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Congenital cardiac defects in trisomy 18 - challenges and dilemmas

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Trisomy 18 also known as Edwards" syndrome is the second most common trisomy syndrome after trisomy 21. The syndrome pattern consists of major and minor anomalies involving multiple organ systems, pre and post-natal growth deficiency and marked psychomotor and cognitive disability. Cardiovascular manifestations are seen in almost 90% of these patients. However traditionally, corrective and palliative cardiovascular procedures are rarely undertaken in view of the "lethal" nature of the syndrome.

A female baby conceived of IVF treatment, was born by LSCS at 35 weeks of gestation with a birth weight of 2.1 kg, head circumference of 30cm and length of 40cms. She had a stormy neonatal course with respiratory distress syndrome and moderate sized patent ductus arteriosus (PDA) requiring mechanical ventilation for 7 days and oxygen dependence for much longer. Subsequently, marked failure to thrive continued; weight remained static at 2.8 kg after 3 months. At 5th month, when the child was reviewed again, presence of characteristic dysmorphic features and severe growth failure prompted a chromosomal analysis, the karyotype of the child was 47,XX,+18 which confirmed the diagnosis of trisomy 18. Cardiovascular examination revealed no cardiomegaly and no murmur, with a palpable second heart sound and mild lower limb desaturation. Chest radiograph suggested mild peripheral pruning. Echocardiogram showed a severe pulmonary hypertension with bi directionally shunting PDA suggestive of early pulmonary vascular obstructive disease. She was started on dual pulmonary vasodilator therapy. Prior to a planned reassessment a month later, she deteriorated due to aspiration pneumonia and required intensive care and was stabilized and discharged after 5 days. A month later she had a second episode of bronchopneumonia that was complicated with sepsis and prolonged oxygen dependence. After a prolonged hospital stay, she was stabilized and discharged home on oxygen therapy, but succumbed at home after a few days.

As in Trisomy 21, early development of pulmonary hypertension is a known entity even in Edwards" Syndrome. Simple and common cardiac defects like PDA and septal defects are quickly rendered inoperable due to pulmonary vascular disease. Traditionally, cardiovascular procedures are rarely undertaken in these patients in view of the known poor survival beyond infancy. Amongst the multitude of anomalies and restricted potentials of this chromosomal syndrome, cardiac defects are one of the most correctable and may help to achieve reduction in morbidity, reduce the number of hospitalizations, facilitate discharge from hospital and may help in improving growth. Despite the high neonatal mortality rates and poor survival, early cardiovascular diagnosis and prompt intervention may have a role in improving the quality of life of these babies and their families, if not in prolonging it.

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