Recent advances in molecular medicine: Changing the practice of neurology

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The study of the nervous system dates back to ancient Egypt. Neurological diseases comprise a vast range of diseases that vary in clinical presentation, genetic influence, and therapeutic response. Many have a specific genetic basis. High throughput techniques such as micro array, comparative Genomic Hybridization (CGH), whole exome and genome sequencing allow for one-step interrogation of multiple genes for an even larger range of diseases. The recognition of these genes and their associated proteins has led to new approaches in treatment. There have been recent achievements in genomic medicine in many neurological conditions. Here we will briefly discuss 2 new things in the field of neurogenetics. Clinicians made the diagnosis of autism spectrum disorders populations and then walked away since there was little to offer for effective therapy. Recently, mutations in the BCKDK gene were detected in affected individuals from consanguineous families who had epilepsy, autistic features, associated with intellectual disability. The encoded protein is responsible for phosphorylation-mediated inactivation of the branched-chain ketoacid dehydrogenase enzyme where the potential treatment is obvious and simple. The second is chromosomal microarray studies that can now identify duplications or deletions of the genome associated with intellectual disability. High consanguinity rates and inbreeding coefficients increases the number of recessive disorders in the Middle East such as Joubert syndrome. Much progress has been made finding genes responsible for Mendelian syndromes involving intellectual disability. A number of new tools for efficient subtelomere screens based on array CGH technologies have recently been published. Telomere analysis has identified cryptic subtelomeric translocation segregating through 5 generations using array-CGH analysis. Subtelomeric rearrangements may be a major cause for unexplained mental retardation and/or multiple congenital anomalies. As these techniques become more accessible to researchers and clinicians, it would build up more knowledge about molecular - behavior relationship, thus changing the understanding of neurological disorders and allowing definite diagnosis which is the cornerstone of genetic counseling.

Biography

Meguid is a Professor of Human Genetics at national research center located in cairo. she is a senior geneticist at the genetics institute in pasadena, california. she is a fellow of medical genetics at uppsala university in sweden. she got prize foe excellence in medical science from national research centre in 2001. in 2002 she was awarded as a L’Oreal-UNESCO for womens in science. she also achieved distinctive arabn scientist prize in genetics from bahrain and national award for scientific excellence in advanced technology in 2009. again in 2011 she got a prize for scientific appreciation from national research centre.

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