

LAUREL BRAGG, and BRUCE ANTHONY., Department of Chemistry/Biochemistry, West Virginia Wesleyan College, Buckhannon, WV, 26201.
Detection of splice variants/deletions within the gene spastin.

Hereditary spastic paraplegias (HSPs) are an inherited set of diseases whose main phenotype is progressive stiffness and contraction in lower limbs as a result of peripheral nerve damage. This genetic disorder follows inheritance including autosomal dominant or recessive, or x-linked recessive. The mode has a direct impact on the chances of inheriting the disorder. Long nerves are affected due to long distances to transport, and are particularly sensitive to defects. Genes that contribute to the disorder include; 72 spastic gait disease-loci and 55 spastic paraplegia genes (SPGs). Several genetic mutations have been identified which underlie various forms of HSP, and genetic testing is available and used to confirm clinical diagnosis. Testing does not include all genes or identify all the mutations of genes known to cause HSP. Many cases involve a mutation in the SPG4 (Spastin) gene, which controls the spasticity of the lower extremities, yet the extent of mutation or variance is unknown.

After previous research, which determined the average expression levels between two different exons, further analysis was performed using nested primers within exons 5 and 11 to narrow down the location for splice variants in the chosen gene. Buccal swabs were collected from several consenting patients (control and affected) and PCR used to amplify differences. A decrease in expression levels of the tested gene indicated missing genomic sequence while increases mean extra DNA. We suggest this to be a possible diagnostic tool for specific forms of HSPs.