Epidemiology and therapeutic outcomes of patients with Spinal Muscular Atrophy
results from a 12-year real-world study based on the French National Healthcare database (SNDS)

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Poster 114P
WMS 2024 - 8/12 oct 2024
Prague, Czech Republic

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Background

Spinal Muscular Atrophy (SMA) is a genetic disease affecting the central nervous system, peripheral nervous system, and voluntary muscle movement.

In the past years, 3 innovative therapies have been launched in France, improving the management of patients with SMA: nusinersen with an access via nominative early access program since mid-2016, onasemnogene abeparvovec since 2019 and risdiplam since 2020.

Objective

In the context of these dramatic changes to the SMA treatment landscape, this study was designed to provide real-world data regarding the epidemiology, the management of therapeutic options, the healthcare pathways, the impact on overall survival, and the costs (not included in this presentation).

Methods

Data sources

SNDS

DCIR
Outpatient reimbursement data

PMSI
Hospital reimbursement data

The SNDS covers about 99% of the population living in France

Study population and period

Index date : first hospital stay or Long Term Disease status (ALD) or innovative drug initiation

01/01/2011 — 31/12/2022

Patients were included and followed up over 12 years.

***Proxys used to classify in SMA subtypes**

SMA1*
(G12.0# or G12.1#) OR (G12.8# or G12.9# with at least one administration of one of the 3 innovative therapies) and age < 12 months at index date

SMA2*
(G12.0# or G12.1#) OR (G12.8# or G12.9# with at least one administration of one of the 3 innovative therapies) and 12 ≤ age < 27 months at index date

SMA3*
With at least one administration of one of the 3 innovative therapies AND not treated by riluzole (used only for amyotrophic lateral sclerosis
G12.0# AND → 24 years old at index date

#ICD-10 codes were searched in hospitalizations and Long Term Diseases diagnoses

G12.0 Infantile spinal muscular atrophy, type I; **G12.1** Other inherited spinal muscular atrophy; **G12.8** Other spinal muscular atrophies and related syndromes; **G12.9** Spinal muscular atrophy, unspecified

Statistical analysis

Epidemiological indicators were standardized based on age/sex of the general French population.

In addition to descriptive analyses, process mining methods were used to describe healthcare pathways.

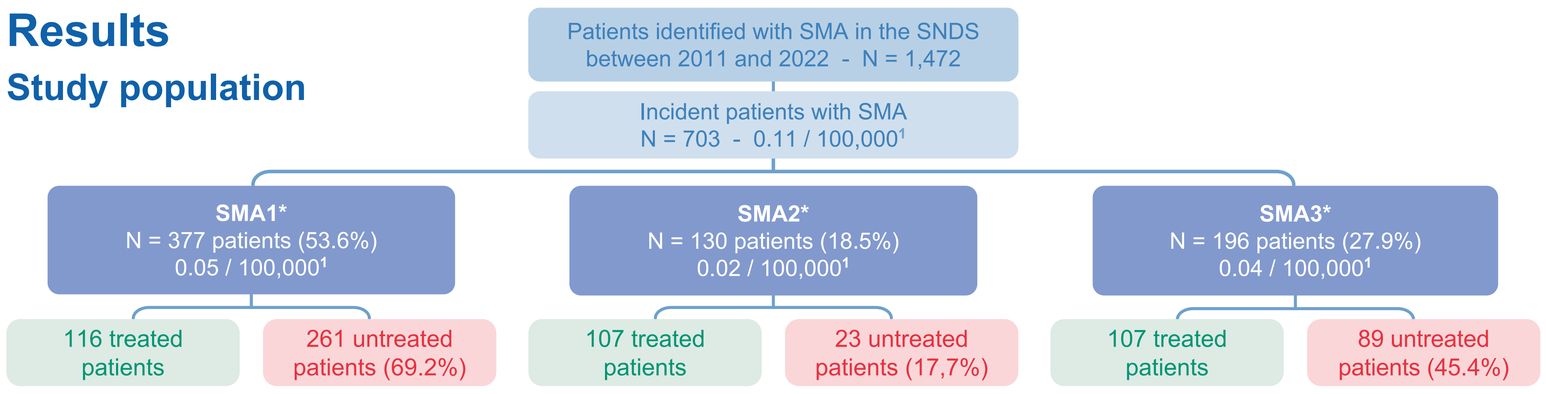
Conclusion

This real-world study using the SNDS confirms that SMA therapies meaningfully increased the survival of SMA patients, particularly in the most severe patients.

This study provides a visualisation of SMA healthcare pathways by subtype SMA1, SMA2 and SMA3, even if differentiating these patients proved to be difficult as there is a continuum in the disease progression.

Results

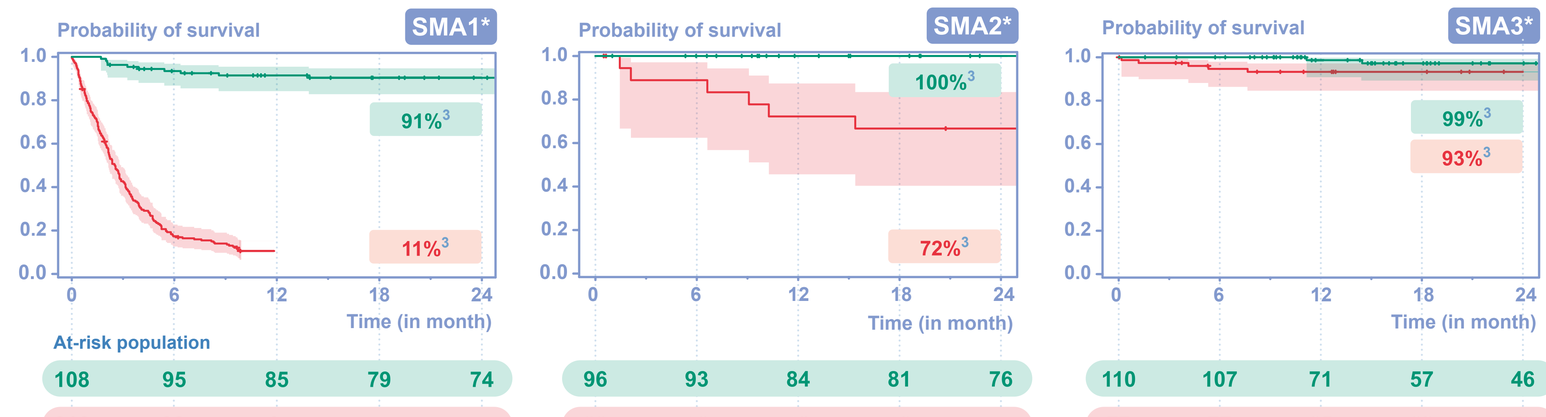
Study population



Characteristics of the study population
(incident patients)

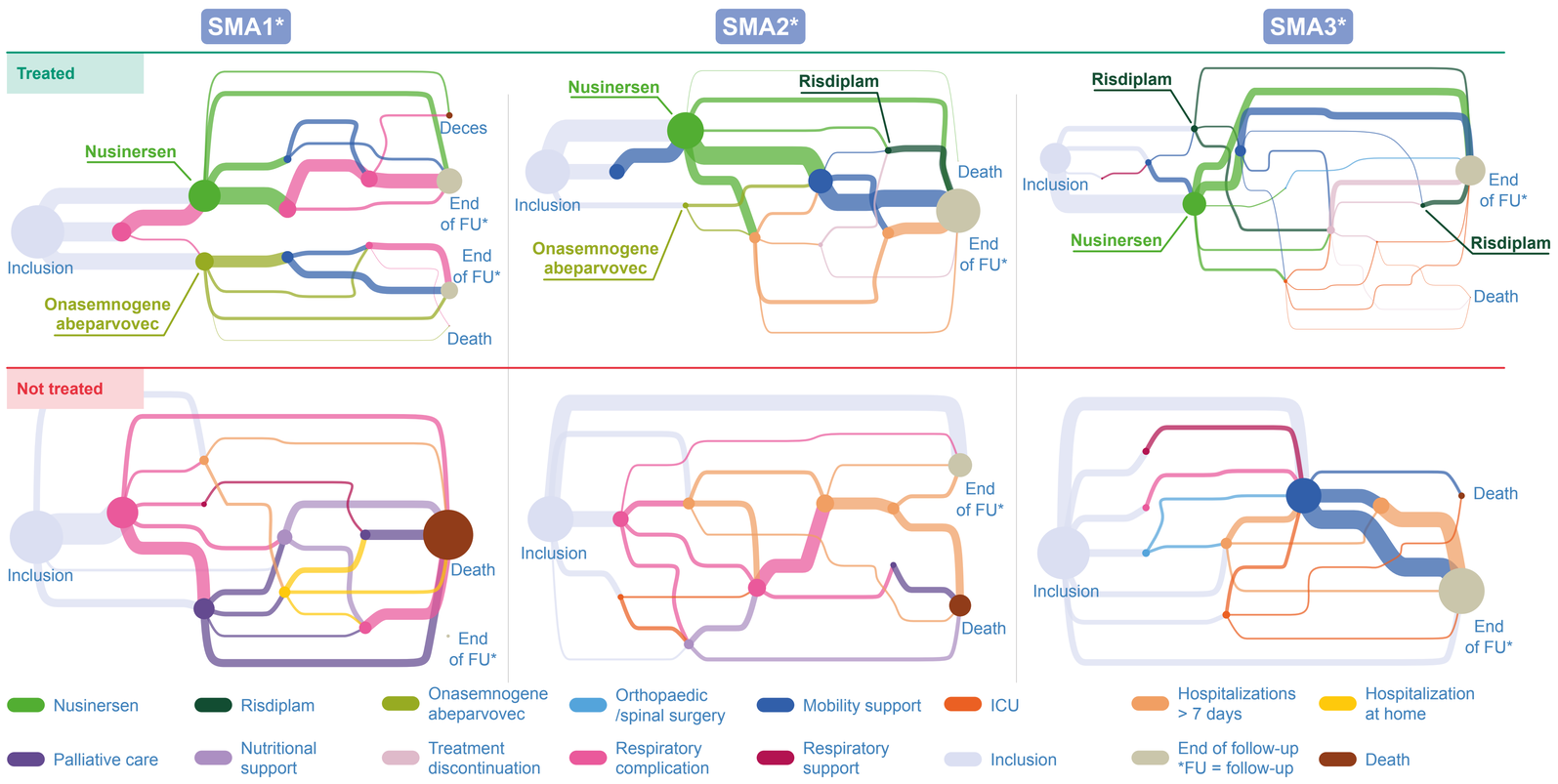
	SMA1*	SMA2*	SMA3*
Median age (Q1;Q3) at index date	4.0 months (2.0; 6.0)	18.0 months (14.0; 22.0)	159.0 months (71.5; 232.5)
% female	46.2%	55.4%	48.0%
Median follow-up time (Q1;Q3)	0.2 year (0.1; 0.4)	2.3 years (0.2; 9.5)	6.0 years (2.2; 10.3)
Median age (Q1;Q3) at end of follow-up[2]	1.1 year (0.2; 2.4)	6.1 years (3.4; 8.5)	18.7 years (11.2; 27.4)

Overall survival (Kaplan Meier) for patients affiliated to the General Scheme
about 75% of the French population

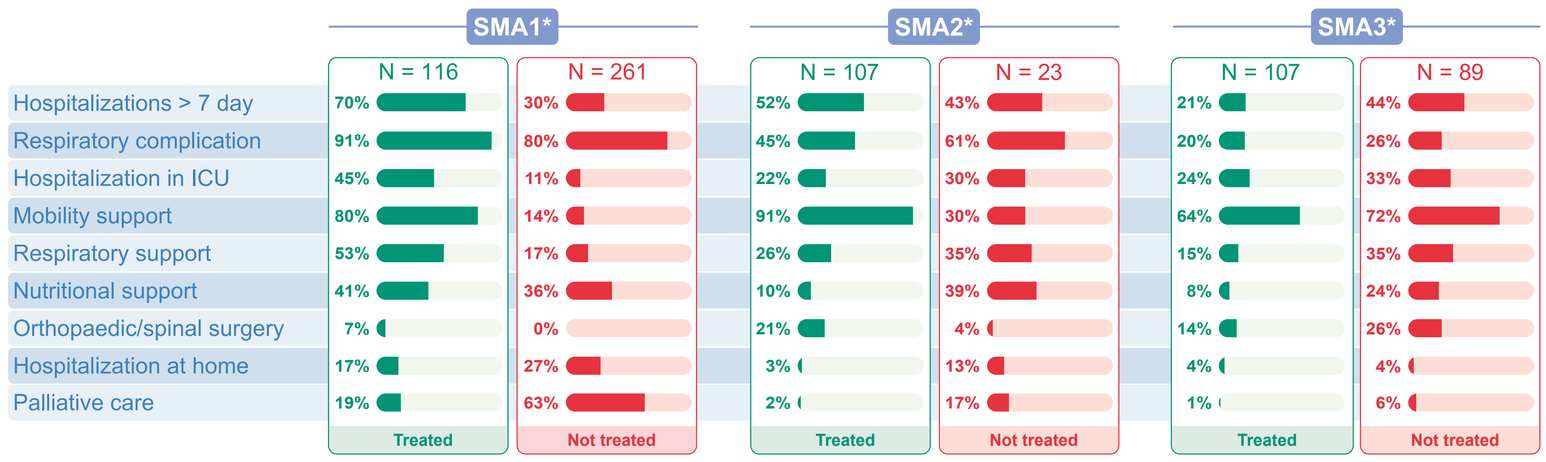


Graphical representation of the most common healthcare pathways observed

The size of each node and edge is proportional to the number of patients.



Percentage of patients presenting the events of interest at least once during the follow-up



Foot note explanations

1. 2022 incidence rates;
2. i.e. end of study period (31/12/2022) or death if it occured before end of study.
3. Probability of survival at 12 months

Regulatory statement

This SNDS study was registered with the HDH under the reference T94955362022081, was approved by CESREES on 22 September 2022 and authorised by the CNIL on 30/11/2022. (DR-2022-258 (request 922250)) - CNAM agreement signed on 12/09/2023.

Acknowledgments

The authors would also like to thank the Direction de la Stratégie, des Études et des Statistiques (DSES), Département Accès, Traitement et Analyse de la Donnée (DATAD), and Cellule de la CNAM en Charge de l'accompagnement des Demandes D'extraction (DEMEX) teams at the Caisse Nationale de l'Assurance maladie (CNAM) for the data extraction.

Conflict of interest

The presenting author was part of the scientific committee of this study, which was financed by Novartis Gene Therapies.