CENTER FOR NEUROSCIENCE AND CELL BIOLOGY

RARE JOURNEYS WITH INSOMNIA

A PROJECT FROM THE CENTER FOR NEUROSCIENCE AND CELL BIOLOGY OF THE UNIVERSITY OF COIMBRA (CNC-UC)

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SLEEPING WELL IS FUNDAMENTAL TO OUR PHYSICAL AND PSYCHOLOGICAL WELL-BEING.

HOWEVER, THERE ARE TIMES WHEN WE CAN'T SLEEP, AND THAT IS CALLED **INSOMNIA**.

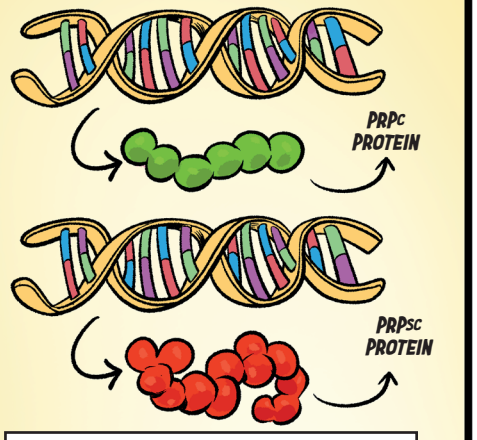
INSOMNIA IS RELATIVELY COMMON, WITH 1 IN 10 PEOPLE SUFFERING FROM CHRONIC AND SEVERE INSOMNIA.

WHAT IS RARE IS **FATAL FAMILIAL INSOMNIA** (OR FFI).

AS ITS NAME SUGGESTS, IT IS A FATAL DISEASE WITH A KNOWN **GENETIC CAUSE**

FFI IS A NEURODEGENERATIVE DISEASE THAT AFFECTS AROUND **50 FAMILIES** WORLDWIDE.

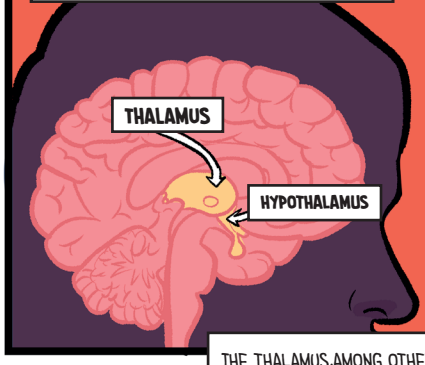
FFI IS A RARE **PRION** DISEASE THAT LEADS TO A SIGNIFICANT LOSS OF PHYSICAL AND MENTAL FUNCTION, WITH A MAJOR IMPACT ON THE QUALITY OF PATIENTS' LIVES AND THEIR FAMILIES.



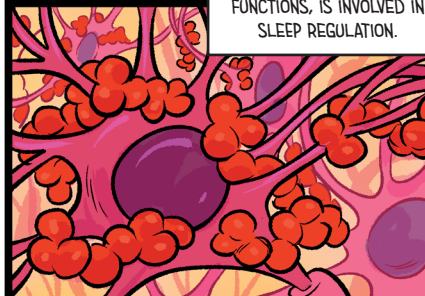
THE DISEASE IS CAUSED BY A MUTATION IN THE **PRNP** GENE THAT ENCODES THE SCRAPIE PRION PROTEIN "PRPSC", AN ABNORMAL PROTEIN, INSTEAD OF THE PRION PROTEIN "**PRPC**".

THIS MUTATION IS TRANSMITTED DOMINANTLY, WHICH MEANS THAT IF ONE OF THE PARENTS HAS THE MUTATION, THERE IS A 50% CHANCE THAT THE CHILD WILL ALSO HAVE IT.

THE MUTATION THAT CAUSES **FFI** LEADS TO THE FORMATION OF AN ABNORMAL FORM OF THE PRION, WHICH FIRST ACCUMULATES IN A REGION OF THE BRAIN CALLED **THALAMUS**, AFFECTING ITS FUNCTIONING.

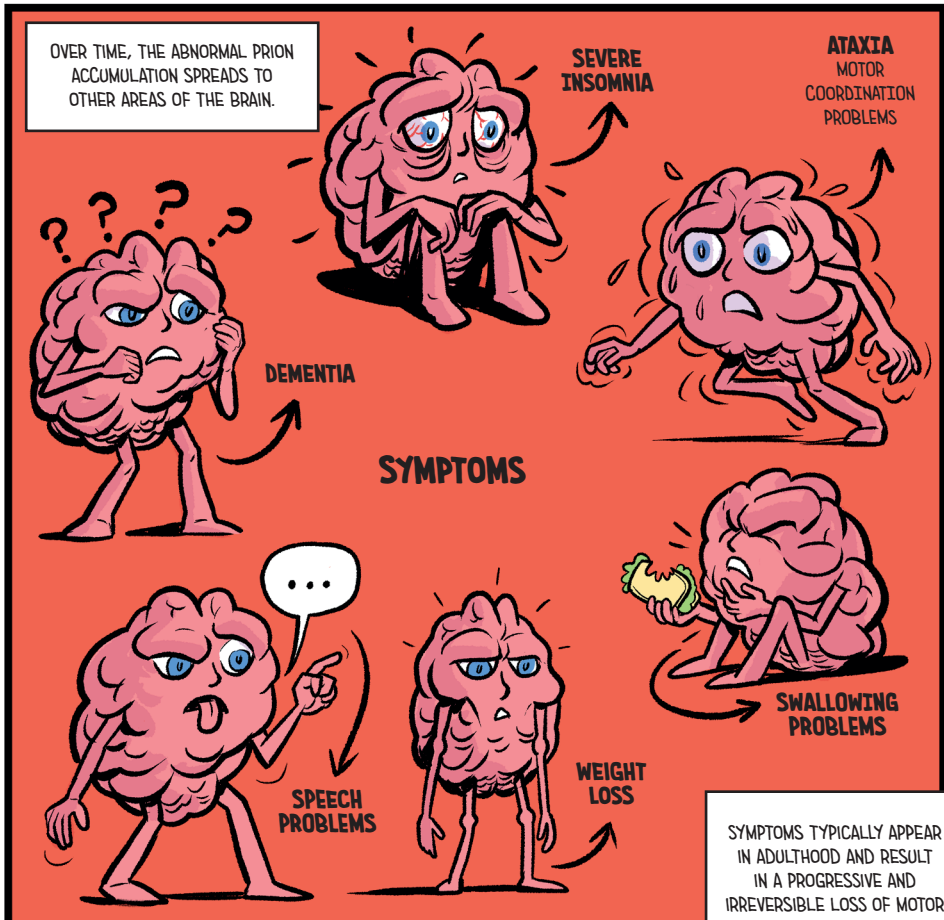


THE THALAMUS,AMONG OTHER FUNCTIONS, IS INVOLVED IN SLEEP REGULATION.

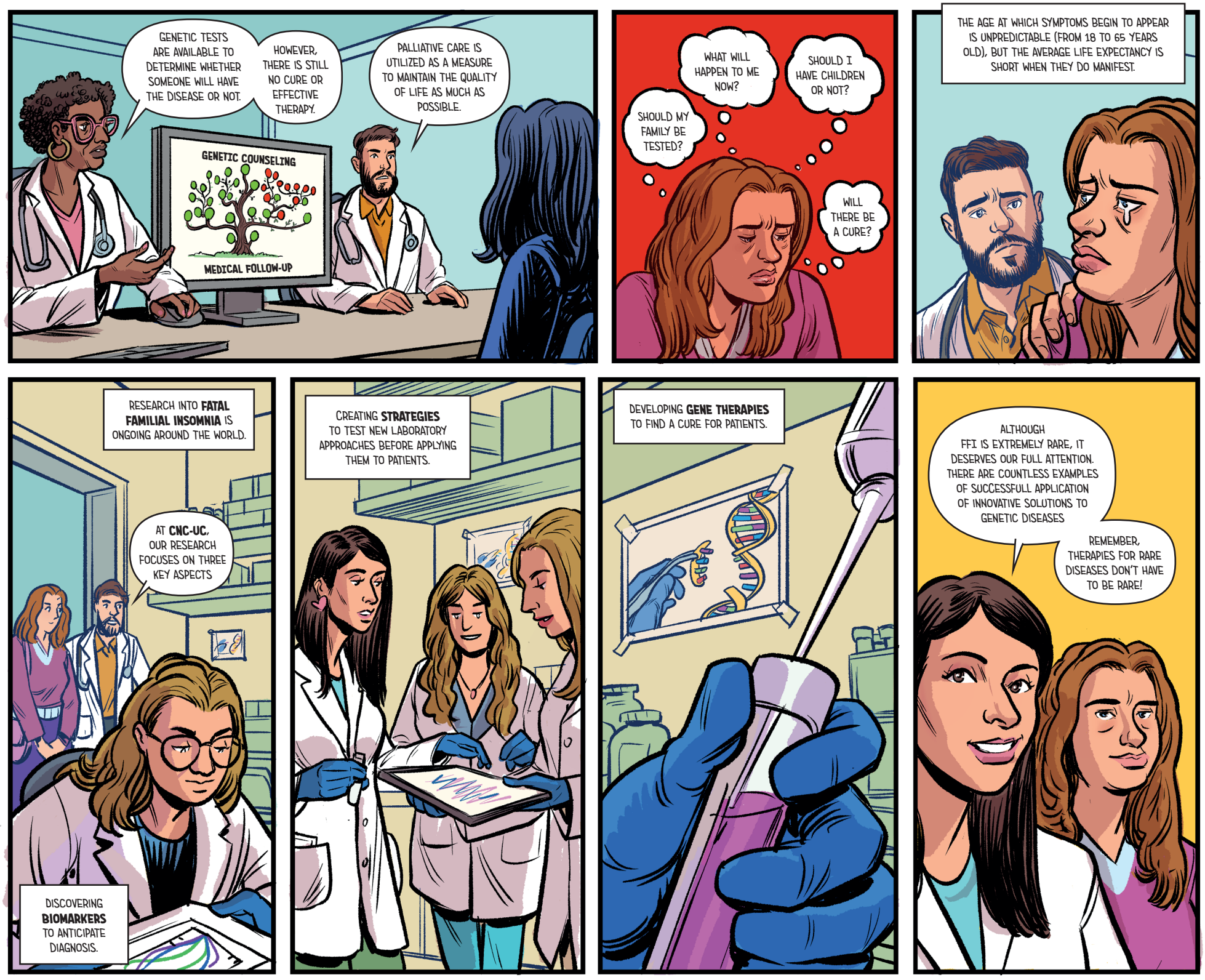


ITS DYSREGULATION CAUSES INSOMNIAS IN PATIENTS WITH FFI

OVER TIME, THE ABNORMAL PRION ACCUMULATION SPREADS TO OTHER AREAS OF THE BRAIN.



SYMPTOMS TYPICALLY APPEAR IN ADULTHOOD AND RESULT IN A PROGRESSIVE AND IRREVERSIBLE LOSS OF MOTOR AND INTELLECTUAL ABILITIES.



PROJECT DONE IN COLLABORATION AND CO-FINANCED BY:

ciibb CENTRE FOR INNOVATIVE BIOMEDICINE AND BIOTECHNOLOGY

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RARE DISEASE DAY®

March 15, 2024 World Sleep Day Sleep Equity for Global Health