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Case Report

Cardiac care in trisomy 18: A path to improved outcomes (case report)

Abdullghany Dowaikh, MD [a], Atif Alsahari, MD [b], Saad Khoshhal, MD [c] and Tarek Momenah, MD, FRCP(C), FSCAI [b,d,*]

[a] *Pediatric Cardiology Department, Prince Sultan Cardiac Center, Riyadh, Kingdom of Saudi Arabia*
[b] *Structural Heart Interventionist, Pediatric Cardiology Department, Prince Sultan Cardiac Center, Riyadh, Kingdom of Saudi Arabia*
[c] *Pediatric Cardiology, Medical College, Taibah University, Al-Munawarah, Kingdom of Saudi Arabia*
[d] *Adjunct Associate Professor, College of Medicine, Alfaisal University, Riyadh, Kingdom of Saudi Arabia*

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الملخص

أهداف البحث: تعتبر متلازمة تريزومي 18 أو متلازمة إدواردز اضطرابا كروموسوميا يتميز بشذوذات تنموية شديدة وعجز إدراكي. تعتبر المضاعفات القلبية من الأسباب الرئيسية للوفيات في هؤلاء المرضى، والدور الذي تلعبه التدخلات القلبية لا يزال محل جدل.

عرض الحالة: نقدم حالة لطفلة حديثة الولادة كاملة المدة تعاني من تريزومي 18، ولدت عن طريق القيصرية الاختيارية. ظهر لدى الوليدة انسداد شرياني رئوي ومجموعة من التشوهات القلبية الأخرى، مما استدعى التدخل الفوري. على الرغم من التحديات الأولية، بما في ذلك نوبة وجيزة من انخفاض التشبع بالأكسجين بعد التدخل، استجابت المريضة بشكل إيجابي لتوسيع الصمام الرئوي بالبالون وإدخال قسطرة طارنة في قناة الشريان الشرياني، مما يوضح الفوائد المحتملة للتدخلات القلبية في مرضى تريزومي 18.

المناقشة: تسلط هذه الحالة الضوء على التطبيق الناجح للتدخلات القلبية في مريض تريزومي 18، مما يتحدى فكرة الرفض العام لمثل هذه العلاجات لهذه الفئة. تشير نتائجنا إلى أن التدخلات الجزئية المختارة يمكن أن تحسن جودة الحياة وتثبت الحالة، مما يدعم الحاجة إلى المزيد من البحوث لوضع إرشادات واضحة للعلاج في هذه الديموغرافية.

الاستنتاجات: تضيف هذه الحالة إلى الأدلة المتزايدة التي تدعم جدوى وفوائد التدخلات القلبية المحتملة في مرضى تريزومي 18، مما يدعو إلى نهج أكثر تخصيصا لمعالجة هؤلاء المرضى.

الكلمات المفتاحية: تريزومي 18؛ متلازمة إدواردز؛ التدخلات القلبية؛ انسداد الشريان الرئوي؛ جودة الحياة؛ الرعاية التلطيفية

Abstract

Background: Trisomy 18 (also known as Edwards syndrome) is a chromosomal disorder characterized by severe developmental anomalies and cognitive deficits. Cardiac complications are a leading cause of mortality in these patients, and the role of cardiac interventions remains controversial.

Case Presentation: We report a case of a full-term baby girl with trisomy 18, born via elective cesarean section. The neonate presented with pulmonary atresia and a series of other cardiac abnormalities, necessitating immediate intervention. Despite the initial challenges, including a brief episode of desaturation post-intervention, the patient responded positively to a balloon pulmonary valvuloplasty and emergency patent ductus arteriosus stent insertion, illustrating the potential benefits of cardiac interventions in patients with trisomy 18.

Discussion: This case highlights the successful application of cardiac interventions in a patient with trisomy 18, challenging the notion of universally denying such treatments to this population. Our findings suggest that selective interventions can improve quality of life and stabilize the condition, supporting the need for further research to establish clear guidelines for treatment in this demographic.

Conclusion: This case adds to the growing evidence supporting the feasibility and potential benefits of cardiac interventions in patients with trisomy 18, advocating for a more individualized approach to treatment.

* Corresponding address: P.O. Box 300-598, Riyadh, 11372, KSA.
E-mail: Tarekmomenah@me.com (T. Momenah)

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Keywords: Cardiac interventions; Edwards syndrome; Palliative care; Pulmonary atresia; Quality of life; Trisomy 18

Introduction

Trisomy 18 syndrome (also known as Edwards syndrome) is a distinctive autosomal chromosomal disorder characterized by the presence of an additional copy of chromosome 18. Among the autosomal trisomy syndromes, it stands as the second most prevalent, with Down syndrome (trisomy 21) being the most common. The seminal elucidation of Edwards syndrome was provided by Edwards et al., in 1960, who meticulously delineated the clinical manifestations of a neonate presenting with multiple congenital anomalies and cognitive deficits. Subsequent research by Smith et al. definitively established the extra chromosome 18 as the root cause of this syndrome.[1]

Cardiac-related complications stand as a leading cause of mortality in individuals with trisomy 18. While the consideration of interventions remains a subject of debate, emerging evidence suggests potential benefits from select minor interventions.[1–4] Within this case report, we present a patient born within our institution, diagnosed with trisomy 18, who underwent successful cardiac intervention. This case serves as a noteworthy illustration of the potential advantages and considerations surrounding such interventions in the management of patients with trisomy 18.

In considering the ethical dimensions of treating patients with trisomy 18, recent literature and evolving medical practice suggest a shift towards a more interventionist approach. Evidence is accumulating in favor of treatments that can meaningfully extend life and enhance its quality, even in the face of complex, life-limiting conditions such as trisomy 18.[5,6] Ethical deliberations are increasingly informed by these findings, aligning medical decisions with the fundamental values of beneficence and non-maleficence. Moreover, within our cultural and religious framework, there is a strong imperative to offer care that may alleviate suffering and improve patient outcomes. This perspective is grounded in the principle of preserving human dignity and underscores the responsibility of healthcare providers to explore all viable options to optimize the quality of life for their patients. Our case reinforces this evolving ethical paradigm, illustrating how tailored cardiac interventions, guided by both medical insight and compassionate care, can yield positive results in the context of trisomy 18.

Case report

We present the case of a full-term baby girl, who was delivered in our hospital by elective cesarean section. Her mother was 38 years old and had gestational diabetes. Her fetal ultrasound showed pulmonary atresia with a hypoplastic right ventricle.

Immediately after delivery, the infant was flat and had cyanosis. The neonatal resuscitation protocol was started for less than 1 min and then the patient's condition improved; her Apgar score was 4 and 8. At 10 min of age, she was still in respiratory distress so she was moved to the Neonatal Intensive Care Unit and given noninvasive mechanical ventilation. Urgent cardiology consultation was done. Echocardiography showed membranous pulmonary atresia, hypoplastic tricuspid valve with a z score of −2.2, hypoplastic right ventricle, small restrictive ventricular septal defect, large atrial septic defect with right-to-left shunt, and moderate to large patent ductus arteriosus (PDA) with left-to-right shunt (Figure 1). Prostaglandin E1 (PGE1) was started immediately.

The patient was noted to have some dysmorphic features in the form of micrognathia and low-set ears, so genetic testing was conducted. To this end, whole exome sequencing was performed, but the results were delayed as it was conducted abroad.

The patient was kept on PGE1 until her weight reached 2 kg. At the age of 1 month, she underwent successful balloon pulmonary valvuloplasty. The patient did well on this intervention and was referred to the Pediatric Cardiac Intensive Care Unit. Her hospital course was smooth, and she was kept for further investigation and imaging for any other noncardiac association. The patient received multidisciplinary care. She suddenly developed desaturation down to 65% 1 month after the initial intervention, so PGE1 was resumed and she underwent emergency stenting of the PDA (Figures 2 and 3).

Subsequently, the patient improved tremendously. She was discharged in a stable condition on the heart failure medications captopril, furosemide, and spironolactone.

Post-stent whole exome sequencing

The patient, now 9 months old, continues to receive follow-up care in our clinic. Despite experiencing failure to thrive and global developmental delays, her cardiac function has shown improvement, her oxygen saturation levels range between 80% and 85%, and she was successfully weaned off the medications, with the family expressing satisfaction with her progress. Neurologically, she has been stable throughout her hospital stay and follow-up. She also faced feeding challenges, common in trisomy 18, which have been managed effectively with nasogastric tube feeding, ensuring adequate nutrition and growth.

Discussion

In conclusion, our case study highlights a potential avenue for improving the quality of life in patients with trisomy 18 through the judicious application of minor palliative interventions. Specifically, cardiac catheterization has emerged as a pivotal intervention, demonstrating notable benefits in terms of reduced hospitalization frequency and a more manageable disease trajectory.[2] Although our patient was not fully diagnosed with trisomy 18 before the procedure, this procedure allowed the patient to be discharged from the hospital to give her more time with her family and reduce the amount of stress to her family. The family has been on board with giving the patient all medical care available since before the results of whole exome sequencing were available.

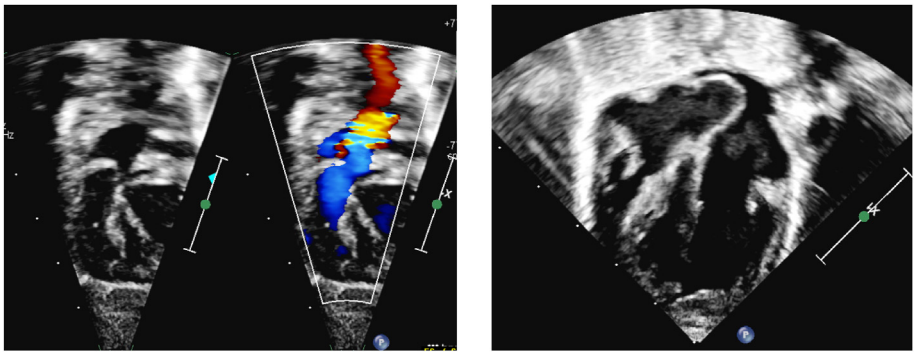

Figure 1: Echocardiography images showing membranous pulmonary valve atresia and hypoplastic right side.

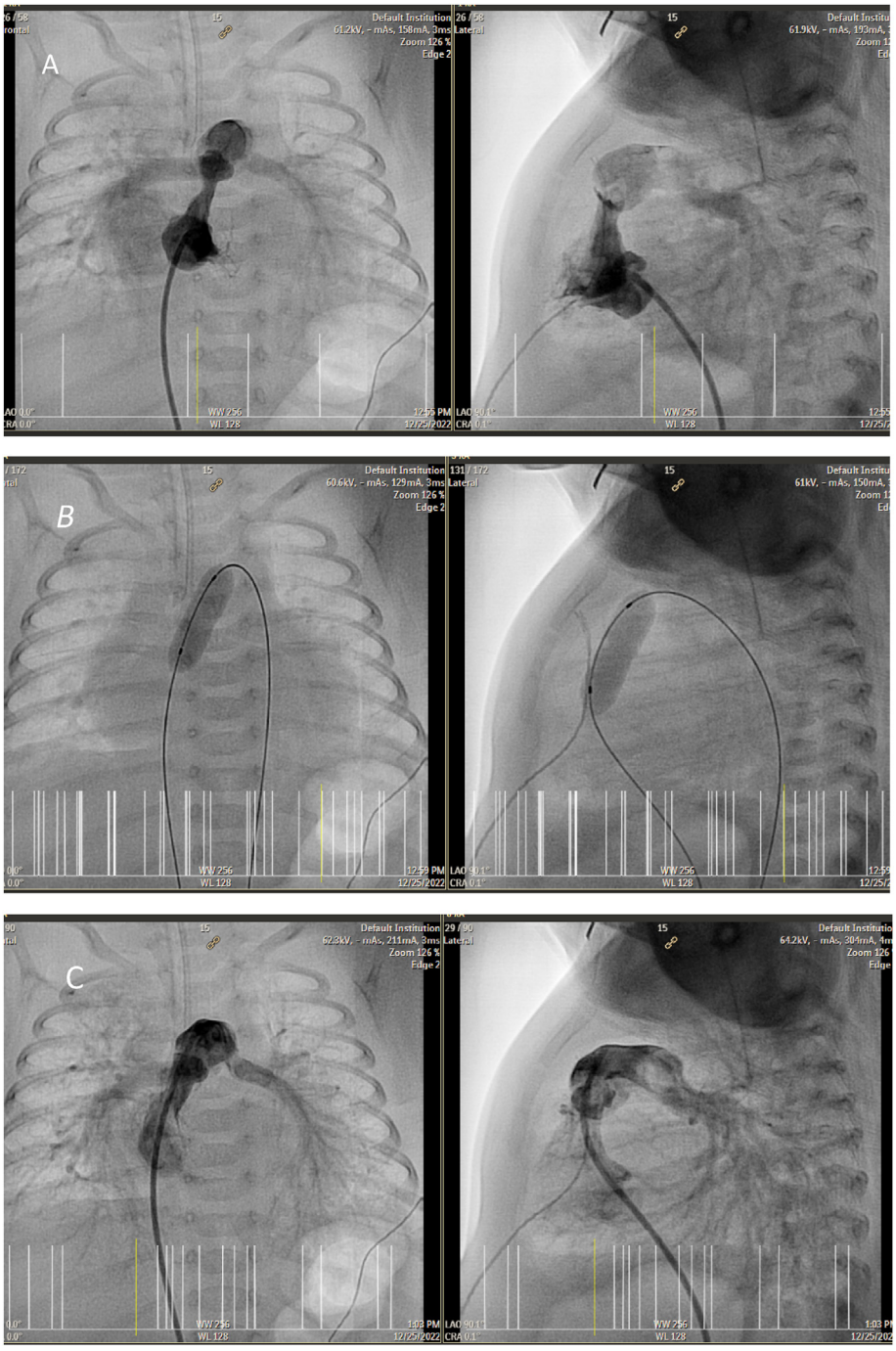

Figure 2: (A) Angiography showing no significant retrograde flow across the right ventricular outflow tract (RVOT). (B) Ballooning of the RVOT. (C) Post-intervention angiography.

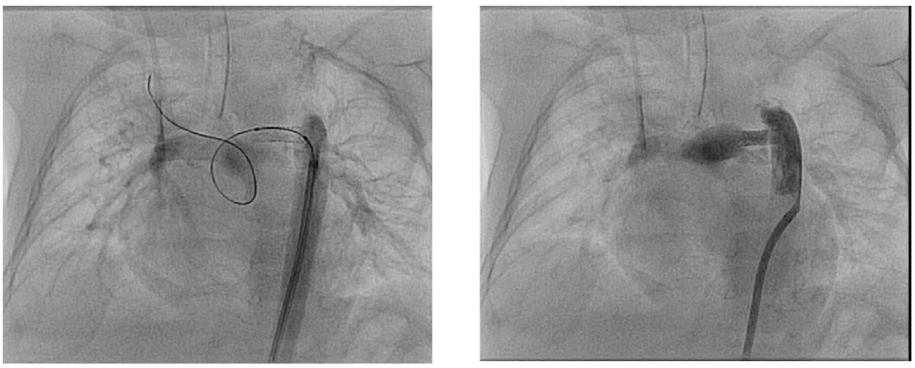

Figure 3: PDA stent insertion.

The patient was initially discharged on captopril, furosemide, and spironolactone to manage episodes of desaturation and heart failure symptoms following stent placement. These medications aimed to improve cardiac function and fluid balance. Remarkably, as her condition improved, she was weaned off these medications. She remains off medication at her latest follow-up, underscoring the success of our tailored approach to her care.

This procedure, while not a panacea, holds promise in ameliorating the overall well-being of select patients within this challenging population. Further research and prospective studies are warranted to delineate the precise criteria for patient selection and ascertain the broader implications of such interventions in trisomy 18 management.

Throughout the process, the family was engaged in a shared decision-making model. We provided compassionate and continuous support, ensuring that they were well informed and active participants in the care of their child. This approach aligns with our ethical commitment to respect patient autonomy and family rights.

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Conflict of interest

The authors have no conflicts of interest to declare regarding the publication of this case report.

Ethical approval

Ethical approval was not required for a case report.

Authors contributions

A.D. was involved in patient care, data collection, and manuscript drafting. A.A. contributed to the conception and design of the study. S.K. assisted in critical revision of the manuscript. T.M. played a key role in the overall coordination and drafting of the manuscript. All authors (A.D., A.A., S.K., T.M.) have critically reviewed and approved the final version of the manuscript. All authors have critically reviewed and approved the final draft and are responsible for the content and similarity index of the manuscript.

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