



Perspectives on the care and management of infants with trisomy 18 and trisomy 13: striving for balance

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Purpose of review

At the time of diagnosis of the trisomy 18 and trisomy 13, parents and care providers face difficult and challenging decisions regarding management. Because of the increased infant mortality and developmental outcome associated with both conditions, the conventional approach to management has been to withhold technological support. In recent years, an active dialogue on this topic has emerged. The purpose of this review is to summarize the literature on the outcome of infants with trisomy 18 and 13 and to discuss the key themes in this emerging dialogue.

Recent findings

In recent years, several important studies have appeared that have analyzed the issues relevant to this topic, including parental autonomy, best interest of the child standard, and quality of life. Some authorities state that in areas of ambiguity it is best to defer to parents' views, whereas others indicate concern that the best interest standard has given way to parental autonomy. Information on the actual experience of parents of children with trisomy 18 and 13 has been limited until recently.

Summary

The author recommends a balanced approach to counseling families of the newborn with trisomy 18 and 13 at the time of diagnosis. The counseling process should include presentation of accurate survival figures, avoidance of language that assumes outcome, communication of developmental outcome that does not presuppose perception of quality of life, and respect for the family's choice, whether it be comfort care or intervention.

Keywords

congenital malformations, developmental disability, infant mortality, lethal conditions, trisomy 13 syndrome, trisomy 18 syndrome

INTRODUCTION

The trisomy 18 and trisomy 13 syndromes comprise two important pediatric conditions that – despite their well-recognized neonatal mortality – deserve close attention and interest among those of us who care for children. I make this point for two reasons: after trisomy 21/Down syndrome, trisomy 18 and trisomy 13 represent the second and third most common autosomal trisomy syndromes; their combined total prevalence (elective termination of pregnancies, stillbirths, and live births), is approximately one of 1800, which makes the experience of having a baby with either a very common event (over 2000 US families annually) [1,2^{***}]; both syndromes consist of a recognizable pattern of multiple congenital anomalies with increased neonatal and infant mortality and significant

intellectual disability in older children, making care challenging for the family, the primary care practitioner, and the specialist [3].

The conventional approach to management of neonates with the trisomy 18 and 13 syndromes has been a withholding of technological support and surgery with the provision of comfort care, more recently under the guidance of palliative care teams.

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Curr Opin Pediatr 2012, 24:000–000

DOI:10.1097/MOP.0b013e3283595031

KEY POINTS

- The conventional approach to management of neonates with the trisomy 18 and 13 syndromes has been a withholding of technological support and surgery with the provision of comfort care, more recently under the expert guidance of palliative care teams.
 - Since 2003 and especially in the last 4 years, an active dialogue on the choices in the care and management of infants with trisomy 18 and 13 has emerged in the pediatric and bioethics literature.
 - The themes that are relevant to this discourse include parent autonomy, best interest of the child standard, futility, burden of treatment, allocation of resources, and quality of life.
 - The author recommends a balanced approach to counseling families of the infant with the syndromes that includes presentation of accurate figures for survival, avoidance of language that assumes outcome, accurate communication of developmental outcome that does not presuppose a family's perception of quality of life, and the recognition of the family's choice, whether it be comfort care or interventions.
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