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Single-stage reconstruction of supravalvular aortic stenosis and severe peripheral pulmonary artery stenosis in an infant with Williams syndrome: a case report

Williams syndrome (WS) is a rare multisystem disorder caused by a 7q11.23 microdeletion, leading to elastin deficiency and generalized arteriopathy. The most clinically significant manifestations include supravalvular aortic stenosis (SVAS) and peripheral pulmonary artery stenosis (PPAS). Severe, multilevel PPAS in early infancy presents a major surgical challenge due to near-systemic right ventricular (RV) pressures and the technical demands of reconstructing small-caliber vessels in low-body-weight patients.

Case presentation. We report a 3-month-old infant (4.9 kg) with WS presenting with severe bilateral PPAS and SVAS. Preoperative cardiac catheterization demonstrated an RV-to-aortic (RV/Ao) pressure ratio of 0.95 (RV systolic pressure 95 mm Hg). Single-stage surgical correction was performed via median sternotomy: Doty aortoplasty for SVAS and extensive bilateral patch augmentation of the main, branch, and lobar pulmonary arteries using autologous pericardium. The postoperative course was complicated by reperfusion syndrome, hemodynamic instability, and transient acute renal failure requiring peritoneal dialysis. Despite these challenges, the patient recovered fully. At 6-month follow-up catheterization, RV systolic pressure had decreased to 30 mm Hg (RV/Ao ratio 0.30), with sustained hemodynamic improvement.

Conclusions. Early single-stage reconstruction of combined SVAS and multilevel PPAS is feasible and can provide excellent, durable relief of RV pressure overload in small infants with Williams syndrome, even in the presence of complex vascular anatomy and significant early postoperative morbidity. This approach warrants consideration in selected high-risk cases.

Key words: Williams syndrome, supravalvular aortic stenosis, peripheral pulmonary artery stenosis, pulmonary arterioplasty, infant cardiac surgery, single-stage repair

Williams syndrome is a rare multisystem genetic disorder caused by a microdeletion on chromosome 7q11.23 involving the elastin (ELN) gene [1, 2, 9], which encodes elastin – a key structural protein of the vascular extracellular matrix. Elastin deficiency leads to a generalized arteriopathy characterized by progressive arterial stenoses [2, 4], most commonly affecting the supravalvular region of the aorta and the pulmonary arterial tree.

Cardiovascular abnormalities are the most clinically significant manifestations of Williams syndrome [3], with supravalvular aortic stenosis (SVAS) and peripheral pulmonary artery stenosis (PPAS) occurring frequently. The pulmonary artery involvement demonstrates a broad anatomical spectrum, ranging from mild branch stenoses that may regress over time to diffuse multilevel narrowing of lobar, segmental, and subsegmental branches

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that can result in significant right ventricular pressure overload.

Management strategies for PPAS remain challenging and depend on the severity and distribution of stenoses. Mild lesions may be managed conservatively or treated with catheter-based interventions; however, severe or diffuse disease often requires surgical reconstruction. Contemporary surgical approaches increasingly emphasize extensive pulmonary arterioplasty targeting multiple stenotic segments to achieve optimal hemodynamic outcomes [10].

Simultaneous repair of SVAS and PPAS in early infancy remains technically demanding, particularly in patients with low body weight and complex pulmonary arterial anatomy.

We present the case of a 3-month-old infant with Williams syndrome who underwent single-stage surgical correction of supravalvular aortic stenosis and peripheral pulmonary artery stenosis. This case highlights the challenges of surgical decision-making in infants with complex pulmonary arterial involvement and illustrates the potential postoperative course following early combined vascular reconstruction.

Case presentation

A 3-month-old infant with a body weight of 4.9 kg and a diagnosis of Williams syndrome was referred to our center for evaluation of complex cardiovascular anomalies. Clinical examination revealed tachypnea with dyspnea both during feeding and at rest, accompanied by mild peripheral cyanosis.

Transthoracic echocardiography demonstrated supravalvular aortic stenosis with a peak gradient of 20 mm Hg. Significant peripheral pulmonary artery stenosis was also noted, with a left pulmonary artery diameter of 2.5 mm and a right pulmonary artery diameter of 2.6 mm. Doppler gradients across the right and left pulmonary arteries were 91 mm Hg and 84 mm Hg, respectively. The right ventricular outflow tract gradient was measured at 47 mm Hg.

Cardiac catheterization confirmed severe peripheral pulmonary artery stenosis *(Figure 1)*. The diameters of the right and left pulmonary arteries were 1.8 mm and 2.3 mm, respectively. The right ventricular systolic pressure was 95 mm Hg, while systemic arterial pressure measured in the ascending aorta was 100 mm Hg, corresponding to a right ventricular-to-aorta (RV/Ao) pressure ratio of 0.95.

The patient underwent surgical repair through a median sternotomy. Prior to initiation of cardiopulmonary bypass, the main and branch pulmonary arteries were extensively mobilized in order to expose all identifiable stenotic segments. Following systemic heparinization, cardiopulmonary bypass was established using aortic and bicaval cannulation. The aorta was cross-clamped and systemic cooling to 32 °C was initiated. Antegrade cardioplegia was administered via the aortic root.

An aortotomy was performed extending from the ascending aorta toward the right and non-coronary sinuses. The thickened sinotubular ridge was excised, and supravalvular aortic stenosis was repaired using the Doty aortoplasty technique with augmentation of the aortic wall using a glutaraldehyde-treated autologous pericardial patch. Reconstruction of the pulmonary arteries was then performed. Longitudinal incisions were made along the inferior and medial surfaces of the main pulmonary artery and extended distally toward the inferior lobar branches beyond the stenotic regions. Two elongated autologous pericardial patches were used to augment the main, branch, and lobar pulmonary arteries using 7–0 polypropylene sutures *(Figure 2)*. Segmental ostial arterioplasty was not performed in this case because the dominant stenotic lesions were located in the lobar branches. Additionally, the complexity of the procedure and the lack of distal vascular control devices limited further distal reconstruction. After completion of the reconstruction, the heart chambers were carefully de-aired and the aortic cross-clamp was removed. Rewarming was initiated. Shortly after initial separation from cardiopulmonary bypass, severe systemic hypotension developed with arterial pressure dropping to 50

Figure 1. **Preoperative cardiac catheterization**

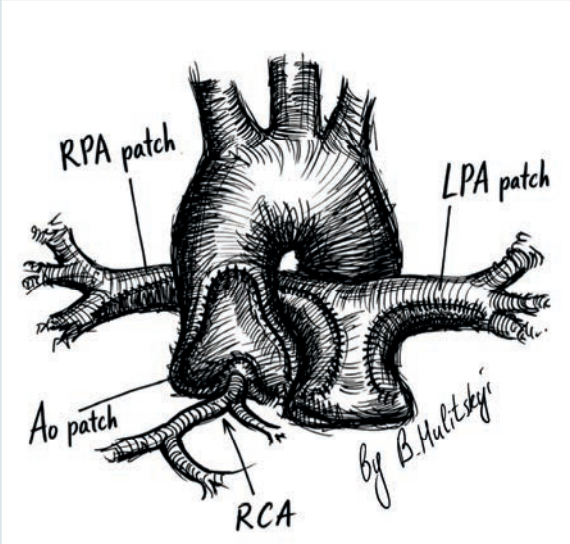

Figure 2. **Surgical reconstruction of the pulmonary arteries and supravalvular aortic stenosis. RCA – right coronary artery; RPA – right pulmonary artery; LPA – left pulmonary artery**

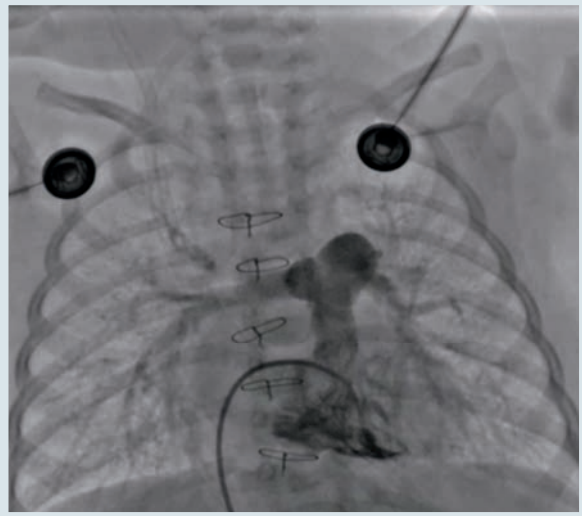

Figure 3. **Postoperative cardiac catheterization**

mm Hg. Cardiopulmonary bypass was therefore resumed for an additional 60 minutes of myocardial reperfusion support. Subsequently, the patient was successfully weaned from bypass. The aortic cross-clamp time was 124 minutes and the total cardiopulmonary bypass duration was 188 minutes, with an additional 60 minutes of reperfusion support.

The postoperative course was complicated by significant reperfusion syndrome and hemodynamic instability requiring inotropic support with epinephrine (0.12 μg/kg/min), dobutamine (5 μg/kg/min), and milrinone (0.3 μg/kg/min). Severe oliguria (0.4 ml/kg/h) developed in the early postoperative period. A peritoneal dialysis catheter was placed on the first postoperative day, and dialysis was initiated with hourly exchanges using 50 ml dialysate. On postoperative day 4, epinephrine infusion was discontinued. By postoperative day 6, urine output increased to 0.8 ml/kg/h and dialysis frequency was reduced to every 4 hours. The first extubation attempt on postoperative day 7 was unsuccessful, and the patient required reintubation after 3 hours. At that time, urine output further improved to 1.09 ml/kg/h. Peritoneal dialysis was discontinued and the dobutamine dose was reduced to 3 μg/kg/min. Successful extubation was achieved on postoperative day 8. Chest drains were removed on postoperative day 7, and the peritoneal dialysis catheter was removed on postoperative day 10. Dobutamine infusion was discontinued shortly thereafter. The patient was transferred from the intensive care unit to the ward on postoperative day 18.

Postoperative echocardiography demonstrated a significant reduction in right ventricular pressure to 21 mm Hg. Residual gradients across the right and left pulmonary arteries were 20 mm Hg and 25 mm Hg, respectively.

The patient was discharged home on postoperative day 28.

At 6-month follow-up cardiac catheterization, the right ventricular systolic pressure was 30 mm Hg, with systemic arterial pressure of 100 mm Hg, corresponding to a RV/Ao pressure ratio of 0.30, indicating significant hemodynamic improvement following surgical reconstruction *(Figure 3)*.

Discussion

Peripheral pulmonary artery stenosis represents a common component of the generalized arteriopathy associated with Williams syndrome [3]. Elastin deficiency leads to abnormal arterial wall architecture and progressive narrowing of medium- and large-sized arteries. This condition occurs in up to 40–60 % of patients with Williams syndrome. Involvement of the pulmonary arterial tree may range from mild focal stenoses to diffuse multilevel disease affecting lobar, segmental, and subsegmental branches.

The clinical spectrum of PPAS in Williams syndrome is highly variable. Mild stenoses may remain hemodynamically insignificant and have been reported to regress spontaneously during childhood [3]. In selected patients, catheter-based interventions such as balloon angioplasty

may provide temporary relief of obstruction. However, in patients with diffuse or severe disease, catheter interventions are often associated with limited efficacy and a significant risk of restenosis or vascular injury [5]. For these patients, surgical reconstruction remains the most definitive treatment option.

Several surgical strategies for reconstruction of the pulmonary arterial tree have been described. Contemporary approaches emphasize extensive mobilization of the pulmonary arteries and augmentation of multiple stenotic segments using patch arterioplasty [6–8]. Some authors advocate reconstruction extending to lobar and segmental branches in order to relieve all hemodynamically significant obstructions. Large surgical series have demonstrated that aggressive multilevel pulmonary artery reconstruction can significantly reduce right ventricular pressure and improve long-term hemodynamic outcomes. In a contemporary 2-decade experience including 145 patients, L.M. Felmly et al. reported significant postoperative improvement in right ventricular pressure following extensive pulmonary artery reconstruction [6]. In that series, the right ventricular-to-systemic pressure ratio decreased from approximately 1.0 preoperatively to 0.30 postoperatively, which is comparable to the hemodynamic improvement observed in our patient. Different patch materials have been used, including autologous pericardium and pulmonary homograft tissue. In addition, short ostial stenoses may be addressed using targeted techniques such as V-plasty. The heterogeneous anatomical distribution of peripheral pulmonary artery stenosis has been highlighted in recent surgical studies. A. Al-Khaldi et al. analyzed late outcomes following surgical reconstruction of peripheral pulmonary arteries and proposed an anatomical classification reflecting the extent of pulmonary artery involvement at different levels of the pulmonary arterial tree. Their results demonstrated that extensive patch augmentation of the pulmonary arteries can provide durable hemodynamic improvement in patients with complex multilevel disease [7].

In the present case, the dominant stenotic lesions were located in the lobar pulmonary artery branches. Surgical repair therefore consisted of patch augmentation of the main and branch pulmonary arteries extending distally toward the affected lobar segments. Although not all distal stenoses were addressed, this strategy resulted in a substantial reduction in right ventricular pressure, with the RV/Ao pressure ratio decreasing from 0.95 preoperatively to 0.30 at follow-up.

The postoperative course in our patient was complicated by significant hemodynamic instability, prolonged mechanical ventilation, and acute renal dysfunction requiring temporary peritoneal dialysis. These complications likely reflect the severity of the preoperative hemodynamic burden and the physiological impact of extensive pulmonary artery reconstruction. Reperfusion of previously underperfused pulmonary vascular territories may lead to transient hemodynamic instability and increased postoperative support requirements [11].

Despite these early postoperative challenges, the patient demonstrated marked hemodynamic improvement with significant reduction in right ventricular pressure and acceptable residual gradients across the pulmonary arteries. This case illustrates both the technical feasibility and the potential postoperative complexity associated with early combined repair of supravalvular aortic stenosis and peripheral pulmonary artery stenosis in infants with Williams syndrome. The hemodynamic improvement observed in our patient (RV/Ao ratio reduction from 0.95 to 0.30) is comparable to results reported in large surgical series [6] of peripheral pulmonary artery reconstruction.

Several limitations should be acknowledged. As a single-case observation, the results may not be representative for all patients with Williams syndrome and diffuse PPAS. Furthermore, reconstruction was limited to the main, branch, and lobar pulmonary arteries without addressing more distal segmental stenoses, primarily because the dominant obstructive lesions were located at the lobar level, combined with the technical challenges of extensive distal repair in a 3-month-old infant and the absence of specialized distal bleeding control tools (such as neuroclips).

Conclusions

Early surgical management of combined supravalvular aortic stenosis and peripheral pulmonary artery stenosis in infants with Williams syndrome is feasible but may be associated with a complex postoperative course. Individualized surgical strategies based on the anatomical distribution of pulmonary artery stenoses are essential to achieve optimal hemodynamic outcomes.

Доступність даних:	Data availability:
Дані взято із джерел, що є у вільному доступі. Всі посилання на використані джерела наведено в статті.	*The data have been obtained from freely available sources. All references to the sources used are listed in the article.*
Джерела фінансування:	Sources of funding:
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Конфлікт інтересів:	Conflict of interest:
Конфлікту інтересів немає.	*There is no conflict of interest.*
Участь авторів:	Authors' participation:
Ідея та дизайн дослідження – С.М., В.К., Д.Г., Б.Г.; лікування хворих – В.В., А.К., Н.Р., Б.Г.; збір і обробка матеріалів, написання тексту – Б.Г.	*Study concept and design – S.M., V.K., D.H., B.H.; patient treatment – V.V., A.K., N.R., B.H.; collection and processing of materials, writing text – B.H.*
Етичне схвалення:	Ethical approval:
Не передбачене темою і концепцією роботи.	*Not provided for by the topic and concept of the work.*
Інформована згода:	Informed consent:
Від батьків пацієнта отримано інформовану згоду на публікацію випадку.	*Informed consent to publish case report was obtained from parents of the patient.*
Подяка:	Gratitude:
Автори статті дякують персоналу клініки та колегам, відповідальним за ведення пацієнта.	*The authors of the article thank the clinic staff and colleagues responsible for patient management.*

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Одноетапна реконструкція надклапанного аортального стенозу та тяжкого стенозу периферійної легеневої артерії в немовляти із синдромом Вільямса: клінічний випадок

Синдром Вільямса – рідкісне мультисистемне генетичне захворювання, спричинене мікроделецією ділянки 7q11.23, що призводить до дефіциту еластину та розвитку генералізованої артеріопатії. Найбільш клінічно значущими проявами є надклапанний стеноз аорти та периферійні стенози легеневих артерій. Тяжкі багаторівневі стенози легеневих артерій у ранньому дитячому віці становлять значну хірургічну проблему через системний тиск у правому шлуночку та технічну складність реконструкції судин малого діаметра в пацієнтів із малою масою тіла.

Клінічний випадок. Представлено випадок 3-місячного немовляти (маса тіла 4,9 кг) із синдромом Вільямса, в якого було діагностовано тяжкі двобічні периферійні стенози легеневих артерій та надклапанний стеноз аорти. Передопераційна катетеризація серця показала співвідношення тиску правого шлуночка до аорти (RV/Ao) 0,95 (систолічний тиск у правому шлуночку – 95 мм рт. ст.). Виконано одномоментну хірургічну корекцію через серединну стернотомію: пластику надклапанного стенозу аорти за методикою Doty та широку двобічну пластику стовбура, гілок і дольових легеневих артерій латками з автоперикарда. Післяопераційний перебіг ускладнився синдромом реперфузії, гемодинамічною нестабільністю та транзиторною гострою ниркової недостатністю, що потребувала проведення перитонеального діалізу. Незважаючи на ці ускладнення, стан пацієнта поступово стабілізувався. Через 6 міс під час контрольної катетеризації серця систолічний тиск у правому шлуночку знизився до 30 мм рт. ст. (RV/Ao = 0,30), що свідчило про значне та стійке гемодинамічне покращання.

Висновки. Рання одномоментна хірургічна реконструкція при поєднанні надклапанного стенозу аорти та багаторівневих периферійних стенозів легеневих артерій у немовлят із синдромом Вільямса є технічно можливою та може забезпечити виражене й тривале зниження перевантаження правого шлуночка тиском навіть за наявності складної судинної анатомії та значних ранніх післяопераційних ускладнень. Такий підхід може бути доцільним у ретельно відібраних пацієнтів з високим ризиком.

Ключові слова: синдром Вільямса, надклапанний аортальний стеноз, периферійний стеноз легеневої артерії, легенева артеріопластика, кардіохірургія немовлят, одномоментне відновлення