DRAVET Syndrome UK

Hope for families with life-limiting epilepsy

Intractable Epilepsy: **Could it be Dravet?**

A DIAGNOSIS GUIDE FOR HEALTH PROFESSIONALS

- **Dravet Syndrome** is rare, life-long and life-limiting neurological condition, encompassing treatment-resistant epilepsy, intellectual disability and a spectrum of associated comorbidities.
- The condition occurs in one in around every 15,000 live births in the UK[1].
- Dravet Syndrome typically presents in during the first year of life in an otherwise healthy child with prolonged, febrile and afebrile, focal clonic (usually hemiclonic), or generalized clonic seizures; other seizure types appear over time and continue throughout life.
- From the second year of life, development slows, and cognitive, motor and behavioural difficulties become apparent.
- Comorbidities associated with Dravet Syndrome include autism, ADHD, challenging behaviours and difficulties with speech, mobility, eating and sleep.
- There is an increased risk of SUDEP with Dravet Syndrome (15 times higher than other childhood-onset epilepsies).
- Dravet Syndrome is a clinical diagnosis, supported by the identification of variants in the SCN1A gene (found in approximately 85% of cases).
- **Finding the right treatment plan is critical for children and adults with Dravet Syndrome.**

Diagnosing Dravet syndrome

Dravet Syndrome is under-diagnosed. Being aware of the typical features of the syndrome and intervening early to request confirmatory genetic testing is critical to enable patients and their families to access appropriate treatment and support.

Genetic testing is normally a simple blood test, available free of charge on the NHS in the UK. Testing can be carried out by the Epilepsy Genetic Service in Glasgow and in other centres in the UK through local clinical genetics departments.

Even if the SCN1A test comes back negative, Dravet Syndrome should not be ruled out if the patient fits the diagnosis clinically. Further advice can be sought from the Glasgow Epilepsy Centre.

Why diagnosing Dravet Syndrome matters

Previously known as severe myoclonic epilepsy in infancy (SMEI), Dravet Syndrome has a devastating impact on patients and family members, but it can be effectively managed with appropriate treatment and support. Diagnosis at any age – including late adulthood – and getting the right treatment in place improves health and quality of life and can help reduce the risk of SUDEP. Healthcare professionals at the frontline of treating seizures and other symptoms of Dravet Syndrome have a key role to play in improving the diagnosis and management.

"Certain types of gene tests, with the clinical picture, allow doctors to be quite confident of a Dravet Syndrome diagnosis. Genetic testing allows us to think about what the right treatments might be for someone, at an early stage"

– Professor Sameer Zuberi, Consultant Paediatric Neurologist and member of the Dravet Syndrome UK Medical Advisory Board

For more information, contact us at www.dravet.org.uk

Living with Dravet Syndrome

Symptom patterns change during a lifetime... for intractable epilepsy patients don't stop asking

Could it be Dravet?

Key characteristics of Dravet Syndrome include:

Age of onset is typically between 3-9 months

- Average age of onset is 6 months, Rarely, some children can present as early as one month old or as late as 20 months. Previously well, developmentally normal.

Initial seizure types include

- Febrile seizure, hemiclonic, generalised clonic/tonic, status epilepticus (>30 minutes); triggered by high temperature or water.

Normal development until 1 year

- Development slows, walk a little later (e.g. 17–18 months), unsteady for longer; language slower to acquire; sometimes regress.

EEG normal in first 1–2 years then

- Generalised spike wave, polyspike, multifocal discharges, may be photosensitive.

In the second year myoclonic and atypical absences may appear

- Other seizure types emerging from the second year include: myoclonic seizures, focal seizures with impairment of awareness (complex partial/ dyscognitive seizures), atypical absence seizures, atonic seizures.

After 2–5 years, episodes of status settle

- Convulsive status much less frequent but can still occur; TCS evolve to focal seizures with impairment of awareness and then primarily nocturnal generalised clonic/tonic seizures.
- Myclonia more prominent as older, may fluctuate or become more prolonged.

Long-term development

- Physiological ataxia does not improve in normal time course, pyramidal signs develop.
- Unusual crouch gait appears after 13 years (bony malalignment with significant functional impairment).
- Variable and autistic features in some, many children have extremely challenging behaviour.
- Intellectual disability (severe 50%, moderate 25%, mild 25%), children and adults are long-term dependent, requiring 24/7 supervision and care (often 2:1).

Adults with intellectual disability and epilepsy

- **Don't rule out Dravet!** The right diagnosis and treatment can still reduce seizure and disease burden even in intractable adult patients[2].

References

1. Symonds et al. *Brain* 2021
2. Catarino et al. *Brain* 2011

Five things you can do if you suspect Dravet Syndrome

1. Check for characteristics of Dravet Syndrome

Dravet Syndrome is a clinical diagnosis, diagnosed from reported characteristics, outlined in this leaflet. It's important to check for these as soon as you suspect Dravet Syndrome to enable early and appropriate treatment.

2. Refer for genetic testing

Genetic testing (with counselling) has benefits at any age. The right diagnosis and treatment can reduce seizure and disease burden even in intractable adult patients. It is worth retesting someone for Dravet Syndrome if they received a negative test more than 5 years ago, as the accuracy of testing has improved. If negative, don't rule out Dravet Syndrome if the patient fits the clinical diagnosis.

3. Avoid prescribing sodium blocking medication

Exercise caution with carbamazepine, gabapentin, lamotrigine, oxcarbazepine, phenytoin, pregabalin, tiagabine, and vigabatrin. These medications can make seizures worse. Prolonged use can increase the risk of poorer intellectual outcomes.

4. Put emergency protocols in place and discuss SUDEP with caregivers

Those with Dravet Syndrome are at increased risk of premature death due to the frequency of status epilepticus and high rates of SUDEP. It's vital to work with caregivers to implement emergency protocols. Discussing SUDEP is also important; getting the right treatment in place and an effective seizure control plan can help reduce the risks of SUDEP.

5. Signpost to Dravet Syndrome UK

Dravet Syndrome UK is a registered charity dedicated to improving the lives of those affected by this devastating condition. We provide emotional, practical, and financial support for families affected by Dravet Syndrome, as well as educating and funding research. When families register with us, they will receive a free comprehensive guide to living with Dravet Syndrome.

Interdisciplinary management

Dravet Syndrome causes children cognitive developmental impairment – often severe. Multidisciplinary management is required in addition to drug treatment tailored to the specific needs of each patient. For example, in children, expressive and receptive language is often impaired; early intervention with speech therapy optimises potential. Ensure developmental assessments begin as early as possible and are repeated regularly.

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