Mendelics' new approach makes Fabry disease's diagnosis easier and faster

Diagnosis of Fabry Disease will be simpler and more accurate using the Sequencing First strategy

Mendelics, the first and largest Brazilian laboratory specialized in Next Generation Sequencing (NGS), in partnership with Takeda, a pharmaceutical company committed to developing and delivering life-changing treatments, started an innovative program for the diagnosis of Fabry Disease.

Fabry Disease is a rare and progressive hereditary condition characterized by the buildup of a type of fat (called glycosphingolipids), leading to severe systemic symptoms, mainly in the heart, nervous system and kidneys. The disease is caused by mutations in the *GLA* gene, which produces the alpha-galactosidase A (α-Gal A) enzyme, responsible for processing fat molecules inside the cells.

The disease's manifestation and severity vary widely among patients, which makes the diagnosis challenging. Generally, Fabry disease is underdiagnosed worldwide. In addition to the diagnosis difficulty due to nonspecific symptoms, the tests commonly adopted fail to detect it in affected women. Due to the inheritance pattern of the disease, women do not show signs and symptoms as men and they require additional testing, which further delays the diagnosis and, consequently, the start of treatment.

With the Sequencing First approach, the diagnosis of Fabry Disease will be performed primarily by genetic (molecular) testing of the *GLA* gene in search for changes associated with the disease. If any pathogenic variants or variants of uncertain significance are detected (following international protocols for genetic variant classification), complementary biochemical tests will be performed. Such strategy speeds up the diagnosis of the disease, as it bypasses the waiting time for the results of biochemical tests and the subsequent mandatory confirmation by genetic testing, in addition to reducing the chance of false-positive and false-negative results caused by pre-analytical errors and limitations of the techniques used.

"Sequencing First is an innovative approach for the diagnosis of some rare genetic diseases, which consists of DNA sequence analysis as the first diagnostic approach, using biochemical or functional tests as complementary analysis, after an eligible genetic result. We reversed the order of exams so that the patient benefits from a faster and more accurate diagnosis", explains David Schlesinger, Mendelics' CEO. "*The test performs the sequencing of all regions of the GLA gene with greater sensitivity, shorter logistics and execution time*", adds Schlesinger, who is also a geneticist.

Another benefit involves the sample collection for the exam, which is performed using a buccal swab: it is painless, simple and can be done in any clinic, in less than a minute. After DNA sequencing in the laboratory in São Paulo (Brazil), data analysis is performed by the medical team using Abracadabra®, a software developed by Mendelics, following international protocols for the analysis and classification of genetic variants (Guidelines of the American College of Genetics and Genomics, ACMG). The result is available in up to 30 days, a much shorter waiting time when compared with the traditional strategies, and consists of a medical report describing all genetic alterations found.

About Mendelics

Mendelics is the first and largest Brazilian laboratory specialized in Next Generation Sequencing (NGS). It was founded in 2012 with a mission to democratize fast, accurate, data-driven genetic diagnoses. With the largest laboratory structure for genetic sequencing in Latin America, more than 100,000 processed genomic samples, a team of over 300 employees, and pioneering technical and analytical processes with international quality standards, Mendelics has consolidated as a reference in genetic analysis. It is the only Latin American genomic laboratory to simultaneously obtain accreditations from CAP (American College of Pathologists - # 8671464), INMETRO (NBR / ISO-15189), and PALC (Programs of Acceptance for Clinical Laboratories - Brazilian Society of Clinical Pathology - #32290508). Mendelics has also been awarded by MIT for the development of Abracadabra®, an exclusive artificial intelligence platform that performs genetic analyses accurately and efficiently. Since its foundation, Mendelics continues to develop innovative products in healthcare, such as the Genomic Newborn Screening, a test that detects more than 340 early-onset treatable diseases. For more information, visit mendelics.com.