



Genetic Variant (rs2304672) in 5' UTR region in PER2 gene and its association to Parasomnias and other sleep disorders in Puerto Rico

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ABSTRACT

Sleeping is one of the main physiological processes of restoration. Inadequate sleep patterns can limit this process and lead to chronic health conditions. In 2014, both World Health Organization (WHO) and the Central Disease Control (CDC) in the United States indicate sleep problems as a public health crisis. Approximately 40% of the general population suffers from sleep disorders, additionally 60% of these use medication to induce sleep. External factors, such as stress, sounds, and light quality have been theorized to contribute to lack in sleep quality. Yet, little is studied on internal factors affecting sleep quality. In this study, we referenced data from the 1000 Genomes Project to study the genetic variant (rs2304672) of the PER2 gene in Puerto Rico. The variant is linked to poor sleep quality and a sleep disorder known as Parasomnia. Characterized by sleep walking, sleep talking, bedwetting, nightmare disorders, and sleep paralysis principally. As an admixed population, Puerto Rico's three main ethnic groups have an influential factor in the distribution and prevalence of said genetic variant in the island. Our results found that the genetic variant is common in the southwestern and southeastern coastal regions of Puerto Rico. Additionally, we found that the genetic variant frequency in the entire island is of 3.83% in the general population of Puerto Rico.

INTRODUCTION

- Sleep is a process of neurological, cellular, and systemic regeneration in the human body.
 - PER2 gene is the principal gene that regulates sleep.
- In 2014, the World Health Organization (WHO) and the Central Disease Control (CDC) indicate a Public Health Crisis regarding poor sleep quality.
- Genetic variant rs2304672 associated to poor sleep and Parasomnia
 - Parasomnias: Sleep disorder categorized by abnormal activity in any stage of the sleep cycle.
- Puerto Rico is an admixed (mixed) country of various ethnic groups and is theoretically favored to house genetic variants
- This study is aimed at identifying the genetic variant in Puerto Rico and its distribution

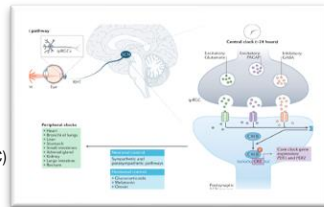


Fig 1. Circadian Cycle (Sleep/Wake Cycle) diagram explaining the activation or inhibition of sleep by genes and biochemical pathways mediated by light stimulus

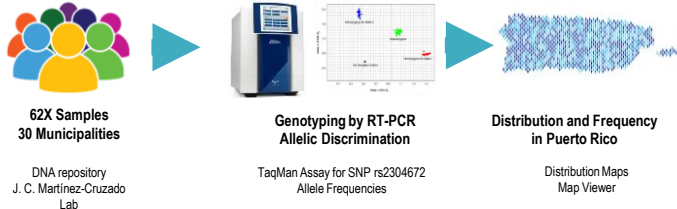


Fig 2. Countries chosen as Representative marker populations in the study 1000 Genomes

OBJECTIVES

- Confirm the Presence of the genetic variant of PER2 gene, rs2304672, in Puerto Rico
- Determine distribution patterns and frequency of the genetic variant in Puerto Rico
- Compare statistical data from this study and the 1000 Genomes Project

METHODOLOGY



RESULTS

Region	Genetic Variant Frequency
North	1.64%
South	2.38%
Center	2.66%
East	8.39%
West	5.07%
Island-wide	3.83%
1000 Genomes Project	8%

Table 1. Percentages of the distribution of the genetic variant rs2304672 in the island of Puerto Rico, compared to the 1000 Genomes Project data

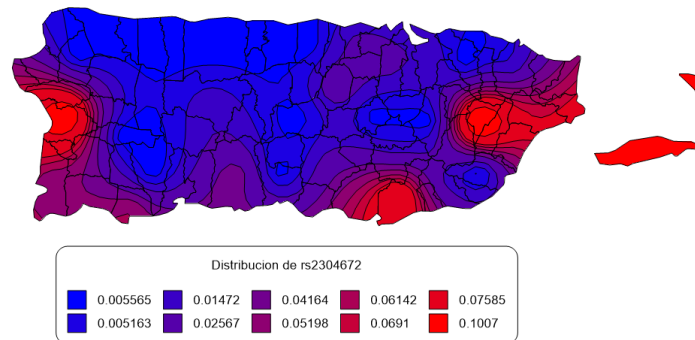


Fig 3. - Dispersion and frequency of the genetic variant rs2304672 in the PER2 gene in the global population of Puerto Rico.

PRELIMINARY CONCLUSIONS

Our gathered data suggests that the presence of the genetic variant of the PER2 gene (rs2304672) is present in Puerto Rico. Furthermore, we were able compare our data with the data from the 1000 Genomes Project. In Puerto Rico, we found that the genetic variant exists in 3.83% in population while the 1000 Genomes Project reports 8% of the general population contains the genetic variant. This difference in percentage is principally attributed to the number of samples tested; a difference of 1:5 between the 1000 Genomes project and this study, respectively. Additionally, we found that the genetic variant is common along the coastal southwest and southeast of Puerto Rico.

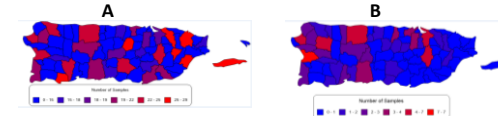


Fig 4. Municipality sample distribution for our study and Puerto Ricans in the 1000 Genomes Project. Our study sample distribution by municipality is shown in A (n= 622) and in B municipality distribution of Puerto Rican volunteers for the 1000 Genomes Project until October 10, 2009 (n=77).

FUTURE PLANS

Further testing must be done to differentiate presence of heterozygotes and speculated homozygotes for genetic variant. Equally, the genetic variant must be tested on volunteers that share symptomatic characteristics or familial history to Parasomnias. Including:

- Sleep Paralysis
- Narcolepsy
- Insomnia
- Sleep Talking
- Sleep Walking
- Recurrent Nightmares
- Bedwetting (Enuresis)
- Leg/Arm Movements during sleep
- Teeth Biting During Sleep (Bruxism)
- Night Terrors
- Panic Attacks
- Anxiety Attacks
- Anxiety
- Depression
- Neurological Conditions
- Neuromuscular Conditions
- Alzheimer's Disease
- Bipolar Disorder
- Parkinson's Disease
- Celiac Disease
- Professions of High Stress
- PTSD
- Sleepy in absence of light
- Sleep sweating

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