

Current understanding on the genetic basis of key metabolic disorders: A review

ABSTRACT

Advances in data acquisition via high resolution genomic, transcriptomic, proteomic and metabolomic platforms have driven the discovery of the underlying factors associated with metabolic disorders (MD) and led to interventions that target the underlying genetic causes as well as lifestyle changes and dietary regulation. The review focuses on fourteen of the most widely studied inherited MD, which are familial hypercholesterolemia, Gaucher disease, Hunter syndrome, Krabbe disease, Maple syrup urine disease, Metachromatic leukodystrophy, Mitochondrial encephalopathy lactic acidosis stroke-like episodes (MELAS), Niemann-Pick disease, Phenylketonuria (PKU), Porphyria, Tay-Sachs disease, Wilson's disease, Familial hypertriglyceridemia (F-HTG) and Galactosemia based on genome wide association studies, epigenetic factors, transcript regulation, post-translational genetic modifications and biomarker discovery through metabolomic studies. We will delve into the current approaches being undertaken to analyze metadata using bioinformatic approaches and the emerging interventions using genome editing platforms as applied to animal models.