BioCore Exploring Scientific Community

International Journal of Pediatrics and Neonatal Health

Case Report ISSN 2572-4355

Sildenafil Citrate: An Effective Agent in the Management of Idiopathic Pulmonary Hypertension in a Child with Rubinstein-Taybi Syndrome

Bryan Sibley*, Jonathan Joseph

Doctor of Medicine, Louisiana State University School of Medicine, Shreveport, USA

Corresponding Author: **Bryan Sibley**
Doctor of Medicine, Louisiana State University School of Medicine, Shreveport, USA. Tel: 337-289-0042,
Email: bsibley@drsibley.com

Citation: Bryan Sibley et al. (2020), Sildenafil Citrate: An Effective Agent in the Management of Idiopathic Pulmonary Hypertension in a Child with Rubinstein-Taybi Syndrome. Int J Ped & Neo Heal. 4:1, 01-02

Copyright: ©2020 Bryan Sibley et al. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited

Received: August 28, 2019
Accepted: September 12, 2019
Published: February 24, 2020

Case Report

We report a case of a patient who had Rubinstein-Taybi Syndrome (RTS), with multiple associated complications, including pulmonary hypertension. The symptoms and sequelae of pulmonary hypertension were abated through the use of Sildenafil Citrate.

This patient was born at 39 weeks gestation to a 28 year old Gravida 3, Para 2 female whose pregnancy was complicated by preterm labor and polyhydramnios. The infant was initially admitted to the Neonatal Intensive Care Unit (NICU) where he was treated until 30 days of life. While in the NICU, he was diagnosed with Rubinstein-Taybi Syndrome, respiratory distress syndrome, patent ductus arteriosus, anemia, grade II intraventricular hemorrhage, hypotonia, gastroesophageal reflux, undescended testis, possible hypothyroidism, right coloboma, and dysphagia.

His physical examination was remarkable for low anterior hairline, depressed bridge of nose, marked hirsutism, bilateral wide thumbs, bilateral dysplastic nails of his hands and feet, and significant generalized hypotonia. He later exhibited speech, gross motor, fine motor, and cognitive delays.

Throughout his life, the patient was evaluated and treated on multiple occasions for both routine childhood illnesses and complications of RTS. The primary complication of RTS at the end of his life was cardiac failure secondary idiopathic pulmonary hypertension. Six months prior to his demise, while admitted to the Pediatric Intensive Care Unit and treated for heart failure, his clinical condition improved significantly secondary to the use of Sildenafil Citrate.

For the next 6 months his symptoms remained well controlled and this was directly attributed to the use of Sildenafil Citrate.

Eventually he developed respiratory compromise secondary to complications of viral bronchiolitis; this led to worsening cardiac failure and death at the age of 3 years.

Discussion

Rubinstein-Taybi Syndrome, also known as Broad Thumb-Hallux syndrome, is an autosomal dominant disorder with a prevalence of 1 in every 100,000-125,000 live births.1 Michail et al. first described the disease in 1957.[2] Although there are no exact criteria for diagnosing RTS, the disease is often characterized by mental retardation, poor post-natal growth, microcephaly, broad thumbs and toes, and facial abnormalities.

Some cases of RTS are due to microdeletions in chromosome 16 p13.3 or mutations in the cyclic adenosine monophosphate-response element-binding protein (CREBBP), which is located at 16 p13.3. 55% of RTS patients have a mutation at the CREBBP or E1A binding protein p300 (Ep300) gene.[3] 45% of RTS patients are diagnosed on clinical features alone.

CREBBP is a protein that is important in the regulation of cell division and growth and is crucial in normal fetal development. A mutation in this protein results in a disruption of normal development before and after birth.

About one third of patients with RTS are affected with congenital cardiac abnormalities including atrial septal defect, ventricular septal defect, patent ductus arteriosus, coarctation of the aorta, pulmonary artery stenosis, bicuspid aortic valve, aortic stenosis, dextrocardia, vascular rings, and dysrhythmia.[4] Pulmonary arterial hypertension is characterized by vascular proliferation and remodeling of the small pulmonary arteries. Continual increases in the vascular resistance leads to right ventricular heart failure and eventual death.[5] Pulmonary hypertension has been associated with obstructive sleep apnea and patent ductus arteriosus in patients with RTS.

Our patient was also diagnosed with idiopathic pulmonary hypertension, congenital patent ductus arteriousus (PDA), reactive airway disease, and recurrent respiratory distress syndrome. Complications of pulmonary hypertension ultimately led to heart failure and the patient's subsequent demise. Shortly after the diagnosis of idiopathic pulmonary hypertension was made, this child was treated with a variety of commonly used medications, none of which were effective in controlling heart failure. Sildenafil Citrate (Sildenafil Citrate) therapy was initiated and his symptoms temporarily abated. Six months later, he developed worsening heart failure which led to his demise.

The life expectancy of a patient with Rubinstein-Taybi Syndrome is usually into early childhood due to the many defects that arise as a result of the disease. Our patient developed severe idiopathic pulmonary hypertension, the symptoms of which were controlled with the use of Sildenafil Citrate. Even though this patient's pulmonary hypertension was the ultimate cause of death, Sildenafil Citrate provided

symptomatic relief resulting in an improved quality of life for an additional 6 months

References

1. Milani, Donatella, et al. "Rubinstein-Taybi Syndrome: Clinical Features, Genetic Basis, Diagnosis, and Management." Italian Journal of Pediatrics 41.4 (2015): n. pag. Print.

2. Michail, J., J. Matsoukas, and S. Theodorou. "Arched, Clubbed Thumb in Strong Abduction-Extension & Other Concomitant Symptoms." Revue de Chirurgie Orthopédique et Réparatrice de l'Appareil Moteur 43.2 (1957): 142-46. Print.

3. Fadly, A. Mohd, et al. "Chromosomal 16p Microdeletion in Rubinstein-Taybi Syndrome Detected by Oligonucleotide Based Array Comparative Genomic Hybridization: A Case Report." Journal of Medical Case Reports 6.30 (2012): n. pag. Print.

4. Wiley, Susan, et al. "Rubinstein-Taybi Syndrome Medical Guidelines." American Journal of Medical Genetics 119.A (2003): 101-10. Print.

5. Demerouti, MD PhD, Eftychia A., et al. "Complications Leading to Sudden Cardiac Death in Pulmonary Arterial Hypertension." Respiratory Care 58.7 (2013): 1246-54. Print