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DIAGNOSIS OF NEUROMETABOLIC DISORDERS THROUGH NEXT GENERATION SEQUENCING PANELS AND BIOINFORMATICS TOOLS: TWO YEARS OF EXPERIENCE

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Genomic research in neurodegenerative and metabolic disorders is essential for: 1) diagnosis, prognosis and treatment of these disorders, 2) the application of new preventive treatments and/or their inclusion in clinical trials of new drugs, 3) provide, within a reasonable period of time, a good family genetic counselling, and 4) avoid diagnostics odyssey.

The new sequencing technologies require us to leap toward a new medication and to a new paradigm in which genomic tools will be essential in the diagnostic process of the patient and, in many cases for early diagnosis of diseases that could eventually only be diagnosed by clinical signs and symptoms developed by the patient.

In recent years, there has been an exponential growth of knowledge about the genetic basis of disorders. This exponential growth of knowledge is now one of the biggest challenges faced by health systems and physicians to their patients. For this reason, the construction of personal genomic skills is essential, especially in a reference unit, and is based on already developed projects (diagnosis of lysosomal disorders, and monogenic diabetes by massive sequencing).

Since October 2012 we have developed and implemented in our unit a total of 17 genetic panels based on massive sequencing technology primarily concerned with the diagnosis of neuropaediatric and metabolic disorders. So far we have analyzed a total of 205 patient samples from up to 20 different hospitals in the country, reaching a diagnosis rate between 40-45%. In this communication we intend not only to relate the results obtained with the application of these paediatric diagnostic panels, but also the challenges we need to face to achieve increase speed, efficiency and safety of these diagnostic tools.

Keywords: metabolic and neuropaediatric disorders, massive sequencing