CANADIAN PAEDIATRIC SURVEILLANCE PROGRAM

Study aims to shed light on Fragile X Syndrome

Even though Fragile X Syndrome (FXS) is the most common inherited form of intellectual disability, little is known about the prevalence, geographic distribution, or how it is managed across Canada.

A new Canadian Paediatric Surveillance Program (CPSP) study will investigate the number of new FXS cases in Canada as a first step toward ensuring proper access to care for children and families.

Data from the United States suggest that FXS affects 1 in 5,000 boys and 1 in 6,000 to 8,000 girls. But with only two FXS clinics in Canada, comparable national data are limited and suggest far fewer Canadian cases.

This potential discrepancy raises many questions. Are there fewer children with FXS in Canada? Are doctors not asking for the test? Or are there barriers to health care that interfere with diagnosis and FXS management? Without answers, making decisions about policy is difficult, said Dr. Jonathan Down, co-principal investigator of the study, which runs from April 2012 to April 2014.

"[For instance], when talking about screening at a population level, it's not a small undertaking. The more information that's available regarding the prevalence, the better we are able to make that informed decision," said Dr. Down, a developmental paediatrician with the Vancouver Island Health Authority.

Children with FXS typically show symptoms of developmental delay at about a year of age. Because they have problems with speech and language, behaviour and social interaction, many are also diagnosed with autism and/or attention deficit hyperactivity disorder by toddler or school age.

Currently there is no cure, and options to manage FXS consist primarily of medications to address behavioural issues, and services such as speech and occupational therapy. Genetic counseling, which allows families to make more informed choices about having another child, is also an important aspect of care for affected families.

Population screening

FXS is an inherited condition passed from mother to baby. Mothers with the FXS allele may have the pre-mutation, meaning they have few or no symptoms. Or they may have the full mutation, meaning they too may have some degree of intellectual disability.

About 25 percent of families will have another child before the first child is diagnosed. That's one reason why population screening for FXS has become a popular topic in genetics circles, despite some debate about the most appropriate timing. Newborn screening prevents a delayed diagnosis and provides access to early intervention services. Alternatively, screening women of reproductive age lets them know whether they are FXS carriers before becoming pregnant.

While there may be benefits to each approach, more Canadian data is needed before Canadian universal screening guidelines can be implemented, said Dr. Gudrun Aubertin, study co-principal investigator and clinical geneticist with the Vancouver Island Health Authority.

"Is there a benefit to identifying FXS at a younger age and is there the ability to really serve those families? The idea of screening reproductive-aged women would be very different. You would be screening primarily for the pre-mutation but the reproductive decision-making would be for women. You wouldn't be presenting a parent of a new baby with this potentially devastating diagnosis," said Dr. Aubertin.

Clinical trials

Clinical trials for medications that specifically target the underlying neurological pathways of FXS are also underway in Toronto and Vancouver, leading to hope there may someday be targeted treatment available. To provide equal access to families who may want to participate in clinical trials and, more importantly, to provide appropriate access to future treatment, decision-makers need more information about the number of FXS cases in Canada and other related demographic information.

"We saw the opportunity for this project as really the first step in working toward a future program or system to serve these families," said Dr. Aubertin. "It's going to inform all sorts of calculations in the future that need to be done as to how to develop and deliver services."

For the study protocol visit www.cps.ca → Surveillance → Canadian Paediatric Surveillance Program → Studies

eCPSP: Improving paediatric surveillance

Launched in November 2011, the CPSP's online reporting system is quickly becoming a popular option among program participants.

Nearly half of the program's 2,591 paediatricians now use eCPSP, which has helped to speed up the CPSP's reporting cycle by 43% from start to finish. With less monthly printing of reporting forms, there are also environmental benefits.

Participants in eCPSP receive a unique, secure link via e-mail to the monthly reporting form. No usernames or passwords are required, and they can respond from any location with internet access.

Participants also have access to case definitions, full protocols and up-to date study statistics through eCPSP. The electronic system means detailed questionnaires, when required, are available more quickly.

To sign up or for more information, contact cpsp@cps.ca or visit www.cps.ca → Surveillance → CPSP → Electronic Reporting.