Epilepsy

Epilepsy is a chronic neurological disorder in which clusters of nerve cells in the brain signal abnormally and cause seizures. There are many causes of epilepsy; therefore, diagnosis varies and can depend on medical history, blood tests, brain imaging tests, and electrical activity monitors such as electroencephalogram (EEG). Generally, a diagnosis requires that an individual experiences or has the propensity to experience more than one seizure. The effects of a seizure can be focal—meaning localized to a single region of the brain — or generalized, with electrical activity affecting the whole brain. To determine which type of seizure a patient has experienced, care providers observe symptoms and use the diagnostic tools outlined above to identify the origin. Epilepsies originate from many causes, so predicting and preventing seizures are very difficult and highlight the necessity for research to further understand this disease.

Epilepsies affect around 65 million people worldwide, making them one of the most prevalent neurological disorders.

Research Delivers Solutions

There are many causes for epilepsies, including immune system issues, tumors, metabolism, and genetic mutations. Identifying mutations in people with epilepsy can guide research for biomarkers; hundreds of affected genes have been reported. A gene called SCN, which makes proteins that help cells convey messages, has mutations linked to multiple types of epilepsies and intellectual disabilities. New DNA sequencing techniques have sped research progress about these mutations. One SCN gene, called SCN1A, has been linked to the development of generalized epilepsy with seizures, as well as to Dravet Syndrome. However, some mutations in another SCN gene, SCN2A, do not lead to seizures, showing that mutations may have many different impacts.

Other biomarkers are used to examine electrical activity to identify epilepsies. Since the timing of a seizure is difficult to predict, researchers have focused on electrical activity between seizures. High frequency oscillations (HFOs), or fast spikes in brain activity, have been identified using EEG recording. HFOs can be used to determine where a seizure may originate next.

Other research examines microRNAs (miRNAs) in epilepsy. MiRNAs are molecules that regulate the function of proteins. In epilepsy, sometimes miRNAs are abundant, and sometimes they are scarce, like with one called miR-15r-5p. This particular miRNA helps proteins clear cell waste. When impaired, the cell gets confused and increases activity that may lead to a seizure. Biomarkers such as these are helpful to learn the full picture of this disease and to cure it.

Incidence of Epilepsy-Related Deaths in the U.S.

Heightened reporting accuracy is now shedding light on the previously unknown toll of Sudden Unexpected Deaths in Epilepsy (SUDEP). A total of 1 in 1000 adults living with epilepsy today will succumb to SUDEP, highlighting the burden that patients face on a day to day basis.
Effect of Socioeconomic Status

The incidence of reported epilepsy is higher in low and middle income areas (6.68 per 1,000) compared to high income areas (5.49 per 1,000). In the U.S., people on Medicaid are more likely to have epilepsy. When reporting biases such as cultural stigmatization are accounted for, the WHO estimates that those in low or middle income countries make up 80% of people living with epilepsy.

Prevalence of Active Epilepsy Per 100,000 People in the U.S.

4. “What is Epilepsy?” CURE. 2018
8. “What is Epilepsy?” CURE. 2018
10. “A Novel SCN1A Mutation Associated with Generalized Epilepsy . . .” AJHG. 2001
13. “Potential Role of miRNAs as Theranostic Biomarkers of Epilepsy.” Molecular Therapy Nucleic Acids. 2018
15. “Epilepsy Associated Genes.” Seizure. 2017
19. “GBD Data Visualizations”. IHME. 2018
20. “Lets Talk SUDEP.” NIH. 2019

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