

BURDEN OF DISEASE IN PATIENTS WITH FABRY DISEASE: HEALTH RELATED QUALITY OF LIFE AND ASSOCIATED FACTORS

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INTRODUCTION: Fabry disease (FD) is an X-linked lysosomal storage disorder, caused by an accumulation of glycosphingolipids due to alpha galactosidase A deficiency¹. The multisystem involvement, including renal, cardiovascular and neurologic damage, that characterizes Fabry's disease and the need for lifelong treatment interfere with the patient's daily life. However, physical and psychological factors that impact on health related quality of life (HRQoL) are poorly understood².

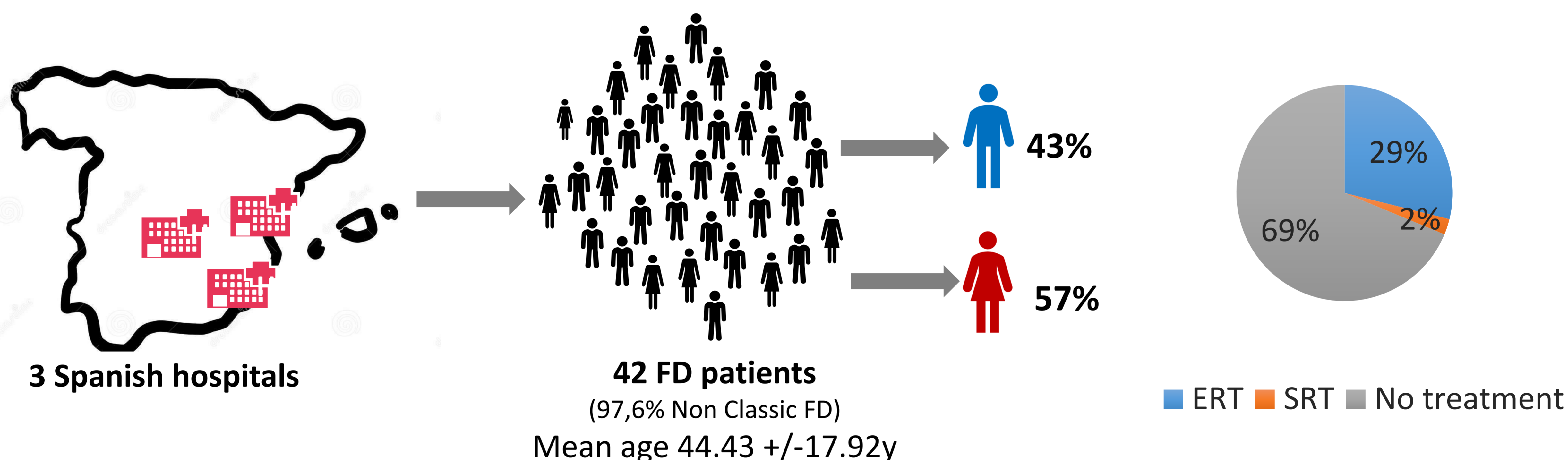
OBJECTIVE: To describe the impact of FD on HRQoL, analyzing the influence of severity of FD, pain, anxiety and depression.

METHODOLOGY: The study is cross-sectional study was carried out in three Spanish Hospitals, and included patients from nephrology consultation, that were older than 14 years with genetic confirmation of FD. We excluded patients with renal replacement therapy, and organ transplantation. The study was approved for a local ethics committee, and all the patients, or legal representatives, agreed to sign the informed consent.

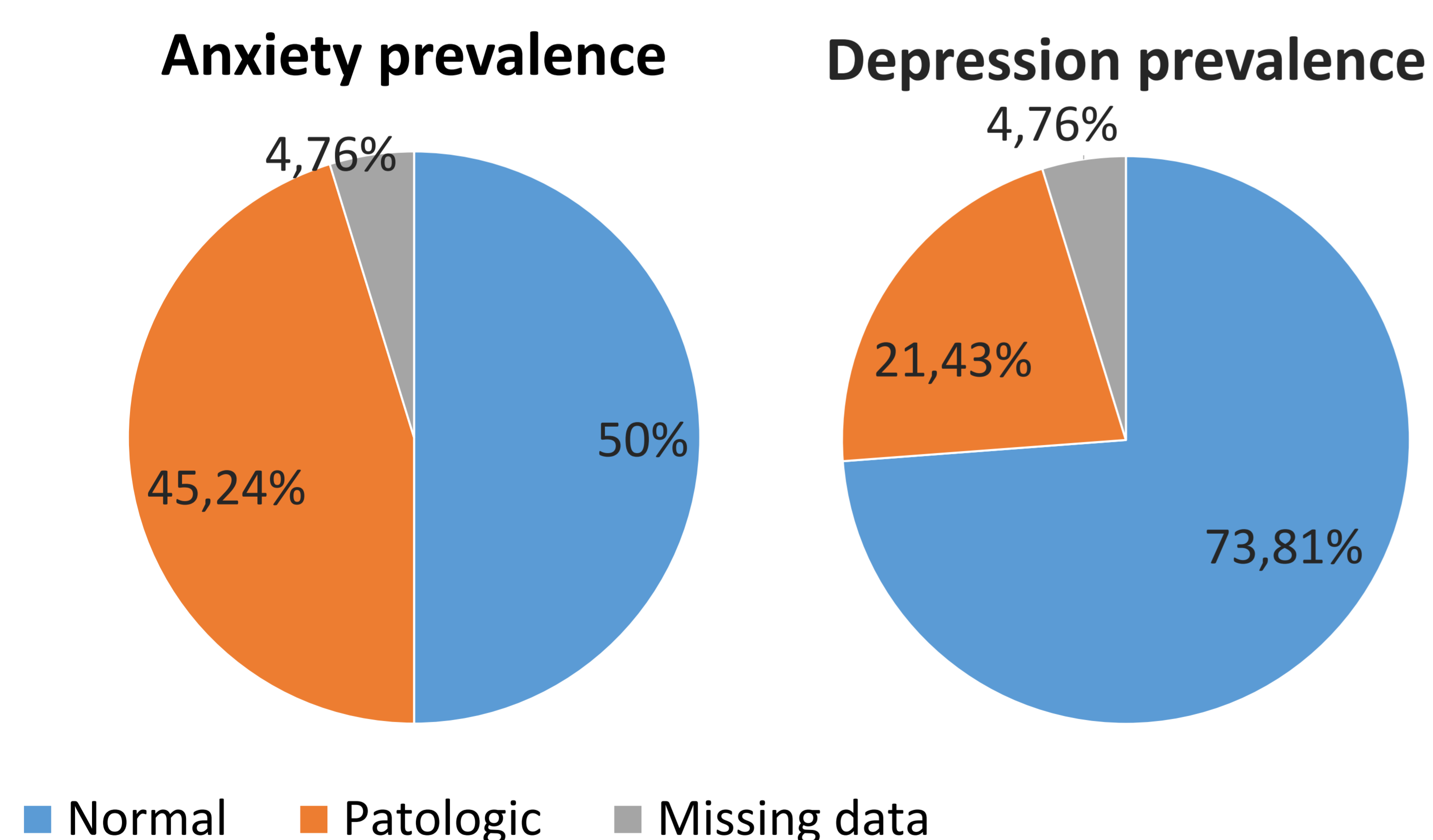
Demographic data, clinical variables of target organ involvement, Mainz Severity Score Index (MSSI), variables related to quality of life, pain and anxiety depression were collected using the EQ5D5L, Brief Pain Inventory (BPI) and Hospital Anxiety and Depression Scale (HADS).

Statistical analysis was performed using parametric and non-parametric test.

RESULTS:



Hospital anxiety and Depression Scale (HADS)

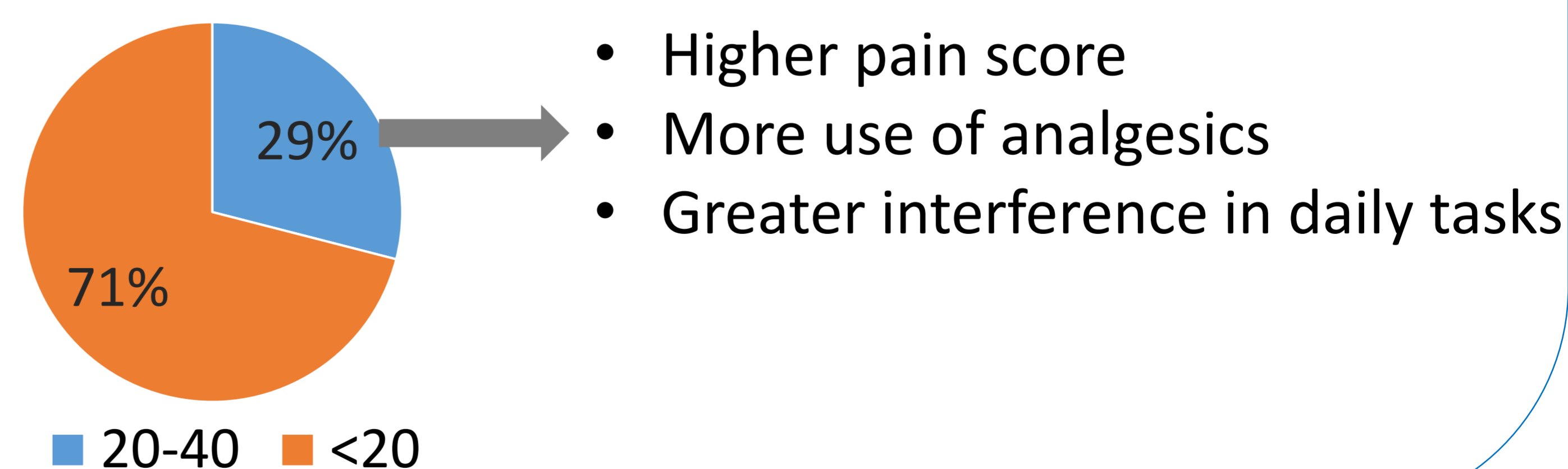


Mainz Severity Score Index (MSSI)

Table1. Patients classification MSSI Score

	Adult patients	Pediatric patients	TOTAL
Mild	27	3	30
Moderated	12	0	12
TOTAL	39	3	42

MSSI Score: 11.93 +/- 11.06



CONCLUSIONS:

- In our series of patients with a non-classic form of FD, the prevalence of anxiety and depression was 1 in 2 and 1 in 5 patients.
- Pain affected 2 in 5 patients, with an impact on the quality of sleep and the ability to enjoy life.
- An active search for anxiety, depression and pain should be performed in all patients with a nonclassical form of FD.

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