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Brief Report

Hypoplastic left heart in Turner's syndrome: a primary indication for transplant?

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Abstract Survival for hypoplastic left heart syndrome patients following the Norwood procedure is 71–90%. Mortality in patients with Turner's syndrome and hypoplastic left heart syndrome after conventional palliation (Norwood operation) has been reported as high as 80%. This questions the approach of traditional staged palliation. Here, we report a patient with hypoplastic left heart syndrome and Turner's syndrome bridged to orthotopic heart transplantation following a hybrid procedure.

Keywords: Turner's syndrome; hybrid palliation; heart transplant; hypoplastic left heart syndrome

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HYPOPLASTIC LEFT HEART SYNDROME CURRENTLY has a survival rate of 71–90% following the Norwood procedure.[1] The occurrence of Turner's syndrome in association with hypoplastic left heart syndrome generally carries a poor prognosis with high surgical mortality.[2–4] Here, we report a patient with hypoplastic left heart syndrome and Turner's syndrome who was successfully bridged to orthotopic heart transplantation following a hybrid Stage I procedure.

Case report

We present a full-term neonate born to a mother with a prenatal diagnosis of hypoplastic left heart syndrome with Turner's syndrome. Prenatal echocardiogram demonstrated evidence of hypoplastic left heart syndrome with mitral and aortic stenosis. The atrial septum was intact but there was a levoatrialcardinal vein that allowed unobstructed pulmonary venous drainage to the superior caval vein. Prostaglandin was initiated immediately after delivery and initial echocardiogram confirmed the diagnosis. CT angiography demonstrated a large tortuous levoatrialcardinal vein decompressing the left atrium to the superior caval vein and the right upper- and middle-lobe pulmonary veins drained independently to the levoatrialcardinal vein.

Initial course after delivery was complicated with metabolic acidosis, pulmonary overcirculation, and poor systemic perfusion. Subsequently, the patient developed acute kidney injury secondary to poor systemic output and adrenal insufficiency. Because of poor prognosis of hypoplastic left heart syndrome with Turner's syndrome, the patient underwent a hybrid Stage I procedure on day of life 13 (Bilateral pulmonary artery bands with 3.5 mm Gore-Tex grafts (W. L. Gore & Associates, Flagstaff, Arizona, United States of America) and patent ductus arteriosus stent placement with 8 mm × 17 mm VisiPro stent (Medtronic, Minneapolis, Minnesota, United States of America)) with subsequent listing for heart transplantation.

Postoperative course was complicated by emergent mediastinal exploration on postoperative day 4 due for tamponade. Chest closure was performed after 2 weeks. The baby developed intermittent episodes of haemoptysis which required mechanical ventilation. Cardiac catheterisation showed mild retrograde arch obstruction with 16 mmHg gradient which was relieved by balloon angioplasty, and large effective left to right and right to left shunts (Table 1).

The baby underwent successful orthotopic heart transplantation with aortic arch reconstruction,

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repair of partial anomalous pulmonary venous return, and removal of bilateral pulmonary artery bands 6.5 months after initial palliation. The baby was discharged after a prolonged hospitalisation (327 days total stay, 120 days post-transplant) in stable condition and is currently on stable immune suppression 24 months post-transplant.

Discussion

CHD in Turner's syndrome includes varying degrees of left heart obstruction. The poor prognostic association of Turner's syndrome and hypoplastic left heart syndrome has been well described (Table 2). In a recent query of the Texas Birth Defect registry, 11 patients were identified. Of these, six patients died before Stage 1 palliation, three died during the interstage, and two patients survived past infancy, one of whom died at 1.7 years.[3] The series by Reis et al[4] showed an 80% mortality and mean age at death of 39 ± 47.8 days following the Norwood procedure. The two survivors in their cohort of 10 patients were Turner's mosaics who survived to the Fontan procedure. All patients in this series underwent the Norwood procedure with a systemic-to-pulmonary artery shunt.

The series by Lara et al combined with the series by Reis et al includes 21 patients.[3,4] Of these, six died before the Norwood procedure, 11 died during the interstage period, two after the Glenn procedure, and two (Turner's Mosaics) were alive after the Fontan procedure. The series by Cramer et al from Wisconsin demonstrated better outcomes with a 75% survival to Glenn and 50% survival to discharge post-Glenn; however, 75% of their patients died before 1 year and the one remaining patient survived with multiple comorbidities.[5] The postoperative complications included right ventricular dysfunction with severe acidosis and renal failure likely from low cardiac output. The high likelihood of poor outcome in this patient was the rationale behind pursing a hybrid Stage I procedure.

The increased morbidity and mortality in the postoperative period in these patients questions the approach of traditional staged palliation for hypoplastic left heart syndrome. The challenges encountered in the management include fluid overload, capillary leak, slow wound healing, and risk for infections. These are likely related to poor lymphatic drainage into the venous system. Loscalzo et al[6] demonstrated a strong association between lymphatic anomalies and CHDs in patients with Turner's syndrome. They also propose lymphatic anomalies might be the primary event leading to heart defects by impinging on developing vascular and cardiac structures. The increased severity of this comorbidity creates a challenge for postoperative care impeding fluid mobilisation from tissues, diuresis and wound healing which creates additional comorbidity.

Table 1. Haemodynamic during second catheterisation 6 months after hybrid Stage I palliation.

Parameters	Value
Atrial pressures (mmHg)	11–12 (no gradient between right and left atria)
Right ventricular end-diastolic pressure (mmHg)	10
Oximetry (saturation %)	Mixed venous: 64% Aortic/pulmonary arteries: 78% Left atrium 92%
Flows (L/minute/m^2)	Qp: 6.3 Qs: 6.3 Qep: 3.2 Qp:Qs ratio 1:1
Mean PA pressure (mmHg)	RPA: 27 LPA: 20
PVR (Woods units/m^2)	2.4 (using mean PA pressure of 27)

LPA = left pulmonary artery; PA, = pulmonary artery; PVR = pulmonary vascular resistance; RPA = right pulmonary artery

Table 2. Review of current literature describing outcomes in patients with hypoplastic left heart syndrome (HLHS) with Turner 's syndrome (TS).

	University of Michigan 1990–1997[4]	Children's Hospital of Wisconsin 1999–2011[6]	State of Texas 1999–2007[3]	Overall
Total HLHS + TS	10	4	11	25
Death before Stage 1 palliation	0/10 (0%)	0/4 (0%)	6/11 (54%)	6/25
Survival to Stage 1 palliation	10/10 (100%)	4/4 (100%)	5/11 (45%)	19/25
Death during interstage	8/10 (80%)	1/4 (25%)	3/11 (27%)	12/25
Survival to Stage 2 palliation	2/10 (20%)	3/4 (75%)	2/11 (18%)	7/25
Discharged alive from Stage 2 palliation	2/10 (20%)	2/4 (50%)	2/11 (18%)	6/25
Survival to 1 year of age	2/10 (20%)	1/4 (25%)	1/11 (9%)	4/25
Overall survival	2/10 (20%)*	1/4 (25%)	1/11 (9%)	4/25 (16%)

Madriago et al show a mortality of 90.4% for HLHS (19/21) but does not provide with a description of what stage these patients were on at time of death

*45, X mosaic

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In comparison, the hybrid Stage I procedure avoids cardiopulmonary bypass and makes recovery quicker.

We hypothesise that, in patients with Turner's syndrome born with hypoplastic left heart syndrome, primary orthotropic heart transplant should be considered in alignment with published literature, which indicates poor prognosis with conventional single ventricle palliation. A hybrid Stage I procedure may be the preferred bridge to transplant considering the high mortality following the Norwood procedure. We understand that this is only one patient but considering poor outcomes following initial palliation in multiple series, we propose the need for a different strategy which can be Stage 1 palliation using a hybrid as a bridge to transplant.

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Conflicts of Interest

None.

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