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Perspectives on the Care of Children with the Trisomy 18 and 13 Syndromes

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The trisomy 18 and 13 syndromes represent the second and third most common autosomal chromosome disorders in humans after Down syndrome. The conditions are particularly unique among birth defect syndromes because of the combination of the high neonatal and infant mortality, the increased frequency of major structural defects especially of the heart, and the consistent occurrence of significant psychomotor and cognitive disability among older children. The conventional approach to care and management of children with these trisomy syndromes is pure comfort care and the avoidance -and sometimes "denial"- of technological intervention. Population studies of mortality from the 1980s through the last decade showed that about 50% of infants died in the first week of life, and only 5 -6% of children were alive at their first birthday. Two studies in North America published in 2016 documented an increase in survival to 12-14% by 12 months of life. The authors of the investigations suggested that increased intervention and procedures during care in recent years could account for this change in survival in the children with the syndromes. Additionally some authors have suggested that there has been a" shift in the paradigm of care" with the model of shared decision being frequently implemented in management. The author recommends involving parents in decision making from the onset, counseling parents in non-biased ways free of their own view towards the quality of life, and using current outcome data in counseling. Avoidance of terms such as "lethal" and "incompatible with life" is important and appreciated. Future studies could investigate the hypothesis that increased intervention accounts for the recent improved survival figures and analyze the quality of life of parents of children and their perception of the child's quality of life. Parents appreciate a goal-directed approach that includes a treatment plan informed by the child's individual physiological problems rather than by the diagnosis of the trisomy syndrome..

Biography

John C. Carey, MD, MPH, FAAP, FACMGG is Professor and formerly Vice Chair of Academic Affairs, Department of Pediatrics, at the University of Utah. Throughout his career, Dr. Carey has been interested in birth defect syndromes and the care of children with these conditions. Dr. Carey graduated from Villanova University in 1968 with an A.B. and obtained his M.D. from Georgetown University School of Medicine in 1972. He trained in pediatrics, & genetics and dysmorphology as a resident and fellow at the University of California San Francisco, 1972-1979. Dr. Carey obtained an M.P.H. from the University of California at Berkeley in 1976 in between his residency and fellowship years. Dr. Carey joined the faculty at University of Utah Health Sciences Center in 1979. He became Chief of the Division of Medical Genetics in 1985 and remained in that leadership position until 1999 when he stepped down to assume the role as Editor-in-Chief of the American Journal of Medical Genetics. He held that editorial position from 2001-2016 and is currently Editor-in-Chief Emeritus. Dr. Carey established the Medical Genetics Fellowship Program at the University of Utah and continues as a mentor in the Program. Dr. Carey's research focus has been in congenital malformations, neurofibromatosis, and syndrome delineation. He has authored or co-authored over 300 papers, chapters, invited articles, and editorials for scientific journals. He co-authored the textbook, "Medical Genetics," by Jorde, Carey, & Bamshad, now in its 6th edition. The book is a widely used text in schools of medicine throughout North America and Europe. Dr. Carey has served as medical adviser and "founding professional" for the Support Organization for Trisomy 18, 13 and Related Disorders (SOFT) since 1980. The medical and ethical aspects of care of infants and children with these important conditions are currently one of his major academic interests.

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