The role of genetics in determining diurnal preferences

Have you felt more forgetful and less focused after a sleepless night? Have you noticed that some of your friends are active already early in the morning, while others would sleep till noon and prefer to work late at night? These are but a few examples of the effect sleep may have on us, our productivity and even our health.

Sleep is still one of the greatest mysteries in science. Its exact function is unknown, but according to the most common theories we need it for the healing of the body, normal functioning of immune and endocrine system, energy replenishment, and memory consolidation. Sleep is believed to be determined by genetic mechanisms shared by all species ranging from insects to mammals.

The timing of sleep is established by 24-h rhythm determined by the **circadian system**, which is activated by light and food intake. Sleep pattern is most likely regulated by sleep homeostasis. The external signals (light, food, sound, temperature, exercise, drugs, etc.) push the homeostasis system to synchronize the internal signals (blood glucose, temperature adaption, hormone production, etc.) accordingly to ensure optimum functioning of the body. Constantly forced desynchronization (shift-work or sleep deprivation, abnormal eating rhythms; also artificial light) requires the body to make extra effort, which may have negative consequences such as reduced physical and mental performance, health problems, weight gain, etc. On the other hand, certain eating and training patterns help to avoid desynchronization in case of shift-work or travelling between different time zones.

Sleep pattern also depends on our **chronotype** that determines if our productivity is greater in the hours of early morning or later in the day, which in most cases refers to our regular rising and bedtimes. Based on chronotype, people are usually classified as morning types (“larks”), evening types (“owls”), or intermediate types. Chronotypes differ by sleep time, melatonin production (hormone that causes increase of sleepiness when it gets dark) and serum cortisol levels (hormone released in response to stress). They also respond differently to sustained wakefulness – “larks” find it much more difficult to focus after a sleepless night than “owls”. However, “owls” have higher risk for delayed sleep phase syndrome. Chronotype determines the peak of physical performance and influences our eating pattern, emotional status, and intelligence. Chronotype can be identified by using self-assessment questionnaires e.g. by *Horne-Östberg*), but they are subjective and might in fact reflect the conventional and not internal clock. Or you can test different rising and bedtimes for a certain period and compare the results. But that is very time-consuming and not always possible due to social obligations. Genetic test, however, provides quick objective results.

Circadian clock genes are believed to have the greatest effect on sleep timing and homeostasis, i.e. chronotype. As they also impact other genes, disturbances in sleep pattern may impact almost all our genes via various signal paths. Genetic effect on diurnal types is approximately 50%, which makes it a hereditary feature. However, chronotype is largely independent of ethnicity, gender and socioeconomic position.

Genetic tests study variations in our DNA sequence and interpret the information available from scientific research. There are different forms of variations – single nucleotide polymorphisms or longer variations such as variable number tandem repeat

(VNTR), where blocks with similar sequence are repeated several times. One of widely studied VNTRs is located in *PER3* gene – this 54 base pair long variation can occur in either four ("short allele") or five ("long allele") repeats and has been associated to diurnal preferences – evening types tend to have two short alleles, intermediate types one short and one long and morning types two long alleles. Knowing that, we can sync our internal and conventional clock if necessary. Based on chronotype, we can also assess various lifestyle-related risks and predispositions – the impact of sleep deficiency, risk for delayed sleep phase syndrome, eating habits, including emotional eating, peak time for physical abilities, emotions and intelligence.

Genetic tests are easy to order and samples are collected from saliva, not blood. The results can be received within a couple of weeks. The information gained from the DNA sequencer is converted into an understandable format and then we can make our own conclusions and make relevant changes to our lifestyle where necessary.

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