# What is Dravet Syndrome?


**Dravet Syndrome** is a genetic disorder of Chromosome 2 usually affecting a gene called SCN1A, causing epilepsy and other neurological and developmental problems. Dravet Syndrome accounts for **1%** of the epilepsy population.


dravet.ca

The condition gets its name from the doctor who first recognised and described it - Dr Charlotte Dravet.

## What are the features?

Seizures starting in infancy
Multiple seizure types
Developmental delays emerging in early childhood
Motor and behavioural disorders increasing with age

## What are the Comorbidities?

Autonomic dysfunction
Autistic traits
Attention deficit and hyperactivity
Ataxia (unsteadiness)
Orthopedic conditions
Sleep disturbances

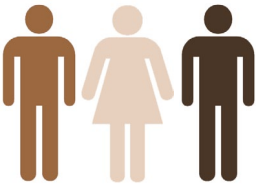
## How common is it?

**1:30,000** people worldwide are born with **Dravet Syndrome**

*About* **1000** in Canada will have Dravet Syndrome but many will be ***undiagnosed***.

## Who does it affect?

It affects males and females equally.



It is present in all nationalities, races and ethnic groups.

## Why did it happen?

Genetic disorders occur at the **point of conception** and nothing the mother or father did could have caused it or prevented it from happening.

**80%** of people with Dravet Syndrome have an SCN1A mutation. Of these, 95% have de novo mutations and **5%** have inherited mutations. Mutations of SCN9A, SCN2B, PCDH19 and GABRG2 genes are also seen in Dravet Syndrome.

**5%** *cases are inherited*

**95%** *cases are completely random*

Scientists are researching new medications and treatments, and gaining greater understanding of the genetics of ***Dravet syndrome***.

## Can it be treated?

Dravet syndrome is a lifelong disorder. Anti-epilepsy medications and special diets can reduce the seizures. Speech therapy, physiotherapy and occupational therapy can alleviate some of the other symptoms.

**1978**
Severe Myoclonic Epilepsy of Infancy first described by Dr Charlotte Dravet

**1982**
42 cases of SMEI were presented at the XIII$^{th}$ Epilepsy International Symposium

**1987**
SMEB Borderline forms described

**1989**
SMEI classified by ILAE

**2000**
Stiripentol tested for treatment of SMEI

**2001**
SCN1A mutations discovered in Belgian patients with SMEI and SMEI formally renamed as Dravet Syndrome

**2006**
Micro deletions in SCN1A discovered, accounting for 2-3% of Dravet cases and Mosaic SCN1A mutations identified in parents having more than one child with Dravet Sydrome

**2008**
Screening test identified for early diagnosis of Dravet Syndrome

**2009**
The SCN9a gene is identified as a possible modifier of SCN1a gene in Dravet Syndrome

**2013**
Mutations in other genes discovered in rare cases of Dravet Syndrome