Diagnostic work-up and review of a CYP11B2 defect in a patient with Failure to Thrive

Manuel Schaller

Master Thesis in Master of Medicine

A defect of CYP11B2 is rare. Nonetheless, a deficiency of aldosterone synthase remains a possibility in infants and toddlers with Failure to Thrive. This Master Thesis aims to present a patient‘s diagnostic work-up that lead to the diagnosis of a defect of CYP11B2 in reflection of what the literature recommends for each stage of the diagnostic procedure. Furthermore, there is a short review of the literature concerning the symptomatology, pathophysiology, therapy and differential diagnosis of patients with a suspected or confirmed defect of Cyp11B2.

Supervisor: Prof. Dr. med Anna Lauber-Biason, University of Fribourg
Expert: Dr. med. Mariarosaria Lang-Muritano