



**LEADING SCIENTIST**  
NEURODEGENERATIVE DISEASE AND  
NEUROLOGICAL DISORDERS



**Simon G. Gregory, PhD**  
**David H. Murdock Research Institute (DHMRI)**  
Director, Genomics Core Laboratory  
Director, Discovery MS

**Duke University School of Medicine**  
Professor of Neurology



## EXPERTISE

Genetics, genomics  
and epigenetics of  
complex disease

## DHMRI

Integrated analytical  
platforms, knowledge-  
building capabilities,  
and academic-industry  
partnerships, focused on  
human health, nutrition  
and agriculture

## DISCOVERIES

- » Used novel approaches and resources to construct a map on human chromosome 13 to establish a role for mutations in the BRCA2 gene with familial breast cancer
- » Led the construction of the mouse genome map using human sequence homology, which facilitated the mapping, sequencing and evolutionary assessment of other organisms
- » Directed the coordination of the mapping and sequencing of human chromosome 1, as part of the Human Genome Project, which led to the identification of mutations responsible for 35 Mendelian disorders, developmental disorders or cancer
- » Used convergent analysis of gene expression, linkage and genetic association in humans, and QTL and gene expression in model organisms to identify the genetic association of interleukin receptor 7 alpha chain (IL-7R $\alpha$ ) gene with multiple sclerosis (MS)

## RESEARCHING

- » Identification of the genes and mechanism behind IL-7R $\alpha$  splicing and their putative role in the development of MS and other autoimmune diseases
- » Characterization of novel biomarkers of chronic pathology in the development of MS and establishment of novel drug treatments for the disease
- » Identification of predictive biomarkers of response and the effect of drug exposure in a novel drug trial of oxytocin in autism spectrum disorder





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## C A S E S T U D Y

### Finding Mechanisms, Pathways and Genetic Regulation of Autoimmune Diseases

**Challenge:** The laboratories of Dr. Simon Gregory, PhD, at the David H. Murdock Research Institute and the Duke Molecular Physiology Institute are identifying the mechanisms of disease development and novel methods of treatment in multiple sclerosis. Like all autoimmune diseases, MS is caused by the body's inability to 'recognize self.' The result is an individual's immune system attacking constituents of the central nervous system. Further, MS is caused by both genetic risk factors and environmental influences. Dr. Gregory's lab is trying to understand these mechanisms.

**Research:** IL-7R $\alpha$  was the first gene outside of the MHC to be associated with MS in more than 20 years. Dr. Gregory's laboratory has since identified genes that regulate IL7R and are also associated with MS. Genetic and functional association with DDX39B was published in *Cell* opening a novel pathway and mode of gene regulation in other autoimmune diseases. Analyses are currently expanding to other genes that are regulated by DDX, which promises to provide significant impact in the field of autoimmune research.

**Importance:** MS is a disease for which there is no cure. Dr. Gregory's lab, through collaboration with groups across the US, is trying to understand the mechanisms of how the disease develops, the development of novel signatures that would permit an earlier diagnosis and exploring a novel drug therapy using naturally occurring compounds within the body to trigger a reverse of the pathology of the disease.

**Publications:** *Cell*. Human Epistatic Interaction Controls IL7R Splicing and Increases Multiple Sclerosis Risk. Galarza-Muñoz G, Briggs FBS, Evsyukova I, Schott-Lerner G, Kennedy EM, Nyanhete T, Wang L, Bergamaschi L, Widen SG, Tomaras GD, Ko DC, Bradrick SS, Barcellos LF, Gregory SG, Garcia-Blanco MA., March 2017.

### About The North Carolina Research Campus

The North Carolina Research Campus, located in Kannapolis, NC, near Charlotte, is a scientific community of eight universities, the David H. Murdock Research Institute, global companies and entrepreneurs. Research and development focus on safer, more nutritious crops, healthier foods and precision nutrition.

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