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Clinical Pearl:
Trisomy 13 and Trisomy 18: Current Approach

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> *"She had a single central eye and a proboscis, which did not work; as long as she cried, she was pink. She also had polydactyly, and we sent chromosomes, as this was about 45 years ago. She had trisomy 13 and lived about 4 or 5 hours."*

It was my first morning as a neonatal attending, and my senior resident called and asked me to come to see this female newborn who had just been born. I said, "sure. I will be right there!" When I arrived at the ISCU, I heard the baby crying, and then she stopped. She had a single central eye and a proboscis, which did not work; as long as she cried, she was pink. She also had polydactyly, and we sent chromosomes, as this was about 45 years ago. She had trisomy 13 and lived about 4 or 5 hours. Later that month, we had a stillborn fetus with trisomy 13 and a liveborn newborn with Trisomy 18. Each time I called our excellent, thoughtful geneticist, a general pediatrician, Dr. Ira Salafsky, to see each newborn infant and counsel the families. This was before prenatal evaluations, including quad screens and evaluation of free fetal cells, were being evaluated. This was 1982 or 1983.

> *"What is clear from the literature is that each case should be individualized, and the family should be provided clinical information about their fetus and newborn as early as possible, along with outcome data depending on the potential clinical approach to management (1-3). It is also essential to provide parents with data about survival, quality of life, and developmental outcome for these infants (1-5)) so families and clinicians can make thoughtful decisions about management and approaches. (6)"*

I decided to talk with my colleagues, and we started a multidisciplinary care group to evaluate each anomalous fetus and talk with each family, just as mentioned in the article by Cortezzo and colleagues and the editorial by Dr. John Carey regarding diagnostic evaluation and therapeutic approach (1,2). There are several excellent articles from around the world on clinical approaches to fetuses and newborns with Trisomy 13 and 18 referenced in the Cortezzo article, and I think the basic principles of the approach include serial discussions with families with relevant clinicians beginning in the prenatal period with the goal of formulation of a clinical plan. What is clear from the literature is that each case should be individualized, and the family should be provided clinical information about their fetus and newborn as early as possible, along with outcome data depending on the potential clinical approach to management (1-3). It is also essential to provide parents with data about survival, quality of life, and developmental outcome for these infants (1-5)) so families and clinicians can make thoughtful decisions about management and approaches. (6)

> *"A total of 92% were offered postnatal resuscitation (1). Seventy-two (58%) fetuses were liveborn. Forty seven% of families elected comfort care, and 43% elected to pursue invasive treatment to extend life (l). The remaining 10% chose noninvasive therapies (1). The median length of survival for Trisomy 13 and 18 infants was 7 and 29 days, respectively (1). "*

To give each of us an idea, here are some outcome data from the 125 pregnancies from Cortezzo and colleagues. One hundred twenty-five pregnancies in this retrospective study, and 6% ended in a spontaneous loss, 19% in intrauterine fetal demise, 17% in termination of pregnancy, and 58% in a live birth (1). Univariate analysis, male fetal sex, lower maternal gravitate, and fetal hydropower resulted in higher spontaneous fetal loss (1). A total of 92% were offered postnatal resuscitation (1). Seventy-two (58%) fetuses were liveborn. Forty seven% of families elected comfort care, and 43% elected to pursue invasive treatment to extend life (l). The remaining 10% chose noninvasive therapies (1). The median length of survival for Trisomy 13 and 18 infants was 7 and 29 days, respectively (1).

Of the four infants with T 13 and 23 infants with T 18 who were alive at one month of age, 50% of T 13 and 48% of T 18 infants were alive at one year of age, with a median length of survival of 111 days and 590 days respectively. The one-month and one-year survival rates of children managed initially with comfort care were 15% and 6%, respectively (1). 74% ofT 18 infants and 44% of T 13 infants were discharged from initially from the NICU. Of the 32 infants discharged home, nearly 1/2 were readmitted, frequently with respiratory insufficiency and apnea (1). 90% had evidence of congenital heart disease (1). Consistent with our study data, a model shows that certain variables, including mechanical ventilation, cardiac surgery, gastrostomy, and parenteral nutrition, predict survival to 6 months (1). "Despite profound neurodevelopmental impairments, these patients meet early developmental milestones and have meaningful interactions with their families (1)."

References:

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