

PROFESSIONAL VIEWPOINT: Journal Club

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Guon J., Wilfond BS, Farlow B, Brazg T, Janvier A. 2013. *Our children are not a diagnosis: The experience of parents who continue their pregnancies after a prenatal diagnosis of trisomy 13 or 18. Am J Med Genet Part A*, December 5. Epub ahead of print.

Nishi E, Takamizawa S, et. al, and Kosho T. 2013. *Surgical intervention for esophageal atresia in patients with trisomy 18. Am J Med Genet Part A*, December 5. Epub ahead of print.

Two important articles have recently appeared on the online version of the *American Journal of Medical Genetics*, and both will be published in print in the February 2014 issue. These 2 papers provide new knowledge that is helpful and significant in the care and management of children with trisomy 13 and 18.

The first of the two papers (cited above) is authored by the group led by Barb Farlow and Dr. Annie Janvier, who have extended their questionnaire studies of parents of persons with trisomy 13 and 18 belonging to various internet-based support groups, some of which include SOFT families. The authors surveyed parents where the baby had been diagnosed to have trisomy 13 or 18 during pregnancy, and the women chose to continue their pregnancy. Since pregnancy termination rates in Europe for prenatally diagnosed pregnancies of trisomy 13 and 18 are over 90%, investigating the impressions and experiences of the families who continue is crucial to our understanding of the challenges that the families face. The majority of parents did perceive some pressure to terminate, and most were told that the baby “would likely die before birth.”

What is most important for healthcare professionals and providers is that the parents describe “special” caretakers who gave balanced and personalized information, respected their choice, and provided support. The insights gained from this article are likely familiar to many families reading this piece, but this information is crucial – I would assert – to healthcare providers.

The second paper by Nishi and colleagues is authored by the Japanese group led by Dr. Kosho and is additionally helpful in care and management. About 5% of children with trisomy 18 will have the gastrointestinal malformation called esophageal atresia (EA) with a tracheoesophageal fistula (TEF). There have been a number of ethical discussions and published articles over the years about the management of newborns with trisomy 18 who have the EA and TEF as part of the syndrome. Dr. Nishi and Kosho and their coauthors looked at the outcome of newborns with trisomy 18 who underwent a palliative surgical approach compared to those who had received more “radical” surgery for the EA/TEF. The outcome in the two groups was different with those receiving the radical surgery having 27% 1-year survival rate compared to 0% in the palliative surgery group. This work – like Dr Kosho’s seminal paper in 2006 – suggests (although based on only 24 children) that, when more intervention occurs, 1-year survival of infants with trisomy 18 increases. This issue is important, because, prior to Dr.’s 2006 paper on intensive respiratory care, the question of what would happen if an infant with trisomy 18 was treated with more intervention was essentially unknown. This work addresses that question for one specific group (those with EA/TEF- about 5% of children) and adds more knowledge to this ongoing puzzle.