

Surveillance of rare genetic disorders: No longer orphan diseases

Mapping of the human genome has facilitated the discovery of more and more rare disorders. Today, more than 6,000 rare genetic disorders affect approximately three million Canadians. Many of these conditions have very little Canadian data and are unpopular projects for research funding. These orphan diseases are individually few in number, but collectively are an important cause of morbidity and mortality. Accordingly, the challenge becomes one of ascertaining sufficient numbers to enable meaningful analysis and interpretation, which is essential for the development and implementation of timely interventions. The CPSP affords a unique opportunity of addressing the issue of too few numbers by providing a mechanism for national and international data collection. For example, interna-

tional collaboration results in accelerated knowledge acquisition and exemplifies a perspective that can best be described as one of enhanced 'global village' surveillance. The success of the CPSP in collecting sufficient data on rare genetic disorders is illustrated by the results from ongoing studies on Smith-Lemli-Opitz syndrome (SLO) and CHARGE association/syndrome (CHARGE A/S). It is anticipated that the upcoming study on Prader-Willi syndrome (PWS) will prove to be just as successful. More information on both of these study results and related publications is available in CPSP 2001 Results <<http://www.cps.ca/english/proadv/CPSP/2001Results.pdf>> <<http://www.cps.ca/english/proadv/CPSP/2001Results.pdf>>.

LEARNING POINTS

- Three new DHCR7 mutations have been identified in Canadian patients.
- Study results may suggest the cost effectiveness of prenatal screening for SLO.
- The age of diagnosis for CHARGE A/S has decreased from 11 years to 1.5 months.
- Earlier diagnosis and intervention of CHARGE A/S in children affected by combined sensory impairment (deafness or blindness) has huge implications for their developmental outcome.
- Data collected from the new study on PWS will help to document whether the diagnosis is made clinically or genetically.
- Early diagnosis of PWS, followed by appropriate management, can have a positive impact on the patient's life, particularly regarding treatment of morbid obesity and modification of behavioural problems, such as violent aggressive outbursts.

The Canadian Paediatric Surveillance Program (CPSP) is a joint project of the Canadian Paediatric Society and Health Canada's Centre for Infectious Disease Prevention and Control that undertakes the surveillance of rare diseases and conditions in children. For more information visit our Web site at <www.cps.ca/english/cpsp> or <www.cps.ca/francais/pcsp>.

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