Heart Transplantation in a Pediatric Patient with Bardet–Biedl Syndrome

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Bardet–Biedl syndrome (BBS) is a rare autosomal recessive, non-motile ciliopathy caused by mutations in genes encoding proteins localized to the basal body of the primary cilium. It is a multisystem disorder with an estimated prevalence of approximately 1 in 125,000 individuals. Core clinical features include truncal obesity (70%), retinal dystrophy, postaxial polydactyly (approximately two-thirds of patients), hypogonadism (40%), renal abnormalities (40%), and intellectual disability (50%) (Tsang et al., 2018). The reported prevalence of congenital and acquired cardiac anomalies in BBS is approximately 29.8%, with unspecified cardiomyopathy highlighted in about 7% of patients (Melluso et al., 2023). However, there are no guidelines addressing pediatric heart transplant eligibility in this population, as life-threatening heart failure secondary to BBS-associated cardiomyopathy has not been previously reported.

This case focuses on a 13-year-old female with Bardet-Biedl Syndrome who developed end-stage systolic and diastolic heart failure and underwent orthotopic heart transplantation at the age of 11. She had no history of graft rejection. She developed significant comorbidities post-transplant, including diabetes mellitus, hypertension, hyperlipidemia, and metabolic dysfunction–associated steatotic liver disease (MASLD). Two years after transplant, the patient exsanguinated and died in her sleep from a severe gastrointestinal hemorrhage secondary to advanced liver cirrhosis. Autopsy showed no evidence of cardiac graft rejection.

It is known that patients with BBS are predisposed to obesity, liver disease, and related metabolic syndrome. The adverse effects of immunosuppression are well-studied, including hyperglycemia, hyperlipidemia and hypertension (Bhat et al., 2021). In this case, the progression of liver disease was attributed to BBS-associated susceptibility, which was likely accelerated by worsening metabolic disease related to long-term immunosuppressive therapy. Recognition of these potential post-transplant complications is critical for informed risk–benefit assessment when evaluating heart transplantation candidacy in patients with BBS.

References:

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