PB2361 PREDIGA, EDUCATIONAL AND DIAGNOSIS PROJECT IN ACID SPHINGOMYELINASE DEFICIENCY DISEASE (ASMD) AND GAUCHER DISEASE (GD): RESULTS TO THE OBJECTIVE ACHIEVED OF 200 PATIENTS ANALYZED.

Topic: 35. Quality of life, palliative care, ethics and health economics

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Background:

Acid sphingomyelinase deficiency disease (ASMD) and Gaucher disease (GD) are lysosomal disorders caused by deficiency of lysosomal enzymes acid beta-glucosidase and acid sphingomyelinase. These enzyme deficiencies cause accumulation of glucocerebroside and sphingomyelin, respectively, mainly in macrophages, inducing deterioration of the organs in which they accumulate. Theoretical prevalence of ASMD is 0.4 per 100,000 and GD is 1 per 40,000. The absence of epidemiological studies in Spain leads us to believe that ASMD is highly underdiagnosed, and that the incidence of GD data is still to be defined. The consequences of undertreatment and the availability of effective therapies make it increasingly necessary to promote awareness and knowledge of these diseases and their diagnostic algorithms.

Aims:

The main objective of this study is to identify patients with idiopathic splenomegaly or splenectomy as well as to promote awareness and knowledge of ASMD and GD and their diagnosis algorithm through a national medical education program. Prevalence of ASMD and GD in patients with idiopathic splenomegaly or splenectomy will be also established.

Methods:

Regarding educational program, the participating centers lead clinical sessions as part of the PREDIGA education program. It was planned to impart about 100 clinical sessions within the medical education program and reach 200 patients fulfilling diagnostic criteria for ASMD and GD in the recruitment process.

In addition, PREDIGA project included an epidemiological, clinical, observational, non-interventional, multicenter, retrospective and prospective, cross-sectional with a single cohort study. The study population included adults and children with idiopathic splenomegaly or splenectomy. We performed a retrospective (5 years) and prospective review (2 years) of medical records to detect sustainable patients. In a single visit, the informed consent form was signed and the patient’s clinical data were recorded. We used the usual clinical procedures to collect a blood sample for Dry Blood Spot (DBS) to perform enzymatic, genetic, and biomarkers analyses.

The analysis of the samples was performed in the Archimed laboratory using ArchimedLife platform. The diagnostic algorithm for the analysis of the samples included two steps: first, enzymatic analysis was performed and if the result was positive or doubtful, genetic analysis was performed to confirm the positive result for ASMD or GD. In addition,
samples which results were positive for ASMD or GD were subjected to analysis of the biomarkers Lyso-Gl1 or Lyso-SM.

**Results:**

52 HCPs (22 Hematologists/ 18 Internal Medicine/ 12 Pediatricians) from 34 hospitals had implemented the medical education program and epidemiological searching in their health area of influence for 2 consecutive years. To date, 49 educational sessions, on-site and virtual, have been held by research team with 1000 assistants.

A total of 206 patients' blood samples have been collected to perform a DBS. Results to date have revealed the diagnosis of 2 ASMD and 4 GD patients derived from the epidemiological program. Thanks to implementation of this educational program 2 ASMD and 1 GD patients were diagnosed. Therefore, in this study the prevalence of ASMD and GD in patients with idiopathic splenomegaly or splenectomy is 0.97% and 1.9% respectively.

**Summary/Conclusion:**

The medical educational plan and the epidemiological study derived of this project have increased awareness of these conditions allowing the diagnosis of new ASMD and GD patients and better define the prevalence of these diseases in this cohort.