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Abstract Topic: - Genetic, genome and epigenome databases and resources

Abstract Title: - Whole-Genome Sequencing of 100 Genomes Identifies a Distinctive Genetic Susceptibility Profile of Qatari Patients with Hypertension

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Aims: - this study aimed to screen for variants that are associated with EH in 100 hypertensive/100 control patients comprising Qatari individuals using GWASs of whole-genome sequencing and compare these findings with genetic data obtained from more than 10,000 published peer-reviewed studies on EH.

Methods: - Whole genome Sequencing, GWAS

Results: - The GWAS analysis performed with 21,096 SNPs revealed 38 SNPs with a significant ≥4 log-p value association with EH. The two highest EH-associated SNPs (rs921932379 and rs113688672) revealed a significance score of ≥5 log-p value. These SNPs are located within the inter-genic region of GMPS-SETP14 and ISCA1P6-AC012451.1, respectively. Text mining yielded 3748 genes and 3078 SNPs, where 51 genes and 24 SNPs were mentioned in more than 30 and 10 different articles, respectively. Comparing our GWAS results to previously published articles revealed 194 that are unique to our patient cohort; of these, 13 genes that have 26 SNPs are the most significant with ≥4 log-p value. Of these genes, C2orf47-SPATS2L contains nine EH-associated SNPs. Most of EH-associated genes are related to ion gate channel activity and cardiac conduction. The disease-gene analysis revealed that a large number of EH-associated genes are associated with a variety of cardiovascular disorders. The clustering analysis using EH-associated SNPs across different ethnic groups showed high frequency for the minor allele in different ethnic groups, including Africans, East Asians, and South Asians.

Conclusions: - The combination of GWAS and text mining helped in identifying the unique genetic susceptibility profile of Qatari patients with EH. To our knowledge, this is the first small study that searched for genetic factors associated with EH in Qatari patients.

Keywords: - The GWAS analysis performed with 21,096 SNPs revealed 38 SNPs with a significant ≥4 log-p value association with EH. The two highest EH-associated SNPs (rs921932379 and rs113688672) revealed a significance score of ≥5 log-p value. These SNPs are located within the inter-genic region of GMPS-SETP14 and ISCA1P6-AC012451.1, respectively. Text mining yielded 3748 genes and 3078 SNPs, where 51 genes and 24 SNPs were mentioned in more than 30 and 10 different articles, respectively. Comparing our GWAS results to previously published articles revealed 194 that are unique to our patient cohort; of these, 13 genes that have 26 SNPs are the most significant with ≥4 log-p value. Of these genes, C2orf47-SPATS2L contains nine EH-associated SNPs. Most of EH-associated genes are related to ion gate channel activity and cardiac conduction. The disease-gene analysis revealed that a large

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