

Rapid whole genome sequencing in the neonatal intensive care unit of Brazilian hospitals.

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Background & Objectives: Rare Mendelian diseases can be caused by a variety of variant types what makes the molecular diagnosis of Mendelian diseases a challenge because many investigation methods may be used for the identification of disease-causing variants. Traditionally, a series of genetic tests are used step wise for the cytogenetic or molecular diagnosis of these diseases. Mainly in low- and middle-income countries, the access to these tests is difficult, they are very expensive and the results are frequently delayed. Because of that precise timely diagnoses are not made and opportunities for prevention, anticipatory guidance, and treatment are missed or delayed.

Method (s) and Results: To address these difficulties related to performing multiple genetic tests we will perform rapid whole genome sequencing (WGS) on 100 neonatal patients (and their parents) being treated in the intensive care units of Brazilian hospitals with suspected rare Mendelian diseases that fit a pre-established selection criteria. WGS libraries will be sequenced on the NovaSeq 6000 platform at DASA laboratory. The final report will be returned in 15 days and will include the analysis of CNVs, indels, SNVs and a number of selected non-coding variants known to cause rare diseases.

Conclusions (Significance and Impact of the Study): With this project we expect to decrease the time for precise diagnosis of neonatal patients with rare Mendelian disease and in critical medical conditions. Precise timely diagnoses may improve management and treatment of these patients and decrease the costs to the health care system in general.

Conflict of interest disclosure: The authors are employees of the DASA laboratory.