

Title	Rare Disease Management - Experiences from Abroad
Agency	INESSS, Institut national d'excellence en santé et en services sociaux 1195 Av. Lavigerie, bureau 60, Québec, QC, G0S 1E1 ; Tel: 418 643-1339, Fax: 418 643-8220; inesss@inesss.qc.ca, inesss.qc.ca
Reference	ETMIS 2011 7(6) Printed French edition 978-2-550-62800-2, English summary (PDF)978-2-550-62799-9 http://www.inesss.qc.ca/fileadmin/doc/INESSS/Rapports/OrganisationsSoins/ETMIS2011_Vol7_No6.pdf http://www.inesss.qc.ca/fileadmin/doc/INESSS/Rapports/OrganisationsSoins/INESSS_Summary_RareDisease_EN.pdf

Aim

In 2010, Québec's Minister of Health and Social Services expressed the intention to establish a strategy for the management of rare diseases in Québec. To that end, he began by mandating the Institut national d'excellence en santé et en services sociaux (INESSS) to review what is being done abroad in terms of rare disease management.

Conclusions and results

What emerged from the experiences with developing national plans in Europe was that the process initially requires analyzing what is already being done and performing a needs analysis, then setting up a committee to develop a plan, and finally implementing and monitoring the plan. Measures must be accompanied by outcome indicators to allow for the periodic evaluation of the plan. This process must be carried out with rigour and in collaboration with all community stakeholders, including experts, practitioners, institutions, industry, and also patients and their associations.

This report synthesizes the main action areas in the field of rare diseases observed outside Québec, whether or not part of an overall plan. These areas cover screening and diagnosis, epidemiology, patient management and access to care, improvement of treatments and access to appropriate drugs (including orphan drugs), health professional development, research and information.

Recommendations

Québec has the opportunity to benefit from the content of these different initiatives and from several experiences in Europe with both developing and implementing a rare disease plan or strategy. It could also consider sharing expertise with others, whether regarding information on the diseases, clinical practice guidelines or the use of drugs or other products, by participating in Orphanet (recent materialization of Orphanet-Québec project) or in other collaborations especially with the professional or patient associations concerned.

Methods

Given that rare disease management is more advanced in Europe than in the rest of the world, the literature search focused especially on the national plans of the different European countries and on the official documents issued by the European Union. Although the United States, Australia and the Canadian provinces do not have national or provincial plans for managing rare diseases, their experience, particularly with orphan drugs, was examined.

Written by

Stéphanie Elger, INESSS, Canada