

Quality of Life in Hemophilia

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Assessment of health-related quality of life (HRQoL) is vital to evaluating the well-being of patients with hemophilia and their individual experiences of the condition, Dr. Alessandro Gringeri said.

Perception is a subjective process, mediated by consciousness, with each individual perceiving an object or event differently from other people. Subjectivity and objectivity are indistinguishable in the perception of oneself, one's body, and the surrounding world, Dr. Gringeri said. Health and the perception of well-being matter equally; therefore, it is essential to take into account quality of life and not just objective science. "The overall well-being as subjectively perceived by the individual, affected or not by a disease, must not be ignored or considered secondary to other parameters — it should be equal."

Over the past three decades, a number of generic instruments have been developed internationally to assess HRQoL in children and adults. More recently developed disease-specific instruments, including for children and adults with hemophilia, better capture and quantify various aspects of patients' well-being during different developmental phases. Given the high cost of hemophilia treatment, this information helps illustrate the benefits and improvements of treatment to quality of life for patients, Dr. Gringeri said.

In its 1946 constitution, the World Health Organization (WHO) stated that health should be considered "a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity." The United Nations Development Programme's Human Development Index rests on a single value that puts together health, longevity, knowledge (literacy and school enrolment), and standards of living (per capita GDP). Alternatives to this purely economic approach include the measurement of non-economic aspects of quality of life, such as social and environmental variables of well-being, Dr. Gringeri said. More recently, in 1994, the WHO defined quality of life as "the individual's perception of his position in life in the context of the culture and value system in which he lives, and in relation to his goals, expectations and concerns."

HRQoL assessment can be used for individuals in good health, though it is especially helpful for individuals with chronic illnesses because it measures specific health-related factors that affect quality of life, Dr. Gringeri said. HRQoL can therefore be defined as the "qualitative dimension of functioning" — that is, the individual experience of illness together with the psychosocial response to disease-related and treatment-related symptoms.

HRQoL instruments should take into account the multidimensional nature of HRQoL, both the physical and the psychological — emotional, mental, social, and behavioural — components of the patient's perception of well-being and functioning, and if relevant, the perceptions of observers.

In the past five years, a number of disease-specific HRQoL questionnaires have been developed for individuals with hemophilia. Some of these questionnaires have been developed

internationally, such as the pediatric Haemo-QoL in 6 European countries, the adult MedTap questionnaire (USA, Canada, Spain, Germany) and the adult Haemo-Latin QoL (South America), whereas others have been developed only on a national level, such as the Canadian Cho-KLAT, the American QoL questionnaire for young patients, the Italian Haem-A-QoL and the Spanish Haemophilia-QoL. Hemophilia-specific HRQoL assessment tools include questions about individual treatment, physical health and symptoms, psychosocial and emotional functioning, socio-economic conditions, and demographic profile (age, gender, marital status).

It is essential to design HRQoL instruments for a range of age groups, since concepts of well-being differ among young children, adolescents, young adults, and the elderly, Dr. Gringeri noted. The Italian HRQoL instrument developed by von Mackensen et al. and known as the Haemophilia Quality of Life Questionnaire for Adults (Haem-A-QoL) is notable because it features shared variables (Haem-A-QoL Core Instrument) with the Haemophilia Quality of Life Questionnaire for children (Haemo-QoL), allowing for comparison of HRQoL between adults and children. The questionnaire for adults with hemophilia has been linguistically validated in more than 20 languages, and a questionnaire for elderly patients is to be introduced at the Hemophilia 2008 World Congress.

HRQoL questionnaires provide an opportunity for patients to report their own experiences of their physical and mental functioning and well-being, Dr. Gringeri said. Disease-specific HRQoL instruments help evaluate the benefits of existing and new treatments or strategies from the perspective of patients' values and expectations. Knowing patients' perceptions of their health and treatment-related outcomes ultimately benefits their care. The information also benefits epidemiological and clinical research to improve the overall quality and delivery of hemophilia assessment and care.

HRQoL should be considered a vital part of medical management, Dr. Gringeri said. Reliable, valid and standardized instruments are needed to monitor the progress or deterioration of patient well-being. They must be specific and sufficiently sensitive to capture different responses in specific circumstances and over time, similar to the instruments that measure objective parameters, like joint health, he added.

Given the numerous questionnaires available, choosing the right one depends on factors such as the following:

- Type and scope of the study (e.g., prospective, cross-sectional, national, international)
- Study population (children, adults, or both)
- Study administration (delivery, time frame, follow-up)
- Study outcomes (e.g., clinical results, physical health, psychosocial functioning, costs and benefits of treatment, patient satisfaction, perception of quality of life)

Increasingly, patients are assessed with both generic and disease-specific instruments, to obtain the optimal results from each type of instrument. This combination allows, on one hand, a comparison with the general population (generic,) and on the other hand, it provides a comprehensive picture of the specific health condition (disease-specific). An important issue is ensuring that questions are immediately comprehensible, unambiguous, and appropriate to the

age level and culture of the patient being interviewed. Therefore, HRQoL instruments must be psychometrically, linguistically, and cross-culturally validated, said Dr. Gringeri.

Data from generic and disease-specific instruments can be compared to measure HRQoL in different health conditions, as Dr. Gringeri described. In Europe, the generic questionnaire for children (KINDL) revealed lower scores for children with chronic diseases and other health conditions — including obesity, asthma, and atopic dermatitis — in the dimensions “body,” “family,” and “chronic disease”. The Haemo-QoL study also revealed regional variations in patient perception of quality of life.

Children with hemophilia similarly tend to perceive themselves as having a lower quality of life — particularly in the physical domain, because they cannot participate in certain sports or activities, Dr. Gringeri noted. At the same time, he said, children with chronic diseases tend to have a higher perception of family than others. This highlights the value of providing educational and psychosocial support to families in improving quality of life for children with hemophilia.

HRQoL assessments can also be used to compare the healthcare systems, resources, and patient quality of life in different countries. Hemophilia-specific quality of life information is useful in advocating for governments to harmonize their hemophilia treatment programs with those of other countries.

More recently, an Italian quality of life study comparing patients with inhibitors and those without inhibitors found similar quality of life, but small disparities in terms of physical functioning. The data also show a steep decline in quality of life for elderly hemophilia patients compared with that for the general population.

A current study in Europe, known as the European Study of Clinical, Health Economic and Quality of Life Outcomes in Haemophilia Treatment (ESCHQoL Study) and led by Wolfgang Schramm, is assessing, in addition to other clinical, socio-economic and quality of life parameters, patient well-being in relation to factor consumption levels, Dr. Gringeri reported. Preliminary findings indicate that higher national factor availability, which can allow prophylaxis and immunotolerance treatment, provides a better quality of life to patients with hemophilia, compared to lower amounts that cannot support state-of-art care.

Health-related quality of life is measurable and quantifiable, Dr. Gringeri said. Many valid and reliable generic and disease-specific instruments are now available, and healthcare practitioners must learn how to use them.