Study of twins discovers gene mutation linked to short sleep duration

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Researchers who studied 100 twin pairs have identified a gene mutation that may allow the carrier to function normally on less than six hours of sleep per night. The genetic variant also appears to provide greater resistance to the effects of sleep deprivation.

Results show that a participant with p.Tyr362His – a variant of the BHLHE41 gene – had an average nightly sleep duration of only five hours, which was more than one hour shorter than the non-carrier twin, who slept for about six hours and five minutes per night. The twin with the gene mutation also had 40 percent fewer average lapses of performance during 38 hours without sleep and required less recovery sleep afterward – sleeping only eight hours after the period of extended sleep deprivation compared with his twin brother, who slept for 9.5 hours.

According to the authors, this is only the second study to link a mutation of the BHLHE41 gene – also known as DEC2 - to short sleep duration. The study provides new insights into the genetic basis of short sleep in humans and the molecular mechanisms involved in setting the duration of sleep that individuals need.

"This work provides an important second gene variant associated with sleep deprivation and for the first time shows the role of BHLHE41 in resistance to sleep deprivation in humans," said lead author Renata Pellegrino, PhD, senior research associate in the Center for Applied Genomics at The Children's Hospital of Philadelphia. "The mutation was

associated with resistance to the neurobehavioral effects of sleep deprivation."

Study results are published in the Aug. 1 issue of the journal *Sleep*.

The study group comprised 100 twin pairs – 59 monozygotic pairs and 41 dizygotic pairs – who were recruited at the University of Pennsylvania. All twin pairs were the same sex and were healthy with no chronic conditions. Nightly sleep duration was measured at home by actigraphy for seven to eight nights. Response to 38 hours of sleep deprivation and length of recovery sleep were assessed in a sleep lab. During sleep deprivation, cognitive performance was measured every two hours using the Psychomotor Vigilance Test.

Although individual sleep needs vary, the American Academy of Sleep Medicine recommends that adults get about seven to nine hours of nightly sleep. However, a small percentage of adults are normal short sleepers who routinely obtain less than six hours of sleep per night without any complaints of sleep difficulties and no obvious daytime dysfunction.

"This study emphasizes that our need for sleep is a biological requirement, not a personal preference," said American Academy of Sleep Medicine President Dr. Timothy Morgenthaler. "Most adults need at least seven hours of quality sleep each night for optimal health, productivity and daytime alertness."

According to the AASM, most people who regularly get six hours of sleep or less are restricting their sleep and suffer from insufficient sleep syndrome, which occurs when an individual persistently fails to obtain the amount of sleep required to maintain normal levels of alertness and wakefulness. Data from the Centers for Disease Control and Prevention indicate that 28 percent of U.S. adults report sleeping six hours or less in

a 24-hour period. Insufficient sleep results in increased daytime sleepiness, concentration problems and lowered energy level, and it increases the risk of depression, drowsy driving, and workplace accidents.

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