TRIS Project: Trisomy 9 results

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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.
Origins of the TRIS project

- Principal Investigator’s experience with young children with full Trisomy 18 in early 1990’s
- Discussions on Tri-family and Tri-med lists including prenatal issues, family needs, working with professionals & medical concerns
- TRIS was “born” in 2003; Advisory Committee and a planning group formed; pilot data collected in 2005-06; online TRIS Survey launched 2/1/07
TRIS project components

- Database development through parent-to-parent contacts, links with rare trisomy groups and online searches
- TRIS Survey, Full and Modified versions, include items about birth history, medical issues, developmental progress and support needs. In addition, annual TRIS Follow-up Survey for annual updates in key areas
- Dissemination through presentations & publications
- Collaboration with experts in field
- Outreach for raising awareness and recruitment
201 TRIS Full Surveys completed since February 1, 2007 including 52 with full Trisomy 18 (26%) and 31 with full Trisomy 13 (15.5%)

166 were still living at time of survey completion (some in adulthood)

166 live in the United States

169 married and 146 identified as middle income

Majority completed 13 or more years of formal education
Available literature on Trisomy 9 types

- Much of the available literature provides results of prenatal testing (e.g., Benacerraf, Pauker, Quade, Bieber, 1992; Schwartz, Ashai, Meijboom, Schwartz, Sun & Cohen, 1989) or fetal autopsy (e.g., Smoleniec, Davies, Lunt, Berry & James, 1993; Yeo, Waldron, Lashley, Day-Salvatore & Vintzileos, 2003)

- There are also case studies of living children such as Sanchez, Fijtman & Migliorini (1982). The largest sample to date is sonographic findings for six infants described by Schwendemann and colleagues (2009)
TRIS project database: Trisomy 9 participants

- 35 children and adults with a type of Trisomy 9 (17.5%): 15 with Trisomy 9 mosaic, seven with partial Trisomy 9, eight with Trisomy 9p and five with partial Trisomy 9p; one passed away at age of nine months
- Two represented in TRIS Modified Survey (infants living 60 days or less): one with Trisomy 9 mosaic and one with partial Trisomy 9 – both were stillborn
- Mothers age ranged 24 to 41 years ($M=32.9$, $Sd=5.04$ years); range for entire group ($n=200$) was 17 to 45 years ($M=31.2$, $Sd=6.07$)

($M$ = mean, $Sd$ = Standard deviation)
Marital status: 30 married, one single, one separated, two divorced and one widowed

Educational level: three had 10-12 years of schooling, 16 with 13-16 years of schooling, 14 with 17-20 years and two with more than 20 years

Income level: 25 identified as middle income, five low income and five high income; no $$ ranges specified due to international scope of project
• 29 live in the United States, three in Canada and one each in Belgium, England and Sweden illustrating the global reach of the project
Overview of all T9 types (n=26 April 2010)

- Age at time of survey completion ($M=89.8$ months, range=2 - 468 months, SD=108.58 months); results indicated the presence of low set ears, small jaw and cleft palate for some children in the sample.

- Birth information: Gestational age: $M=38.9$ weeks; Birth weight: $M=2681.5$ grams and Length: $M=48$ centimeters

($M = \text{mean}$)
Overview of all t9 types

- Few cardiac anomalies were reported: five of 24 with ASD, five of 24 with PDA and two of 24 with VSD (data not available for remaining two participants)

- Immediate postnatal period
  - Feeding (18 of 25)
  - Respiratory difficulties (seven of 25)

Fourteen long-term survivors with trisomy 9 mosaic ($M=78.6$ months, range=2 - 293 months, $SD=89.32$ months; 3–99% mosaicism) (one deceased at time of survey completion); Results indicated the presence of low set ears and small jaw for some children in the sample.

Gestational age: $M=38.2$ weeks, Birth weight: $M=2228.6$ grams and length: $M=46.3$ centimeters
Few cardiac anomalies were reported (n=4 ASD, n=2 PDA, n=1 VSD). Feeding (n=11) and respiratory difficulties (n=5) in the immediate postnatal period.

Data from TRIS Developmental Matrix indicated a wide range in functioning level.

“Results presented here are the largest series focusing on long-term survivors with trisomy 9 mosaicism” (p. 1036)
Developmental Matrix results (n=13)

- Wide range of skill levels; all demonstrate preferences for people and objects
- Strengths in language and communication
  - Imitates gestures
  - Says, signs or gestures to indicate “no”
  - Asks/signs “wh” questions
- Strengths in social skills
  - Engages in social play with parent
  - Demonstrates responsibility for own belongings
  - Plays cooperatively with same age peers
Developmental Matrix results examples

- 28 months at time of survey completion
  - Scribbles with crayon or pencil
  - Attempts to throw or kick a ball
  - Walks unassisted

- 293 months at time of survey completion
  - Associates representation with object
  - Climbs stairs
  - Able to transfer in and out of chair and on and off couch
Implications for further research

- Areas for further study: long-term survivors in this sample exhibited several cardiac defects with ASD was most identified. Almost all children were identified with feeding difficulties and one third of the sample experienced respiratory difficulties.
- Developmental information indicated a wide range in functioning level; need for longitudinal study using the TRIS Follow-up Survey.
- Some items specific to the physical characteristics and presenting medical conditions of T9 types were not included in TRIS Survey development (e.g., micrognathia [small jaw], bulbous nose, skeletal anomalies); need for T9 specific data collection instrument for further study.
Implications and Conclusions

- Development of a care book similar to SOFT publications for T18 and T13, including medical conditions, developmental outcomes etc.
- Continued outreach through family support groups, social media and parent contacts to increase number of T9 families participating in TRIS project
- *Raise awareness of this unique subgroup*
For additional information

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TRIS project website:
http://web.coehs.siu.edu/Grants/TRIS/