INTRODUCTION

- Dravet syndrome is a rare, refractory epilepsy typically involving multiple co-morbidities including motor, cognitive and behavioural impairments.1
- The wide scope of co-morbidities combined with frequent, treatment refractory seizures can be expected to result in a high impact on patients and caregivers affecting all aspects of their lives, with many aspects not reported.2

OBJECTIVES

- The Dravet syndrome caregiver survey (DISCUSS) sought to determine the social and financial impact of Dravet syndrome on patients and their caregivers and explore healthcare resource utilisation associated with its current management.

METHODS

DISCUSS was an online, anonymous survey conducted from 23 June – 4 August 2016 comprising 195 questions about:
- caregiver household (demographic information), health status of the patient,
- current and past treatment
- experiences of diagnosis
- quality of life, including the standardised EQ-5D-5L instrument.3
- social and financial caregiver impact and
- health services use.

Only fully completed surveys4 were accepted for submission. Gate questions ensured negative responses were not probed further.

Participants were recruited through email invitations to approximately 1,000 members of patient advocacy groups (PAGs) of different countries associated with the Dravet Syndrome European Federation (DSEF) as well as through internet based sources (Facebook and Twitter).

Survey versions were available in English, Spanish, Latin American Spanish, Portuguese, Brazilian Portuguese, French, German, Italian and Polish (translated from English by specialist translators), and Croatian, Dutch and Romanian (translated by local language PAG members). All language versions were tested by local speakers before survey launch.

For descriptive statistics, patient ages were grouped as infant (<2 years), pre-school (2-5 years, inclusive), middle childhood (6-11 years, inclusive), adolescent (12-17 years, inclusive) and adult (18 years and older). Statistical significance (p<0.05) of differences between frequencies was determined using a two-proportion test for two proportions with a 95% confidence interval on the difference between the proportions using XLSTAT in Excel.

1) with the exception of one question about the cost of non-pharmaceutical treatments

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REFERENCES


RESULTS

Demographics

- 584 fully completed surveys were submitted (mothers 86%, fathers 12.3% and other caregivers 1.6%) of which the majority (92%) lived in Europe.
- 20% of caregivers lived in a single and 77.6% in a two-parent household with more than one adult.
- The mean patient age was 10 years (median, 9 years).
- The middle childhood group was the largest, most others comprised 25%-15% and infants 5%-6% of total submissions (Figure 1).

Disease severity

- Less than 10% of patients reported no seizures in the previous 3 months. Whereas only 3% of infants were seizure-free, this proportion increased slightly by age, to 14% of adolescents (p<0.05 compared to infants) and 11% of adults (p<0.05 compared to infants).

Comorbidities

- Patients older than 5 years had on average 4.3 (IQR 1.7) of the 6 surveyed diagnoses or co-morbidities.
- Nearly all (91%) patients older than five years reported at least one other co-morbidity or impairment in addition to seizures (Figure 3).

Disease management

- Reported treatment patterns were broadly consistent across all age groups, and in line with clinical guidance. Valproic acid, typically used as first-line treatment, is currently taken by 76% of patients. Phenytoin, stiripentol, topiramate or the ketogenic diet are currently taken by 53%, 47%, 34% and 6.5%, respectively.

Patient quality of life

- The mean EQ-5D-5L index value for all patients two years or older (completed by caregivers in proxy) was 0.42 (SD 0.29) and ranged from less than zero to one. No large difference across age groups was observed.

Experience of diagnosis

- While the time to diagnosis was very long (>4 years) for the majority (83%) of adult patients, diagnosis was not diagnosed at first instance, less than 20% of middle childhood patients waited that long (Figure 4).

Impact on the family

- Most (80%) caregivers reported that caring for a child with Dravet syndrome had influenced their career choices. Nearly a third (30%) were unemployed, many (81%) giving up their job because of their caregiver responsibilities.

Sources of support

- Caregivers reported that sources of support are mostly the family and sometimes the employer, with relatively few (23%) taking up support from social services. While personal costs are partially or fully covered for most (80%) families, these may be high for those paying out of their own pocket.

CONCLUSIONS

- Totalling 584 full submissions, this survey captured about 15% of the Dravet syndrome patient population under the age of 18 in the European Union Five (France, Germany, Italy, Spain and the United Kingdom) making it the largest and most comprehensive of its kind carried out to date.30
- Families caring for a Dravet syndrome must manage multiple impairments in addition to epilepsy symptoms.
- This survey highlights diagnostic delays and considerable social and financial impacts on families.

(30) assuming a prevalence of 1/45700 in a population of 340 million of which about 20% are under the age of 18.