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Rare Disease Management

Experiences from Abroad

Summary

Prepared by

Stéphanie Elger

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*Institut national
d'excellence en santé
et en services sociaux*

Québec 

SUMMARY

RARE DISEASE MANAGEMENT: EXPERIENCES FROM ABROAD

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.

Rare diseases encompass genetic diseases, rare cancers, auto-immune disorders, congenital anomalies, toxin-induced diseases, infectious and other diseases. These diseases are often severe and debilitating, most often appear very early in life, may be associated with chronic pain and may not have any curative treatment. They often reduce people's autonomy and quality of life, while placing a heavy burden on families and caregivers, and also affect life expectancy. According to the definition prevailing in Europe, a disease is considered rare when it affects no more than one in 2000 people.

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.

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.

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.