Fragile X syndrome: the role of molecular diagnosis and screening in an integrated approach to services

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Record Status
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Citation

Authors' objectives
This assessment summarizes the state of knowledge regarding the genotypic analysis of individuals with the syndrome and screening for asymptomatic carriers of a dynamic mutation on the FMR1 (fragile X mental retardation 1) gene. This report also discusses the usefulness of developing or maintaining such a service in Quebec's health-care system and the management of affected individuals by medical, social and educational services, and explores the ethical issues involved.

Authors' conclusions
Agency believes that:

1. The necessary medical, social and educational resources should be available to meet the needs of affected families in a timely and appropriate fashion.

2. The different players in the health and social services and educational systems should examine the possible ways of improving early identification and the diagnostic workup of children with signs consistent with fragile X syndrome, devoting special attention to the services available for developmentally delayed children.

3. One or two laboratories should be available to perform, for all of Quebec, the molecular tests for fragile X syndrome in the following situations: - The molecular diagnosis of fragile X syndrome in a symptomatic individual with either an indication recognized in the medical association guidelines or signs consistent or associated with the syndrome, in the opinion of the ordering physician. - Cascade screening of an affected individual's relatives. - Confirmation of carrier status in a pregnant woman with a family history of signs associated with the syndrome. - Prenatal diagnosis, if the mother is a carrier of a premutation or a full mutation.

4. All laboratory services should be subjected to quality control.

5. The different players in the health and social services system, the educational system and the job sector should improve intersectorial collaboration at the regional level in order to improve the coordination and continuity of the services available to affected individuals and their families.

6. The issues of the accessibility, continuity and complementarity of services for fragile X syndrome reflect, in part, an organizational problem that also affects children with developmental delays of other etiologies and their families, with the result that the required efforts should be part of a coherent approach that will benefit all these families.

7. The public authorities should give preference to a mode of service organization that promotes the respect of individuals, ensures equal access to services in all regions and prevents discrimination, especially in the area of insurance.

8. Research should continue, here and elsewhere, to better document the following: - The epidemiology of the...
syndrome in the general population. - The risk of hereditary transmission of the syndrome. - Phenotype prediction. - The development of genetic tests better suited to wide-scale use. - The psychosocial impact of diagnosing, screening and genetic counselling.

9. It would be essential to evaluate, by means of pilot projects, any high- or low-risk-population diagnostic and screening strategy whose implementation is being considered, on the basis of the following criteria: - Its technical, organizational and economic feasibility. - Its efficacy in terms of the number of individuals or couples who have received genetic counselling and follow-up that meet their needs. - Its usefulness in terms of the services that are already available. - Its ethical and social acceptability.

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**INAHTA brief and checklist**

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