



SKIN MANIFESTATIONS OF INTERNAL DISEASE

## **RARE SKIN DISEASES WITH SKIN INVOLVEMENT: A RESTROSPECTIVE STUDY**

*M Doda<sup>(1)</sup> - M Fida<sup>(1)</sup> - S Rista<sup>(1)</sup> - E Vasili<sup>(1)</sup>*

*University Of Medicine, Department Of Dermatology, Tirana, Albania<sup>(1)</sup>*

**Introduction:** Diseases defined as rare, affect less than 5 in 10,000 individuals. Due to their complexity, severity and duration, they can lead to degenerative changes and a significant reduction in the quality of life of patients. However, if detected in time, many of these diseases can be treated and kept under control.

**Objective:** We aim the recognition of epidemiological-clinical features of rare diseases with skin involvement, thus contributing to literature, serving as basis for further in-depth studies. We intend as well to increase awareness of rare diseases among general practitioners, dermatologists, pediatricians.

**Materials and methods:** In this study, the time interval included, is from January 2012 to December 2017. We have analyzed the records of patients hospitalized in the UHC "Mother Theresa" Tirana, to select those who suffer from rare diseases. The list of these rare diseases was downloaded from European Organization for Rare Diseases.

**Results:** 80 patients were diagnosed with rare diseases (49% males and 51% females). 17 rare diseases with skin involvement were identified and divided into three groups to facilitate data analysis: I- group of neurocutaneous pathologies (22.5%), II- group of congenital diseases (43.7%), III- group of diseases with different etiopathogenesis (33.8%). Neurofibromatosis was the most common disease with 23%. Age range is from 14 to 81 years old. Most affected age group is 21-30 years old. Xeroderma pigmentosum had the highest number of days of hospitalization (41 days), followed by pyoderma gangrenosum (29 days).

**Conclusions:** Rare diseases require special attention from family doctors and dermatologists to carry out an early diagnosis, that will be followed by the possibility of keeping the disease under control and improving the quality of life of patients.

