

PROFESSIONAL VIEWPOINT: Journal Club Perspectives on the Care and Advances in the Management of Children with Trisomy 13 and 18 , *American Journal of Medical Genetics, Part C, Seminars in Medical Genetics, September 15, 2016* (whole issue) by Drs. John C Carey and Tomoki Kosho

On September 15 of this year, the *American Journal of Medical Genetics, Part C* published an entire special issue devoted to the trisomy 13 and trisomy 18 syndromes. The issue included 9 papers, consisting of an Introduction by the two Guest Editors of the issue (Carey and Kosho), two Commentaries, one Editorial, and five Original Research Articles.

The main purpose of the collection of articles in this issue of the Seminars in Medical Genetics series is to continue and ideally expand the ongoing dialogue that has emerged in the medical literature in the last decade regarding the care of infants with the two conditions. This dialogue relates to the controversy that is very familiar to the readership of this newsletter: on one side is the conventional approach of initiating comfort care and holding off on intervention for the management of newborns and infants while the opposing view argues for a more balanced approach to ongoing care that offers intervention when appropriate depending on the individual child (summarized by Carey, 2012). These contrasting views have created what was referred to by one of the papers in this series (Andrews and colleagues) as "a palpable tension."

The first group of papers in this issue deals with perspectives on care and research studying interventions in children with trisomy 13 and 18. The second group of papers documents advances in the management of specific medical manifestations, i.e., seizures and tumors. In the rest of this piece we will highlight the papers in the issue. (The following summary is adapted from the Introduction by Carey & Kosho, 2016).

The opening Commentary by Dr. McCaffrey plunges immediately into the dialogue regarding the level of care in individuals with trisomy 13 and trisomy 18. He argues eloquently against the use of the terms "lethal" and "incompatible with life", still commonly applied labels after initial diagnosis. The essay summarizes the key papers in recent years regarding interventions and makes a "plea for truth, transparency, and recognition of our prejudices regarding patients" with the two syndromes.

The other Commentary is by Andrews and colleagues, including SOFT's own Scott and Vivian Showalter. These authors propose a model for care in the trisomy 13 and trisomy 18 syndromes that uses "shared decision making as a foundational principle" and the pathways approach as a method. The paper reviews the chronology of the thought process that led us to the current controversy about care. The authors provide a detailed Table (we would consider to be the centerpiece of this work) that is designed to be a guide for applying the approach. (The Conference that helped lead to this article was partly funded by SOFT.)

The next three papers involve interventions, procedures, and events in children with trisomy 13 and 18. Josephsen and colleagues (including Dr. Steve Braddock) from St Louis University perform a single center review of procedures performed in children with trisomy 13 or 18 over a 15-year period. These authors show an increasing rate of procedures per patient over this period of time.

In the next article Dr. Debbie Bruns and colleagues utilize the Tracking Rare Incidence Syndrome (TRIS) database and examine medical interventions and survival rate in 82 children with full trisomy 18. The authors discuss their results and its implications for future research.

The last paper in this group of studies on interventions is the questionnaire study of Drs. Janvier and Barrington and Barbara Farlow. These authors invited parents of children with trisomy 13 and 18 to answer a survey online; 261 participated. Parents demonstrated "common hopes" when they received a diagnosis of one of these conditions. The authors suggest that "rigorous transparency regarding specific interventions and outcomes may help personalize care for these children."

The last two papers discuss specific manifestations that can occur in infants and children with trisomy 18. The first of these by Matricardi and colleagues from Italy is a multicenter study of the clinical aspects, EEG features, and neuroimaging in children with full trisomy 18 and associated epilepsy. The last article is the paper by Satge and co-authors, who reviewed the literature and present the occurrence of tumors in trisomy 18. The authors show that children with the syndrome are at increased risk for a liver tumor (hepatoblastoma) and a kidney tumor (Wilms tumor).

Carey and Barnes accompany this article with an Editorial that summarizes the evidence for the association of Wilms tumor in the trisomy 18 syndrome and uses the SOFT Registry, a parent-reported outcome database (created by Ann Barnes), to estimate the risk for a child with the syndrome to develop the tumor.

**The cover figure of the issue is a composite group of photographs of seven SOFT children taken by esteemed photographer, Rick Guidotti, at the recent SOFT Conference in Tacoma, WA. All of these papers mentioned above will be accessible on www.trisomy.org in early October.

REFERENCE Carey JC. 2012. Perspectives on the care and management of infants with trisomy 18 and trisomy 13: Striving for balance. *Curr Opin Pediatr* 24:672-678.