

Tracking Rare Incidence Syndromes (TRIS) project
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Began in 2007, the Tracking Rare Incidence Syndromes (TRIS) project seeks to increase the knowledge base on rare incidence trisomy conditions, and to make this information available to families and interested educational, medical and therapeutic professionals. Related outcomes will be the development of appropriate services, advance treatment options, and supports during the prenatal, newborn and subsequent developmental periods. Children affected by T18, T13, and other rare syndromes possess unique needs requiring education of those involved in their daily, educational and medical care.

Below is a list of publications based on TRIS project data (reference information and Abstract; PDF copies are available by request to dabrunsi@siu.edu):

2014

Brunsi, D. A. & Campbell, E. (2014). Twenty-two long-term survivors with full trisomy 18: Presenting and current medical conditions. *American Journal of Medical Genetics Part A*. DOI: 10.1002/ajmg.a.36318

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.

Brunsi, D. A. (2014). Caring for an infant with trisomy 18: A case study and guidelines. *Clinical Nursing Studies*, 2(1), 30-36. DOI: 10.5430/cns.v2n1p30

Much of the literature describes the prognosis for infants with full trisomy 18 as bleak. The case study offered here provides an overview of an infant with this diagnosis who lived for 88.5 days in a Level III neonatal intensive care unit. Care decisions (medical and basic care) and their outcomes are described along with implications for care for infants with a diagnosis often characterized as incompatible with life. The need for information about treatment options and collaboration among an infant’s medical team is emphasized.

2013

Brunsi, 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. DOI: 10.1097/01.TIN.0000437408.58941.d4

Research indicates that approximately 40% to 70% of children with disabilities are identified and treated for feeding difficulties such as reflux. The available literature on children with trisomy 18 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.

Bruns, D. A. (2013). Erring on the side of life: Children with rare trisomy conditions, medical interventions and quality of life. *Journal of Genetic Disorders and Genetic Reports*. DOI: 10.4172/2324-9331.1000103

The prevailing viewpoint on children with rare trisomy conditions such as trisomy 18 (t18) and trisomy 13 (t13) is almost uniformly negative. Yet, case studies offer information about long-term survivors. What is missing in the discussion is an unbiased examination of surviving children within the context of necessary, rather than “aggressive”, medical interventions and overall quality of life. A move beyond palliative or comfort care must be an option for this population. There must be a move toward valuation of life and corresponding provision of treatment and examination of developmental gains rather than limited intervention or palliative care for infants with lethal fetal abnormalities. This article presents a call to examine the individual child rather than decision making by diagnosis framed by recommendations from the Convention on the Rights of the Child (CRC) and Convention on the Rights of Persons with Disabilities (CRPD) Medical professionals and parents must work together to ensure medical needs are met and a positive quality of life can be achieved.

2012

Bruns, D. A. & Schrey, C. (2012). Examining in-home care needs and work responsibilities for parents with children with a rare trisomy condition. *International Journal of Developmental Disabilities*, 58(3), 159-175. DOI: 10.1179/2047387712Y.0000000002

Balancing in-home care needs and work responsibilities can present many challenges for families. This can be especially true with a family member with a disability. Mothers with children with severe disabilities are often not able to continue working in the job they held before their children’s birth or had hoped to attain after becoming a parent. The present study explores parent perspectives related to in-home care needs and work responsibilities for their child with a rare trisomy condition, themselves, and their spouses. Results describe the interplay of children’s daily caregiving needs, time management, mothers’ change in work status, and spouse’s work outside the home. Daily caregiving largely contributed to mothers not working outside of the home due to their children’s basic and medically related needs as well as a lack of suitable caregivers. In addition, parents in this study often had to reduce work hours or leave the workforce to care for their child. A call for additional research is coupled with implications for service providers working with families with a member with a rare trisomy condition.

2011

Bruns, D. (2011). Birth history, physical characteristics, and medical conditions in long-term survivors with full trisomy 13. *American Journal of Medical Genetics Part A*, 155(11), 2634-2640. DOI 10.1002/ajmg.a.34283

The purpose of the study is to provide data about long-term survivors with full trisomy 13 (t13). Mothers of 30 long-term survivors with full t13 completed an online survey. Survey data were downloaded into an SPSS database. Descriptive statistics were used to analyze survey data. Tracking Rare Incidence Syndrome (TRIS) Survey data on survival, birth information including maternal and paternal age at conception, physical characteristics, and medical conditions were compared. Data indicate longer mean survival rates (48.4 months for those living at the time of data collection, 40.8 months for those who died prior) than described in the literature. Means for gestational age, birth weight, and length are 38.11 weeks, 2,789.34 g and 48.45 cm, respectively. Long-term survivors presented with syndrome related physical characteristics (e.g., low-set ears,

cleft lip and palate) and medical conditions (e.g., ventricular septal defect (VSD), feeding difficulties). We conclude that data indicate longer survival and a range in birth information (gestational age, birth weight, and length) along with presence of common presenting physical characteristics and medical conditions of long-term survivors with full t13.

Bruns, D. (2011). Presenting physical characteristics, medical conditions and developmental status of long-term survivors with trisomy 9 mosaicism. *American Journal of Medical Genetics Part A*, 155(5), 1033-1039. DOI 10.1002/ajmg.a.33928

Much of the available literature on individuals with trisomy 9 mosaicism focuses on reports concerning results of prenatal testing or fetal autopsy with limited reports of long-term survivors. Data from the Tracking Rare Incidence Syndromes (TRIS) project offer the largest series to date examining the presenting physical characteristics and medical conditions at birth for 14 individuals. Results indicated the presence of low-set ears and microcephaly for some children in the sample. Cardiac anomalies were reported along with feeding and respiratory difficulties in the immediate postnatal period. In addition, developmental status data indicated a wide range in functioning level with examples of demonstrated skills provided. Implications for professionals caring for patients with trisomy 9 mosaicism are offered.

Bruns, D. & Foerster, K. (2011). 'We've been through it all together': Supports for parents with children with rare trisomy conditions. *Journal of Intellectual Disability Research*, 55(4), 361–369. DOI 10.1111/j.1365-2788.2010.01381.x

Parenting a child with a developmental disability can be a positive experience. A salient part of this outcome is support at the time of diagnosis and in an ongoing manner from immediate and extended family members. Studies are sparse on this topic for parents with a child with a rare trisomy condition. The present study examined the support needs of parents with a child or adult with a rare trisomy condition (n = 20). Participants were recruited from the Tracking Rare Incidence Syndromes (TRIS) project. The TRIS Family, Friends and Finances Protocol was the data collection instrument. The protocol included primarily open-ended items. Qualitative analyses were conducted to identify themes from the protocol and follow-up phone contacts. Support from immediate and extended family members varied from very positive to participants describing very negative interactions with specific individuals. Many in the sample reported affirming experiences with spouses and difficulties with grandparents and other extended family members. Results both confirmed the literature and reflected the unique circumstances of the participants. It is critical to raise awareness of the similar and disparate support needs of this unique population, as the affected children are living longer and their families require continuing support to meet their and their children's needs.

2010

Bruns, D. A. (2010). Neonatal experiences of newborns with full Trisomy 18. *Advances in Neonatal Care*, 10(1), 25-31.

PURPOSE: To provide information about neonatal experiences for newborns with full trisomy 18 (t18).

SUBJECTS: Mothers of 21 newborns with full t18 (13 survivors; 8 who died prior to mothers' participation in the study).

DESIGN: Mixed method, descriptive, online survey.

METHODS: Subjects completed an online survey. Data were downloaded into an SPSS database. Descriptive statistics were used to analyze resulting data.

MAIN OUTCOME MEASURES: Subjects' responses on Tracking Rare Incidence Syndromes survey.

PRINCIPAL RESULTS: Newborns presented with syndrome-related physical characteristics (e.g., low-set ears, small jaw) and medical conditions (e.g., heart defects). Gestational age was generally older than what is described in the existent literature. In the NICU, newborns were

provided with a variety of equipment (e.g., nasal cannula, pulse oximeter) and received treatments including blood transfusions and echocardiograms. Data also indicate longer survival rates (103.3 months for survivors, 37.5 months for non-survivors) than described in the literature. CONCLUSIONS: Data indicate presence of common presenting physical and medical characteristics and varied medical assistance to newborns with full t18.

2008

Bruns, D. A. (2008). Pregnancy and birth history of newborns with Trisomy 18 or 13: A pilot study. *American Journal of Medical Genetics Part A*, 146A(3), 321-326.

DOI: 10.1002/ajmg.a.32147

Data presented here (20 children; seven with trisomy 18, 13 with trisomy 13) are an initial step of the TRIS project to increase the knowledge base for families with newborns and older children with rare trisomy conditions. The TRIS project also plans to provide professionals and families with data and resources to facilitate decision-making on behalf of surviving children and young adults. (Research Letter)

2006

Bruns DA. 2006. Tracking rare incidence syndromes (TRIS) project. *American Journal of Medical Genetics Part A*, 140A, 2510.

The TRIS project is aimed at both families and professionals in the hopes of increasing and expanding information related to daily care, medical interventions and developmental accomplishments as a means toward greater understanding in decision-making in children and adults with rare trisomy syndromes. We will begin by examining families with a child or adult with trisomy 18 or 13 and then expand to include rarer trisomies. The TRIS survey is currently in revision and will be available in print and electronic formats by the end of the year. The project will begin in the United States and expand to other English-speaking countries and, ultimately, be launched on a worldwide scale.