

# ACCESS TO ORPHAN DRUGS AND QUALITY OF LIFE IN RARE DISEASE

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## INTRODUCTION

Over 7000 rare diseases (RD) affect around 300 million patients worldwide. More than 80% of RDs are genetic and appear early in life<sup>1</sup>, resulting in a 30% mortality rate in children diagnosed before their fifth birthday.

To date, there has been no locally conducted study about the healthcare needs of people living with RDs.

Research on RDs focuses on treatment and care of RDs with limited focus on health related quality of life and accessibility to Orphan Drugs (ODs).<sup>2</sup>

## AIMS

To analyse what regulations and policies related to OD accessibility are available in Malta and internationally.

To assess the Quality of Life of RD patients.

## METHOD

A retrospective analysis was carried out to observe features of OD policies in RD patients in Malta and internationally.

Six main themes<sup>3</sup> were used to assess the accessibility of ODs and were used in a systematic literature review.

The themes were: 1) OD policies 2) Marketing Authorisation 3) Incentives Offered 4) Pricing of ODs 5) Reimbursement 6) Pharmacovigilance.

A self-administered Health Related Quality of Life (HRQOL) 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 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 the 18 years of age.

## RESULTS

There were OD specific legislations in 29 countries. Between January 2000 and December 2017, there were 1,952 ODs in the EU<sup>4</sup> and 3,642 ODs in the US.<sup>5</sup>

In the US ODs have a market exclusivity of 7 years and in the EU ODs have a market exclusivity of 10 years, which is extended for ODs used in paediatric patients. Accessibility of ODs depended on pricing, re-imburement policies and drug availability.

Two hundred and twenty five responses were gathered when using the HRQOL. 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.

Patients from the US faced 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 and mental and social health.

## CONCLUSION

All the countries in this study had an OD regulation in place.

There were differences between countries in pricing, licensing and reimbursement of ODs, which have an impact on accessibility.

There is a need for improvement in the quality of life of RD patients.

## REFERENCES

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