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.

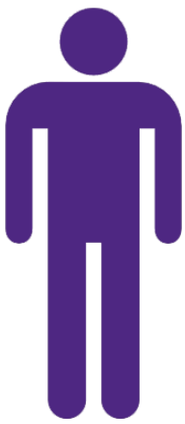
1978

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

dravet.ca

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 XIIIth 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

www.dravet.ca