Special Issue

Citrin Deficiency: Molecular Pathogenesis and Novel Targets for Therapy

Message from the Guest Editor

Citrin deficiency is caused by mutations of SLC25A13, which encodes mitochondrial aspartate glutamate carrier, or citrin. Citrin deficiency causes many diseases, including CTLN2 or adult-onset type 2 citrullinemia at adult age, neonatal intrahepatic cholestasis caused by citrin deficiency (NICCD) in the neonatal period, failure to thrive and dyslipidemia caused by citrin deficiency (FTTDCD) in the adolescent period and many other disease states and symptoms, including liver failure, hepatocellular cancinoma, pancreatitis, preference for protein- and fat-rich foods and aversion to sugars and carbohydrate-rich foods, anorexia and so on. This indicates the importance of functions of asratate glutamate carrier in many metabolic processes. Therapeutic procedures for citrin deficiency are very important not only for the treatment of citrin deficiency, but also the understanding of coordinate metabolic processes. Finally, the mouse model of the disease, citrin/mitochondrial glycerol 3-phosphate dehydrogenase double-knockout mouse, will be very important for creating useful treatment methods.

Guest Editor

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