UMBRELLA-SUMMA LEGACY: UNIFIED PLATFORM FOR A BETTER INTEGRAL EVALUATION OF MYELODYSPLASTIC SYNDROMES IN SPAIN-STRATEGY FOR UNRAVELING PERSONALIZED GENOMIC MEDICINE IN PUBLIC HEALTH SYSTEM INCORPORATING PATIENT REPORTED OUTCOMES

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Background & aim

Myelodysplastic syndromes (MDS) are a heterogeneous group of clonal diseases characterized by a **difficult diagnosis**, **complex prognostic stratification**, and **unsatisfactory treatment**.

The **Spanish MDS Group (GESMD)** aims to **enhance clinical management** and **personalized medicine** for **MDS patients in Spain** by improving these aspects and facilitating future research.

Methods

A **coordinated** and **collaborative model** was established to **optimize resources** within the **GESMD: Three consecutive funded projects** have been **developed since 2021.**

Results

1 UMBRELLA

Unified platforM for a Better integral Evaluation of MyeLodyspLastic Syndromes in SpAin

2021-2023
902,055 euros
4 Institutions

Mutational studies of **541 patients** to be included into **Spanish Registry of MDS (RESMD)**. **450 samples** stored for research. **+20** published **studies.**

Research subprojects

- Molecular mechanisms involved in MDS evolution to AML.
- The role of transcriptional regulatory mechanisms in the pathogenesis and treatment of MDS.
- Genetic characterization of therapy-related myeloid neoplasms.
- Clonal hematopoiesis of undetermined potential in cancer patients: impact on immune dysfunction, treatment, and secondary malignancies.

2 UMBRELLA-SUMMA

Unified platforM for a Better integral Evaluation of MyeLodyspLastic Syndromes in SpAin-Strategy for Unraveling personalized genoMic Medicine in public heAlth system

2024-2026
901,250 euros
4 Institutions

23 hospitals have requested sequencing services from the 4 institutions of the coordinated project.

352 patients sequenced by the 4 institutions in **2024.**

19,436 patients with clinical information +/- molecular data included in the **RESMD.**

Validation of new diagnostic tool **Optical Genome Mapping**



Research subprojects

- Pathophysiological mechanisms of MDS evolution in three different cytogenetic subgroups: normal karyotype, +8 and del(5q).
- Identification of transcriptional mechanisms of progression and resistance to treatment in patients with high-risk MDS.
- Multiomics analysis of clonal and bone marrow niche cells interations in low-risk MDS: implication for disease heterogeneity and therapeutic outcomes.
- Characterization of the germinal and somatic landscape of therapy-related myeloid neoplasms (TRMN).

3 UMBRELLA-SUMMA Legacy

Unified platforM for a Better integRal Evaluation of myeLodyspLastic syndromes in spAin-Strategy for Unraveling personalized genoMic Medicine in public heAlth system incorporating patient reported outcomes

2025-2026
3,394,050 euros
25 Institutions



Research in 3 MDS groups of interest

TP53 mutated patients
TRMN
MDS with germline predisposition

25 Institutions
116 Researchers
26 New contracts

Enhancing of RESMD data by incorporating:



Optical Genome Mapping



Digitalization of bone marrow images



Patient-Reported Outcomes (PROMs) and Experience Measures (PREMs)



Artificial Inteligence (AI)

WORK PACKAGE 1	WORK PACKAGE 2	WORK PACKAGE 3	WORK PACKAGE 4	WORK PACKAGE 5
Coordination	Data integration and biobank management	Molecular/genomic studies: MDS/CH with TP53, TRMN and patients with germline predisposition	AI development for clinical data integration and treatment personalization	Training and dissemination of results

Conclusions

UMBRELLA, **UMBRELLA-SUMMA**, and **UMBRELLA-SUMMA Legacy** have fostered a **multidisciplinary approach** that is **transforming MDS patient care** and **research** in **Spain.**

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