Abstract: PB2068

Title: RARITY MATTERS! THE EXPERIENCE IN OUR ADULT HEMATOLOGY SERVICE.

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Background:

Rare diseases are a group of pathologies that affect a small number of the population compared to other diseases. Even since the symptoms can be general, they can sometimes hide the real rare pathology that lies behind them, thus concluding in a misdiagnosis or even in a failure to diagnose at the right time. Currently, to define a disease as rare, we must rely on some criteria defined in the United States of America, the European Community, and Japan. And based on the American Rare Disease Act of 2002, a rare disease is defined as a pathology that affects less than 200,000 Americans, or less than 6 people per 100,000 inhabitants. On the other hand, if we rely on the European Orphan Regulation No. 141/2000, of 1999, any pathology that has a prevalence of less than or equal to 5 cases per 10,000 inhabitants is considered a rare disease. Meanwhile, the Japanese pharmaceutical Orphan drug law of October 1993 qualifies as a rare disease any pathology that affects less than 50,000 inhabitants in Japan, in other words with a prevalence of less than 1 case in 2500 inhabitants. The field of medicine is thus suffering from the lack of scientific medical knowledge as these diseases are still not well studied and the possibility of clinical studies is very limited due to their very low incidence. Therefore, a key point remains the epidemiology of these pathologies. It is reported that in the world there are 260 - 440 million individuals who suffer from rare diseases that reach the figure of nearly 7000 types and that not all of them have found the cause, mechanism, or cure.

Aims:

Based on the fact that in Albania there is still no register on rare pathologies and taking into account the difficulties in diagnosis and treatment, in order to give the best overview of patients with rare diseases who have been diagnosed or treated in the Service of Hematology, where they were diagnosed as rare events with the adult presentation, we have conducted a retrospective study on the incidence and prevalence of rare hematological diseases that have been diagnosed in the Hematology Service since 2005 until now.

Methods:

The study was based on the registers of the Hematology Service as well as on the records of the Statistics Service and included all the patients over the age of 18 years old who were admitted to the Hematology Service and were diagnosed with rare diseases from 2005 to December 2022.

In the research, the ICD 9 nomenclature of rare hematological pathologies was used, and the data were analyzed in EXCEL 16.0 and SPSS 25.0

Results:

Based on the data collected in the Hematology Service, there were a total of 64 patients with rare diseases, in the interval of age at the moment of the presentation, from 56.8 to 79.3 years and with an average age of 69 years. The largest percentage was obtained from Waldenström Macroglobulinemia (MW) cases and Gaucher Disease (GD) cases with respectively 16 cases of MW and 10 cases of GD, and, we had ultra-orphan diseases such as one patient with Nieman Pick, or chronic Eosinophilic Leukemia etc. were diagnosed. (table 1)

Summary/Conclusion:

The diagnosis and treatment of rare diseases remain a real challenge because we face real diagnostic and therapeutic difficulties. In the world, there are nearly 7,000 types of rare diseases that require a definitive curative treatment, but only 5% have a specific cure approved by the FDA. In the European Community, rare cancers make up to 22% of all neoplasms, and among these are many hematological neoplasms that require a cure.

Epidemiological exploration remains a key link in the field of a cure for them.

Keywords: rare diseases, rare neoplasms, orphan

Rare Diseases	Worldwide (yearly)	Albania (total cases 2005 - 2022)
Castleman Disease	21 -25 /per million	3
Chronic Eosinophilic Leukemia	0.18 - 0.36/ 100,000	2
Cold Agglutinin Disease	1 in million	3
Common Variable Immunedeficiency	1 / 25,000 to 50,000	1
Erythropoetic Porphiria	1 / 74,300 individuals	3
Factor X deficiency	1/45,000-286,000	2
Factor XIII Deficiency	1 in 2,000,000-5,000,000	1
Fanconi Anemia	1 in 100000 to 250000 births	3
Gaucher Disease	1 / 50,000 to 100,000	10
Histiocytosis	1-2/200,000 people	6
Mastocytosis	1 /10,000	2
Nieman Pick Disease type C	1/45,000-286,000	1
PNH	0.5-1.5 / million	5
Primary Myelofibrosis	0.3/100,000 person	122
Waldenstrom Macroglobulinemia	3 /million	16

Keywords: Screening, Prevalence, Adult