November 6th, 2017 - Brenda Degerald, Senior Array and Genotyping Specialist, Illumina & Jonathan Bibliowicz, PhD, Senior Sequencing Specialist, Illumina - *Genomic Solutions for the Study of Complex Neurodegenerative Disorders.*

The ADRC invited Illumina to give a special seminar about the newest available genomic sequencing and array technology for study of neurodegenerative diseases at the molecular level. Jonathan Bibliowicz, PhD, of Illumina spoke about how a 'Multi-Omics' approach can help researchers gain more detailed insight into the complex genetics of neurodegeneration, and also generate findings that reflect causation instead of correlation.

Illumina sees promise in following the current successful approaches in cancer research, which feature the use of WES and WGS to search for gene drivers and pathways altered in specific tumor types. Illumina offers TruSeq PCR Free for WGS; Nextera Exome for WES; and the Amplicon Panel, custom or designed. Notably, sequencing costs have steeply declined with the use of NovaSeq 6000.

In the area of gene discovery in neurodegeneration, Illumina offers the TruSeq Neurodegeneration Panel, an application used to identify novel variants in known genes related to neurodegenerative diseases and screen different population samples for known variants. Validation of known variants detected in GWAS and WSG/WES helps researchers to investigate the genetic pathways underlying multiple diseases, such as ALS, PD, AD, FTD, DLB, and EOD.

The rationale behind 'Multi-Omics' is that multiple methods build confidence in results and can help researchers publish more findings of causation than correlation. For example, Multi-Omic analysis has already led to observations about the molecular mechanisms driving aging of the immune system, such as a global difference and transcriptional differences between young and old T cells. Multi-Omics comprises mutation detection technologies (genome, exome, arrays, panels); RNA-Seq (Total RNA, mRNA, small RNA); and epigenetics (Methyl-Seq, ChIP-Seq, ATAC- Seq).

In the quest to understand genetic networks, ATAC-Seq enables the study of gene regulation through epigenetic profiling. This method improves the input amount and reduces prep time, and uses a simple cell input protocol. The Nextera based library preparation can be used for studying open-chromatin structure.

The Illumina speaker emphasized the importance of single cell sequencing for the study of complex cellular dynamics. Illumina sequencers offers a full wide of applications: miR-seq, RNA-seq, exome, genomic enrichment, targeted panels, genome sequencing, rare sequence detection, MG meta genomics, bisulfite sequencing, and ChiP-seq.

Turning to the subject of genotyping arrays, Illumina's Brenda Degerald laid out the advantages of this mature technology: cost per sample cost, sample through-put, statistical power, and data management. Illumina's genomic portfolio provides flexible solutions for all approaches, from global screening array (population scale screening and discovery) to multiethnic genotyping array family (large scale cohort studies), and from arrays to next generation WGS and targeted sequencing. The content of the focused genome portfolio includes the specific type of disease, condition, or classification, including the neuro-consortium array; drug consortium array; psych array; immunoArray v2, QC Array, Exome, and OncoArray.

Furthermore, the Infinium NeuroArray-24 provides deep replication and fine mapping for neurological diseases. Infinium NeuroArray-24 includes a Core exome-24 backbone, with known content from ExAC and the NHGRI GWAS Catalog and novel content from whole-genome and whole-exome sequencing.

To close this presentation of the "Illumina genetic continuum," Degerald introduced methylation analysis technology. Methylation arrays are cost effective for large-scale screening, while methylation sequencing provides deep information from across CpG rich regions.