Gene identification in HSP, the handling of large scale genomic data, and sharing of genomic results across countries

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Next-generation sequencing has significantly increased the pace of gene discovery in hereditary spastic paraplegias and neuromuscular diseases. Further, it increasingly allows for the delineation and precise description of phenotypic spectra of disease genes, the resulting phenotypic overlap between clinical entities, and in some areas this is challenging the existing classification of disorders. With the decreasing price for genome sequencing, we will witness an even larger expansion of genetic data in the coming years. Going forward it appears inevitable that intelligent and safe sharing of data on genetic variation is a precondition to overcome the challenges of determining pathogenicity and to allow for the unbiased determination of validity of new disease genes. Local and global efforts are now taking shape to address this pressing issue, which will eventually lead to a global understanding of genetic variation in our species, and hence, improved and standardized diagnostic approaches in HSP. This presentation will review these issues and highlight work done in the Zuchner lab, the spastic paraplegia network, and recent efforts of the Global Alliance for Genomics and Health (GA4GH).