Development of a Canadian Voluntary Population-Based Registry on Down Syndrome: Preliminary Results (2000-2002)

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Abstract

In 2000, the Down Syndrome Research Foundation initiated the formation of a Canadian voluntary population registry. The purpose of this article is to describe the background, development, and implementation of the pilot study for this registry. The pilot study collected responses to specific questions from participating families from across Canada, with a view to determining the feasibility of using both paper and web-based questionnaires for collecting the data. Though preliminary, these results indicate that families are interested in being part of the registry and its accompanying research. This registry, once established, could be useful as an access point for further research.

Down syndrome (DS) is characterized by the presence of additional chromosome 21 matter, and occurs in approximately 1 out of every 900 live births in Canada (Wyatt, 2000), making it the most prevalent genetic cause of developmental disabilities. Over the last 20 to 30 years, early detection of DS and surgical and medical management of congenital heart disease have led to a significant increase in the life expectancy of individuals with DS (Baird & Sadovnick, 1987). However, the syndrome is associated with a number of medical, cognitive, and developmental factors that tend to have a significant impact on health and quality of life. The associations between Down syndrome and specific health related problems - including cardiovascular and respiratory problems, childhood leukemia, and early onset Alzheimer's disease - have long received attention (Boker et al., 2001; Hasle, Clemmensen & Mikkelsen, 2000; Leonard, Bower, Petterson & Leonard, 1999; Stoll, Alembik, Dott & Roth, 1998; Van-Allen, Fung & Jurenka, 1999). Increasing attention is now also turning to broader issues, and these often involve interdisciplinary research.

To date, however, there is little in Canada in the way of an established infrastructure to collect and manage data on the overall health, education, and social status of individuals with DS. This is due, in part, to the fact that there is no national database that is specific to DS. Researchers interested in studying various aspects of DS have, in the past, relied primarily on support from local DS support groups in their respective areas to recruit interested participants. Often, this has not led to high response rates. To enhance the capacity of clinicians and health researchers to conduct research, the Down Syndrome Research Foundation (DSRF) initiated a modest first step in the development of such an infrastructure.

In April 2000, a pilot population-based registry was initiated. The long-term goals of this registry are to: 1) establish and maintain a voluntary registry of individuals (or guardians) interested in participating in specific projects or research pertaining to all aspects of DS; and 2) establish a longitudinal database on general health, social, and educational trends in individuals with DS. The DSRF also developed, in parallel, a Research Review Committee consisting of practitioners interested in DS, health researchers interested in population health, and a parent representative. The Committee was established to determine appropriate usage and access guidelines, security and privacy measures, and a process for giving access to interested researchers. We report here the results from the first survey that was conducted between April 2000 and April 2002.

Method

Development of the questionnaire

The primary data-gathering instrument was a multi-sectioned questionnaire that was developed by DSRF in consultation with primary health care providers, researchers (specializing in the areas of developmental medicine, disability research, and psychology), and families of individuals with DS. This initial questionnaire was pilot tested on families and health care professionals in the greater Vancouver area, and on DS support groups across the country. The final version of the questionnaire was amended in accordance with ideas that emerged during this testing.

Description of items in the questionnaire

The final questionnaire consisted of 6 sections: 1) basic demographic information; 2) medical conditions; 3) treatments and therapies; 4)

behavioural and/or psychological related conditions; 5) social, family, and cultural issues; and 6) an open-ended question to encourage comments on issues or concerns was included.

Distribution of questionnaire

The questionnaire was mailed by DSRF to community support groups across Canada. A web based format was also constructed and placed on the DSRF website. Information about the web-based format was included in the letter sent to community groups to give families the option of using either format.

Analysis

All responses were entered into a database management system, and standard basic qualitative and quantitative data analysis methods were used (e.g., Patton, 1990).

Results

Demographics and Medical/Health related problems

A total of 247 families participated in the registry. Of these, 213 questionnaires were completed by female parent/guardians, 14 by male parent/guardians, and 20 by both female and male parent/guardians. There were 35 web-based responses and 212 paper-based responses. The web-based forms were used mostly by families living in Ontario and BC. Table 1 shows the general demographic characteristics of the participants. Table 2 summarizes the medical problems reported for the participants of all ages.

Table 1. Demographic Characteristics of the Participants

Characteristic	*% of all participants*
Gender	
Male	56%
Female	44%
Age group	
0-4 years	14%
5-12 years	45%
13-18 years	22%
19 years and over	19%

cont'd

Table 1. (Cont'd.)

Characteristic	*% of all participants*
Residential status	
Living with parents	96%
Living on own	1.3%
Living with roommate/spouse	1.8%
Living in a group home	.9%
Geographical area	
British Columbia	29%
Alberta	10%
Saskatchewan	1.6%
Manitoba	2%
Ontario	50%
Quebec	5%
New Brunswick	.8%
Nova Scotia	.4%
Prince Edward Island	.4%
Yukon	.8%

Table 2. Percentage of Reported Medical/Health Problems

Medical / Health Problem	*% of all participants*
Hearing	54%
Cardiac	49%
Visual	43%
Respiratory	38%
Dental	31%
Gastrointestinal	30%
Sleep	26%
Behaviour	19%
Thyroid	16%

Treatments and therapies

Sixty-seven percent reportedly had been involved in an infant development program. Almost half had been involved in Physical Therapy (49%), and 45% in Occupational Therapy. Interestingly, only 6% reported having been involved in Speech and Language Therapy.

Involvement in exercise and recreational program. Forty percent of individuals between the ages of 5 and 65 were involved in a regular exercise program. Fifty-seven percent were involved in a regular social or

recreational program. Across all age groups, there was more involvement with social/recreational programs than with regular exercise programs.

Employment. Over 40 people had some vocational or volunteer experience, typically involving manual or basic clerical labour (e.g., in restaurants, bookstores, coffee shops, recreational and community centres). Several respondents described situations of steady and/or long-term employment. One or two responses described their son or daughter as being too young for employment, but noted that the prospects of volunteering were already being considered.

Qualitative analysis of issues and concerns

Eighty parents/guardians (37%) completed the final question highlighting major issues and concerns in relation to their son/daughter.

Health. In the 0-4 and 5-12 age groups, the primary issues identified were related to feeding and toilet training. In the 13-18 age group, issues regarding behaviour problems, family stress, society's perception of the individual with DS, sexual development, and finding primary health care professionals who were experienced with the needs of young adults with DS were identified as concerns. In the over 19 age group, the main concerns included eating habits, weight gain, speech problems, and concerns about sexual activity and birth control.

Education. In the 0-4 age group, the primary issues were expectations of schools and educational goals. In the 5-12 age group, a concern identified was a lack of qualified teachers and supports. Pressure to put their child into a segregated classroom was also a concern raised by a number of parents. In the 13-18 age group, lack of appropriate programming, and lack of understanding of inclusion were predominant issues. In the over 19 age group, the principal need identified was how to find a balance between educational expectations and necessary life skills.

Age specific issues. In the 0-4 age group, issues of concern were attitudes of professionals, lack of support and funding for services, and expectations for the future. In the 5-12 age group, two main issues were identified: lack of support and funding for services, and social development and friendships. For those 13-18 years old, the main issues identified were planning for post school options and making and maintaining friendships. In the over 19 age group, the main issues identified were independent living, employment, social development, and sexuality.

Discussion

The main objective of this pilot project was to test whether or not it is feasible to establish an infrastructure to collect and manage data on the overall health, education, and social status of individuals with DS across Canada. The results presented here indicate that this is indeed feasible.

The largest number of responses came from the print based version of the questionnaire. Since the questionnaire did not have any responses related to socio-economic status or accessibility to the internet, we cannot determine whether these factors played a role in the number of responses received from the Web based forms. Anecdotally, several families did report that security and confidentiality were of concern in using the internet. In order to increase the security of the registry, a two-part security and registration process has now been instituted.

In terms of responses from across the country, the largest number were received from Ontario and British Columbia - both provinces where the parent support groups were instrumental in distributing the forms and providing additional information for parents. Given that such support groups generally involve parents with children who are between birth to approximately 16 years of age, it is not surprising that most of the responses came from families in this age range. Clearly, further outreach and information will be required to garner support from families in other provinces as well as parents who have children in the older age range.

In general, the information obtained from the registry corroborates and/or expands upon current reports in the literature about medical and health of individuals with DS. For example, from birth to preschool age, a large percentage of children were reported to have medical problems, with cardiac conditions reported most frequently. The incidence of heart disease reported in the literature is 35-50% (Baird & Sadovnick, 1987). Recently, Freeman et al. (1998) reported the results of a population-based study of infants born with DS. They found that 44% of infants had congenital heart defects. Interestingly, in spite of the number of medical problems of the individuals with DS, the issues of concern to parents were principally not medical in nature but rather related to attitudes and expectations of health professionals who were involved with their child and issues of access to services. The attitude of professionals towards children with DS is an area that continues to require attention and can perhaps be further investigated using this registry.

In the school age group, medical problems continue to be a concern, with hearing problems reported most frequently. It is estimated that more than 50% of individuals with DS have some degree of hearing loss (Balkany, Mischke, Downs & Jafek, 1979). In this survey, we did not distinguish between hearing loss and hearing problems (such as ear infections). Further research using this database will help to identify the incidence of hearing loss and ear infections, as well as the impact on speech/language development and on behaviour and learning.

Another finding was the relatively high percentage of sleep and behaviour problems reported in the school age group. There is a high incidence of both obstructive (Marcus et al., 1991) and central (Ferri et al., 1998; Levanon, Tarasiuk & Tal, 1999) sleep apnea reported in children with DS. However, the relationship between sleep apnea and behaviour has yet to be elucidated. In our survey, 23 individuals reportedly had both sleep and behaviour problems.

Parents/guardians of children in the school age group strongly voiced concerns regarding lack of access to services and support. Social development and lack of or difficulty in making and maintaining friendships were clearly identified as major needs, and this raises questions about the impact of these issues on the family. Leonard et al. (1999), in their survey of parents of school aged children, suggested that chronic non life-threatening conditions (such as hearing and bowel problems) impose a burden on families although they do not threaten quality of life. In contrast, our findings suggest that quality of life may be affected, not due to medical problems, but to lack of support and friendships. This view is supported by Siklos (2002), who, in a recent needs survey of parents of children with DS (mean age of child 11 years, SD 5 years), reported that one of the most important needs of parents was "for my child to have friends of his/her own" but that this same need was mostly rated as being only "partly met" or "unmet."

In the 13-18 age group, hearing, dental, and visual problems were reported most frequently. Recently, Hennequin, Allison and Veyrune (2000) evaluated oral health problems in children with DS (mean age of 9.6 years) and found that in comparison with their siblings, children with DS had a greater frequency of oral health problems. In terms of visual deficits, recent reports in the literature suggest high rates of vision related problems, such as strabismus (Haugen & Hovding, 2001) and amblyopia (Tsiaras, Pueschel, Keller, Curran & Giesswein, 1999) in children with DS. Research on early identification of visual problems and impact of visual problems on learning

are obvious issues for further study. During these years, social factors appear to take precedence, with integration, life skills, employment, and friendships identified as key issues. Several authors have shown that, in comparison to their non-disabled peers, young adults with Down syndrome lack peer friendships (Buckley & Sacks, 1987; Carr, 1995).

In the last age group, dental and vision problems were reported most frequently. In a longitudinal study (8.5 years) of individuals with DS aged 21 to 40 years, Gabre, Martinsson and Gahnberg (2001) reported that there was increased bone loss and that these individuals were at higher risk for impaired oral health. Issues for further study here include identifying factors that result in increased risk, strategies for maintaining good oral health practices, and studying the impact of providing education/intervention on oral health. In terms of parental needs, concerns related to sexuality, independence and the future of the individual with DS were identified. Access to, and information about, additional programs were also identified as strong concerns across the lifespan.

The initial response received from the registry is promising. Development of proactive strategies to reach families in the central provinces and territories are now in progress. The registry appears to be a useful access point to develop research questions with a multidisciplinary perspective.

Acknowledgement

The authors would like to thank all the families who participated in this project.

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