

ACCESS TO ORPHAN DRUGS AND QUALITY OF LIFE IN RARE DISEASE

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Background: Over 7000 rare diseases (RD) affect around 300 million patients worldwide. The majority of RDs are genetic and appear early in life, resulting in a 30% mortality 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.

Purpose: (1) To retrospectively analyse regulations and policies related to Orphan Drugs (ODs) accessibility in the EU and The US and propose methods to improve patient access (2) To create a Quality of Life assessment tool specific to RD patients and explore issues of diagnosis, information provision at the time of diagnosis, use of health and support services and general quality of life

Methodology: A retrospective analysis was carried out to extract features of various OD policies to help identify areas that can improve accessibility. A self-administered Health Related Quality of Life (HRQOL) Assessment tool was created, validated by seven experts, and published online. Different patient groups in European Union countries the United States of America were contacted to invite their members to participate.

Results: There were OD specific legislations in all of the EU countries and in the US. Accessibility of ODs depended on pricing, re-imburement policies and product availability. Two hundred and twenty-five responses given by RD patients were analysed. Accessibility issues were a hurdle for RD patients as 52% reported that medication is available in other countries but not in their country. Forty percent received a misdiagnosis and 34% were waiting over 1 year to receive a diagnosis. In terms of mental health, 74% complained of stress and anxiety problems.

Discussion: Although all the countries examined 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 given the high cost of illness, mental health problems and poor accessibility to medications.