Abstracts to be presented at ASHG 2022				
Date	**Time (PT)**	**Lead Author**	**Title**	**Program #/ID**
October 25th	4:55 - 5:15 pm	Sarah L. Stenton	Performance of diagnostic methods in identifying disease-causing variants: assessment of the Rare Genomes Project CAGI challenge	52
October 26th	9:00 - 9:15 am	Marafi, Dana	A reverse genetics and genomics approach to gene paralog function and disease: Myokymia and the juxtaparanode	90
	11:30 - 11:45 am	Moez Dawood	Saturation genome editing reveals 10% of missense SNV alleles in functional domains of PALB2 as functionally abnormal	147
	3:00 - 4:45 pm	Scott Barish	ITGB8 is a candidate disease gene for autosomal dominant and recessive trait forms of muscular dystrophy and neurological disease	PB1895
		Haowei Du	The Multiple de novo Copy Number Variant (MdnCNV) phenomenon: peri-zygotic DNA mutational signatures and multilocus pathogenic variations	PB2703
		Hadia Hijazi	TCEAL1 loss-of-function results in an X-linked dominant neurological syndrome and drives the neurological disease trait in Xq22.2 deletion	PB1981
October 27th	3:00 - 4:45 pm	Stylianos Antonarakis	FOXI3 pathogenic variants cause one form of craniofacial microsomia	PB2586
		Bonner, Devon	Multi-omic approach identifies a novel non-coding deletion at Xq28 in a patient with X-linked primary immunodeficiency.	PB1922
		Daniel Calame	Monoallelic variation in the DExH-box helicase DHX9, a product of the DHX9 gene paralog, perturbs neurodevelopment & causes peripheral nerve axon degeneration	PB1920
		Zain Dardas	Genome-wide investigation of potentially pathogenic copy number variants & mechanisms fomenting their origins	PB2303
		Ruizhi (Vince) Duan	Large-scale multimodal genomic analyses of 150 consanguineous kindreds from the Middle East and North Africa uncover novel neurodevelopmental disease mechanisms	PB1898
		F. G. Frost	Deleterious SNAPC4 Variants are Associated with a Neurodevelopmental Disorder	PB1810
		A. Garde	SRSF1 haploinsufficiency is responsible for a syndromic developmental disorder with intellectual disability and variable marfanoid habitus	PB1974
		Grochowski, Christopher M.	A multiomics approach to resolving small supernumerary marker chromosomes	PB2248
		Roni Zemet Lazar	Biallelic RAD51C loss-of-function variants drive perizygotic SNV/indel hypermutator phenotype in a subject with Fanconi anemia complementation group O	PB1771
		Andy Rivera	Integrating Genomic and Phenotypic Analyses of Autonomic Nervous System Dysfunction in a Rare Neurological Disease Cohort	PB1888
		Walker, Kimberly	Exome technology innovations advancing personalized medicine	PB2988
October 28th	2:00 - 2:15 pm	Liu, Pengfei	Application of RNA sequencing on transdifferentiated patient fibroblasts for genetic diagnosis of neurological disorders	434
	3:15 - 3:30 pm	Avinash Dharmadhikari	Bi-allelic variants in SPOUT1, an RNA methyltransferase functioning in spindle organization, cause a novel neurodevelopment disorder	463
October 29th	9:15 - 9:30 am	Goddard, Pagé and Ungar, Rachel	Genome reference impacts RNA-seq interpretation and rare disease diagnosis	553

**Titles link to program/poster submission on GREGoR Consortium website (gregorconsortium.org)*