Guidelines on rare diseases: Methods on handling evidence neither identified nor required

April 28 2011

People with rare diseases have the same right to high-quality health care in line with current medical knowledge as other patients do. However, relevant and reliable clinical studies on rare diseases are often lacking. Among other things, this makes the development of corresponding guidelines more difficult, but precisely such guidelines could help improve treatment quality.

The German Federal Ministry of Health therefore commissioned the Institute for Quality and Efficiency in Health Care (IQWiG) to answer the following question: What methodological approaches are used by guideline developers and health technology assessment (HTA) agencies in the handling of evidence (clinical study results) for the development of guidelines on rare diseases? In addition, IQWiG was to investigate whether physicians and researchers pose different requirements for the evidence in guidelines on rare diseases than for guidelines on other, more common diseases.

Methodological approach hardly addressed

For this purpose, IQWiG systematically searched for and evaluated manuals on the development of treatment guidelines, methods papers by important HTA agencies, as well as guidelines on selected rare diseases. The findings show that the handling of evidence on rare diseases is hardly addressed in these documents. Explicit references to or

instructions on the handling of such evidence was found only sporadically; in any case, uniform methodological requirements cannot be inferred from the information retrieved. However, neither do the manuals, methods papers, and guidelines analysed provide indications that a fundamentally different approach and different requirements should be adopted for rare diseases than for more common ones.

Evidence on more common diseases is also often poor

"Little or no evidence is not only a problem of rare diseases," explains IQWiG's Director Jürgen Windeler. "It is still possible to develop guidelines, and in principle there is no good reason to adopt a different approach or pose different requirements for rare diseases than for more common ones. This applies to the handling of evidence (as investigated in the report) as well as to the planning and conduct of the clinical studies themselves. If people with rare diseases are to be provided with high-quality health care, then studies of high methodological quality are required. The number of patients affected is usually large enough to enable such studies - even if in some cases international cooperation is needed."

Provided by Institute for Quality and Efficiency in Health Care