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SUPPLEMENTARY INFORMATION

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Integrating behavioural health tracking in human genetics research

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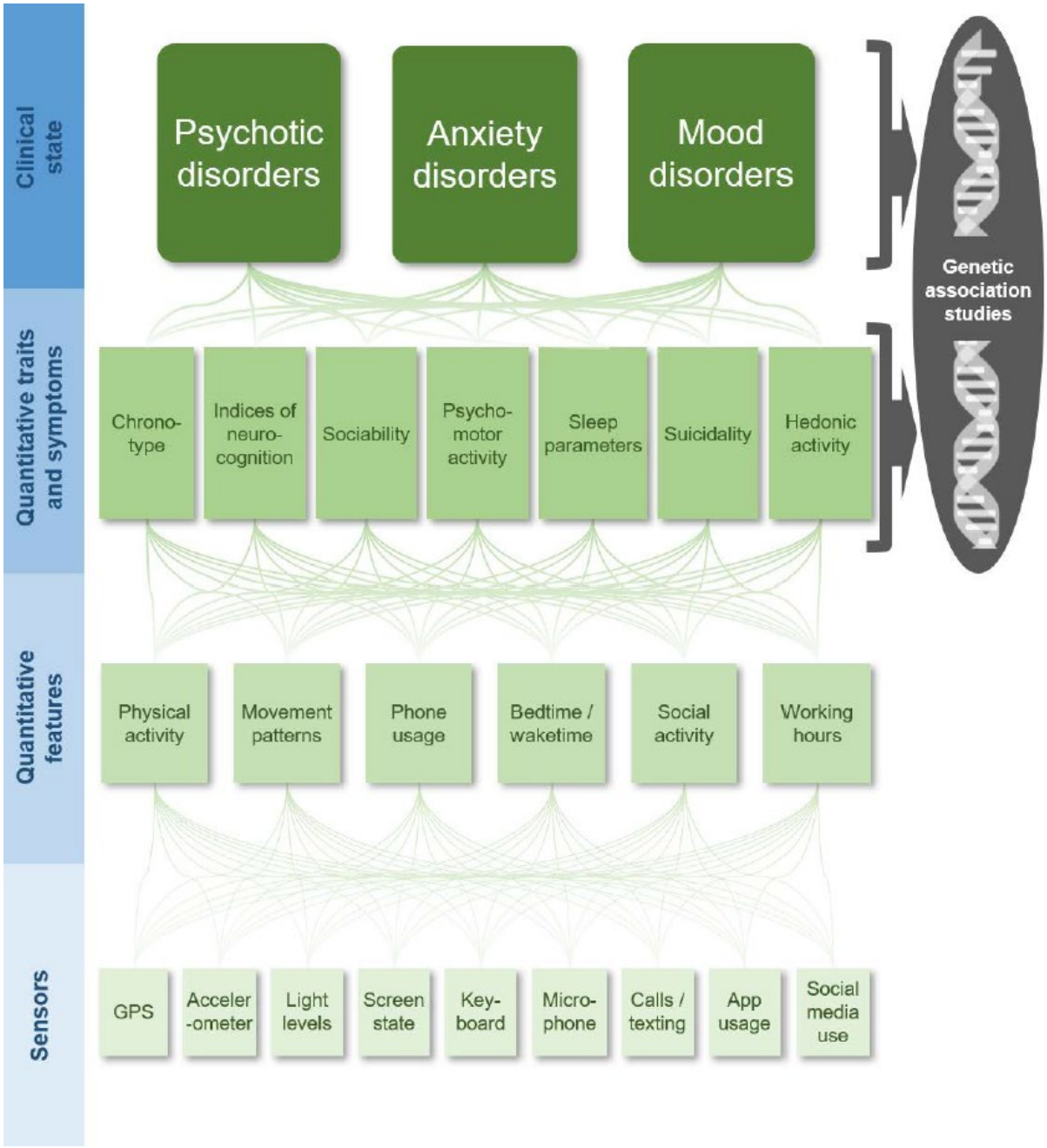
Supplementary Box 1. Potential research impact of behavioural tracking of sleep measures.

Severe mood disorders, including bipolar disorder (manic–depressive illness) and major depressive disorder, are associated with disturbances in several parameters of sleep. However, current diagnostic criteria (and hence most genetic studies undertaken to date) consider only the most basic information about sleep. For example, self-reported abnormality in the amount of sleep is among the core criteria for depressive episodes, but is not defined quantitatively, nor characterized longitudinally (beyond the requirement that it must occur for at least 1 month). Furthermore, these criteria do not distinguish between hypersomnia and insomnia, despite evidence that such information might provide a means to identify distinct subtypes of mood disorder[1]. Behavioural health tracking will enable objective determination of the amount of sleep experienced by an individual, cross-sectionally and longitudinally, both within and outside of depressive episodes. But of even greater potential value, behavioural tracking can provide the means for large-scale longitudinal assessment of a wide range of sleep parameters that have been related to mood disorders, for example, variability across different days in the length of sleep bouts[2], and incorporate these measures as phenotypes for genetic studies.

UK Biobank used a self-report questionnaire about sleep duration and quality to establish multiple sleep phenotypes in >400,000 participants. Because of the anticipated inaccuracies in these data, and to enable analyses of a broader range of sleep parameters, the project obtained objective cross-sectional sleep phenotypes by providing ~85,000 individuals with actigraphy watches from which they collected continuous accelerometer data for 7 days. Smartphones, by contrast, yield sleep data that approach in richness and accuracy the information obtained from actigraphy devices but do so without requiring the provision of any hardware beyond what the participants are already using; they thus can generate data continuously over periods of months to years from possibly several hundred million people worldwide, in conjunction with a wide range of similarly longitudinal behavioural, physiological, and environmental data.

References

1. Harvey, A. G. Sleep and circadian functioning: Critical mechanisms in the mood disorders? *Annu. Rev. Clin. Psychol.* **7**, 297–319 (2011).
2. Pagani, L. et al. Genetic contributions to circadian activity rhythm and sleep pattern phenotypes in pedigrees segregating for severe bipolar disorder. *Proc. Natl Acad. Sci. USA* **113**, E754–E761 (2016).



Supplementary Figure 1. Schematic of behavioural health tracking.

The figure illustrates the steps involved in using sensor data from connected devices for behavioural health tracking. By extracting a set of reliably measured features from different types of sensor data, it is possible to construct heritable quantitative traits and symptoms (endophenotypes) that are associated with categorical diagnoses. Both the endophenotypes and categorical diagnoses are used for genetic association studies.