



Spanish and European Dravet syndrome population estimates

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Why we need population estimates

The prevalence of Dravet syndrome is estimated to be of 1 in 20,000 to 1 in 40,000 people. According to these figures, a country like Spain should have around 1,500 Dravet patients.

We estimate that 60-70% of the Spanish patients with a Dravet syndrome diagnosis are affiliated with the Dravet Syndrome Foundation. With a current number of 124 affiliates, we estimate that only 200-250 of patients have a diagnosis of Dravet in Spain, **less than 20% of the estimated actual Dravet population.**

When we created the Dravet Syndrome Foundation in Spain in 2011 these numbers were much lower, with less than 5-10% of the estimated Spanish Dravet population having a correct diagnosis. The availability of a free genetic test for Dravet syndrome, developed and sponsored by the Foundation, has led to a 5% grow in diagnosis per year. We estimate this percentage will continue to grow at a similar rate.

Only the UK in Europe has a percentage of diagnosis over 20%. We estimate only 2-3 additional European countries reach percentages of 10-20%, with most having a **diagnostic rate below 10 or even 5%.** The main contributor to this moderate success is the availability of a public genetic test. As an example, 85% of the Spanish Dravet patients younger than 4 years old (11/13) have joined the Foundation during the last year following their genetic diagnosis. Therefore availability of a free (public or privately-funded) genetic test has a major role in identifying more and younger patients.

We estimate the diagnostic rate for Dravet in Europe will grow at a similar rate to that of Spain, approximately 5% per year.

While several European countries have Dravet patient registries at different stages of development, much of these data are not yet available. This immature patient registry stage, combined with the elevated rate of underdiagnosis, mean actual population data numbers are elusive. In the mean time, patient surveys can help us get a better picture of the European Dravet population.

Basic principles of data collection

- **DSF Spain registry (n=127 patients)**
- **European survey*(Europe n=278, Spain n=61)**
Electronic survey developed and administered by DSF Spain, and distributed in collaboration with the Dravet Syndrome European Federation. Duration: 1 month (ended June 2014).
- **Facebook survey (Spain n=63)**
Administered in July 2014 through a private Spanish Dravet families group to get further clarification on seizure freedom duration.

***Note:** the European survey data is part of a larger unpublished study that will be published in a peer-reviewed article.

FIGURES

PATIENT AGE STRATIFICATION

DSF Spanish Registry

July 2014

124 Spain, 3 Portugal

| DSF Spain Registry | | |
|--------------------|-----|--------|
| n : | 127 | 100% |
| > 18 years old | 21 | 16.50% |
| 14-18 years old | 9 | 7% |
| 8-14 years old | 32 | 25% |
| 4-8 years old | 49 | 38.60% |
| < 4 | 13 | 12.60% |
| no date of birth | 3 | 2.40% |

European survey (electronic) created and administered by DSF Spain

June 2014

| Europe (including Spain) | | |
|--------------------------|-----|------|
| n | 278 | 100% |
| > 18 years old | 43 | 15% |
| > 4 years old | 239 | 86% |
| 2-18 years | 234 | 84% |
| Spain only | | |
| n | 61 | 100% |
| > 18 years old | 9 | 15% |
| > 4 years old | 56 | 92% |
| 2-18 years | 50 | 82% |

MORTALITY

DSF Spanish Registry

N= 127

124 Spain, 3 Portugal

1 death out of 127 patients in a 5-year follow up (0,78%).

| Follow up time | | |
|-------------------|-----------|-------------|
| average | 31 months | 1-55 months |
| more than 4 years | | 22 |
| more than 3 years | | 31 |
| more than 2 years | | 36 |
| more tha 1 year | | 21 |
| less than 1 year | | 20 |

DRUG RESISTANCE

European survey (electronic) created and administered by DSF Spain

June 2014

Asked to report "average number of seizures per month during the last 6 months".

| Europe (including Spain) | | |
|---------------------------------|-----|------|
| n | 278 | 100% |
| > 4 tonic-clonic seizures | 102 | 37% |
| ≥ 4 tonic-clonic seizures | 125 | 45% |
| 0 tonic-clonic seizures | 64 | 23% |
| 0 seizure (any type of seizure) | 26 | 9% |
| Spain only | | |
| n | 61 | 100% |
| > 4 tonic-clonic seizures | 15 | 25% |
| ≥ 4 tonic-clonic seizures | 21 | 34% |
| 0 tonic-clonic seizures | 14 | 23% |
| 0 seizure (any type of seizure) | 7 | 11% |

Facebook survey by DSF Spain

July 2014

Asked to report "duration of seizure freedom".

| Spain | | |
|--|----|--------|
| n | 63 | 100% |
| Patients seizure free more one year | 1 | 1.58% |
| Patients seizure free 6-12 months | 6 | 9.52% |
| Patients seizure free 3-6 months | 3 | 4.76% |
| Patients seizure free less than 3 months | 53 | 84.12% |

Comments on seizure freedom duration:

We estimate that all of the patients who were seizure free for more than 6 months could be classified as (formerly known) "atypical Dravet" or Generalized Epilepsy with Febrile seizure plus (GEFS+)

We estimate that some (2/3) of the patients reporting seizure freedom for more than 3 months are also those formerly known as "atypical Dravet".