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## “Rare or Not So Rare, That Is the Question”: Orphan Diseases in Children

Guest Editor:

**Dr. Wilfredo De Jesús Rojas**

Department of Pediatrics,  
University of Puerto Rico, San  
Juan, PR 00921, USA

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### Message from the Guest Editor

More than 6000 rare diseases affect more than 25 million of individuals in the United States of America. A disease that affects less than 200,000 individuals is considered to be a rare disease. The exact prevalence of pediatric rare diseases is still unknown. Rare and orphan diseases include several syndromes, most of them with a genetic etiology. The diagnostic challenges to identify and confirm a rare disease are significant. The complexity of treating and managing a patient with a rare disease may be difficult for the general medical provider without adequate diagnostic tools and specialist referrals. An interdisciplinary approach for rare-disease management is critical to address symptoms and for patients to have access to effective medical treatments. A better understanding of the basic science, medical pathophysiology, and clinical diagnosis for current and future treatments is needed. Additionally, new strategies to involve caretakers in the community to participate in pediatric rare-disease research is important to understand rare etiologies of the disease, decrease medical barriers, improve the access to healthcare and avoid health disparities in this population.



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### **Prof. Dr. Paul R. Carney**

Departments of Child Health and Neurology, University of Missouri, 400 Keene Street, Columbia, MO 65211, USA

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*Children* Editorial Office  
MDPI, Grosspeteranlage 5  
4052 Basel, Switzerland

Tel: +41 61 683 77 34  
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