

# Dravet Syndrome Patient Journey



## 1. FIRST SYMPTOM

**Timeline:** 1/4 month -12 months

### Clinical Presentation / Symptoms

Generalised seizure, possibly fever-related, possible long duration

#### Identify Patient Needs

- Parents need reassurance (the first szr may have been traumatic for parents)
- Parents need basic information about epilepsy and seizures
- They need to know what to do if it should happen again.
- If the seizure was vaccination-related, parents need to know how to proceed with further vaccinations in a safe way

#### Ideal Outcome / Support

- The worries of the parents are taken seriously and reassurance is offered.
- Family receives instructions on how to handle a seizure if it should happen again; description of emergency meds, when to go to the hospital
- Vaccination plan.

## 2. DIAGNOSIS

**Timeline:** 3 months- 8 year (normally 2 years)

### Clinical Presentation / Symptoms

Development is different from child to child.  
The disease affects the entire life of the person  
People with Dravet won't be ever autonomous  
Child presents more seizures and of different types: absence, partial or generalised tonic clonic/ myoclonic seizures, with or without trigger factor.  
Possibly fever-related, possible long duration, cluster of seizures and status epilepticus  
Overall development can be normal until age of 2 or even 6-7.  
Comorbidities like mental retardation, speech, motor, behavior and orthopedic problems may occur.

#### Identify Patient Needs

- Parents need a proper explanation of the diagnosis and prognosis with psychological support
- Parents need to be offered a genetic test
- Parents need to understand that development change from child to child
- Parents want to be sure about diagnosis (GEFS+ or Dravet?)
- They need instructions on how to deal with/treat seizures and what other non-pharmacological therapies their child may require
- Parents need to know whether there are clinical trials in Dravet going on in their countries and whether their child is eligible for them.
- They need to know how to keep their child safe (harm from falls; seizure detection at night, dealing with fever...)
- Parents need to know what social help is available from government.

#### Ideal Outcome / Support

- Family is explained that a prognosis is difficult to give.
- Genetic counseling; explaining the cause and chances of reoccurrence.
- Professional support is offered to cope with diagnosis and family is referred to parents group/Dravet organization.
- Parents receive clear instructions, emergency protocol, explanation of risks and how to minimize them.
- Parents receive clear information about putative clinical trials in Dravet where their child is eligible, with deep informing about benefits and risks of participating in them
- Family is explained that education and rehabilitation are extremely important for child's development and should be very closely monitored
- Family is given a document that summarizes the social support that may be required and offered for the disease

## 3. TREATMENT

**Timeline:** All along the disease

### Clinical Presentation / Symptoms

- Dravet syndrome is drug resistant – AEDs only work for a while if at all. Treatment should aim at best possible seizure control and less possible side effects.
- Special attention to triggers and how to prevent seizures.
- Status epilepticus should be prevented as much as possible and may need aggressive treatment.
- Paramedic treatment may be needed to optimize physical and mental development

#### Identify Patient Needs

- Parents need support and respite care from professionals.
- They need information on the prescribed medication, side effects, how to monitor long-term side effects
- Advise on how to deal with triggers, illness, photo- and patterns sensitivity, etc.
- They need help/medical advise concerning the non-epileptic symptoms.
- School advise; advise for respite care
- Acces to trials/new treatment options

#### Ideal Outcome / Support

- Regular counseling with professionals is offered.
- Up to date information is available for parents at any time.
- Parents are informed about medication side effects and the timing for control blood test
- Optimal treatment of non-epileptic symptoms is given.
- Parents receive help finding a school/daycare and respite care.
- Centres involve Dravet patients in trials
- Family nurse is offered to educate family to manage seizures

## 4. FOLLOW UP

**Timeline :** 2-16 years

### Clinical Presentation / Symptoms

Other problems may occur like mental retardation, motor, behavior and orthopedic problems, parkinsonism, etc.

#### Identify Patient Needs

- Parents need evidence-based advise and expertise when it comes to these additional symptoms.
- Parents need emotional support.
- Evidence of enabling needs (psychomotricity, speech therapy, postural rehabilitation, behavioural therapy)

#### Ideal Outcome / Support

- Monitoring of these problems and offer treatment where possible;
- Developing guidelines
- Developing standards for quality of life adult patients
- Availability of home care and/or institutional care of excellent quality
- Definition of the rehabilitation pathway (psychomotricity, speech therapy, postural rehabilitation, behavioural therapy)

## 5. FOLLOW UP ADULT

**Timeline:** 16 years and up

### Clinical Presentation / Symptoms

Transition to adulthood.  
Convulsive seizures are still present, whilst hemiclonic seizures become less common, and absence seizures and myoclonic seizures tend to disappear. Convulsive status epilepticus are less frequent.  
Temperature-sensitivity and, in general, reflex seizures usually decrease.  
Comorbidities continue.  
In some healthcare centers, lack of coordination between pediatrics and adult services may entail an insufficient support for patient and family.

#### Identify Patient Needs

- Parents need advise and support in transition to adulthood
- Adult management
- Monitoring of seizures and development, new therapeutic opportunities, neuropsychological behavioural situation

#### Ideal Outcome / Support

- A process for transition from pediatric to adult physician should be put in place
- Definition of a rehabilitation path for maintenance
- Occupational therapy
- Taking charge of possible aggravation of the various motor, swallowing, behavioral, social and cognitive problems.