Molecular Charazterization and Screening of Metabolic Diseases Ornithine Transcarbomylase Deficiency (OTC) and Citrullinemia (CIT) among Puerto Ricans

Rivera - Corona, Andrea

Newborn screening is a vital process that defines the course that a baby's life could take. Identifying metabolic diseases at early stages gives the opportunity of early treatment, making the chances of survival higher. Amongst the metabolic diseases tested with newborn screening the urea cycle diseases are one of the most essential. The Urea Cycle is responsible for metabolizing nitrogen in the form of ammonia which can be toxic in high levels and so affected newborns can develop coma after 4 days of life. Although the usage of tandem mass spectrometry is the most common way of performing newborn screening for a vast number of metabolic disorders by identifying elevations on chemical compounds in the dried blood spot (DBS) specimen from the newborns, some factors can induce false negatives and false positives. Therefore, several molecular methods have been incorporated to NBS, using the disorders most common mutations as models. These vary due to the ethnic background and those considered "common" mutations by other programs might not be of use to our population. Therefore the aim will be to identify common mutations amongst the population conducting characterization procedures. After characterizing a urea cycle condition patient's genome and comparing it to another baby without the diseases results confirmed that there are indeed two previously unpublished mutations found. Further investigations would confirm if these are specific to our population.

Awards Won: