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Molecular diagnostics and personalized genomic medicine applied to neurological disorders

Alexis Trott

Federal University of Santa Maria, Brazil

Abstract

The efficiency of molecular tests provides some accurate diagnoses for patients and is essential in the process of treatment and genetic counseling, and is very important for Personalized Genomic Medicine. The early and accurate diagnosis of a particular disease can improve quality of life of many patients, mainly with neurological morbidities. Our studies involve two investigative lines, including (1) the molecular diagnosis of genetic neurodegenerative diseases; (2) the role of single nucleotide polymorphisms (SNPs) in increasing the risk of neuropsychiatric diseases that affect human health, lifespan and longevity. From our genetic studies, we have detected many affected patients with autosomal dominant spinocerebellar ataxias (SCAs), a complex group of neurodegenerative diseases that share a similar molecular mechanism involving expansion of CAG repeats (dynamic mutations). Over the generations, the symptoms can start early with more severe progression, depending on the size of the CAG repeats, a phenomenon called anticipation. In addition, we have developed a non-invasive early prenatal fetal gender determination, which is important in cases with risk of transmission of X-linked diseases. Our tests involve analysis of DYS14 gene and RhD diagnosis by rapid low-cost cell-free DNA extraction with conventional and multiplex PCR that are simple, fast, inexpensive, and highly sensitive techniques. Currently, we are also performing investigations involving some SNPs related to the serotonin signaling pathway that is considered a major modulator of primate higher-order executive tasks. Serotonergic signaling is altered in many psychiatric disorders such as schizophrenia, depression and addiction. Previous investigations, including from our research team, found association between SNPs occurring in the 5-HT2a receptor as T102C polymorphism and smoking habit, alcoholism, food behavior, short lifespan and longevity. In the future, results from these studies may help us detect subjects susceptible to neuropsychiatric disorders and also to develop safer and more efficient pharmacological treatments (pharmacogenetics). Therefore, molecular methods to the diagnosis of diseases can be used as effective and low-invasive tools in the prediction, prevention and therapeutic care to many neurological diseases.

Biography

Alexis Trott received a Bachelor’s Degree in Biological Sciences and a Master's degree in Genetics and Molecular Biology and a Ph.D. in Biochemistry (Medical Genetics) from the Federal University of Rio Grande do Sul (UFRGS), Brazil. He is currently Research Associate at the Federal University of Santa Maria (UFSM), Brazil". He is currently Professor and Principal Investigator at the University of Western Santa Catarina (UNOESC), Brazil. He has published several papers in high-profile journals such as Cytokine and Clinical Genetics, and currently serves as Scientific Journal Referee to journals, including Molecular Neurobiology, Cytokine, Atherosclerosis and the American Journal of Perinatology, as well as Scientific Referee for several Research Foundations.

alexistrott@hotmail.com