Should a Down Syndrome Child With a Failing Heart Be Offered Heart Transplantation?

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Introduction

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The term "futility" has been frequently discussed in the medical and ethics literature over the last 30 years and has been generally dismissed as impossible to define, that is, the term has been applied to so wide a variety of clinical situations that it is virtually incoherent [1]. Nevertheless, it is still used in some situations to justify withholding curative or life-saving treatments. A technically complex and costly procedure that might or might not be used in a challenging clinical situation could be called "futile," "clinically nonbeneficial," or "medically inappropriate," but regardless of the label, differing viewpoints often lead to clashes about whether such a procedure should be done.

Problematic situations are made even more difficult when a child's life is at stake. Such a clinical situation is described in the following fictional vignette, which was the focus of the ethics debate at the 2017 annual meeting of The Society of Thoracic Surgeons.

Case

Eight-year old Angela Downing was born with trisomy 21 and complete atrioventricular (AV) canal; she currently weighs 25 kg with body surface area of 1.0 m^2. At age 5 months she underwent repair, including closure of the mitral valve "cleft." She was left with residual moderate left AV valve (mitral) insufficiency and moderately depressed left ventricular function. Her parents noticed easy fatigability and nighttime snoring when she was 4 years old. After months of scheduling issues due to Angela's parents' work schedules and the necessity of finding a caregiver for her younger sister, a sleep study found obstructive sleep apnea, and continuous positive airway pressure (CPAP) was prescribed. Angela resists her CPAP and often removes the mask during the night. Referral to a behavioral psychologist does little to facilitate her tolerance, and Angela's follow-up with her pulmonologist is often precluded by her parents' report that her breathing while asleep "isn't any worse than it has been." Echocardiography at that time showed pulmonary hypertension, for which she was prescribed sildenafil daily.

Over time, Angela's atrioventricular valve insufficiency has progressed and is now severe, and her left ventricular function has remained moderately depressed. Her pulmonary hypertension appears to have worsened qualitatively on echocardiogram, and her pulmonary vascular resistance is 4 Wood units by catheterization. She has been referred to Dr S. M. Bradford for mitral valve repair.

After rescheduling the procedure twice at Angela's parents' request, Dr Bradford operates and repairs the mitral valve. After weaning from cardiopulmonary bypass proves difficult, transesophageal echocardiography shows depressed ventricular function and residual moderate mitral valve insufficiency. Bypass is resumed, and the patient undergoes mitral valve replacement with a 21-mm St. Jude mechanical valve. After valve replacement, she fails to wean from bypass owing to poor left ventricular function and is placed on venoarterial extracorporeal membrane oxygenation (ECMO) by way of chest cannulation. The chest is left open.

Over the next 7 days Angela fails to wean from ECMO support owing to poor left ventricular function, and her course is complicated by reexploration for bleeding and difficulty achieving adequate heparinization. She has had thromboemboli to several fingers and toes. Her chest remains open. Angela's parents have been unable to be at her bedside for daily rounds, but have tried to visit her as much as possible during evening nursing handovers. As decision time nears, the parents want "everything" done. The surgical team believes the likelihood of successful weaning from ECMO approaches zero. The only reasonable options seem to be withdrawal from ECMO or placement on a ventricular assist device (VAD) as a bridge to heart transplantation (HT). Dr Bradford wonders whether he should follow the parents' wishes and embark on the VAD-HT pathway.

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Pro

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Dr Bradford should implant the VAD and list Angela for heart transplantation.

Background

Down syndrome (DS) is the most common genetic syndrome with an incidence of 1 to 2:1,000 live births. Children present with a wide spectrum of cognitive delay ranging from mild to severe. Most children with DS are in the mild to moderate range with an IQ in the 50 to 70 range. Accordingly, their functional status also varies widely—many high-functioning DS children have an IQ in the 70 to 90 range. The long-term prognosis for persons born with DS is generally good, with survival to age 60 to 70 years [2]. Fifty percent of children with DS have cardiac malformations, most commonly AV septal defects.

Multicenter studies on children undergoing pediatric cardiac surgical repairs have shown a lower mortality among children with DS. A large national database study comparing more than 4,000 DS children with non-DS children showed that the DS group had a significantly lower mortality in all risk categories except single ventricles [3]. The likely explanation for the better outcomes in DS patients undergoing AV septal defect repairs is that non-DS children have more dysplastic left AV valves and therefore typically have more postoperative residual regurgitation and stenosis requiring reinterventions [4]. In addition, DS children are also more resistant to ischemia-reperfusion injury after cardioplegic arrest and bypass, probably because of higher levels of antioxidants [5].

Conversely, DS children who underwent aortopulmonary and bidirectional Glenn shunts had a higher mortality [3]. That is likely because DS children generally have a higher incidence of airway abnormalities, lung abnormalities, and pulmonary hypertension that translates into worse outcomes for single ventricle physiology [6, 7].

Historically, organ transplantation in DS patients was considered a contraindication until the landmark case of Sandra Jensen in 1995 [8]. This 34-year-old DS woman, who was living independently with heart failure, was denied heart transplantation despite being approved for the procedure by California Medicaid. After a successful lawsuit, she underwent a successful heart-lung transplant. Since then, solid organ transplants in patients with intellectual disabilities, including DS, have been performed; and studies reveal that the short-term and long-term outcomes are similar to those of non-DS recipients. Of 5 DS heart transplantation recipients, 4 survived to 16 years. The single bad outcome was related to poor compliance and compares well with the noncompliance rate in non-DS recipients [9].

Mechanical support as a bridge to transplantation in children is now commonplace and has become the standard of care in managing children with end-stage heart failure [10].

Ethical Analysis

When faced with the decision of whether to offer mechanical support or heart transplantation to DS children, ethical analysis can be based on the principles on which the physician-patient relationship is built, and which are the pillars of medical and surgical ethics.

Respect for Autonomy

This principle asserts the parent's right to hold views, make choices, and take actions on behalf of their children based on their personal values and beliefs [11]. It acknowledges the decision-making rights of parents that enable them to act autonomously. Conversely, disrespect for autonomy involves attitudes and actions that ignore, insult, demean, and are inattentive to the parent's right of autonomous action. Importantly, this principle underlies informed consent, in which complete disclosure and understanding, without coercion or manipulation, is required before any elective surgical intervention. Based on this important ethical principle and the relevant literature, a surgeon who is experienced and skilled at caring for children with heart failure and trisomy 21 should discuss all available options with the patient's parents. Ideally the process of informed consent should begin early with prenatal and preoperative counseling and indepth discussion of the risks, benefits, and alternatives, which include the options of surgical repair and comfort care.

Beneficence and Nonmaleficence

Beneficence requires physicians to take positive steps to help others. Nonmaleficence imposes a negative obligation, to avoid causing more harm than good. These principles are closely related and interact with each other during surgical decision making. A surgeon often has to produce pain and other harms for the benefits of potential palliation or cure. This dichotomy is illustrated by the "rule of double effect," which posits that a single act can have two foreseen effects, one beneficial and one harmful. Four elements may make such an act morally justifiable: the nature of the act, which must be good or morally neutral; the physician's intent, which desires only the good effect and does not desire the bad effect, even though it can be foreseen; the bad effect must not be a means to the good effect; and the proportionality between these two effects is such that it weighs the good effect more heavily than the bad [12].

Implanting a VAD as a bridge to transplantation for Angela fulfills all these conditions, so the VAD-HT option may be justified.