# SCN9A is Mutated in Familial and Sporadic Febrile Convulsions

Published date: March 30, 2011

## Technology description

**Invention Summary**

Febrile seizures are the most common seizure disorder of infancy with a prevalence of 2-5% in European and North American infants. The impact of seizures brought on by elevated body temperature is considerable because individuals who experience them have a 2-7% incidence of developing afebrile seizures and epilepsy later in life. This technology implicates mutations in the sodium channel gene scn9a (nav1.7) in familial and sporadic cases of febrile convulsions and its associated severe complications such as idiopathic generalized epilepsy and intractable epilepsy. It is postulated that up to 20% of all febrile seizures are caused by mutations in this sodium channel gene. This will be beneficial both for diagnosis of patients at risk for febrile seizures, with potential preventative measures, as well as a possible drug target for antiepileptic drugs.

## Application area

An estimated 2.3 million Americans are afflicted by epilepsy, a neurological disorder that can lead to frequent seizures. Despite a long history of pharmacological research, there is no single drug that works for every seizure type and every individual suffering from the disorder. The market has annual sales of as much as $150 million.

## Advantages

Diagnose patients at risk for febrile seizures.
Use as a drug target for epilepsy.

## Institution

The University of Utah

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