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THE IMPACT OF RARE DISEASES ON HEALTH RELATED QUALITY OF LIFE

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INTRODUCTION

Over 7000 Rare Diseases (RDs) affect around 60 million patients living in the European Union (EU) and the United States (US).[1] More than 80% of RDs are genetic and appear early in life, resulting in a 30% mortality rate in children diagnosed before their fifth birthday.

RDs are chronic and result in a decreased Quality of Life (QOL).

AIMS

To develop a QOL assessment tool for RD patients to identify issues of accessibility, diagnosis, information provision at the time of diagnosis, personal care and independence and mental health.

METHOD

A self-administered Health Related QOL assessment tool was developed. The assessment tool consisted of 30 questions which were divided into 4 main sections: 'Demographics', 'Personal Care and Independence', 'Mental and Social Health' and 'Accessibility to Orphan Drugs.'

The tool was validated by seven experts: 3 pharmacists, 2 researchers, 1 clinician and 1 RD patient.

The developed and validated tool was published online. RD alliances and support groups were contacted locally and internationally to request that they disseminate the assessment tool to their members and patients.

The tool was disseminated to RD patients and guardians who could read and write in English and who were above 18 years of age.

RESULTS

Two hundred and twenty five responses were gathered. Eighty-two of the respondents were male. One hundred and thirty five patients stated that they received a misdiagnosis in relation to their condition. Fifty four patients stated that it took 5 years or more for them to receive a correct diagnosis of their condition.

Forty four patients claimed that it was difficult for them to be able to afford orphan drugs prescribed to them and 71 patients claimed that it was almost impossible for them to afford their medication. Twenty nine patients claimed that no medications were available for their condition.

US patients face larger financial burdens and have greater accessibility issues than EU RD patients ($p<0.05$). There was a significant QOL difference between EU and US RD patients ($p<0.05$) as the Europeans reported having a better QOL in relation to personal care and independence (Table 1) and mental and social health.

Table 1: Patient Personal Care and Independence (N=225)

How difficult was the following?	Group	Mean Likert Scale Score*	Std. Dev.	p-value
Eating, drinking or being fed	EU	3.58	0.502	0.000
	US	2.38	0.500	
Bathing, washing or general hygiene	EU	3.44	0.512	0.116
	US	2.94	0.998	
Getting in and out of bed	EU	4.44	0.512	0.000
	US	2.84	0.454	
Moving about in the home	EU	3.56	0.512	0.000
	US	2.19	0.946	
Visiting public places	EU	2.50	0.516	0.232
	US	2.68	0.909	
Understanding parent, caregiver or people around you	EU	4.50	0.516	0.463
	US	4.61	0.495	

** 1= Extremely difficult; 5= Very easy*

CONCLUSION

RD patients face challenges related to diagnoses of their condition and QOL. Accessibility of Orphan Drugs depended on pricing, re-imbursement policies and product availability. There is a need for improvement in the QOL of RD patients given the high cost of illness, mental health problems and poor accessibility.

REFERENCE

1. Bograt K, Irvin V. Health-related quality of lif e among adults with diverse rare disorders. Orphanet J Rare Dis. 2017;12(1):177