DISCUSS Dravet – Key socioeconomic findings from a large multinational survey of Dravet syndrome caregivers

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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 carried out from 23 June -4 August 2016 comprising 150 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 surveys(a) 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-tailed z-test for two proportions with a 95% confidence interval on the difference between the proportions using XLSTAT in Excel.

(a) with the exception of one question about the cost of non-pharmacological treatments

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 household with more than 1 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 6% of total submissions (Figure 1).

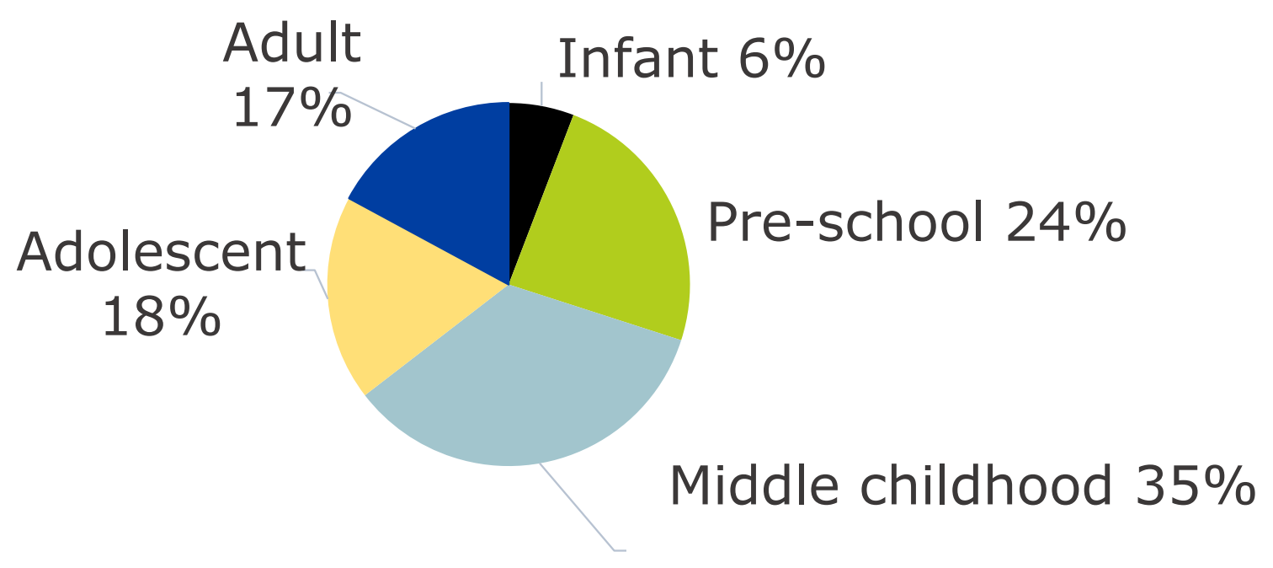

Figure 1. Age distribution of patients

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 steadily by age, to 14% of adolescents ($p<0.05$ compared to infants) and 11% of adults (Figure 2)

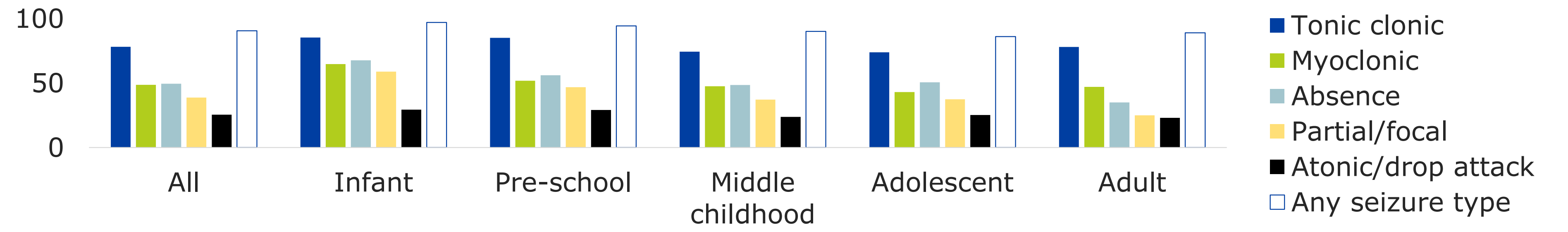

Figure 2. Percentage of patients experiencing at least one seizure in the previous 3 months.

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Disease severity (continued)

- Half of patients required at least one emergency admission and 46% at least 1 ambulance call in the past 12 months. Younger patients reported more emergency admissions than older patients (94% infants and 76% pre-school patients compared to 30% adolescents and 28% adults (both $p<0.001$). A similar trend was observed for ambulance calls.

Comorbidities

- Patients older than 5 years had on average 4 (3.7 ±1.2) of the 6 surveyed impairments 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)

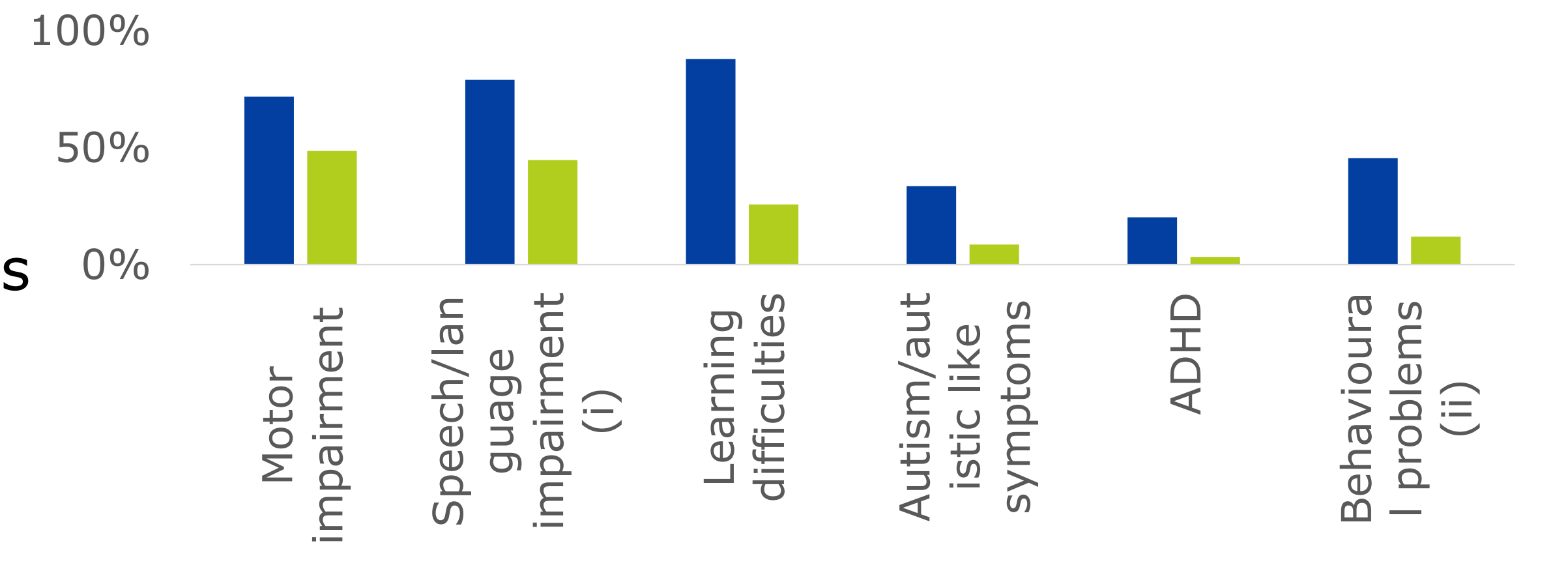

Figure 3. Percentage of patients reported having a co-morbidity (blue bars) and in therapy for the co-morbidity (green bars). Abbreviation: ADHD, attention deficit hyperactivity disorder; (i) includes patients that do not talk at all (ii) excluding autism and ADHD

Disease management

- Reported treatment patterns were broadly consistent across age groups and in line with clinical guidance. Valproic acid, typically used as first-line treatment[4] is currently taken by 76% of patients. Clobazam, stiripentol, topiramate or the ketogenic diet are currently taken by 53%, 47%, 34% and 6.5%, respectively.

Patient quality of life

- The mean EQ5D-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 not diagnosed at first instance, less than 20% of middle childhood patients waited that long (Figure 4).

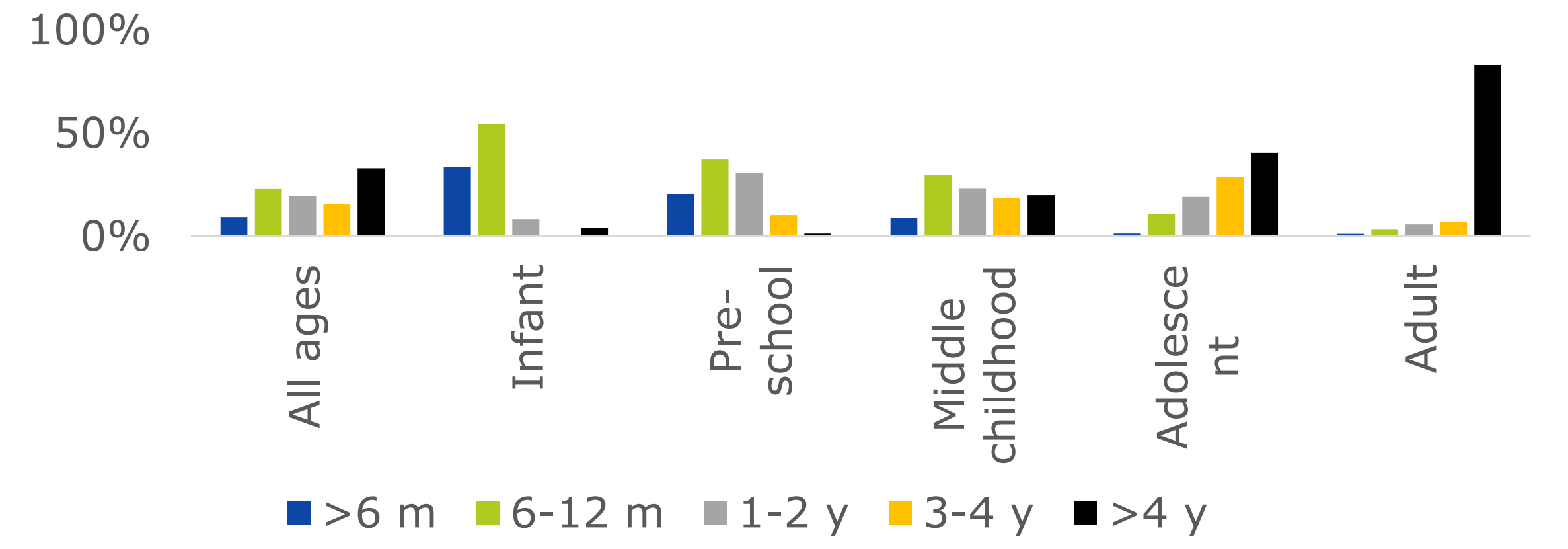

Figure 4. Time to diagnosis for patients not diagnosed at first instance (expressed as a percentage of patients waiting the indicated time)

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.
- Most (77%) caregivers reported having less than one hour per day completely for themselves, for example for relaxing or for social activities. This was true across all age groups, dropping only slightly for adult patients (Figure 5).

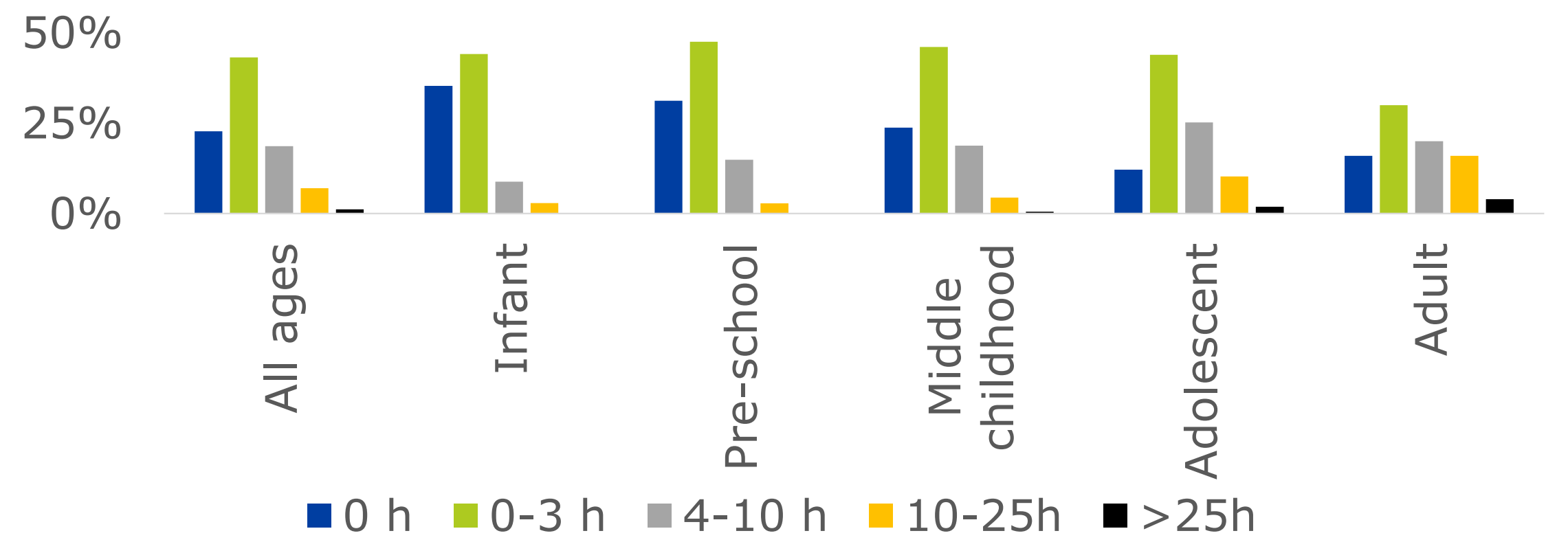

Figure 5. Time per week that caregivers have completely to themselves (for example for relaxing or social activities)

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.(b)
- Families caring for a member with Dravet syndrome must manage multiple impairments in addition to epilepsy symptoms.
- This survey highlights diagnostic delays and considerable social and financial impacts on families.

(b) assuming a prevalence of 1/45700[5] in a population of 340 million[6] of which about 20% are under the age of 18.[7]

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