Abstracts to be presented at ASHG 2023				
Date	**Time (ET)**	**Title**	**Lead Author**	**Program #/ID**
Thursday, November 2nd	**9:00 - 9:15 am**	Heterozygous Loss-of-Function Variants in SMC3: Lessons for the 'Medium-Hanging Fruit' Era of Mendelian Disease Gene Discovery	Boone, Philip	2023-A-1559-ASHG
	9:15 - 9:30 am	Break-induced replication mediated by inverted repeats underlie formation of pathogenic inverted triplications	Grochowski, Christopher	2023-A-3165-ASHG
		MECP2 copy number variants studied by multiple approaches reveal impact of genomic structure to disease variability	Bengtsson, Jesse	2023-A-2794-ASHG
	2:00 - 2:15 pm	Novel molecular diagnoses in individuals with holoprosencephaly and prior negative sequencing	Cohen, Andrea J.	2023-A-3728-ASHG
	3:00 - 5:00 pm	Elucidating the genetic etiology underlying septo-optic dysplasia (SOD)	Beheshti, Shaghayegh	PB4737
		Making the call: Trends among successful strategies to conclude the diagnostic odyssey for participants at the Pacific Northwest Undiagnosed Diseases Network clinical site.	Blue, Elizabeth	PB3237
		VizCNV: An integrated platform for CNV detection and analysis of genome sequencing data	Du, Haowei	PB3523
		Complex genetic architecture underlying craniofacial microsomia	Gogate, Nikhita	PB4698
		Expanding the range of WNT signaling syndromes: a promoter variant in WNT9B is a candidate in a case with Femoral Facial syndrome.	Marvin, Colby	PB4755
		Investigating the genetic and phenotypic landscape of Ectodermal Dysplasia	Munderloh, Chloe	PB1618
		A multi-omics approach to the characterization of a novel repeat expansion in FAM193B in a family with oculopharyngodistal myopathy	Reuter, Chloe	PB4596
Friday, November 3rd	**3:00 - 5:00 pm**	Unraveling the Intrafamilial Phenotypic Variability in Sibling Pairs with Neurodevelopmental Diseases	Bozkurt-Yozgatli, Tugce	PB4933
		Biallelic FLVCR1 variants cause a disease spectrum from adult neurodegeneration to severe neurodevelopmental disorders through disrupted choline transport	Calame, Daniel	PB4645
		Genomic analyses of 281 consanguineous kindreds from the Middle East and North Africa facilitate the discovery of novel recessive neurodevelopmental rare disease traits	Duan, Ruizhi	PB4786
		Data sharing in the GREGoR Consortium to support rare genetic disease research.	Heavner, Ben	PB4711
		Evaluation of missing disease-causing variation in autosomal recessive conditions using long-read sequencing	Patterson, Karynne	PB3205
		Genomic Rare Variant Mechanisms for Congenital Cardiac Laterality Defect: A Digenic Model Approach.	Rai, Archana	PB1568
		A VUS re-analysis: Understanding the likely disease-mechanism of a de novo missense variant in RALA using public bioinformatic tools	Wiel, Laurens	PB1019
Saturday, November 4th	**10:45 - 11:00 am**	Driver project for advancing long-read de novo genome assembly methods in clinical research	Délot, Emmanuèle	2023-A-4146-ASHG
	11:00 - 11:15 am	Clinical exome sequencing efficacy and phenotypic expansions in Congenital Anomalies of Kidney and Urinary Tract (CAKUT)	Rivera-Munoz, Andres	2023-A-1625-ASHG
	2:15 - 4:15 pm	NODAL variation is associated with a continuum of laterality defects from simple D-transposition of the great arteries to heterotaxy	Dardas, Zain	PB4300
		Long-read sequencing of 1000 Genomes Project samples to catalog normal patterns of human genome structural variation	Gustafson, Jonas	PB4524
		BCM-GREGoR: A rare disease program to solve the unsolved with novel methods and analytical approaches	Jhangiani, Shalini	PB4643
		Investigating the molecular mechanism of a complex genomic rearrangement causing 6X amplification at 13q33.3	Kaur, Parneet	PB3233
		ARHGAP1 identified as a candidate gene for a novel autosomal dominant syndromic neurodevelopmental disorder.	Mendez, Hector Rodrigo	PB4634

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