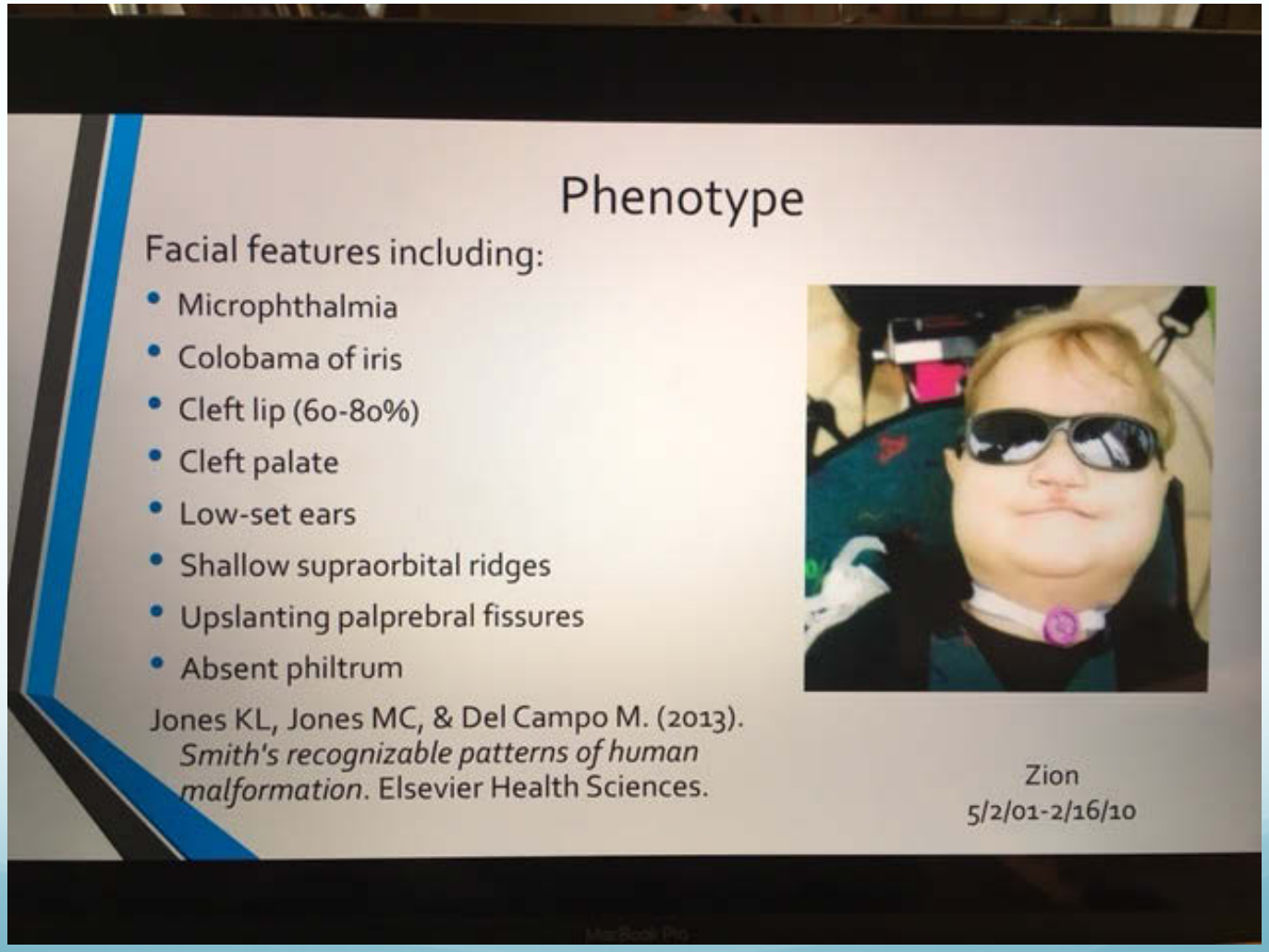
Online modules continued

Sample slide from t18 module



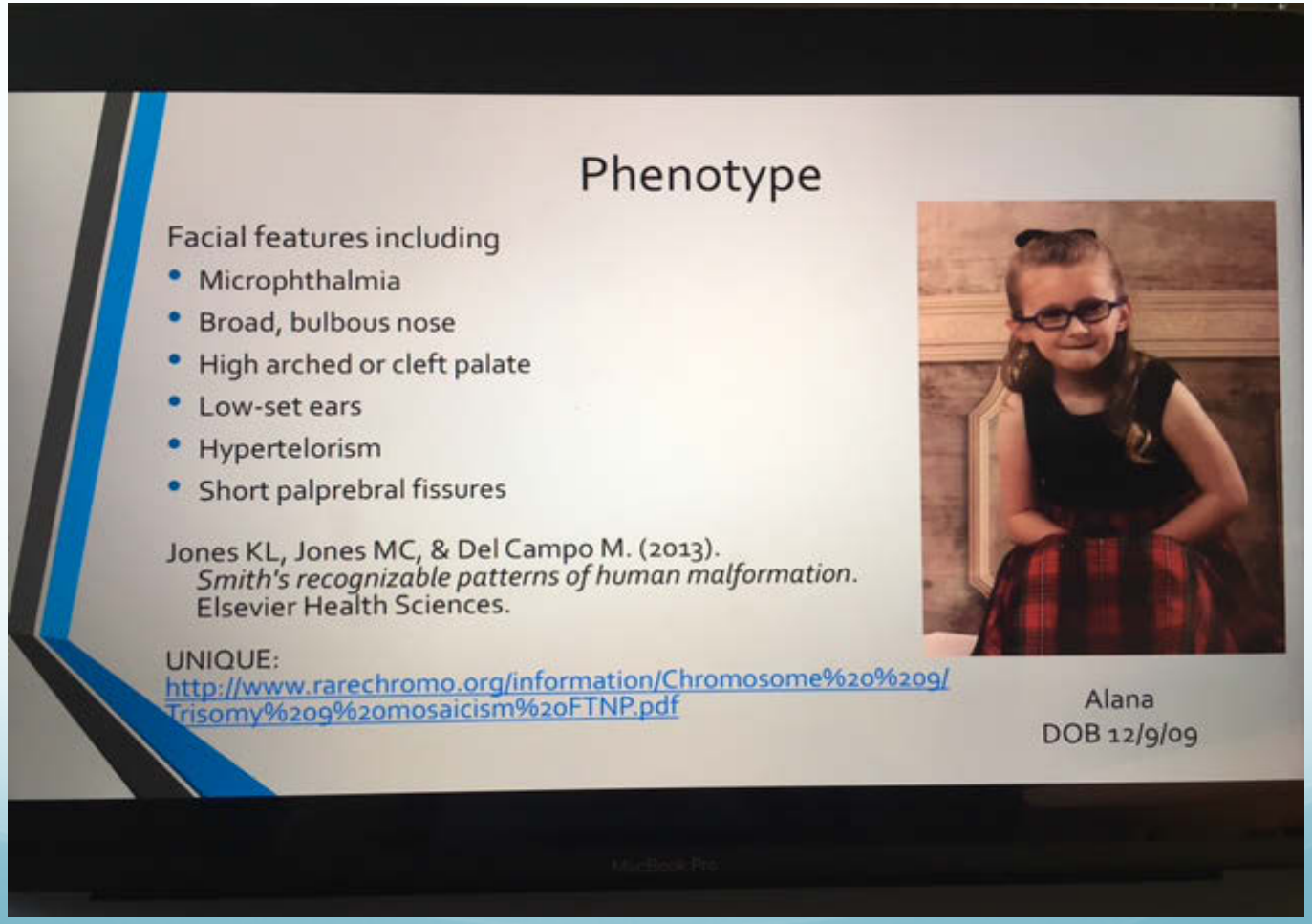
Online modules continued

Sample slide from t13 module



Online modules continued

Sample slide from t9m module



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