Burden of Disease and Cost of Illness of Spinal Muscular Atrophy in Portugal

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1 - Introduction:

– Spinal muscular atrophy (SMA) comprises a diverse group of genetic disorders that causes a progressive loss of motor neurons. Autosomal recessive proximal SMA or 5q-SMA is the most common form, accounting for up to 95% of cases.[1]

– SMA is a heterogeneous genetic disease with different degrees of severity.[2] It ranges from very compromised neonates (type 0) to adults with minimal manifestations (type IV).

– SMA types 0 and IV are uncommon –SMA Types I to III are the representative phenotypes seen within the general population with SMA.[3]

– This study aimed to estimate the cost and disease burden associated with SMA types I, II and III, in mainland Portugal during 2019.

2 - Methods

– The burden of disease and cost of illness were estimated using a prevalence approach.

– SMA prevalence was estimated assuming the prevalence rate reported by Kekou et al. [4] – 1.5 per 100.000 inhabitants.

– The main sources of information were the hospital morbidity database (base de dados de morbilidade hospitalar, BDMH) and other official sources concerning planned activity; statistics on drugs consumption [5]; and an expert panel regarding prevalence distribution per phenotype, outpatient resource consumption and labor force participation rate (the panel was composed by 4 neuropediatricians and 3 neurologists). Unit costs were based on official data.

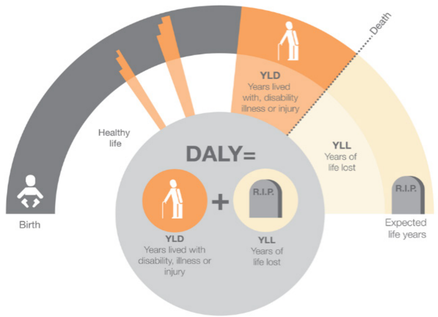
– Disease burden was measured as DALY (disability adjusted life years), i.e., the sum of Years of Life Lost (YLL) due to premature mortality and Years Lived with Disability (YLD) (Figure 1).[6]

– YLL were calculated from the number of SMA related deaths and the respective standard life expectancy at the age of death estimated by the Global Burden of Disease (GBD) Study 2017.[7]

– YLD were computed considering the number of cases of SMA and a disability weight based on the GBD Study 2019 and other sources (Table 1).[4,8]

– Costs of illness were estimated using a societal perspective and included medical and non-medical direct costs as well as patients' productivity losses estimated using the human capital approach.

Figure 1. DALY diagram.



Source: https://publichealthmatters.blog.gov.uk/2015/09/15/gbd-compare-a-new-data-tool-for-professionals/

Table 1. Mean disability weight per patient with SMA, by type.

SMA type	Disability weight
Type I	0.602
Type II	0.500
Type III children	0.046
Type III adults	0.327

Source: Authors' estimation based on experts' responses (adapted from Global Burden of Disease Study [2019]).

3 - Results

– Prevalence of SMA was estimated at 147 patients (Table 2).

Table 2. Estimated SMA prevalence (types I-III), year 2019.

SMA type	Number of patients
Type I	18
Type II	46
Type III	83
Total	**147**

Source: Authors' estimation.

– Six deaths were attributed to SMA, which generated a loss of 345 years of life due to premature death, 75% of which are related to SMA type II and III.

– Mean disability weights by type (Table 1) were applied to the respective estimated SMA prevalence, resulting in a total loss of 58.4 YLD. Most of this loss is attributed to adults with SMA type III (41%), followed by SMA type II (39%).

– Consequently, it is estimated that 403 DALY were lost (86% due to premature death; 14% due to disability) (Figure 2). This equates to a loss of 2.7 DALY per patient with SMA (5.4 DALY per patient with type I and 2.4 DALY per patient with type II or III).

– Direct costs amount to 16.6 million euro (15.0 million euro in medical costs; 1.6 million euro in non-medical costs). Indirect costs (related to productivity) were estimated at 194 thousand euro (Table 3).

– Total costs were 16.8 million euro, representing an average annual cost per patient of 114 thousand euro (395 thousand euro per type I patient; 93 thousand euro per type II patient; and 65 thousand euro per type III patient).

Figure 2. DALY attributable to SMA in mainland Portugal in 2019.

Table 3. Total costs attributable to SMA (2019).

	Average annual cost per patient	Total cost
Direct medical costs	**102,341 €**	**15,044,120 €**
Hospitalization	3,300 €	485,077 €
Ambulatory	5,148 €	756,745€
Pharmacologic treatment	93,893 €	13,802,297 €
Direct non-medical costs	**10,663 €**	**1,567,475 €**
Support devices	626 €	92,066 €
Home adaptations	198 €	29,051 €
Social support	3,167 €	465,497 €
Informal caregiver	5,978 €	878,774 €
Transportation	694 €	102,087 €
Productivity costs	1,325 €	194,736 €
Total	**114,329€**	**16,806,331 €**

4 - Conclusions:

SMA has a relevant socioeconomic impact, despite its low prevalence, namely at an individual level and on family/caregivers, stressing that all stakeholders need to be involved in the definition of national health policies on SMA approach.

5 - Acknowledgements:

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6 - References:

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