A biomarker is a substance or activity that can be measured and serves as a marker of a specific biological activity. Often, biomarkers are used in clinical studies to determine if drugs are having a desirable effect much earlier than one can observe changes in the disease itself. A biomarker may be a substance measured in the blood or urine, for example, or may be a measurement of a parameter such as blood pressure or brain activity. Many types of biomarkers exist, but they generally fall into one of three categories.

1. Biomarkers that give information on disease status or risk. In TSC, an electroencephalogram (EEG) can often be useful in the diagnosis of seizure types or in the assessment of risk of developing seizures.

2. Biomarkers that measure the effect of a drug or other intervention on a biological process. This type of biomarker does not give any information as to whether the disease is being affected, but it does show whether the drug is having its expected activity or not.

3. Biomarkers that measure the direct interaction of a drug with its target molecule, or receptor. This is a challenging measurement to make. Therefore, this type of biomarker is most commonly used in laboratory studies, although some compounds detected by positron emission tomography (PET) scanning can be used in clinical studies.

(over)
The development of biomarkers for use in TSC is important because biomarkers will enable us to know that a drug in a clinical study is being given at the correct dose to ensure that it binds to its intended receptor and has its expected biological response. If those things are not happening, there is no point in continuing the clinical study because the drug is not being adequately delivered. This means that research subjects will not be exposed to an investigational drug that has no chance of working. Similarly, disease biomarkers in TSC might help determine earlier in a clinical study whether a drug is impacting at least one aspect of the disease. The knowledge obtained from biomarkers enables a clinical study to be more scientifically rigorous and safer for research subjects.

**REAL LIFE IMPLICATIONS FOR SOMEONE WITH TSC**

Biomarkers have the potential to positively impact anyone diagnosed with TSC. For example, an EEG biomarker may in time tell us which infants will develop infantile spasms before the seizures actually begin. This knowledge could then help develop preventative treatments for those types of seizures.