

## It Takes a Village: Treatment Success with Subcutaneous Treprostinil in Adult Patient with Trisomy 21 and Severe Pulmonary Arterial Hypertension

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**Background:** Subcutaneous (SQ) treprostinil is a widely used treatment for patients with severe PAH. Although parenteral prostacyclin, such as treprostinil, is considered “gold-standard” therapy, it is offered to patients with adequate cognitive capability and psychosocial support due to its narrow therapeutic window. This case describes a 33 year-old female with trisomy 21 and developmental delay who presented in June 2017 with severe PAH associated with repaired congenital heart defect (AV canal defect repaired in childhood). There have been no previously published case reports of the use of SQ treprostinil in an adult patient with trisomy 21. The aim of this case report is to describe how a multifaceted and collaborative approach can lead to the successful treatment of an adult patient with trisomy 21 and severe PAH using SQ treprostinil.

**Methods:** The patient presented to clinic after a witnessed syncopal episode while exercising with her trainer. Previously the patient had lived semi-independently under supervision of disability services. Due to her severe symptoms, the patient had to move back home with her parents for close monitoring. After diagnostic right heart catheterization in July 2017, it was determined the patient needed parenteral therapy. A team was assembled to provide adequate medical and psychosocial support to the patient. The specialty pharmacy (SP) nurse worked very closely with the patient and her parents to build confidence and trust to initiate and continue with therapy despite side effects. The SP nurse provided home visits twice weekly for three months until independence and safety was achieved. The patient remained under parental care for the first six months of treatment. After the patient’s functional status improved and she demonstrated understanding of basic pump and site issues to report to caregivers, she was able to return to her semi-independent living status with biweekly home health nursing support. SP trained home health nurses on pump and site management and trained in-home caregivers on daily weight and blood pressure monitoring. The family remained available 24/7 to assist with urgent treatment related issues and even moved within minutes of the patient’s apartment.

Treatment initiation involved multiple SP pre-teach visits before admission to improve pump familiarity and decrease anxiety. A dry site was placed to improve the patient’s acceptance of the catheter. During the inpatient stay, SQ treprostinil was rapidly titrated over five days to 7ng/kg/min with minimal systemic and site related side effects. The patient continued to titrate over 10 weeks to 28ng/kg/min. Titration was limited intermittently by diarrhea, leg pain, fatigue, and dizziness. Monthly PH clinic appointments were scheduled for 60 minutes to accommodate ongoing coordination of care and included PH team members (MD, NP, RN, MA), SP nurse, home health nurse, disability coordinators, and the patient’s parents.

**Results:** Repeat RHC completed in August 2017 on SQ treprostinil monotherapy demonstrated improvement in hemodynamics, specifically cardiac output (CO) and cardiac index (CI), however continued moderately severe PAH. The patient was subsequently initiated on tadalafil oral therapy and SQ treprostinil was titrated to 50ng/kg/min over the next 4 months. The patient underwent repeat RHC in January 2019 demonstrating significant improvement in hemodynamics with near normalization of pulmonary artery pressure (PAP) and pulmonary vascular resistance (PVR). She continues on current therapy and receives biweekly home health visits and PH clinic visits every 3 months. Due to her significant improvement in functional status, she was able to return to her “normal” life, which included daily exercise with her trainer, social independence (going to movies), and travel by airplane with family on a two-week



vacation to visit Disney World. The patient did not suffer any significant adverse events related to the treatment including no admissions or infusion site infections.

**Conclusions:** The initiation of SQ treprostinil therapy was the first of its kind in an adult PAH patient with trisomy 21 at our institution. We have shown that successful long-term treatment with SQ treprostinil therapy in this patient population relies upon multidisciplinary team planning and extensive family, medical, and social support. This case emphasizes that close collaboration and partnership between the PH program, family, and extended health care team results in improvement in patient outcomes and quality of life.

Figure 1. Hemodynamic Parameters

Table 1. Right Heart Catheterization hemodynamic data

Date of RHC	6/4/2018	9/10/2018
Hemodynamic setting	Milrinone 0.2 mcg/kg/min	Heart Mate II (8400 rpm, 5.1 L, PI 6.6) Sildenafil 80mg TID
RA pressure (mmHg)	17	12
RV systolic/diastolic/mean (mmHg)	68/13/21	58/6/15
RVEDP (mmHg)	21	15
PA (mmHg)	71/27/42	55/20/34
PCWP (mmHg)	25	13
TPG (mmHg)	17	21
DPG (mmHg)	2	7
RA saturation (%)	63	62.5
Hemoglobin (g/dL)	16	8.9
PA saturation (%)	63.2	59.7
Peripheral saturation	93	100
SBP/DBP/MAP (mmHg)	74/60/64	125/82/94
Cardiac output – Fick (L/min)	2.63	4
Cardiac index L/min/m <sup>2</sup>	1.45	2.6
CVP/PCWP	0.68	0.92
PAPi	2.59	2.92
SVR (dynes/cm <sup>2</sup> )	1429.66	1686.67
PVR (Wood Unit)	6.46	5.25

RA, right atrium; RV, right ventricle; RVEDP, right ventricular diastolic pressure; PA, pulmonary artery; PCWP, pulmonary capillary wedge pressure; TPG, transpulmonary gradient; DPG, diastolic pulmonary gradient; SBP, systolic blood pressure; DBP, diastolic blood pressure; MAP, mean arterial pressure; CVP, central venous pressure; PAPi, pulmonary artery pulsatility index; SVR, systemic vascular resistance; PVR, pulmonary vascular resistance

