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Abstract

Bartter Syndrome: Causes, Diagnosis, and Treatment

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Abstract:

Introduction

Bartter syndrome is an inherited renal tubular disorder caused by a defective salt reabsorption in the thick ascending limb of loop of Henle, resulting in salt wasting, hypokalemia, and metabolic alkalosis. Mutations of several genes encoding the transporters and channels involved in salt reabsorption in the thick ascending limb cause different types of Bartter syndrome. A poor phenotype–genotype relationship due to the interaction with other cotransporters and different degrees of compensation through alternative pathways is currently reported. However, phenotypic identification still remains the first step to guide the suspicion of Bartter syndrome. Given the rarity of the syndrome, and the lack of genetic characterization in most cases, limited clinical evidence for treatment is available and the therapy is based mainly on the comprehension of renal physiology and relies on the physician's personal experiences. A better understanding of the mutated channels and transporters could possibly generate targets for specific treatment in the future, also encompassing drugs aiming to correct deficiencies in folding or plasma membrane expression of the mutated proteins.

Biography:

Tamara da Silva has completed her MD, PhD. Possui Doutorado in the Nephrolithic area of the Federal University of São Paulo (UNIFESP), Brazil

Recent Publications:

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