Journal on Developmental Disabilities
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Review Board

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Journal on Developmental Disabilities is published twice yearly by the Ontario Association on Developmental Disabilities (OADD). The Association has established the following objective for the journal:

> *To foster and promote thoughtful and critical dialogue about relevant issues in the field of developmental disabilities, including those broader social issues that impact on persons with developmental disabilities.*

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Each issue features a selection of research, conceptual, review, and informational articles. An issue may have a central theme. Reviews of relevant books, movies, websites, software, and other resources are welcomed, as are letters to the editor. In this way we strive to collectively enrich our understanding of issues, enourage stimulating debate among those working in the field, and improve services.

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100 College St, Room 511, Toronto, Ontario Canada M5G 1L5
phone: (416) 657-2267
fax: (416) 657-2267
Website: *www.oadd.org*
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Thinking About Research within a Logic Model Framework with Quality of Life as a Long-term Outcome

Guest Editors

Barry Isaacs
Surrey Place Centre

Kristine Ericson
Surrey Place Centre

People with developmental disabilities represent a broad and heterogeneous population. The diversity of the seven papers appearing in this issue of the *Journal on Developmental Disabilities* is only a tiny indication of this fact. One of the problems for service providers is integrating the expanding literature in the area to develop and maintain state-of-the-art services. Organizing research within a *logic model* framework that is used in program evaluation and inserting improved quality of life as a long-term outcome within that framework, is useful for this purpose. This seems reasonable since improving quality of life is one of the ultimate goals of the work that all of us do in the field of developmental disabilities.

In a logic model for program evaluation, important aspects of people being served, the services strategies being used, and the outcomes of service, are identified and linked together (Cooksy, Gill & Kelly, 2001; Hernandez, 2000; Julian, Jones & Deyo, 1995). Knowledge about these three aspects of service, and the links between them, is necessary for a variety of purposes such as: 1) identifying assumptions made in service, 2) making informed data-supported decisions about service improvements, and 3) identifying important needs that are not being met in service. An example will serve to illustrate these points.

An initial logic model for a fictitious behavioral service is shown in Figure 1. In a real evaluation, this flow chart would likely be constructed through a review of service policies and procedures and interviews with the service providers. This flow chart can then be used to identify and

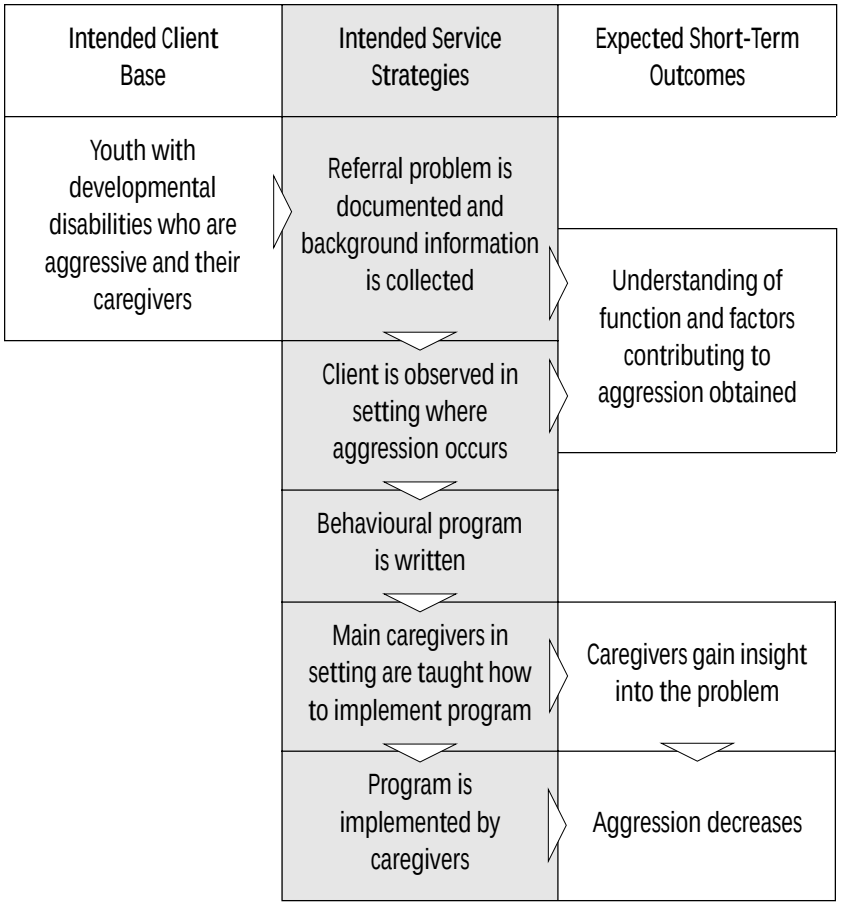

Figure 1
Logic model for a ficticious behavioural service

scrutinize underlying assumptions in the service, and to formulate relevant questions for evaluation.

Some of the service assumptions would already be apparent to the service providers. In this example, these might include: 1) the collection of background information and client observation leads to a proper understanding of the problem, 2) caregivers actually do implement the program as intended, or 3) both caregiver insight and program imple-

mentation contribute to a decrease in the problem behavior. The service providers, however, may not be aware that it is assumed that caregivers gain insight into the problem simply through being taught the behavioral program. A breakdown within any one of the above assumptions is likely to drastically affect the goal of decreasing aggression.

By examining Figure 1, and by considering the identified assumptions, a set of key questions and variables for the evaluation can be identified. Examples of questions would be: are the expected outcomes being achieved? And, how do factors such as caregiver understanding of the problem, treatment integrity, and client characteristics influence outcome? Examples of variables would be a standardized record of referral problems and background information, pre-and post-measurements of the problem behavior, pre- and post-measurements of the caregivers' understanding of the problem, and assessments of caregiver learning and implementation of the treatment program.

If operationalized properly, data on these variables would yield results that demonstrate whether or not what is intended is actually happening in service, and support recommendations for service improvements. For example, a proper analysis might tell the service providers that caregivers who have a good understanding of the problem are also better at learning the treatment program, show better follow-through with treatment, and experience better outcomes for their children. This finding might lead to a future focus on education about the problem behavior to enhance learning and follow-through of treatment programs and improve outcomes.

By linking outcomes with background information it may also be discovered that some clients are not benefiting from service because important issues vital to the resolution of the problem are not being addressed. Depending on what these needs are, service planners might consider addressing them within the current service, linking clients with other services to address the needs, or advocating for the establishment of new services to address these needs.

The kind of information required in logic models of specific services is the same as that needed in the planning and understanding of services designed to improve quality of life for people with developmental disabilities. To improve quality of life for people with developmental disabilities we first must be able to describe the population and the systems with which it is involved. Who and how many people are included in the population? What needs exist? And, what factors stand in the way of improvements? The papers by Hélène Ouellette-Kuntz and Dana Paquette, Maria Medved and Maire Percy, Heather Grant, William Picket, Miu Lam, Michael O'Connor and Hélène Ouellette-Kuntz, and Oded Freidman and Ivan Brown all provide information that contributes to one or more of these questions. Information of this sort is vital for service planning.

Strategies to improve life for people with developmental disabilities must be based on need, with desired outcomes in mind. Beginning with a discussion of need, Jennifer Nachshen and Leah Anderson provide important information about parent advocacy, a potentially powerful means of improving quality of life. Furthermore, the Parent Advocacy Scale they test asks specifically about expected outcomes. Robert King and Robert Barnett compare the response of two systems – Health versus Community and Social Services – to the needs of two sisters and discuss how their quality of life was influenced by these systems. Maria Medved and Maire Percy outline both interventions to meet the needs of individuals with Prader-Willi syndrome and what is known about the outcomes that may be expected from these interventions.

Expected service outcomes should follow logically from knowledge about the population and the service strategies employed to meet the needs of that population. If we view quality of life as something that improves only after some or many other, specific things change, then in most cases it becomes a long-term outcome influenced by a set of short-term outcomes. Many factors that influence quality of life are proposed in the literature. These factors include, housing, recreational opportunities, services, education, personal relationships, levels of inclusion and social policy. In their paper, Robert King and Robert Barnett discuss the

influences of many of these factors on the quality of life of two sisters. Although the other papers in this volume do not present service outcome data, every paper contributes to the important groundwork necessary for good outcome research. As demonstrated in the logic model discussion, a good understanding of outcomes is not possible without knowledge about populations and service strategies. One of the goals in the developmental services sector should be to produce more and better outcome research.

Considering research within the logic model framework, and identifying improved quality of life as a long-term outcome within that framework, is useful for integrating literature from a wide range of areas. Placed within this context, research is given a common goal, and the importance of work that even indirectly affects quality of life is not lost. By placing large bodies of literature within this context, important issues such as gaps in knowledge, service needs, and the relevance of outcomes can be addressed in common terms.

Finally, the papers in this issue of the *Journal on Developmental Disabilities* employ a wide range of methodologies. This methodological diversity parallels current thinking in program evaluation and the use of logic models. Single case experiments can be as important and powerful as control group studies depending on the question that is being asked, and qualitative and quantitative methods are seen as complimentary, rather than incompatible. In the same way, methodological flexibility is critical if we are to fully understand how the services we provide influence the quality of life of people with developmental disabilities.

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The Prevalence of Developmental Disabilities in Lanark County, Ontario

Hélène Ouellette-Kuntz
Queen's University
and Ongwanada

Dana Paquette
Queen's University

Abstract

Knowing the prevalence of developmental disabilities is essential to policy planning. In 1996, an Ontario Government Policy Analyst with MCSS (Ministry of Community and Social Services) estimated the overall prevalence by combining MCSS and MET (Ministry of Education and Training) administrative datasets. This paper compares the MCSS approach to an agency survey conducted in Lanark County. The comparison revealed that both approaches led to gaps in estimating the number of people with developmental disabilities receiving services and supports. When the MCSS and agency survey figures were combined, the "administrative" prevalence in Lanark County was estimated at between 0.8% and 0.9%. Implications for policy planning are highlighted.

Understanding the magnitude and nature of a population in need of services is essential to planning. Persons with developmental disabilities constitute such a population. These individuals have "a condition of mental impairment present or occurring during [their] formative years that is associated with limitations in adaptive behavior" (Developmental Services Act, 1993, p.1). In Ontario, the parents of young children with developmental disabilities (0–5 years) are entitled to financial support in the form of the Handicapped Children's Benefit (HCB), older children (4–21 years) are entitled to an integrated education and adults with developmental disabilities (20 years and older) are entitled to a pension (formally under the Family Benefits Allowance program, FBA, now under the Ontario Disability Support Program, ODSP). Furthermore, a wide array of supports funded by the Ministry of Community

and Social Services (MCSS) have been developed to meet the special needs of this population. Such supports include residential, occupational, vocational, behavioural and family support services.

Despite the extensive array of services designed for persons with developmental disabilities in Ontario, no central registry of these individuals is available. As a consequence, the actual number of individuals with developmental disabilities in Ontario receiving or in need of services is not known. The lack of a registry results in an inability to effectively monitor trends, including shifts in causative factors and need for services over time.

Trends can be reflected in changes in prevalence rate. Prevalence is an epidemiologic term used to designate the number of individuals with a given condition in a given population at a designated time. Prevalence can be "administrative" or "true." The "administrative" prevalence is the proportion of individuals in a catchment area who are receiving services or identified to service providers as requiring services. The "true" prevalence is the proportion of individuals who upon screening of a population are identified as meeting the diagnostic criteria.

In 1988, a Working Group on the Epidemiology of Mental Retardation in Canada concluded that the prevalence of developmental disabilities was at least 0.8% of the population with about equal rates for mild (IQ=50–70) and severe (IQ<50) mental retardation (MR) (Health & Welfare Canada, 1988). Since, the 1988 report, two papers reporting on the "administrative" prevalence of developmental disabilities in Ontario have been found (Nuyen, 1996; Brown, Raphael & Renwick, 1997).

An Ontario government policy analyst estimated that the overall prevalence of "Ontarians labelled as having a developmental disability" was only 0.56% (Nuyen, 1996). This estimate of "administrative" prevalence was derived from a combination of MCSS and Ministry of Education and Training (MET) administrative datasets using the following assumptions:

1. All children between 0 and 5 years of age labelled as having a developmental disability receive HCB.
2. "75% of HCB recipients are labelled as having a developmental disability, for lack of concrete statistics." (p.10)
3. Children aged 0–18 years residing in Schedule I and II facilities are not captured in either HCB or MET statistics.
4. All individuals between 6 and 21 years of age who do not reside in Schedule I and II facilities and who are labelled as having a developmental disability are counted by adding students identified by MET as being "mildly developmentally disabled", "developmentally disabled" or "multi-handicapped" and 5% of those identified by MET as "learning disabled".
5. All adults age 20 years and older labelled as having a developmental disability receive FBA and are labelled as having a "special delay in development", "mild mental retardation", "other unspecified mental retardation", "unspecified mental retardation" or "Down syndrome".
6. The MET figures provide an overestimate.
7. There is double-counting in the 20–21 year age group.
8. The overestimated figures from MET and the double-counting in the 20–21 year are group is offset somewhat by the probable underestimate caused by not adding recipients of Special Services at Home and Community Supports.

The final estimate was obtained by adding items 2, 3, 4 and 5 from the above list of assumptions.

The Centre for Health Promotion (Brown et al., 1997) estimated the number of adults in Ontario who have a developmental disability and who were receiving services funded by MCSS at one point in time in 1995. These authors randomly selected MCSS funded service organizations across Ontario that provided services to geographic areas that represented 56% of the general population of the province, and contacted

each agency by telephone or fax to obtain numbers of people receiving services. From these totals, they computed estimates for the whole of the Ontario adult population. Brown et al. (1997) estimated that 25,473 adults with developmental disabilities in Ontario were receiving services funded by MCSS in 1995. This figure is consistent with that reported by Nuyen.

A critical review of the recent international literature on the prevalence of mental retardation in children (Roeleveld, Zielhuis & Gabreels,1997) concluded that the prevalence of severe MR in children (5 to 19 years) is stable across populations in developed countries at 0.3% to 0.4%. Roeleveld et al (1997) went on to demonstrate that the prevalence of mild MR among children (5 to 19 years) varies greatly across study populations with an average "true" prevalence of 3% and an "administrative" prevalence of 0.5%. In the case of severe MR, Roeleveld et al. concluded that in developed countries the "administrative" prevalence can be said to be the "true" prevalence. The conclusions suggest that among children 5–19 years old, the overall "true" prevalence is 3.3% or 3.4% for all disability levels combined. The "administrative" prevalence however for this age group (all disability levels combined) would tend to be closer to 0.8% or 0.9%.

While national and international reviews have suggested that in Canadian jurisdictions, the "administrative" prevalence of developmental disability should be 0.8% to 0.9%, Nuyen (1996) reported a much lower figure (0.56%). The principal limitation to the approaches used by Nuyen (1996) and Brown et al. (1997) is the inability to use unique identifiers for each client served thereby creating the need to ignore overlap or rely on unproven assumptions regarding overlap among the datasets being combined. An "administrative" prevalence can also be obtained by conducting agency surveys and/or developing central registries of individuals served. These techniques reduce error by removing the need to estimate the extent of overlap between datasets.

The purpose of this paper is to use data from a survey of agencies in

Lanark County, Ontario to determine the size of the population with developmental disabilities requiring services. The prevalence estimates for Lanark County obtained by Nuyen (1996) are compared and combined to the agency survey data in order to evaluate the usefulness of both approaches.

Method

Setting

Lanark County, located in the eastern part of Ontario, has a population of 59,845 people (Statistics Canada, 1997). It is rural with small urban centers. One of the province's largest remaining institutions for persons with developmental disabilities, Rideau Regional Centre, is located in Smiths Falls (a small urban centre with a population of 9395). This institution has a population of approximately 700 residents. The population of Lanark County is primarily white, English speaking, with a roughly equal sex distribution in each age group.

The agency survey approach

All agencies for persons with developmental disabilities in the county were involved in the survey of agencies. Eight agencies, including Rideau Regional Center, contributed information to the study. For Rideau Regional Center, only clients who are originally from Lanark County were included. The investigators worked with service providers to systematically identify without duplication every individual with a developmental disability known to them. Hence, as in the MCSS estimates, the prevalence figures reflect an "administrative" prevalence. One strength of this method is that because each person affected is identified along with details of service requirements, the figures reflect actual numbers of individuals as opposed to "estimated" numbers which are reported by Nuyen (1996) and Brown et al. (1997).

Between November 1994 and July 1995, staff from each agency in Lanark County identified the clients for whom they would be responding and proceeded to collect the information needed. Since some individuals receive services from more than one agency or from more than one

program within an agency, the algorithm found in Figure 1 was used to determine which agency should respond for which client. Basic demographic data was obtained through a telephone interview with the client's family or other primary caregiver. This included the client's date of birth, gender, mother tongue, municipality of residence, living arrangement, sensory impairments, mobility status and communication abilities. Detailed information was subsequently obtained during face to face interviews involving the clients and/or their families and other caregivers. The more detailed information focused on current and future service needs as well as barriers to unmet needs. The interview also included the *Katz Activities of Daily Living Scale* (Katz, 1983) and the *Reiss Screen for Maladaptive Behaviours* (Reiss, 1987). The survey protocol is available from the first author upon request.

Comparing the two approaches

For comparing MCSS estimates and agency survey numbers, MCSS age categories and support designations were used. The numbers are compared for those less than 5 years of age, those who are 5 years old, those between 6 and 21 years of age and those 20 years and older. For the first two groups (<=5 years old) the focus is on Handicapped Children's Benefits' recipients. For the 6 to 21 year old group the focus is on MET enrollments and for the older group, the focus is on Family Benefits Allowance recipients. Since the agency survey includes exact numbers and information on supports received, the MCSS estimated numbers for each age group in Lanark County can be verified and the proportion of individuals not known to service providers can be estimated. Though all categories of individuals are estimated at the provincial level, estimates of the number of children in Schedule I and II facilities and of the number of children 5 years of age receiving Handicapped Children's Benefits are not provided at the county level.

Results

Using 1994 to 1996 data, Nuyen (1996) estimated that 412 individuals in Lanark County were labelled as having a “developmental disability”.

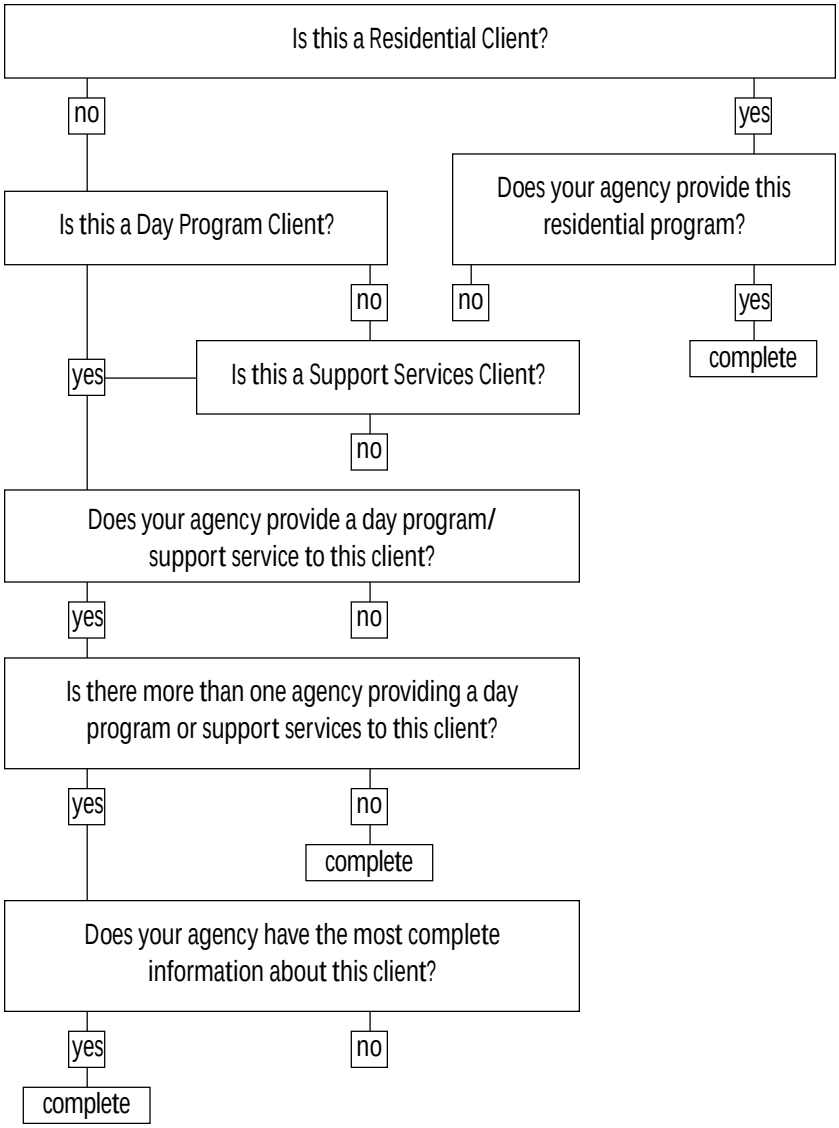

Figure 1
Algorithm for Creation of Client Database

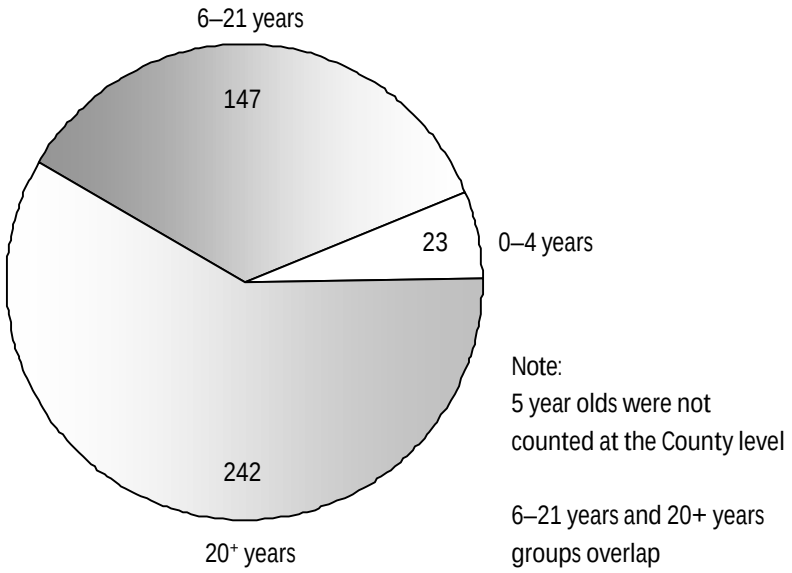

Figure 2
Age distribution of individuals in Lanark County labelled as having a "developmental disability" (Nuyen, 1996)

For a mid-census* population of 57,325, this suggests a prevalence of 0.72%. Figure 2 shows the estimated numbers for the different age groups.

Using agency surveys conducted during the same timeframe (November 1994 and July 1995), Ouellette-Kuntz (1997) obtained basic information for 372 individuals with developmental disabilities receiving services in Lanark County. This represents an overall "administrative" prevalence of 0.65% (using mid-census population estimate). Of the 372 individuals, 90 were children 5 to 19 years of age (Table 1), suggesting a prevalence rate of 0.76% for this age group.

As shown in Table 1, the male to female ratio is approximately 1.4:1.

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* Average of 1996 Census and 1991 Census

Table 1

Demographic distribution of individuals with developmental disabilities in Lanark County known to service providers

Age in years	#	%		#	%
0–4	24	6.5	20	5	1.3
5	10	2.7	21	2	0.5
6–18	75	20.2	22+	251	67.5
19	5	1.3			
Gender	**#**	**%**		**#**	**%**
Male	214	57.5	Female	158	42.5
Living Arrangement	**#**	**%**		**#**	**%**
with own family	142	38.1	foster home	16	4.3
group home	52	14	semi-independent living	14	3.8
institution	51	13.7	family home program	13	3.5
independent living	43	11.6	other	22	5.9
nursing home	19	5.1			
Primary Day Program	**#**	**%**		**#**	**%**
none	60	16.1	Developmental program	29	7.8
MET	83	22.3	Day care	13	3.5
Work experience	43	11.6	Supported employment	11	3
Life skills training	41	11	Other	59	15.9
Activity program	33	8.9			
Financial Assistance	**#**	**%**		**#**	**%**
FBA *	172	67	Special Services at Home †	61	43
HCB *	61	53.5	Employment ‡	15	5.7
own family	53	14.2			

*% for FBA and HCB are % of those eligible

† % of Special Services at Home is % individuals living with own family

‡ % for employment is % of adults

Over a third of the individuals live with their families. While 22.3% are in school, 16.1% are not engaged in a day program. Less than 6% derive financial support from employment. Only 67% of adults who receive services also receive FBA and only 53.5% of children receive HCB.

Although intelligence scores were not available, the high degree of disability among some of these individuals is apparent when reviewing the presence of additional disabilities and maladaptive behaviors (Table 2), and levels of dependence in activities of daily living (Table 3). Almost 20% do not communicate verbally. More than 1 in 6 have the following disabilities: mobility impairment, visual impairment and seizure disorder. One in 10 has a diagnosed psychiatric illness. Maladaptive behaviours, primarily in the form of aggression, are common. As expected, children are likely to exhibit dependence in activities of daily living. A significant number of adults continue to require assistance with such activities as bathing and dressing (Table 3).

Table 2
Additional Disabilities and Maladaptive Behaviors

	affected (n=372)			affected (n=347)	
Condition	#	%	Condition	#	%
mobility impairment			chronic medical problem	107	30.8
all	68	18.3	psychiatric illness		
cerebral palsy	30	8.1	all	37	10.7
seizure disorder	65	17.5	autism	10	2.9
visual impairment	67	18	maladaptive behaviors		
hearing impairment	48	12.9	physical aggression	92	26.5
“Do not communicate			verbal aggression	102	29.4
verbally”	73	19.6	destruction of property	54	15.6
			self-injury	46	13.3
			self-stimulation	63	18.2
			sexual problems	28	8.1
			withdrawal	76	21.9
			other	33	9.5

Table 3

Level of Independence in Activities of Daily Living (96 children, 249 adults)

Activity of Daily Living	requiring assistance		dependent	
	children	adults	children	adults
bathing	38 (40%)	87 (35%)	24 (25%)	37 (15%)
dressing	33 (34%)	49 (20%)	19 (20%)	33 (13%)
feeding	13 (14%)	31 (12%)	16 (17%)	17 (7%)
continence	10 (10%)	23 (9%)	27 (28%)	31 (12%)
toileting	15 (16%)	26 (10%)	28 (29%)	24 (10%)
mobility	20 (21%)	26 (10%)	10 (10%)	22 (9%)

Table 4

Combining Lanark County Database Numbers (Ouellette-Kuntz, 1997) and Numbers provided by MCSS=s estimates (Nuyen, 1996)

Age in years	Nuyen 1996	Ouellette-Kuntz 1997	Both Nuyen and Ouellette-Kuntz	Nuyen only	Ouellette-Kuntz only	Combined Estimate
0–4	23(a)	24(b)	23(c)	-(d)	1(e)	24(f)
5	-†	10(g)	-	-	10(g)	10(g)
6–18	147(h)	87(i)	83(j)	64(k)	4(l)	151(m)
20+	242(n)	251‡(p)	223(q)	19(r)	28(s)	270(t)
total	412(u)	372 (v)	329 (w)	83 (x)	43(z)	455(z)

Calculations

0-4 years	6-21 years	22 years+	Total
c= smaller of a and b	j= smaller of h and	p*=p - §	u= a+h+n
d= a-c	i	q=smaller of p*and n	v=b+g+i+p
e= b-c	k= h-j	r= n-q	w=c+j+q
f= c+d+e	l= i-j	s= p*-q	x=d+k+r
	m= j+k+l		y=e+g+l+s
			z=f+g+m+t

† Nuyen does not report an estimate for 5 year olds

‡ only those 22 years and older are counted

§ those identified in the agency survey as over 22 years and not receiving FBA)

Table 4 which combines the agency survey figures with those provided by MCSS and MET datasets suggests that the 'administrative' prevalence in this county lies between 0.8% and 0.9%. This combined figure reflects the numbers obtained through the survey of agencies and 64 "additional" children suggested by the MET dataset and 19 "additional" adults suggested by the FBA rolls but who are not currently supported by agencies for persons with developmental disabilities.

Discussion

At the county level, the MCSS approach underestimates the number of children under 6 years receiving supports and counts many school aged children not known to service providers. Approximately a third of children under the age of 6 who receive services for persons with developmental disabilities are not counted when 75% of HCB recipients is used to provide an estimate. MET data however estimate almost twice as many school aged individuals than are known to service providers outside of the education system.

The administrative prevalence for children (5–19 years) with developmental disabilities found in Lanark County (0.76%) is in keeping with Roeleveld et al.'s review. While only 90 children (5–19 years) were identified by service providers, Nuyen (1996) reported 147 children (6–21 years) identified by the MET in Lanark County in 1994. While the MCSS data does not allow direct comparison for different age groups, these two figures suggest that only approximately 60% of children identified in the schools receive specialized services from agencies outside the school. The remaining 40% likely constitute what Nuyen describes as the 'transitory' component of the population; those more mildly affected who do not typically receive specialized supports upon leaving school.

Data on level of mental retardation is infrequently used in assessments of support eligibility or individualized program development in Ontario. It is therefore difficult to determine what proportion of the individuals

identified have severe versus mild MR. If Roeleveld et al.'s conclusion regarding the "true" prevalence of mild mental retardation is accurate (3%), as many as 2.2% of children in Lanark County 5 to 19 years of age have an IQ between 50 and 70 and are not known to service providers including MET.

As for estimating the number of adults affected, the use of FBA data suggests that a small proportion of adults receiving disability pensions are not receiving other supports specifically for persons with developmental disabilities (19/242, 7.9%). This group likely consists of more mildly affected individuals.

Conclusion

The usefulness of the current prevalence estimate of 0.72% for planning services and resource allocation in Lanark County is limited. Firstly, since the figure of 0.72% is an estimated 'administrative' prevalence, it does not provide an indication of currently unmet need or future need for support. Secondly, because the figure is derived from a mathematical combination of various datasets, it provides no information about causes of the disability and level of service need, and very limited information about the age of the population.

In Lanark County, the agency survey identified an additional 43 individuals not accounted for in the prevalence estimate. These individuals represent a 10% increase from the MCSS estimated number of individuals. Applied to the province, the 10% increase in population size would represent 6,167 individuals.

Without an accurate registry of individuals receiving or in need of supports from the ministries of Community and Social Services or Education and Training because of a developmental disability, this population is at risk of not being adequately served. As stated by Nuyen (1996), "the (prevalence) rate...its components, and its impact on demand for services is necessary for policy development,... (for making) decisions

around resource allocation, including the equitable distribution of resources across the province…(and) at the local level it can also assist community planning groups as they work towards a comprehensive community-based service system." (p.1)

The MCSS approach for estimating the prevalence of developmental disabilities fails to provide accurate data sufficiently detailed to assist in resource allocation and in developing service systems. While service providers' groups such as the one in Lanark County have resorted to using agency surveys to obtain this information for their own jurisdictions, such surveys are expensive and they are likely to miss individuals who receive financial assistance or special education supports because of their disability but who do not at a specific point in time receive services from agencies for persons with developmental disabilities. Being able to combine agency client information with MET student information and HCB and FBA recipient information would go a long way towards accurately determining the "administrative" prevalence. In order for such information to assist in resource allocation and service system development, detailed information about each individual's needs is required.

Finally, both the MCSS and the agency survey approach capture only the "administrative" prevalence, the "true" prevalence is left unknown. Nuyen (1996) appears to suggest that the "true" prevalence is not of concern to policy makers and planners. Equating those "in need" with those "labelled as having a developmental disability", translates into up to 2.2% of individuals with IQs between 50 and 70 not being recognized as at risk and therefore in need of early intervention and supports in order to maximize their full participation and equality in society. Revealing the "true" prevalence would require applying a consistent assessment tool to a representative sample of Ontarians.

In conclusion, there is currently no evidence to suggest that the "true" prevalence of developmental disabilities in Ontario is less than 3% and that the "administrative" prevalence is significantly less than 1%. Until

accurate data are available, policy makers and service providers should continue to use these figures in planning for a comprehensive service system for persons with developmental disabilities.

Addendum

In December of 1999, the Ministry of Community and Social Services published a report which updates the 1996 estimates of "Ontarians labelled as Having a Developmental Disability". This most recent document estimates that there are 549 such individuals in Lanark County. An agency survey conducted by the author and colleagues in 2000 counted 573 individuals with developmental disabilities known to service providers in that county. These two figures, represent prevalences of 0.92% and 0.96% respectively (based on county population of 59,845; source: 2000 Canadian Almanac & Directory).

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Correspondence

Hélène Ouellette-Kuntz
Ongwanada
191 Portsmouth Avenue
Kingston, ON, Canada
K7M 8A6.
email: oullette@post.queensu.ca.

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Prader-Willi syndrome I: A Literature Review of the Genetic Causes and Physical Characteristics

Maria Medved
University of Toronto

Maire Percy
Surrey Place Centre

Abstract

The aims of this paper are three-fold. The first is to present an overview of the literature on the genetic causes of Prader-Willi syndrome (PWS). Chromosome deletions and uniparental disomy are highlighted as the most common mechanisms. PWS results from the loss of one or more genes normally expressed only by the paternally-inherited chromosome. One of the genes that is consistently silent in PWS is SNRPN. This is the small nuclear ribonucleoprotein particle-associated polypeptide N, which forms part of the spliceosomes in the brain. Recent advances in genetic testing and prenatal diagnosis are also discussed. The second aim of this paper is to review the physical characteristics associated with this syndrome, including the most salient feature of PWS, obesity. Certain physical characteristics may be the result of hypothalamic insufficiency. The third aim is to discuss recent treatment options for individuals with PWS and highlight associated ethical issues.

Prader-Willi syndrome (PWS) is a genetic disorder that exhibits considerable clinical variability. A central characteristic of people with PWS is an apparent insatiable appetite (hyperphagia) leading to severe overeating and the potential for marked obesity and associated health problems and premature death. In addition to hyperphagia, PWS can be associated with obesity and intellectual disability along with oppositional and obsessive-compulsive behaviour. Behaviour characteristic of PWS

is thought to be due to the effects of genetic defects resulting from chromosomal 15 abnormalities. The history of PWS is important, particularly to the field of genetics, for two reasons: one, it is the first recognized instance of uniparental disomy; and two, it is the first recognized instance of a genomic imprinting disorder (Nicholls, Knoll, Butler, Karam, & Lalande, 1989). In conjunction with its unique genetic etiology, PWS is an important disorder to understand so that treatments for this and other imprinting disorders can be developed.

Approximately 1 in 10,000 to 15,000 people are estimated to have PWS (Burd, Vesely, & Martsold, 1990). It seems to affect females and males equally, and it is not associated with any particular ethnic group, socioeconomic class or geographic region (Alexander, Van Dyke, & Hanson, 1993).

Since its first published medical description, clinicians and researchers have continued to seek clarification of diagnostic criteria and etiology (see Table 1). Currently, identification of PWS is based on clinical observation using an additive numerical system – the presence of a characteristic associated with PWS is allotted a score. Characteristics highly suggestive of PWS, called major criteria, receive one point whereas less suggestive criteria, called minor criteria, receive one-half a point (Holm et al., 1993). For a diagnosis of PWS from birth to 3 years, a total of 5 points is required, of which at least 4 must be from the major category. From age 3 to adulthood, a total of 8 points is required, of which at least 5 must be from the major category. In addition to major and minor criteria, there are supportive criteria that are not scored, but if present, increase the certainty of the diagnosis.

In the 1980s, many medical problems associated with PWS were described by various authors (e.g., Cassidy, 1984; Kriz, & Clonninger, 1981). In the last decade, much focus has been on exploring the genetic mechanisms of this syndrome (e.g., ASHG/ACMG Test and Technology Transfer Committee, 1996; Ohta et al., 1999; Saitoh et al., 1997; Sutcliffe, Han, Christian, & Ledbetter, 1997).

In this paper, we first explore the genotypic aspects of PWS including the genetic mechanisms and inheritance factors. This is followed by a description of associated physical features and an exploration of some of the underlying biological mechanisms. Recent treatment options and ethical and legal dilemmas associated with these, will also be discussed. In a second paper, we describe the behavioural and neuropsychological features of PWS (Medved & Percy, this volume).

Genetic Aspects

Genetic mechanisms

PWS can be caused by several different types of genetic mechanisms that result in the same phenotype. In this section, we present the four major genetic mechanisms that cause PWS. We begin with the most common mechanism, and end with the least common one.

Approximately 70% of cases of PWS are caused by what is called a chromosomal "deletion" – more specifically, a deletion of the proximal part of the long arm of chromosome 15 (15q11-q13). Individuals inherit a set of 23 pairs of chromosomes, one from each parent. The deletion associated with PWS is on the chromosome 15 inherited from the father. Although the maternal and paternal chromosome 15 may look the same, they differ due to a phenomenon called "genomic imprinting."

Genomic imprinting refers to a marking of genes that results in the expression of only one of two inherited copies. Whether imprinted genes are active or inactive depends upon the sex of the parent from whom they are inherited (Cassidy, 1993). One mechanism associated with genomic imprinting and gene inactivation is the attachment of a methyl group to certain bases in certain regions of the gene. Normally, certain genes in maternally-derived copies of chromosome 15 are inactivated, and only the corresponding genes in the paternally-derived chromosome 15 are expressed. When these paternal genes are missing, there are no active copies. In the case of paternal deletions of a part of chromosome 15, PWS results.

Table 1
Diagnostic Criteria for Prader-Willi Syndrome

The following criteria for a diagnosis of Prader-Willi Syndrome are based on Holm et al., 1993.) as listed in the Prader-Willi Syndrome (US) Website, www.pwsausa.org. Because infants and young children have fewer symptoms than older children and adults with PWS, the scoring system differs by age group.

Major Criteria *(Count as 1 point each)*

- ❑ Neonatal and infantile central hypotonia with poor suck, gradually improving with age.
- ❑ Feeding problems in infancy with need for special feeding techniques and poor weight gain/failure to thrive.
- ❑ Excessive (crossing two centile channels) or rapid weight gain on weight-for-length chart after 12 months and before age 6; central obesity in the absence of intervention.
- ❑ Characteristic facial features with dolichocephaly in infancy, narrow face or bifrontal diameter, almond-shaped eyes, small-appearing mouth with thin upper lip, downturned corners of the mouth (three or more of these characteristics required).
- ❑ Hypogonadism-includes any of the following, depending on age:
  a. Genital hypoplasia (in males: scrotal hypoplasia, undescended testes, small penis and/or testes; in females: absence or severe hypoplasia of labia minora and/or clitoris).
  b. Delayed or incomplete gonadal maturation with delayed pubertal signs after age 16 (in males: small gonads, decreased facial and body hair, lack of voice change; in females: no or infrequent menses).
- ❑ Global developmental delay in a child younger than 6 years; mild to moderate mental retardation or learning problems in older children.
- ❑ Hyperphagia (excessive appetite)/food foraging/obsession with food.
- ❑ Deletion 15q 11–13 (>650 bands, preferably confirmed by fluorescence in situ hybridization) or other appropriate molecular abnormality in this chromosome region, including maternal disomy.

☐ Sum of Major Criteria Points

Table 1 *(continued)*

Minor Criteria *(Count as ½ point each)*

- ❑ Decreased fetal movement or infantile lethargy or weak cry in infancy, improving with age.
- ❑ Characteristic behavior problems, temper tantrums, violent outbursts, and obsessive/compulsive behavior; tendency to be argumentative, oppositional, rigid, manipulative, possessive, and stubborn; perseverating, stealing, and lying (five or more of these symptoms required).
- ❑ Sleep disturbance or sleep apnea.
- ❑ Short stature for genetic background by age 15 (in absence of growth hormone intervention)
- ❑ Hypopigmentation-fair skin and hair compared with other family members.
- ❑ Small hands (less than 25th percentile) and/or feet (less than 10th percentile) for height age.
- ❑ Narrow hands with straight ulnar border (outer edge of hand).
- ❑ Eye abnormalities (esotropia, myopia).
- ❑ Thick, viscous saliva with crusting at corners of the mouth.
- ❑ Speech articulation defects.
- ❑ Skin picking.

☐ Sum of Minor Criteria Points

☐ Sum of Major and Minor Criteria Points

Supportive Findings

(The following are not scored but increase the certainty of a diagnosis of PWS.)

- ❑ High pain threshold.
- ❑ Decreased vomiting.
- ❑ Temperature instability in infancy or altered temperature sensitivity in older children and adults.
- ❑ Scoliosis or kyphosis (curvature of the spine).
- ❑ Early adrenarche (pubic or axillary hair before age 8).
- ❑ Osteoporosis (demineralization, or thinning, of the bones).
- ❑ Unusual skill with jigsaw puzzles.
- ❑ Normal neuromuscular studies.

Requirements for a Diagnosis of PWS

From Birth to Age 3

- ❑ Five (5) total points are required, of which four (4) must be from the major criteria list.

Age 3 to Adulthood

- ❑ Eight (8) total points are required, including at least five (5) from the major criteria list.

Most paternal chromosome 15 deletions that cause PWS are “de novo” deletions. In other words, the deletion has arisen as a new mutation during the production of sperm. Otherwise, the father's chromosomes are normal in his blood and in other tissues of the body. In these cases, the chance of PWS occurring again is very small.

The second most common genetic cause of PWS is uniparental disomy of chromosome 15. This occurs in 25–30% of cases. Uniparental disomy means that an individual has inherited two copies of a particular chromosome from one parent instead of one copy of the chromosome from each parent. In some cases of PWS, the individual receives two maternally-derived chromosome 15s, instead of one paternal and one maternal chromosome 15. Sometimes the individual receives two maternal chromosome 15s that are exactly the same (called isodisomy), or he or she receives two different maternal chromosome 15s (called heterodisomy). As of yet, it is unclear why disomy 15 occurs, although in some cases this results from the presence of two maternally-derived chromosome 15s in an ovum (egg) instead of one, a phenomenon that is called nondisjunction. (Nondisjunction of chromosome 21, which results in Down syndrome, is the most familiar example of chromosomal nondisjunction.) When compared to cases involving paternal deletions, individuals with PWS caused by maternal chromosome 15 disomy (and hence a missing paternal chromosome 15) tend to lack the typical facial phenotype, but have other characteristics of PWS (Cassidy, 1997).

The next genetic mechanism, an imprinting mutation, is relatively rare, accounting for about 1 to 2% of PWS cases. However, this is an important mechanism to understand, and test for, because transmission is not sporadic as are the mechanisms described above. In the case of an imprinting mutation, the father is a silent carrier of a deletion inherited from his mother. (The term silent carrier means that the father has no clinical abnormalities but carries an abnormal chromosome.) This inherited deletion prevents the resetting of imprinting in the male germline (i.e., in sperm) so that the father transmits a maternal-like imprint to

his offspring. If the father is a silent carrier, he has a 50% chance of having a child with PWS (Brondum-Nielsen, 1997). It is hypothesized that a small deletion in the region of the chromosome 15 gene that controls methylation is the cause of this particular imprinting defect (Saitoh et al., 1997). In the case of an imprinting defect, the phenotype is similar to that in individuals with deletions or disomy, although the frequency of microcephaly is lower.

The fourth mechanism, translocation, accounts for 1% of PWS cases (Cassidy, 1993). Translocations are structural abnormalities resulting from recombination between two different chromosomes. Translocations that cause PWS involve structural rearrangements of the pericentromeric region of chromosome 15 and result in absence or lack of expression of q11.2-q13 of the paternally-derived chromosome 15. (The term pericentromeric refers to the region around the centromere of the chromosome). Translocations often occur sporadically (i.e., in the absence of family history), but about 3 to 5% of cases of PWS caused by translocations are inherited. In the latter case, there is a 25% theoretical risk that an offspring will have PWS. Identifying the breakpoints associated with translocations is important to further research because breakpoints help define the region associated with PWS (Brondum-Nielson, 1997).

In short, there are four known genetic mechanisms that can result in PWS, with paternal deletions and maternal disomy accounting for the majority of the cases.

Relation between Prader-Willi syndrome and Angelman syndrome

It is of interest to mention Angelman syndrome (AS). AS is associated with abnormalities of the same section of chromosome 15 as PWS, but these affect the maternal chromosome 15 rather than the paternal chromosome 15 as in PWS (Glenn, Driscoll, Yang & Nicholls, 1997; Glenn, Porter, & Jong, 1993; Glenn et al., 2000). AS occurs when there is no active maternal copy of this region. About 70–80% of patients with AS have either a maternal deletion in 15q11-q13, mutations on the maternal

chromosome 15 that alter imprinting, or paternal uniparental disomy for the region. Another 20% or so of patients with clinical symptoms of AS have none of these three defects, but have mutations in the maternal chromosome 15 in one or more genes in the region that can be familial (Lalande, 1996). This fourth category of patients with AS have biparental inheritance for chromosome 15 and exhibit normal DNA methylation patterns. Such patients with AS are thought to result from mutations in a single gene. Mutations in the UBE3A gene have been found in a number of biparental patients with AS, strongly suggesting that UBE3A is the AS gene (Glenn et al., 2000). In the case of deletion, the mother has two normal chromosome 15s in her blood, so the deletion is presumed to occur in the ovum (egg). In AS, deletions account for 60 to 70% of the cases. The AS genotype results in severe mental retardation with absence of speech, hyperactivity, frequent paroxysms of laughter, epilepsy and ataxia.

Genetic structure in PWS region

Our knowledge about the genetic structure of the region of chromosome 15 that is associated with PWS continues to increase. Cassidy (1993) suggested there are about three or four genes which are deleted or inactivated in PWS. The list of such genes now includes (small nuclear ribonucleoprotein N (SNRPN), necdin (NDN), zinc finger 127 and IPW (Glenn et al., 2000). All of these genes are expressed by the paternally-inherited chromosome only.

SNRPN is the best described gene that is likely to cause some of the features of PWS (Glenn et al., 1993). This gene is primarily expressed in the heart and the brain. The exact function of this gene is unknown. However, there are indications that it may be involved in the splicing of transcripts (Donaldson, Chu, Cooke, Wilson, Greene, & Stephenson, 1994). SNRPN thus has the potential to affect the nervous system. As described in the next section, discovery of involvement of the SNRPN gene in PWS has led to a major breakthrough in postnatal and prenatal diagnosis of PWS and AS.

In 1997, Sutcliffe and colleagues (Sutcliff et al., 1997) reported that the necdin gene (NDN) is an excellent candidate gene for involvement in the PWS phenotype. Previously, evidence had been found that the NDN gene is imprinted and exhibits paternal-specific expression in tissues (Aizawa, Maruyama, Kondo, & Yoshikawa, 1992). In the Sutcliffe et al. (1997) study, it was found that NDN mRNA was detectable in normal and Angelman tissues, but absent in PWS samples. The authors conclude that their results show that NDN is imprinted with paternal-specific transcription in multiple tissues, including the brain, which is the predominant site of its expression.

In research conducted with mice, NDN had the highest expression in the hypothalamus and midbrain (MacDonald & Wevrick, 1997, as reported in Sutcliff et al., 1997). These findings are important because hyperphagia, among other symptomatology associated with PWS, is thought to be a result of a defect in the hypothalamus. The exact function of the NDN gene is not known, but it has been shown to arrest cell growth, maybe controlling growth of post-mitotic neurons during central nervous system development.

Diagnostic testing

Two different approaches are used in the diagnosis of PWS and AS: chromosomal analysis and DNA analysis. Chromosomal analysis involves the staining of chromosomes to detect deletions and other abnormalities. One particular staining technique is called fluorescence *in situ* hybridization (FISH). In FISH, pieces of nucleic acid that will hybridize to particular regions of chromosomes (probes) are labelled with a fluorescent dye. If a DNA sequence corresponding to the probe is present in the chromosome, the fluorescent probe binds to the chromosome and such binding can be seen as a fluoresent spot when viewed with a special microscope. DNA analysis is used to detect imprinting changes. It includes analysis of the methylation pattern in the PWS/AS region and analysis of microsatellite patterns on chromosome 15 to search for evidence of uniparental disomy. (Microsatellites are regions

of DNA, which contain short sequences of nucleotides that are repeated over and over again. These patterns differ among individuals and are inherited like genes.)

The most recent diagnostic DNA methylation test for PWS and AS involves analysis of a short sequence called a CpG island in the 5' region of the SNRPN gene (Glenn et al., 2000; Sutcliffe et al., 1997).

Prenatal diagnosis of PWS

Prenatal diagnosis is done by analyzing amniotic fluid, chrorionic villus tissue or fetal blood samples. There is a great need for a simple readily available prenatal diagnosis test for detecting as many of the different molecular classes of PWS and AS as possible. The imprinting mutation class is particularly important since this is associated with a recurrence risk as high as 50% in subsequent pregnancies. Because amniocyte and chorionic villus tissue are known to be undermethylated relative to other tissues, and because methylation of fetal DNA is not complete until after 23 weeks of pregnancy, there has been concern that imprinting mutations cannot be reliably diagnosed prenatally. Fortunately, the DNA methylation imprint at the 5' SNRPN locus has been found to be present in a wide range of different tissues including prenatal samples (Glenn et al., 2000). Preliminary studies support the use of analysis of the DNA methylation imprint at the 5'SNRPN locus for prenatal diagnosis, but prenatal tissues must be studied from more affected tissues before this method is considered to be reliable.

Persons considering postnatal or prenatal diagnosis for PWS should consult their local genetics department for details, as methods for prenatal diagnosis may differ from one centre to another.

Risk factors for PWS

Although most cases of PWS are sporadic, there appear to be certain risk factors that are associated with the sporadic mutations. However, this research is suggestive rather than conclusive.

Strakowski and Butler (1987) found an increased frequency (21%) of fathers of children with PWS who were employed in occupations involving exposure to hydrocarbons at the time of conception as compared to fathers of children with Down syndrome or Fragile X syndrome. This finding is corroborated by another study, which found that half of the fathers of children with PWS had been exposed to hydrocarbons (Cassidy, Gaincy, & Butler, 1989). Jobs involving exposure to hydrocarbons include factory workers, machinists, chemists, heavy machine operators, and mechanics. It is not known how hydrocarbon exposure might result in chromosome 15 deletions during spermatogenesis. In the case of disomy causing PWS, parents tend to be older (Cassidy, 1993).

Physical Characteristics

Although the extent or severity can vary, individuals with PWS present with a specific set of morphological characteristics. These are listed in Table 1. Many, but not all, of the physical characteristics may be the result of a defect in the hypothamic-pituitary axis, which is responsible for controlling endocrine levels. Diabetes is extremely common in PWS. Individuals with PWS should be regularly screened for diabetes and treated for this disorder (Nagai & Mori, 1999; Zipf, 1999).

A major defining feature of PWS is the tendency towards obesity, with the bulk of adipose tissue deposited on the trunk, buttocks and thighs, while the lower arms and legs tend to remain lean (Holm, 1981). In infants, obesity is not evident, but in adolescence and adulthood almost all PWS individuals are moderately to severely obese (Wollmann, Schultz, Grauer, & Ranke, 1998). Obesity typically leads to heart failure, which is the major cause of mortality. If obesity is kept under control, regular life expectancy is possible (Cassidy, Devi, & Mukaida, 1994).

The obesity appears to be linked to oxytocin neurons, the putative satiety neurons, in the paraventricular nucleus of the hypothalamus (Swaab, 1997; Swaab, Purba, & Hofman, 1995). More specifically, individuals with PWS have a reduced number of oxytocin-expressing neurons.

Oxytocin, a neuropeptide, has been implicated in satiety as well as aggression, sexuality, sleep-wake cycles and temperature regulation. Individuals with PWS have a reduced number of oxytocin-expressing neurons. They also have elevated levels of oxytocin in their cerebrospinal fluid, as compared to controls (Martin, et al., 1998). Although there is no clear explanation available for the preceding findings, both studies implicate the oxytocinergic pathway as underlying PWS feeding patterns.

As with obesity and short stature, hypogonadism, a major diagnostic criterion, is the result of hypothalamic dysfunction resulting in inadequate levels of sex hormones. The lack of sex hormones results in disordered, incomplete puberty, and both sexes are infertile. Feeding behaviour has been found to be associated with altered levels of metabolites of the neurotransmitter, serotonin. For example, people with anorexia have been found to have low levels of serotonin metabolites (Kaye, Gwirtsman, George, & Ebert., 1991). In PWS, as expected, increased concentrations of serotonin metabolites, have been found in the cerebrospinal fluid regardless of age, body mass, and level of mental retardation (Akefeldt, Ekman, Gillberg, & Mansson, 1998).

The levels of certain proteins also have been investigated as a contributing factor to obesity. Leptin protein, secreted by adipose tissue, is transported to leptin-receptors in the hypothalamus (and choroid plexus) which in turn influences feeding behaviour. High adipose levels result in high levels of circulating leptin which serve to decrease appetite. Researchers have hypothesized that there is a defect in leptin efficacy in individuals with PWS. In PWS, plasma levels fall within normal limits, although, compared to non-affected counterparts, there is a closer association between leptin levels and total body mass index (Pietrobelli et al., 1998) and leptin levels are more homogeneous (i.e., there is less variation in serum levels between individuals) than in non-affected counterparts (Butler, Moore, Morawieki, & Nicolson, 1998). Clearly, further investigations are required to clarify the role of leptin transport and response to leptin to the hypothalamus.

As well as obesity, individuals with PWS often present with small stature (a minor feature) which is the result of low levels of growth hormone. In the first year of life, spontaneous growth is nearly normal, but afterwards, short stature is present in about 50% of the children between the ages of 3 and 13 years, and one-half of individuals with PWS fall below the third percentile of healthy controls (Wollmann et al., 1998).

Characteristic facial features, a major diagnostic criterion (occurring as a cluster) include narrow face, almond-shaped eyes, narrow nasal bridge, and small-appearing downturned mouth with thin upper lips (Aughton & Cassidy, 1990). Hands and feet tend to be smaller than one would expect; for example, the average shoe size for an adult female with PWS is size 3 (Cassidy, 1997). Hypopigmentation (fair compared to other members of the family) is present in about one-third of individuals (Wiesner et al., 1987), and salivary flow is 20% of that found in controls often resulting in thick saliva (Hart, 1998). All of the above characteristics are minor criteria.

There is some evidence that individuals with PWS have decreased sensitivity to pain (Holm et al., 1993). Brandt and Rosen (1998) argue that this might be due to a reduced number of normal axons because nerve action potentials are reduced by 40–50% as compared to non-affected controls. Other authors propose that the reduced pain sensitivity may be due to abnormal auditory brain stem functioning resulting in a disturbance in the cortico-distal modulation system at the peripheral input level (Akefeldt, Akefeldt, & Gillberg, 1997).

No gross anatomical defects have been observed in post-mortem examinations of the PWS brain (Cassidy, 1997). However, a structural magnetic resonance imaging study of individuals with PWS is suggestive of functional abnormalities in the marker region of hypothalamic function. Diffuse minor abnormalities including slightly enlarged ventricles, cortical atrophy and small brain stem are also present (as reported by State and Dykens, 2000). In addition, 12% of children with

PWS have anomalous fissures on the banks of the Sylvian fissure (Broca's and Wernicke's region), the region primarily involved in language processes (Leonard et al., 1993).

Approaches to Treatments in PWS

Management of hyperphagia

Insatiable appetite, a characteristic feature of PWS, begins somewhere between age 2 and 5 years. Since individuals with PWS have metabolism, which is only 60% normal, they require fewer calories to maintain weight. An uncontrollable preoccupation with food usually leads to obesity, serious health problems and premature death. In PWS, length of life, which can extend into the 5th decade, is dependent upon successful weight management. Regulation of the diet of individuals with PWS can help to prevent the serious weight gain and associated health problems such as heart failure and diabetes that result from their insatiable appetite.

At present, there is little evidence that pharmacological agents, such as selective serotonin reuptake inhibitors (SSRIs) (Martin, State, Anderson, et al., 1998; Martin, State, Koening, et al.,1998), neuroleptics, appetite-suppressant medication (Tu, Hartridge, & Izawa, 1992) or surgery, such as biliopancreatic diversion (Grugni, Guzzaloni, & Morabito, 2000) are effective means of weight control. However, the surgical procedure of biliopancreatic diversion (BPD) has been reported to improve respiratory distress, mobility, self-image, alertness and quality of life on a short-term basis in some PWS patients. This approach to treatment still requires monitoring of food intake (Antal & Levin, 1996; Laurent-Jaccard, Hofstetter, Saegesser, & Chapuis Germain, 1991). BPD also requires oral supplementation for iron, vitamin D, vitamin B12 and folic acid deficiency (Laurent-Jaccard, Hofstetter, Saegesser, & Chapuis Germain, 1991). As well, medications such as risperidone may decrease some of the problematic behaviours associated with feeding (e.g., obsessiveness around food), thus making food management easier (Durst, Rubin-Jabotinsky, Raskin, Katz & Zislin, 2000).

Thus, recommended treatment involves a combination of dietary management, behaviour management (this is further elaborated in Medved & Percy, this volume), family intervention and therapy, and pharmacological interventions (Cheetham, Gitta, & Morrison, 1999). However, these approaches are associated with a high failure rate due to the patients' inability to cooperate in changing their behaviours, though successes are possible.

Growth hormone

Because limited growth is a recognized feature of PWS, growth hormone secretion has been studied in this population. Short stature is typically associated with low circulating levels of growth hormone. It is not known if the low level of growth hormone in PWS is due to the hypothalamic dysfunction or if is it secondary to obesity. One retrospective study found that children with PWS had decreased levels of growth hormone regardless of their weight status (Thacker, Hainline, St. Dennis-Freezle, Johnson, & Pescovitz, 1998). The impact of various growth hormone releasers was significantly lower in children with PWS than in non-affected short or non-affected obese participants (e.g., Cappa et al., 1998; Grugni, Guzzaloni, Moro, Bettio, DeMedici, & Morabito, 1998). These studies point to the idea that blunted growth hormone secretion is due to hypothalamic dysfunction.

Recently, there have been numerous studies examining the clinical potential of growth hormone treatment on stature, physical features, and even behaviour in PWS (e.g., Eiholzer, Weber, Stutz, & Steinert, 1997; Hauffa, 1997; Myers, Carrel, Whitman, & Allen, 2000). After treatment with growth hormone, height velocity can double. There can also be a significant reduction in the percentage of body fat (Davies et al., 1998) and facial characteristics (high midface, broad intercalar distance and high chin) can accentuate over time (Butler, Hovis, & Angulo, 1998). In addition to having a physical impact, treatment with human growth hormone is associated with improved physical performance and increased activity levels (Eiholzer et al., 1998). Lindgren et al. (1997) found that after six months, the replacement of growth hormone was

associated with behavioural benefits including increased alertness and interest in other children, stabilized temperament, and better interactions with parents. These behavioural benefits cease once hormone therapy is terminated. The physiological causes of the behavioural improvement are not understood. Overall, the consensus is the administration of growth hormone is promising, although further research exploration of treatment regimen is needed. It has recently been recognized that diabetes mellitus is a frequent complication in PWS, and may develop in 7–20% of individuals with this disorder. Concern has been expressed that growth hormone might increase the propensity to develop diabetes mellitus (Zipf, 1999).

Psychopharmacological agents

PSW may have a specific association with affective disorder and psychotic symptoms that cannot be explained solely by the fact that people with developmental disabilities are more likely to have psychotic symptoms than the rest of the population (Boer, Clarke, Whittington, Butler, & Holland, 2000; Clarke & Boer, 1998). SSRIs can be beneficial in reducing the frequency of skin-picking, hoarding and explosive outbursts (Hellings & Warnock, 1994) and medication such as risperidone has been observed to decrease aggression, impulsivity, and anger outbursts (Durst et al., 2000). Although individuals with PWS may benefit from treatments with psychopharmacological agents, the foundation of most treatment plans remains consistent behavioural limits across multiple environments (Dykens & Hodapp, 1997) (see Medved and Percy, this volume, for further information regarding behaviour and behavioural limits).

Sex hormones

Although sex hormone therapy is effective in producing secondary sex characteristics, most individuals with PWS do not receive sex hormone replacement therapy, thus prompting Muller (1997) to argue that more aggressive endocrine treatment strategies are required. However, as inviting as hormonal replacement therapy may seem, there are some draw-

backs (Cassidy, 1997). Testosterone replacement therapy is associated with an increased rate of behaviour problems in males, and estrogen replacement therapy is associated with an increased risk of stroke in females.

Ethical and Legal Issues of Treatment

Legal and ethical dilemmas about rights and responsibilities arise in the provision of other treatments for individuals with PWS. On the one hand, there should be individual choice, but on the other, care providers have a responsibility to prevent serious medical complications that can compromise the affected individual's health and quality of life (Holland & Wong, 1999). For example, the emphasis on individual choice conflicts with many weight management strategies that are considered too restrictive (e.g., locking refrigerators) and thus violate client rights. Nevertheless, decreased restrictive food practices have resulted in premature death due to complications of obesity (Dykens & Hodapp, 1997). Striking balance between food restrictions and personal autonomy is an ongoing concern that needs to be considered on an individual basis.

There is also concern about treating people with PWS with endocrine replacement therapies, not only because of the physical risks, but also because of the numerous ethical issues involved (Kodish & Cuttler, 1998). Particularly pertaining to PWS, questions such as 'What is the primary emphasis when growth hormone therapy is provided, health or cosmesis?' and 'What is the role of developmental disability in this assessment?' need to be carefully evaluated. Kodish and Cuttler (1996) state that the major argument for the use of growth hormone is increased height and decreased weight which, although associated with decreased obesity, does not necessarily justify its use. Thus, additional questions about the role of parents and other care providers in making treatment decisions need to be considered, as well as the rights of the individual with PWS, and the extent to which they can provide ongoing informed consent.

Conclusion

PWS is a multifaceted genetic disorder that presents with considerable clinical variability. Obesity and medical complications such as heart disease and diabetes are of primary concern. Much of the current biological research has moved beyond descriptions of physical characteristics of PWS, and is focussing on evaluating the efficacy of current medical treatments including hormonal injections and psychotropic medication. Concomitantly, the field of genetics research is exploding. New genetic discoveries are expected to lead to new treatments in PWS (and AS) that may offer alternatives to symptomatic management. Treatment provision in PWS and AS (and indeed all developmental disorders) is not without dilemma since it must be conducted with consideration of rights and responsibilities but within legal and ethical frameworks.

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Prader-Willi Syndrome II: A Literature Review of Behavioural and Neuropsychological Characteristics

Maria Medved
University of Toronto

Maire Percy
Surrey Place Centre

Abstract

This paper presents an overview of the behavioural and neuropsychological characteristics associated with Prader-Willi syndrome (PWS). PWS is a genetic disorder that affects multiple systems and results in a cluster of behaviours that include hypotonia and feeding difficulties in infancy, followed by the emergence of its most striking feature, hyperphagia, in early childhood. Emotional lability, compulsive behaviours such as skin picking and an increased risk of psychiatric disorders, are present. There is support for the notion of PWS behavioural specificity, and that these behaviours constitute a phenotype that is quite challenging for families. Individuals with PWS have an intellectual impairment that is typically in the mild developmental delayed range with poor adaptive functioning. In terms of weight management, environmental and behavioral interventions remain the most effective, although medication may reduce problem behaviours.

Prader-Willi syndrome (PWS) is a hypothalamic-based disorder that can result in, among other characteristics, an altered pattern of growth, obesity, developmental disability, and emotional lability. In 1956, Prader, Labhart and Willi provided the first description of the abnormalities associated with PWS, linking the abnormalities to impaired functioning of the hypothalamus, a small structure that controls the autonomic nervous system. PWS is the consequence of one of a number of genetic alterations, the most common being a microdeletion of chromosome 15q11-q13 (Cassidy, 1997). This syndrome is rather unusual because,

in addition to being caused by microdeletions, it is caused by other types of genetic alterations that lead to a similar phenotype (see part I, this volume).

This review will emphasize recent developments in the literature and will review many, but not all, of the behavioural and cognitive diagnostic criteria. First, we review the behaviours associated with PWS and related issues involving family functioning. Second, we review the literature available on neuropsychological functioning. Where appropriate, treatment and management options will be briefly indicated.

Behavioural Profile

Behavioural presentation

The behavioural presentation of PWS can be divided into two qualitative stages: infancy, and age 2 years to adulthood. In both stages, associated behavioural problems present many challenges to quality of life, both for the individual and families involved (Cassidy, Devi, & Mukaida, 1994).

Stage one is characterized by what is often called "failure to thrive." Infants have difficulty with feeding and decreased motor functioning, resulting in below average weight and hypotonia. Decreased motor function is noticeable *in utero*, the fetus not being very active. At the end of stage one, the hypotonia diminishes and motor functioning improves.

At the beginning of stage two, this feeding predilection radically changes and remains constant throughout the life span; hyperphagia or uncontrollable eating, one of the most striking features of the syndrome, emerges. Many parents initially experience relief because, after all the feeding difficulties, their child is finally starting to eat. However, the magnitude of the eating problem soon becomes evident, and if not vigilantly monitored by caregivers, this hyperphagia quickly results in obesity.

Hyperphagia is associated with intense feelings of hunger. Zellweger

(1981) has categorized the feeding patterns into two distinct types: in one pattern, the person constantly forages for food; and in the other pattern, the person engages in uncontrollable gorging when food is available. Holland, Treasure, Coskeran and Dallow (1995) found that when given unlimited access to food, approximately three-quarters of individuals with PWS ate excessive amounts. Lindgren, Barkeling, Hagg, Ritzen, Marcus, and Rosner (2000) suggest that it is not so much the experience of increased hunger that contributes to the eating patterns associated with hyperphagia, as the decreased sensitivity to satiation.

Although adolescents and adults with PWS understand the purpose of food, and reject eating solely inedible substances, they readily endorse eating contaminated foods involving inedible food combinations (e.g., cake with grass) (Dykens, 2000), and the presence of higher than usual vomiting thresholds often leads to food poisoning. Persons with PWS often fail to easily understand concepts related to contamination, and thus dietary interventions that include education about concepts such as germs and residues may be beneficial in reducing consumption of inappropriate substances (Dykens, 2000).

Many PWS behaviours revolve around food such as searching for, stealing, and hoarding food. Overall, environmental management remains the most effective method of weight maintenance. In order to regulate feeding behaviour, families must implement extreme interventions regarding food – refrigerators and cupboards must be locked, kitchen counters kept clear; individuals with PWS should not have food preparation responsibilities, and the quantity of food consumed at meal time must be monitored (James & Brown, 1993). Behavioral modification interventions have been found to be effective for reducing instances of behaviours such as food theft (Page, Stanley, Richman, Deal, & Iwata, 1983). As outlined in Medved and Percy (this volume), pharmacological agents such as risperidone can indirectly aid in weight management by reducing some of the difficult associated behaviours.

Children with PWS syndrome often start out as friendly, affectionate,

and docile, but these characteristics change as they mature (Greenswag, 1987). Stubbornness replaces affability, and repetitive chattering, verbal aggressiveness, self-assaultive acts and rages become common (Greenswag & Alexander, 1990). There seems to be a continuum of personality types: some people with PWS appear secretive and manipulative, and others appear lethargic and indifferent (Hall & Smith, 1972). An oppositional stance is common; 70 to 90% of individuals with PWS resort to temper tantrums. From early childhood to adolescence, there appears to be an increase in internalization problems or disorders such as depression (van Lieshout, DeMeyer, Curfs, Koot, & Fryns, 1998). Boys are more likely to be depressed than girls, although it is unclear why this might be the case (Dykens & Cassidy, 1995). From adolescence to adulthood, there is a decrease of externalization problems (e.g., aggression) (Dykens, Hodapp, Walsh, & Nash, 1992a). More extreme disorders such as psychosis (Verhoeven, Curfs, & Turner, 1998) are evident in about 5–10% of individuals (Cassidy, 1997). Psychopharmacotherapy can be of benefit (see Medved & Percy, this volume).

Individuals with PWS demonstrate a similar frequency and severity of compulsions as individuals with obsessive-compulsive disorder (Dykens, Leckman, & Cassidy, 1996). Even when compared to individuals who present with "Prader-Willi-like" features, people with PWS demonstrate an elevated number of obsessive-compulsive symptoms (State, Dykens, Rosner, Martin, & King, 1999). As a consequence of these symptoms, individuals with PWS prefer repetitive, stable schedules and experience difficulty with changes in their routines. Common compulsions associated with PWS include ordering, exactness, and arranging; for example, eating foods colour-by-colour, repetitively erasing and re-writing words, and skin-picking.

In fact, skin-picking is the most prevalent form of self-injury (82%) with the head and the front of the legs being the most targeted sites (Symons, Butler, Sanders, Feuer, & Thompson, 1999). Behaviours, such as tantrums and skin-picking, are often related to getting "stuck" in a compulsion or difficulty with routine change; thus extra support dur-

ing transitions in the form of cues can be of benefit in decreasing these behaviours (Dykens & Hodapp, 1997).

Individuals with PWS express awareness of their behavioural difficulties. James and Brown (1992) found that teens and young adults with PWS provided behavioural ratings similar to those supplied by their parents. These individuals also expressed concern over their temper-tantrums, and over one-half requested emotional support.

Behavioural specificity

Researchers have started to explore whether PWS is associated with a unique cluster of behaviours. In other words, can the set of behaviours that typically co-occur in PWS be differentiated from other disorders or syndrome? At present, it remains problematic to draw a definitive conclusion because studies report results which may be the consequence of methodological artifacts such as use of different control groups or assessment tools. To further complicate matters, there is some evidence that there are differences in the frequency of certain maladaptive behaviour between individuals who have PWS due to paternal deletion or maternal disomy (Dykens, Cassidy, & King, 1999). Individuals with a paternal deletion are more likely to skin pick, bite their nails, hoard, overeat, sulk and withdraw than individuals with maternal disomy. Nevertheless, there appears to be a degree of support for the concept of behavioural specificity.

In one study, children with PWS were compared using a maladaptive behaviour checklist (Child Behavior Checklist) to age-and gender-matched children with Down syndrome and children with nonspecific mental retardation (Dykens & Kasari, 1997). Of note, seven behaviours predicted membership in the Prader-Willi group with 91% accuracy: hyperphagia, skin picking, overtiredness, obsessions, compulsions, talking too much, and lack of hyperactivity. This finding strongly supports the claim that PWS includes a specific behavioural phenotype. Clarke and Boer (1998) employed the Aberrant Behavior Checklist and drew upon two other chromosomal deletion syndromes, Cri-du-Chat

and Smith Magenis, in setting up control groups. Specific problematic behaviours in PWS included temper outbursts, inability to delay gratification, repetitive speech, lability of mood, inactivity and self-injury. The authors concluded that "each deletion disorder is associated with a relatively characteristic constellation of problem behaviors, but with some of the behaviors occurring in association with more than one disorder" (p. 270).

When compared to a community sample of individuals with developmental disabilities, participants with PWS were found to be more disturbed overall, particularly in terms of antisocial behaviour (Einfeld, Smith, Durvasula, Florio & Tonge, 1999). When compared to age-matched children at a mental health centre, children with PWS demonstrated an equivalent number of behavioural and emotional problems, although there were differences in intensity and patterning of the various behaviours (van Lieshout, DeMeyer, Curfs, & Fryns, 1998). Moreover, when compared to children from regular schools, lower levels of agreeableness, conscientiousness and motor co-ordination, and higher levels of irritability and dependency were noted in children with PWS (Curfs, Hoondert, van Lieshout, & Fryns, 1995).

Family

Many studies indicate that the behavioural characteristics associated with PWS negatively affect family functioning. The behaviours related to PWS (specifically, overeating, skin picking, over-sleeping and storing things) often lead to high levels of stress in parents, more so than other behavioural and emotional characteristics that are present in developmental disabilities (Hodapp, Dykens, & Masino, 1997). General family problems are also more prevalent in families with a PWS child: members of the family have to do without some amenities and activities more often than in families with children who have mixed mental retardation etiologies.

Parents of children with PWS also report more marital conflict than parents of age and gender matched children with Fragile X or Williams

syndrome (van Lieshout, DeMeyer, Curfs, Koot, & Fryns, 1998). Fathers appear to find dealing with PWS particularly stressful, showing a distress level twice that found in fathers of children with Down syndrome (James & Brown, 1992). Indeed, this study found that couples with a child with PWS have a very high rate of marital separation. The authors hypothesize that the major adjustments required for eating patterns, and the high need for paternal involvement in supervision may be a contributing factor to fathers' high distress levels.

As a consequence of the high stress levels, both parents are at risk for expressing high levels of anger directed towards the child (van Lieshout, DeMeyer, Curfs, Koot, & Fryns,1998); further, "families with a child with PWS seem to be very different [from other families] in the degree to which they suffer from unfavorable family conditions, and parental behaviour is negatively affected by these adverse contextual circumstances" (p. 709). Put another way, the differences in family functioning are linked to the unique challenges of caring for a child with PWS behaviours.

For families with a child with PWS, perhaps more than in families with a child with another type of developmental disability, a key element to ensuring healthy functioning is to ensure that various support mechanisms are available (e.g, caregiver relief, support group).

Neuropsychological Profile

Development and intellectual presentation

There have been a limited number of studies exploring the neuropsychological presentation of PWS. Thus far, the module approach, which involves describing a pattern of cognitive strengths and weaknesses has been the most commonly used. A recent literature search revealed published studies that only explore the possibility that there is a unique neuropsychological phenotype associated with PWS. The reader is referred to PWS I in this issue for a review of the neurological literature.

Achievement of developmental milestones, such as walking and sitting, tends to be delayed in 90 to 100% of infants with PWS. As a general rule, developmental milestones are achieved at about twice the normal time; for example, sitting at 12 months, and walking at 24 months (Cassidy, 1997).

As adults, the majority of individuals with PWS have an intelligence quotient (IQ) which falls in the mild developmental disability range (mean IQ 60s-low 70s). Approximately 40% have borderline developmental disability or low average intelligence, and about 20% have an IQ that falls in the moderately delayed range. Regardless of measured IQ, most children and adults have learning disabilities, and feeding dysfunctions (Dykens, Hodapp, Walsh, & Nash, 1992b); they demonstrate poor academic and adaptive performance, and a high level of maladaptive behaviour (Dykens & Cassidy, 1995). There are no reported findings that cognitive functioning declines over the life span as has been observed in certain other developmental disabilities (e.g., Fragile X).

In order to obtain an overall IQ score, various sub-tests from an intelligence instrument, each assessing a specific aspect of cognitive functioning, are added together. One such instrument provides a verbal IQ score and a visuo-spatial IQ score. In one study, the profiles for about half of the participants tended to be flat; that is, ability levels for different facets of cognitive function were similar (Curfs, Wieger, Sommer, Borghgraef, & Fryns, 1991). If the profile was uneven, a subtest requiring visual-motor ability (Block Design) yielded a higher score compared to those tapping other abilities.

Visuo-spatial tasks involve simultaneous processing, as compared with verbal tasks which require sequential processing of information. A polarization between these processing abilities for persons with PWS has been pointed out by Dykens et al. (1992b) in that simultaneous processing is a relative strength compared to sequential processing. Therefore, since visuo-spatial tasks involve simultaneous processing, this type of

task is a relative strength, and since verbal tasks involve sequential processing, these tasks tend to be a relative weakness. For instance, individuals with PWS have an affinity for jigsaw puzzles, and frequent engagement in this activity is considered a supportive diagnostic criterion. Other studies have substantiated the finding that auditory verbal processing is relatively poorer than visual motor processing (Curfs et al., 1991). Overall these findings are consistent with the profiles of intellectual function in PWS.

It is worth noting that the type of genetic alternation that causes PWS is associated with the level of verbal IQ performance. One study reported that 50% of individuals with PWS due to uniparental disomy had a verbal IQ that was 70 or above, whereas only 17% of individuals with PWS due to a deletion had a verbal IQ that was 70 or above (Roof, Stone, MacLean, Feurer, Thompson, & Butler, 2000).

Polarization between short and long-term memory is also evident (Cassidy, 1997). Once an individual learns new information, he or she is unlikely to forget it, but learning and the retention of new information is problematic.

Another aspect of neuropsychological function is somatosensory and motor ability. There is a dearth of information available, but somatosensory perception appears to be intact (Brandt & Rosen, 1998), but there is a relative weakness in fine motor skills and strength (Levin & Wharton, 1993).

Speech and language

The development of articulation is delayed in PWS, although some individuals will show disordered development such as oral apraxia (oral motor control difficulty) (Akefeldt, Akefeldt, & Gillberg, 1997). Individuals with PWS have voice tones which have a hypernasal quality (Akefeldt et al., 1997). In general, speech is fluent in most individuals with PWS, although there are estimates that as many as 34% of individuals with PWS show dysfluency (Kleppe, Katayama, Shipley &

Foushee, 1990). When present, the most common dysfluencies are interjections (filler sounds) and revisions (using a different word or phrase) (Defloor, Van Borsel, & Curfs, 2000).

Overall, expressive language is more impaired than receptive language (Downy & Knutson, 1993); individuals may understand what is being said, but are unable to verbally respond at a level commensurate with their understanding. Often, this inability results in an underestimation of cognitive ability. Both the inability and others' underestimation of one's cognitive ability, can be extremely frustrating, a fact which should be kept in mind when interacting with individuals with PWS. Levin and Wharton (1993) have suggested that measures of receptive language may be better predictors of cognitive development than traditional measures.

Expressive oral language tends to consist of a noun and a verb, with delays in mastering concepts such as plurality (Akefeldt et al., 1997). In addition, pragmatics or rules of communication such as turn taking and maintenance of a particular topic during conversation are poorly understood.

Achievement and adaptation

A comprehensive neuropsychological evaluation, along with an exploration of intellectual abilities, includes an assessment of achievement. Tests of achievement provide measures of acquired knowledge, and usually deal with academic subjects: arithmetic, reading and spelling. In individuals with PWS, reading tends to be a relative strength; arithmetic is a relative weakness (Cassidy, 1997). Most likely the pattern holds because arithmetic requires sequential processing.

An additional component of a cognitive evaluation is the inclusion of an assessment of adaptive functioning. Most descriptions or definitions of developmental disability include problems associated with adaptive behaviour (e.g., self-care, social skills). For individuals with PWS, coping and social interaction skills are the most problematic adaptive func-

tioning skills (Dykens et al., 1992a). In other areas, difficulty with arousal regulation may be one factor that contributes to the lower than expected adaptive functioning performance. For example, daytime sleepiness occurs in 90% of people with PWS. Thus, taking transit or paying attention during sedentary activities at school or work, for example, is difficult. In general, the adaptive functioning of individuals with PWS rarely reaches the expected level in relation to their IQ level (Martin, State, Anderson, et al., 1998; Martin, State, Koening, et al., 1998), and few individuals achieve independent living status.

Executive functioning

Further research is needed to explore neuropsychological functioning of PWS in all areas; in particular, executive functioning and attentional processing need to be assessed. One investigative team's observations from a single-case psychometric assessment included difficulty with impulse control, perseveration, and impaired executive functioning relative to estimated IQ (Martin, State, Koening, et al.,1998) and such domains are critical for the design of effective educational interventions (e.g., problems with change may be associated with difficulty alternating between plans, difficulty with sequential processing could be due to a planning deficit, and so on).

Conclusion

PWS is a multifaceted genetic developmental disorder that is most saliently characterized by early onset of hyperphagia, together with a wide range of associated behaviour difficulties. In fact, there appears to be empirical support for the idea that there is a cluster of behaviours unique to PWS, and furthermore, that this behaviour cluster is rather stressful for the families of the individuals.

Nevertheless, there is considerable individual variability both in terms of behavioural presentation and intellectual function. An emerging issue is the identification of genetic variables that account for the vast phenotypic variability observed in PWS.

Overall, there is a paucity of literature pertaining to the neuropsychological functioning of individuals with PWS. There is even less literature available on how PWS affects one's sense of self. What we need to know is how individuals understand the world and themselves, including their syndrome. The question, in short, is: What is it like to have PWS? The voice of individuals with this syndrome surely needs to be heard further so that a life with PWS can be better understood as an aspect of lived experienced that we all share.

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Falls Among Persons Who Have Developmental Disabilities in Institutional and Group Home Settings

Heather J. Grant, William Pickett,
Miu Lam and Michael O'Connor
Queen's University

Hélène Ouellette-Kuntz
Queen's University and Ongwanada

Abstract

This descriptive, epidemiological study identified and described fall related injuries among a cohort of 114 adults living in Eastern Ontario Canada who have developmental disabilities. The study period spanned five years from 1991 to 1996. Seventy percent of the cohort experienced a fall event during the study; 79% of these events resulted in injury, and 7% of these resulted in serious injury requiring significant medical intervention. Serious fall injuries occurred most frequently in bathroom and outdoor settings. Persons with comorbid symptoms were statistically significantly more likely to suffer a serious fall injury. Higher injury rates were also associated with being male, mild to moderate levels of disability, group home residential settings, and younger age (<35 years).

Introduction

Falls and their sequlae are important causes of morbidity and mortality in developed countries. Existing research has documented the magnitude of these problems among children and the elderly (Graafmans et al., 1996; Hassan & Dorani, 1999; Lallier, Bouchard, St-Vil, Dupont, Tucci, 1999). To date, there has been little focused attention on the

magnitude of the fall injury problem experienced by persons with developmental disabilities. From the limited fall research that has been conducted with this population (Djurfeldt, 1990; Tannenbaum, Lipworth & Baker, 1989), and the many factors that predispose individuals who have developmental disabilities to falls, it is also likely that injurious falls are a recurrent health problem among persons who have developmental disabilities.

Numerous studies have shown that fall injuries are common causes of morbidity, functional decline and increased health-care use in elderly populations (Monane & Avorn, 1996; Rizzo, Baker, McAvay & Tinetti, 1996). Recent large-scale studies indicate that approximately one out of every three community-dwelling elderly persons fall at least once per year, (Graafmans et al., 1996; Lord, Ward, Williams & Anstey, 1993; Ryynanen, Kivela, Honkanen, Saano & Laippala, 1994), with between 30% and 55% of these falls resulting in injury (Nevitt, Cummings, Kidd & Black, 1989; O'Loughlin, Robitaille, Boivin & Suissa, 1993). Approximately 5–10% of these falls result in serious injuries including fractures, head trauma, severe lacerations and death (Cooper, 1994; Lord, McLean & Stathers, 1992; Rubenstein, Josephson & Osterweil, 1996).

Falls occur because of the interplay between intrinsic factors such as sociodemographic characteristics, lifestyle/behaviours and health-related factors, and extrinsic or environmental-related factors (Hornbrooke, Stevens, Wingfield & Hollis, 1994; Hyman & Arana, 1987; Sasaki, Takagi, Ida, Yamakawa & Ogino, 1999). Chronic illnesses, which occur in approximately 80% of the elderly population, have been shown to be associated with falls. Other health-related risk factors for falls include impairments of muscle strength, balance and gait disorders, visual impairments and medication use (Dolins, Harrison & Andrews, 1997; Nevitt et al., 1989, O'Loughlin et al., 1993; Passant, Warkentin & Gustafson, 1997; Zaleon & Guthrie, 1994).

Existing research with populations of persons with developmental

disabilities has shown that falls and their subsequent injuries are also a frequent and recurring health problem (Ouellette-Kuntz, 1994; Yeh, Nettleton & Verschraehen, 1990). Literature that examined institutionalized populations of adults with developmental disabilities indicated that these groups are highly susceptible to fractures resulting from falls (Lohiya, Crinella, Tan-Figueroa, Claires & Lohiya, 1999; Tannenbaum et al., 1989). Physical risks that resulted from the deinstitutionalisation of children with developmental disabilities have also been documented (Djurfeldt, 1990). The latter research found that falls were the primary mechanism of injury, with an overall rate of 9.12 per 10,000 days in care.

Like the elderly, persons with developmental disabilities also experience many chronic diseases, and possess personal variables such as physical mobility limitations and decreased intellectual abilities that also put them at an increased risk for falling (Konarski Sutton & Huffman, 1997). There are, however, differences between the elderly and developmentally disabled populations which make it important to investigate this group as a unique population. For instance, the presence of a developmental disability in itself has been shown to put an individual at risk for traumatic injury (Poole, Lewis, Devidas, Hauser, Martin & Thomae, 1997). Underlying risk factors include personal characteristics such as seizures and maladaptive behaviours (Konarski, Sutton & Huffman, 1997). Further, altered pain thresholds have been documented among persons who have developmental disabilities (Biersdorff, 1994). People with more severe levels of mental retardation are more likely to display signs of pain insensitivity or indifference. It is plausible that this could contribute to increased opportunities for the occurrence of injury in this population.

There is need for basic research that documents the fall injury experiences among persons who have developmental disabilities. This will assist in the development of preventive interventions, as well as the targeting of these interventions among those at highest risk for injury. Similar research with elderly populations has led to the successful

implementation of fall injury interventions among nursing home and community residents (Myers, Young & Langlois, 1996; Thompson, 1996). Better knowledge of both the descriptive epidemiology and the etiology of fall injuries among persons who have developmental disabilities would benefit health care professionals and planners who ultimately care for these vulnerable adult populations.

This study describes the occurrence of falls and fall injuries in a population-based sample of adults with developmental disabilities in Eastern Ontario, Canada. The objectives were to characterize: 1) the study population in terms of several salient personal characteristics, and 2) the occurrence of falls and fall injuries according to several classic, descriptive parameters including fall/injury location, time of occurrence, the nature and anatomical site of injury, and treatment administered, if any.

Methods

Settings and Participants

Cases for the study were drawn from the client population of Ongwanada, an agency that provides individualized and residential care services to persons who have developmental disabilities and their families in Kingston and Eastern Ontario, Canada. Ongwanada's computerized medical record system was used to identify a cohort of the agency's adult clients. Criteria for inclusion in the cohort were persons who: 1) have a diagnosed developmental disability, 2) resided in an Ongwanada institutional or group home setting on January 1, 1992, and 3) were ambulatory. The period of study covered five calendar years from 1992 through 1996. One hundred and fourteen individuals met these criteria at the start of the study, and a total of 507 person-years of follow-up were available. The time period that the individual subjects were followed for the study ranged from one year and three months to five years, with a mean follow-up time of four years and five months.

Variables

Falls and fall injury data were available for the full study period. As part of its standard approach to care, the agency's staff is required to file a standard incident report each time a fall occurs to one of its residents. Computerized incident reports were used to identify three types of fall outcomes: 1) falls not leading to injury, 2) minor injurious falls, and 3) serious injurious falls. For each fall event that resulted in injury, information was available to describe the injury type (non-injurious, injurious, serious), anatomical site (by body region), nature of injury (e.g., abrasions, fractures) and treatment classification (e.g., emergency department), as well as the date of the injury event. Injuries were considered serious if they resulted in fractures, injury to multiple sites, a laceration that required suturing, an emergency room visit or hospitalization, or further medical treatment/investigation beyond any initial first aid. All other injurious falls were classified as minor injurious falls.

Medical charts were used to gather data on each client for the following variables: age (on January 1, 1992), gender, type of residence (institutional, group home), freedom of movement or confinement to a locked ward (yes/no), level of mental retardation (mild/moderate, severe/profound), physical/sensory impairment (yes/no) and the presence or absence of existing comorbid symptoms that could cause dizziness and/or hypotension. The latter was determined by a written diagnosis of the symptom on the medical chart, or a record of a prescription used to treat the symptom. Symptoms and conditions considered in this list included: circulatory disease, Parkinsonism, and epilepsy. Medications that indicated the presence of these symptoms and conditions included: antiepileptics (anticonvulsants), cardiovascular medications and anti-Parkinsonian agents.

Computerized and chart data were imported and/or entered into a spreadsheet. Individual client characteristics were linked with their fall experiences from 1992–96. All data were reviewed for accuracy and completeness.

Results

The study sample is described in Table 1. Falls and serious injurious falls are described separately according to location, anatomical site, nature, and treatment classification in Table 2. There were a total of 275 reported falls in this cohort. These were distributed as follows: non-injurious falls (21.5%), minor injurious falls (65.0%), and serious injurious falls (13.5%). Seven out of every ten people within this cohort experienced at least one fall, and 79% of these falls resulted in an injury. The vast majority of the falls occurred during daylight hours when individuals are most active. There was, however, a modest peak in fall occurrence around 1:00 a.m., both for falls in general, and for serious injurious falls.

Table 3 describes the various fall severity types according to the personal characteristics of the study sample. Each characteristic is pre-

Table 1
Description of the Study Population by Seven Personal Characteristics (n=114)

Variable	n	%		n	%
Gender			Physical/sensory Impairment		
Male	63	55.3	YES	42	36.8
Female	51	44.7	NO	72	63.2
Age (Years) in 1992			Place of Residence		
18-30	24	21.1	Group Home	28	25.0
31-44	34	29.8	Institution	86	75.0
45-59	35	30.7	Freedom of Movement Restricted		
60+	21	18.4	YES	6	5.3.0
mean age = 43.7			NO	108	94.7
range = 18 – 77			Comorbid Symptoms		
Level of Mental Retardation			YES	81	71.1
Mild/Moderate	68	59.6	NO	33	28.9
Severe/profound	46	40.4	TOTAL	114	100.0

Table 2

Characteristics of All Falls and Serious Injurious Falls

Fall Characteristics	Total Falls n=275	%	Serious Injurious Falls n=37	%
Location				
hallway	72	26.2	4	10.8
outside	58	21.1	10	27.0
bathroom	42	15.3	7	18.9
bedroom	28	10.2	4	10.8
kitchen	14	5.1	2	5.4
living room	10	3.6	2	5.4
stairs	9	3.3	3	8.2
basement	3	1.1	1	2.7
other	15	5.4	0	0
unknown/unspecified	24	8.7	4	10.8
TOTAL	275	100.0	37	100.0
Anatomical Site of Injury				
head/face	70	25.5	7	18.9
arms/hands/fingers	39	14.2	4	10.8
legs/feet/toes	37	13.5	6	16.3
multiple sites	27	9.8	9	24.3
back/buttocks	25	9.1	4	10.8
eyes	12	4.4	2	5.4
unspecified	7	2.5	1	2.7
nose	5	1.8	2	5.4
ears	1	0.4	1	2.7
other	5	1.8	0	0
no apparent injury	47	17.0	1	2.7
TOTAL	275	100.0	37	100.0

Table 2 (continued)
Characteristics of All Falls and Serious Injurious Falls

Fall Characteristics	Total Falls n=275	%	Serious Injurious Falls n=37	%
Nature of Injury				
abrasion	63	22.9	4	10.8
bruise/swelling/redness	62	22.5	5	13.5
laceration (no sutures)	40	14.5	2	5.5
multiple injuries	17	6.2	17	45.9
scratch	13	4.7	0	0
laceration (sutures)	5	1.8	5	13.5
fractures	3	1.1	3	8.1
unknown	1	0.4	1	2.7
others	12	4.4	0	0
no apparent injury	59	21.5	0	0
TOTAL	275	100.0	37	100.0
Treatment Classification				
no action but examination by an RN or MD	121	44.0	5	13.5
no action, no examination by an RN or MD	66	24.0	5	13.5
first aid by RN or MD	56	20.3	5	13.5
further investigation ie. x- ray	15	5.5	9	24.3
hospital emergency room visit	11	4.0	3	8.22
insufficient information	6	2.2	10	7.0
TOTAL	275	100	37	100.0

Table 3
Subject Characteristics According to Fall Types (n=275 falls)

Variable	Total Person Years	Serious Injurious Falls (n=37)	Rates per 100 person years	p value
Gender				0.61
Male	(274.7)	21	7.6	
Female	(232.3)	16	6.9	
Level of Mental Retardation				0.85
Mild/moderate	(291.6)	22	7.5	
Severe/profound	(215.4)	15	7.0	
Age				0.64
³ 35 in 1992	(351.4)	18	5.1	
<35 in 1992	(155.6)	19	12.2	
Physical/sensory Impairment				0.80
YES	(169.7)	13	7.7	
NO	(337.3)	24	7.1	
Place of Residence				0.52
Group Home	(129.1)	14	10.8	
Institution	(378.0)	23	6.1	
Freedom of Movement Restricted				0.22
YES	(60.0)	7	11.7	
NO	(447.1)	30	6.7	
Comorbid Symptoms				0.04
YES	(394.7)	34	8.6	
NO	(112.3)	3	2.7	

sented along with its total person year time in the study. Rates per 100 person years were calculated for each variable by placing the frequency of the fall type in the numerator, the number of person years in the denominator, and multiplying by 100. Chi-square tests were applied to compare rates within variables. Because of the recurrence of falls in

Table 3 (continued)
Subject Characteristics According to Fall Types (n=275 falls)

Variable	Total # Falls (n=275)	Rates per 100 person years	p value
Gender			0.83
Male	151	55.5	
Female	124	53.4	
Level of Mental Retardation			0.86
Mild/moderate	158	54.2	
Severe/profound	117	54.3	
Age			0.76
≥35 in 1992	129	36.7	
<35 in 1992	146	93.8	
Physical/sensory Impairment			0.92
YES	90	53.0	
NO	185	54.8	
Place of Residence			0.89
Group Home	96	74.4	
Institution	179	42.4	
Freedom of Movement Restricted			0.005
YES	38	63.6	
NO	239	53.5	
Comorbid Symptoms			0.003
YES	235	59.5	
NO	40	35.6	

some individuals, it was necessary to include the generalized estimation equation approach, taking into account the dependence between recurrent falls in the same subject, when comparing rates (Glynn & Buring, 1996). Corresponding p-values were presented. Possessing comorbid symptoms and having no restricted freedom of movement in

one's environment were shown to be significant risk factors for falling ($p<0.003$ and $p<0.005$) respectively. As well, persons with comorbid symptoms were significantly more likely to experience a serious injurious fall ($p<0.04$).

Discussion

The present study characterizes the fall injury experience in adults with developmental disabilities who reside in both institutional and group home settings. The results indicate that falls occur frequently among this population, although only a small proportion of these falls result in serious injury and/or medical intervention. Of the 275 fall events documented in this cohort of 114 persons over five years, 37 (13.5%) were classified as serious: 17 resulted in multiple injuries, 9 required further investigation (e.g., xray), 5 resulted in lacerations requiring sutures, 3 involved a fracture and 3 required a hospital emergency room visit.

It is difficult to compare the occurrence of fall injuries within this cohort with existing research due to the lack of common definitions as to what constitutes a fall event and a serious injury. A study that examined institutionalized residents exclusively identified 3.3% of all injuries as serious; 2.8% resulted in lacerations requiring sutures and 0.5% resulted in fractures (Spreat & Baker-Potts, 1983). While the authors do state that the second most likely cause of injury was due to falls, they do not examine the cause of those injuries described as serious. Tannenbaum et al. (1989) examined fractures among residents of an intermediate care facility for the developmentally disabled. Thirty-eight percent of all fractures were found to have occurred following a fall. These included falls while ambulating from bed, on stairs, and during toileting or bathing. While it is difficult to make comparisons between these studies because of the lack of standard definitions and variations between the study populations examined, these studies suggest that serious fall injuries are fairly uncommon among persons who have developmental disabilities.

Rates for non-injurious and injurious falls were highest for clients residing in group homes. During the study period, large numbers of the sample were relocating from a single, institutional setting where they had lived for decades to several group homes across the region. Since the length of follow-up in the group homes varied from a few months for most clients to several years for a few, the observed increased risk to fall injury in group homes may be transient. More falls could occur following a residential relocation due to residents having to adjust to their new physical environments (Bongiono, 1996). In addition, since significant life events involving persons who have developmental disabilities are frequently accompanied by periods of increased agitation, the increased risk may be a function of escalating aggressive behaviors (including pushing and shoving) as clients adapt to change (Sovner, 1989).

Common locations of falls included hallways (26.2%), the outdoors (21.1%) and residential bathrooms (15.3%). Falls that occurred outside may in part be attributable to unfamiliar environmental hazards, as has been established for elderly persons (Parker, Twemlow & Pryor, 1996; Yutterstad, 1996). In addition, gait disorders occur frequently among persons who have developmental disabilities. As these people age, they are at increased risk for falls and subsequent injury in all locations, especially those areas where they spend the most amount of time (Hausdorff, Edelberg, Mitchell, Goldberger & Wei, 1997; Lord et al., 1993). Conceivably, individuals living in institutions walk the corridors and hallways frequently, explaining the frequent fall occurrences in these settings. Psychomotor impairments including decreased mobility and postural instability are also common. This can lead to falls in settings such as the bathroom, where physical agility is especially required (Boonen, Dequeker & Pelemans, 1993).

Although the only characteristic that was seen to be significantly associated with a serious injurious fall was the presence of a comorbid symptom, other characteristics in our sample also corresponded to higher rates of serious fall injuries. They include having a physical/sensory

impairment, living in a group home, being male, having a milder degree of mental retardation, being less than 35 years of age and living on a locked ward. Few studies exist that have examined any of these characteristics in detail. The studies that do exist examined gender and level of mental retardation. They are inconclusive about whether important differences in risk exist between the sexes (Scheerenberger, 1992; Tannenbaum et al., 1989). Moderate and profound levels of mental retardation were associated with higher levels of injury in a different study setting (Spreat and Baker-Potts, 1983).

The current study provides a simple, initial step in defining patterns of fall injuries, but is limited by the lack of certain details in individual case reports. This hindered the ability to describe patterns of injury completely, and identify practical opportunities for prevention. Routine surveillance of falls in larger populations of persons who have developmental disabilities is needed to identify specific high-risk areas, activities and personal risk factors associated with falls and subsequent injuries. Also, areas where individuals do not spend much time but have shown to be locations of high risk for injury occurrence, such as bathrooms, may need to be re-evaluated in terms of safety standards and closer supervision. Studies involving persons who have developmental disabilities living in family homes and in independent settings, as well as studies involving children with developmental disabilities, are also needed to add to our increasing body of knowledge of fall injuries in this population.

Although risk factors for fall injury were explored in the present analysis, etiologic studies that involve case-control or cohort methodologies are required. This could be used to elucidate, for example, whether management and staffing practices contribute to reduced rates of injury. The latter could include a study of the numbers of workers available to staff residences and institutional settings, methods of supervision, and the adoption of maladaptive behavior intervention techniques by staff. Research is needed to suggest how living and recreational opportunities can be arranged so that they minimize risks for injury, while

enriching the lives of persons who have developmental disabilities. Intrinsic risk factors for fall injury are also not well understood. This includes examination of comorbid symptoms and use of psychoactive medications, each of which have been implicated as risk factors in people with developmental disabilities.

Summary

This research has shown that unlike populations of non-disabled persons, serious fall injuries are more common in young adults than the elderly. Other risk factors suggested by this study, however, do reflect a pattern of disability, which is not unlike that of an aging population. These include the presence of a physical or sensory impairment, and comorbid symptoms. For Ongwanada and other agencies serving individuals who have developmental disabilities, these findings suggest a need for careful consideration involving the impact that living environments, activities and supports have on the risk of fall injury, particularly among young adults, males, and those who have additional disabilities.

The results were consistent with past research, which has shown that while fall injuries are common among persons who have developmental disabilities, these injuries are not usually serious in nature. Studies examining characteristics of those who fall have shown results that are inconclusive and contrary to this study's findings. Future research is needed to further classify groups at highest risk amongst this susceptible population, and to identify hypotheses for more focused, etiological studies.

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Correspondence

Heather Grant
Syl and Molly Apps Research Centre
Kingston General Hospital
76 Stuart Street
Kingston, ON
K7L 2V7
email: granth@post.queenu.ca

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Assessing Dementia of the Alzheimer Type in People with Down Syndrome

Oded Friedman and Ivan Brown
University of Toronto

Abstract

Research supports the view that individuals with Down syndrome (DS) develop Alzheimer disease-like neuropathological brain changes by the fourth decade of life. In spite of this, the major clinical sign of Alzheimer disease (AD), progressive dementia, is not always observed in mature and elderly people with DS. This apparent discrepancy suggests that neuropathological brain changes may not serve as accurate indicators of dementia. This study examined dementia of the Alzheimer type (DAT) in adults aged 20–59 with DS using an informant questionnaire that assesses changes in three domains: memory, cognition, and social and emotional behaviour. Fifty people with DS, selected from a social service agency located in Toronto, Canada, were assessed by the caregivers who knew them best. DAT diagnostic criteria were met for 11 (22%) of the 50 people with DS, supporting the view that not all people with apparent neuropathological brain changes associated with DAT show clinical symptoms. Some declines, but not sufficient to meet diagnostic criteria, were reported for another 15 (30%). Dementia decline scores, obtained from the informant questionnaires, were higher for those who met diagnostic criteria than for those who did not. Individual score profiles across the three domains clearly showed individual differences in the pattern of severity of decline, both for those who met diagnostic criteria and for those who did not. Such differences support the views that decline is somewhat unique to individuals and that there may be a "pre-clinical" decline, especially in memory. Moreover, individual differences provide useful information in care provision. The assessment tool developed and tested in this study is described as one useful part of the necessary range of ongoing assessment tools to support both diagnosis and development of care plans.

Introduction

Down syndrome (DS) is a term that is used to describe a set of characteristics that are associated with an increased number of chromosome 21 genes. Most cases of Down syndrome are caused by trisomy 21. It is the most common congenital cause of developmental disability, with a Canadian birth incidence of 1 in every 600 to 700 people (Janicki, 1988). Alzheimer disease (AD) is a degenerative disease that is principally characterized by the brain becoming increasingly riddled with amyloid plaques and neurofibrillary tangles, and lower cerebral cortical levels of acetylcholine (Prasher & Percy, 1999). Clinically, AD is manifested as dementia, the progressive loss of cognitive function independent of the state of attention. Dementia is marked by growing, irreversible declines in memory, performance of routine tasks, time and space orientation, language and communication skills, abstract thinking, and the ability to learn and carry out mathematical calculations (Jozsvai, 1999).

A considerable amount of interesting work has been carried out over the past several years clarifying the links between neurological changes in people with DS and dementia of the Alzheimer type (DAT). It is currently believed that abnormalities associated with the extra chromosome 21 in people with DS may lead to neurological changes – plaques and tangles, and lower cerebral cortical levels of acetylcholine – in the brains of virtually all adults with DS by the time they are 40 years old (Prasher & Percy, 1999; Zigman, Silverman & Wisniewski, 1996). This contrasts with people who have developmental disabilities with causes other than DS, where the incidence of AD-type neuropathology is thought to be the same as that of the general population. It has also become clear, however, that, although all people with DS are at very high risk of developing clinical symptoms of DAT, substantially fewer than 100% actually do so (Jozsvai, 1999; Oliver, Crayton, Holland, Hall & Bradbury, 1998; Zigman, Schupf, Sersen & Silverman, 1996). The available literature reports the prevalence of DAT with the DS population over 40 to be much lower than might be expected, with no

evidence of DAT in many mature and even some elderly people with DS. DAT is assessed for about 22–25% of people with DS who are 40 or more years old (Janicki & Dalton, 2000; Holland, Hon, Huppert & Stevens, 2000), compared to about 2–3% of people with other developmental disabilities who are 40 and over (Janicki, 1995; Janicki & Dalton, 2000). Thus, it appears that, for people with DS, assumed neuropathological brain changes may not always manifest as dementia, even though such changes are usually assumed to manifest as dementia in general populations.

The relationship between DS and DAT has been further explored by examining the age at which DAT is first assessed. For those aged 40–49, the percentages of people with DS who were diagnosed with DAT have been reported to be 9% (Prasher & Filer, 1995), 10% (Holland, Hon, Suppert, Stevens & Watson, 1998), 11% (Visser et al., 1997), 16% (Sekijima et al., 1998), and 22% (Janicki & Dalton, 2000). For those age 50–59, the reported percentages who were diagnosed with DAT are 36% (Prasher & Filer, 1995), 38% (Sekijima et al.,1998), 40% (Holland et al., 1998), and 66% (Visser et al., 1997). Prasher reported that 55% of those aged 60–69 with DS had been assessed with DAT, and 77% of people in the same age group in the Visser et al. study had the same assessment. Janicki and Dalton reported that 56% of their 62 people with DS between the ages of 60 and 79 had been assessed with DAT. Together, these studies support the view that, although DAT is found more frequently as people with DS age, a diagnosis of DAT does not occur in all cases.

Differences among prevalence rates in the literature probably stem from at least four factors. First, accurate assessment of DAT may be impeded by the presence of intellectual impairment in people with DS (Holland, 1999). It may also be that the degree of cognitive impairment associated with the onset of DAT may correspond, at least in part, to the degree of pre-existing cognitive impairment (Oliver et al., 1998; Temple, Jozsvai, Konstantareas & Hewitt, 2001). Second, in the literature reported to date, the number of subjects investigated is often small

because of low numbers of people with DS within populations available for study. Third, most assessments of DAT rely on establishing evidence that memory and other cognitive functions have deteriorated, and sometimes on establishing associated changes in personality. However, there are a number of other factors – such as thyroid dysfunction, depression, autoimmunity, and sensory impairment – that may not have been considered but that produce symptoms similar to those typical of DAT (Holland, 1999; Prasher & Filer, 1995; Prasher & Percy, 1999).

A fourth problem is that the methods of assessing DAT for the purpose of supporting a diagnosis have not been consistent. Approaches to assessing functional changes characteristic of AD-type dementia have included the Folstein Mini Mental State Examination, ICD–10 criteria for the diagnosis of dementia in AD, DSM-IV criteria for the diagnosis of dementia of the AD-type, the Dementia Questionnaire for Persons with Mental Retardation (DMR), the Multi-Dimensional Observation Scale for Elderly Subjects Adapted For Persons With Down Syndrome (MOSES), the Ruocco Geriatric Assessment for Persons with Mental Retardation, the Dementia Scale for Down syndrome (DSDS), CAMDEX criteria for the diagnosis of dementia of the AD-type, various dementia checklists, social skills inventories, and many others (Holland et al., 1998; Visser et al., 1997).

All these assessment tools provide helpful information, but many need to be used in conjunction with other assessment methods to provide adequate information for a diagnosis of DAT. For example, the Mini Mental State Examination does not assess change over time, but rather provides only a snapshot cognitive score with scores less than 24 marking the usual cut-off for the diagnosis of possible dementia (Deb & Braganza, 1999). Other methods and sources make a variety of unique contributions to a more comprehensive assessment approach. The American Association on Mental Retardation (AAMR) and the International Association for the Scientific Study of Intellectual Disability (IASSID), for instance, established criteria for the diagnosis of dementia in individuals with intellectual disabilities that included changes in memory, personality, general mental functioning, and daily living skills

(Aylward, Burt, Thorpe, Lai & Dalton, 1995). Dalton and Crapper-McLachlan (1986) offered an important perspective on language based dysfunction, and Thorpe (1999) identified helpful exclusionary criteria.

The present study developed and pilot tested an assessment approach that used and expanded upon established diagnostic and exclusionary criteria, and that integrated the strengths of a number of the assessment approaches previously used and attempted to minimize the problems associated with them and others. The primary aim of the study was to investigate the extent of clinical change attributed to dementia with age in an unbiased, population-based sample of older people with DS. This investigation involved examining the clinical expression of dementia in persons with DS, with a special focus on the relationship between clinical expression of dementia and age. The implication of this for clinical practice is that it is useful to know the age-specific prevalence rates of DAT for people with DS, to understand the potential for diminishing abilities in such areas as self-care, communication, and orientation skills, and to adapt care and treatment practices to the changing needs of each individual over time (Dalton & Crapper-McLachlan, 1986; Rasmussen & Sobsey, 1994; Roeden & Zitman, 1995).

Method

Participants

The study was carried out in a community agency located in Toronto, Canada that serves about 1,000 people with developmental disabilities and their families. The population studied was adults 20 years of age or older with developmental disabilities due to DS. The diagnosis of DS was confirmed by information in the agency's medical records; this did not necessarily include formal chromosomal confirmation by karyotype analysis.

The 50 participants selected were the main caregivers of this population. These main caregivers were self-identified and included support staff, supervisors, or family members who had prior knowledge of the person with DS for at least six months.

The corresponding 50 adults with DS ranged in age from 20 to 56 years; 15 were female (mean age 40.5, *SD*=6.65), and 35 were male (mean age 40.7, *SD*=9.25). Five were nonverbal (i.e., speech was limited to only a few words or they were unable to understand simple instructions), although it was not established whether they had always been nonverbal or if they had become so. Intelligence test scores were not used as they were available for only 16 of the individuals with DS, primarily because the practice in Canada is not to measure intelligence for adults with developmental disabilities unless specific reasons to do so exist.

Instrumentation

The first phase of the project involved a critical analysis of the content of a number of instruments that are commonly used to assess functional changes characteristic of AD-type dementia. Commonly used assessment domains and foci were noted, and the strengths and weaknesses of various instruments were compared. Three sources from that literature that were considered particularly important were: 1) a report by the American Association on Mental Retardation and the International Association for the Scientific Study of Intellectual Disability working groups for the establishment of criteria for the diagnosis of dementia in individuals with intellectual disability (Aylward, Burt, Thorpe, Lai & Dalton, 1995) that suggested key assessment domains; 2) the work of Dalton & Crapper-McLachlan (1986) that contributed questions regarding language based dysfunction, and 3) work by Thorpe (1999) that shed light on important exclusionary criteria.

An Assessment Protocol was developed from the results of this analysis. The protocol conformed with, but expanded upon, the five DSM-IV criteria for the diagnosis of dementia of the AD type, which are as follows: 1) evidence of both memory impairment and at least one additional cognitive impairment (aphasia, apraxia, agnosia, judgement, or disturbance in executive functioning) for at least six months; 2) cognitive deficits that cause significant impairment in social behaviour, occupational functioning, emotional control, or motivation; 3) continuing and

gradual cognitive and functional decline; 4) other causes of dementia are ruled out (e.g., cerebrovascular disease, hypothyroidism, Parkinson's disease); and 5) disturbance does not occur exclusively during delirium.

The Assessment Protocol has introductory descriptive questions (e.g., age, gender, relationship of the participant to the individual with DS), followed by a general question and a series of more specific questions in each of three main domains that asked participants to report (yes/no) if they have perceived declines in the person with DS.

1. *Memory decline/disturbance*. Specific questions: declines in remembering social arrangements, remembering location of recently placed objects, remembering information told, remembering the day's events, remembering names of family members and friends, finding the way around the neighbourhood, need for prompts in remembering, and failure to recognize family and friends.

2. *Decline in other cognitive functions*. Specific questions: declines in producing or comprehending words, performing complex movements without sign of muscle weakness (e.g., dressing, feeding, brushing teeth), recognizing familiar objects, writing, reading, or calculating, distinguishing between day and night, finding the bedroom or kitchen, appropriate use of everyday objects, level of executive functioning, judgement, self care skills, work skills; use of repeated, stereotyped phrases; and repeating what others say.

3. *Changes in emotional control, motivation and social behaviour*. Specific questions: sleep difficulties, sleep of excessive duration or depth, loss of interest, withdrawal, abnormal behaviours, and inefficient thought.

A second dimension that was assessed for the questions in the three domains was *duration and progression*: Wherever appropriate, the Yes/No questions were followed by the question, “If so, for how long?” In

addition, at the end of the questions in the three main domains, participants were invited to note those characteristics or behaviours that were associated with gradual onset, and those that were associated with rapid onset. In this way, the Assessment Protocol allowed for brief descriptive profiles of each person with DS to be compiled.

A final set of questions in the Assessment Protocol focussed on other possible causes of decline. These include: delirium; depression; untreated hypothyroidism; folic acid abnormalities; hypercalcemia; the presence of more than two risk factors for vascular dementia (i.e., obesity, hypertension, arrhythmia, history of previous stroke, diabetes mellitus, and any smoking history); unaided hearing impairment of six or more months duration; alcohol or substance abuse; Parkinson's disease (related to subcortical dementia and Lewy body dementia); use of medications (e.g., benzodiazepines, psychotropics, antiepileptic drugs, some antibiotics) which could cause mood and behavioural changes leading to a pseudo-dementia picture; and occurrence of recent or past traumatic head injuries (see also Thorpe, 1997; Zigman, Schupf, Havermen & Silverman, 1997).

Meeting diagnostic criteria. To diagnose DAT accurately, both the presence of progressive dementia and a characteristic pattern of neuropathology are required. However, in practice, the diagnosis of DAT is a clinical matter (Trumble, 1999) that involves professional assessment over time of symptoms associated with dementia (Janicki, Heller, Seltzer & Hogg, 1995). In this study, the purpose was not to diagnose DAT, since the study participants were not assessed clinically, but rather to use the information provided by their main caregivers to determine the degree to which they met the diagnostic criteria of the Assessment Protocol. These diagnostic criteria should be one useful source of information for a clinical diagnosis.

Meeting the diagnostic criteria of the Assessment Protocol involved two things: first, meeting the five diagnostic criteria of the DSM-IV, and, second, having one or more “yes” responses (declines observed for at

least a period of six months) in each of the three domain areas of the Assessment Protocol when exclusionary criteria were ruled out. The Assessment Protocol built upon many other assessment approaches by adding three unique features that should provide useful information both for providing appropriate care and making an accurate diagnosis: 1) it was able to compare the degree to which each of the three main domains (memory loss, declines in cognitive functioning, and declines in social, occupational, and emotional behaviour and motivation) contributed to overall declines by comparing the number of "yes" responses within the three domains; 2) it was able to identify specific sources of decline within each of the three domains by asking about several areas of functioning, and 3) it provided a method of describing both the severity of the overall decline, by the total number of yes responses that had a duration of at least six months, and the severity of declines in any one of the three domains, by the number of yes responses within that domain that had a duration of at least six months.

Procedures

The study participants (caregivers) were identified by personnel at the participating agency who were familiar with the adults with DS and their closest caregivers. Information was first collected from each participant regarding the presence or absence of a family history of AD, a history of prior head trauma, and a history of alcohol/drug abuse.

The study participants were then provided with the Assessment Protocol in January 2000. To ensure the greatest possible consistency, participants were given standard instructions for completing the questions, and possible problems were identified and addressed. All Protocols were returned by April 2000. Data were compiled and analyzed by the first author.

Results

Since 40 has been identified in the literature as an age by which people with DS most probably have experienced neuropathological brain

Table 1
Number And Percentage Of People With Down Syndrome Reported Not To Have And To Have Declines In Three Domain Assessment Areas Of DAT

	Age 20–39 (n=26)		*Age 40–59 (n=24)*	
No declines reported	13	(50.0%)	10	(41.7%)
Declines reported	13	(50.0%)	14	(58.3%)
Memory alone	3	(11.5%)	0	(0.0%)
Cognitive alone	0	(0.0%)	2	(8.3%)
Behaviour alone	1	(3.8%)	1	(4.2%)
Memory and cognitive	3	(11.5%)	1	(4.2%)
Memory and behaviour	0	(0.0%)	0	(0.0%)
Cognitive and behaviour	1	(3.8%)	3	(12.5%)
All three domains	5*	(19.2%)	7	(29.2%)

* Five people were reported to have declines in all three domains; of these, only four met the DAT diagnostic criteria. One was reported as having more than two risk factors for vascular dementia, and the study's cautious approach excluded him on these grounds.

changes that are associated with DAT, the data were described for those reported on by the study participants in two general age categories: 20–39 and 40–59. There were no people with DS under 20 or over 59 years of age.

Overall DAT assessment. Using the DSM-IV criteria first, 12 people with DS met the DAT diagnostic criteria – 5 (33.3%) of the 15 people aged 20–39 and 7 (50%) of the 14 people aged 40–59. The five who were under 40 were all between 30 and 39 years of age. Next, data gathered from the more detailed Assessment Protocol were examined to test the diagnostic criteria of the DSM-IV. This testing required that the main caregiver report a decline of at least six months duration for at least one item within each of the three principal domains, and that such decline could not be accounted for by the exclusionary criteria. This testing, which is intended to represent a fairly conservative approach to meeting diagnostic criteria in order to minimize the possibility of misdiagnosis,

Table 2
Decline Scores For People Who Met DAT Diagnostic Criteria And People With Declines But Who Did Not Meet Diagnostic Criteria

	Met criteria		Declines, did not meet criteria	
	20–39	*40–59*	*20–39*	*40–59*
	n=4	n=7	n=9	n=7
Memory	.36	.27	.16	.05
Cognitive	.19	.12	.05	.14
Behaviour	.21	.53	.10	.39
Overall decline score	.25	.37	.10	.19

resulted in one person with DS under 40 being excluded from those meeting the diagnostic criteria. Thus, four people under 40 and seven people over 40 met the DAT diagnostic criteria.

Other declines within domains. Although only 11 of the 50 people with DS in the study met the DAT diagnostic criteria, more than half did have one or more declines in functioning of at least six months duration that could not be accounted for by the exclusionary criteria. However, these declines did not always occur in all three domains. Of the 26 people with DS aged 20–39, 13 (50.0%) were reported to show declines, but only 5 had declines in all three domains. Of the 24 people with DS aged 40–59, 14 (58.3%) were reported to have declined in at least one domain, but only 7 of these had declined in all three domains. The number and percentages of people with DS who had no reported declines, and who had reported declines of at least six months duration in only one of the three domains, in two of the three domains, or in all three domains, are shown in Table 1.

Decline scores were computed for each person with DS for each of the three domains by recording the number of “yes” responses to domain items. Since the three domains had different numbers of items (9, 20, and 7 respectively), the domain scores were weighted arithmetically such that all three contributed equally to the overall decline score. The

scores ranged from a low of 0 (no items reported as declining) to a high of 1.0 (all domain items reported as declining). Table 2 shows these domain and overall decline scores for two groups for whom declines were reported: those who met the DAT diagnostic criteria, and those for whom declines were reported but who did not meet the DAT diagnostic criteria.

To determine if the scores for those who met the DAT diagnostic criteria and those who did not differed statistically, the three domain scores and overall scores for the two groups were contrasted. Since numbers were low in the four groups shown in Table 2, and since the score patterns were quite similar for the two age groups, the age groups were collapsed. As expected, the group that met the diagnostic criteria had higher overall scores ($t=4.30, df=25, p<.01$), memory domain scores ($t=3.34, df=25, p<.01$), and cognition domain scores ($t=2.18, df=25, p<.05$) than the group that showed declines but did not meet these criteria.

The behaviour domain scores showed a different pattern. Among the 14 people aged 40–59, there was wide variation in the number of "yes" responses to the behaviour items in both the group that met the criteria

Table 3
Decline Scores For Eight Individuals With Down Syndrome In Three Domain Areas Of DAT

Study ID#	Age group	Diagnostic criteria status	scores			
			Memory	Cognitive	Behaviour	Overall
#6	20–39	Met criteria	.56	.35	.14	.35
#8	20–39	Met criteria	.22	.05	.43	.23
#11	20–39	Did not meet criteria	.33	.00	.00	.11
#26	20–39	Did not meet criteria	.00	.05	.43	.16
#32	40–59	Met criteria	.56	.20	.14	.30
#37	40–59	Met criteria	.22	.10	.86	.39
#41	40–59	Did not meet criteria	.37	.15	.00	.17
#46	40–59	Did not meet criteria	.00	.00	.43	.14

and the group that did not meet them, ranging from none (3 people) to all (2 people). This resulted in the group that met the diagnostic criteria not having statistically higher behaviour scores than the group that did not meet the criteria. Rather, some people aged 40–59, both who met and who did not meet the criteria, had high behaviour decline scores (2 scores of 1.00 in the group that did not meet criteria, and one score of .86 and two scores of .71 in the group that did meet criteria). When the two groups were collapsed and the two age groups were contrasted, those aged 40–59 had significantly higher behaviour domain scores than those under 40 ($t=2.85, df=25, p<.02$).

These analyses and the data in Tables 1 and 2 suggested that individuals might differ considerably with regard to the onset and severity of symptoms in the three domains. If so, such differences might be very important clinical information for both individuals who met and did not meet the diagnostic criteria. A close inspection of the domain scores for all people with DS in both groups revealed that this was indeed the case. The domain scores of eight individuals from the two age groups and the two diagnostic criteria groups are shown in Table 3, by way of examples. Among those who met the diagnostic criteria, memory decline was a strong aspect of meeting the criteria for #6 and #32, but behaviour decline was the strongest aspect for #8 and #37. Among those who did not meet the diagnostic criteria, memory decline (but little or no other declines) were quite evident for #11 and #41. By contrast, behaviour decline (but little or no other declines) was clearly reported for #26 and #46.

Discussion

The Assessment Protocol developed and pilot tested in the present study appears to be a viable method of carrying out an initial assessment of DAT in adults with DS. It is relatively quick and easy to administer, should be a useful tool to help with a clinical diagnosis, and provides helpful information on specific declines in people with DS who are diagnosed with DAT as well as in those who have not been diagnosed.

A number of interesting findings emerge from this study, and three of them are highlighted here. First, like other studies, the data here support the idea that by no means all people with DS who are apparently developing or have developed neuropathological brain changes associated with DAT actually show clinical symptoms of dementia. Only 29.2% of those aged 40–59, and only 15.4% of those in their 30s, met the diagnostic criteria of the Assessment Protocol. Furthermore, a full 41.7% of those 40–59, and 50% of those under 40, had no reported declines at all. These data lend further weight to the argument that there is a strong need to explore the relationship between clinical symptoms of dementia and neuropathological brain changes in a more detailed way.

Second, declines in some, but not all, domains (29.2% of those over 40 and 30.8% of those in their 30s) suggest that symptoms of dementia may not all appear at the same time. Moreover, information in Table 1 suggests that the onset of such symptoms may differ quite substantially among individuals. On the other hand, the decline scores reported in Table 2 quite clearly suggest that observed memory declines, consistent with findings from Dalton, Mehta, Fedor & Patti (1999), might be the most important clinical symptom for people with DS under 40, and that social and behaviour declines might contribute most strongly to the overall diagnosis for many people over 40. Together, these imply a need to examine closely the onset of symptoms associated with DAT in people with DS, with a view to determining general trends and individual differences. They also support the view that, for people with DS, it is possible that some declines may describe a "pre-clinical" stage of dementia (Devenny, Krinsky-McHale, Sersen & Silverman, 2000).

Third, the decline scores provide an interesting way to describe the severity of the overall dementia in individuals, as well as the severity of contributing domains. Table 3 shows clearly that, even for those individuals who met the DAT diagnostic criteria, the overall scores differ. Moreover, the domain scores contribute to the overall scores in quite different ways for individuals. These data support the frequently-stated view that DAT symptoms often present in unique ways in individuals

(e.g., Prasher & Percy, 1999), but it also underscores the need to try to understand better why this occurs and what the implications for clinical practice and practical support may be.

The present study was limited in a number of ways. First, the use of perceptions of others to identify areas of decline carries some risk of bias and misinterpretation. Still, these are the people who know those with DS best and interact with them on a daily basis, and thus are in a good position to notice declines over time. Second, recognizing changes in behaviour in people who have pre-existing disabilities can sometimes be challenging, especially for those who do not verbally express reasons for their changes. On the other hand, participants who are familiar with the daily routines of those with DS were asked to report perceived declines within the context of those routines over a period of at least six months. Third, the number of people with DS (50) was small in this study, and the number of people who met the DAT diagnostic criteria (11) was very small. It would be interesting to replicate this study using a larger sample of people diagnosed with DAT and people who show declines but who have not been diagnosed. Fourth, the presence of depressive symptoms (e.g., irritability, weight changes, and sleep disturbances) in some of those included in this study was difficult to evaluate, in view of the fact that people with DS sometimes show depression that presents as cognitive dysfunction mimicking dementia but that may also be an early sign of dementia. Finally, little validation of the Assessment Protocol, except for content validation and some comparison with the DSM-IV criteria, could be carried out because of the small number of people with reported declines. Any future work using this diagnostic tool should address validation concerns more fully.

On the whole, people with DS who have been diagnosed with DAT or show declines share service needs with other disabled and non-disabled people with DAT. These needs include adequate housing, specific health care, stimulating and challenging activities, maintaining relationships and contacts with friends and family members, and a range of

social supports within a context that is typical of the community (Janicki, 1988; Salvatori, Tremblay, Sandys & Marcaccio, 1998). At the heart of responding well to such needs is good ongoing assessment that uses a variety of assessment techniques. The diagnostic tool reported on here is an example of one such technique.

Despite the fairly high numbers of people with DS who are diagnosed with DAT, it is surprising how little we currently know about them – especially at the individual level. These people remain, for the most part, socially invisible and undervalued, and are seldom the focus of new policy or program initiatives. Tools such as the Assessment Protocol described in this study can help make people with DS who are diagnosed with DAT or show declines more visible, gather information on symptoms related to their diagnosis or declines, and, in time, help us understand the behaviour associated with DAT more fully.

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Correspondence

Ivan Brown
Centre for Health Promotion
University of Toronto
100 College Street, Suite 512
Toronto, ON Canada
M5G 1L5
email: ivan.brown@utoronto.ca

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The Parent Advocacy Scale: Measuring Advocacy in Parents of Children with Special Needs

Jennifer Nachshen
Queen's University

Leah Anderson
and John Jamieson
Lakehead University

Abstract

Due to conservative fiscal policies and the emergence of the empowerment model of family service delivery, advocacy has become increasingly important to parents of children with developmental disabilities. Despite the fact that advocacy training programs such as "Partners in Policymaking" (Zirpoli, Wieck, Hancox, & Skarnulis, 1994) and "Tomorrow's Challenge" (Hixson, Stoff, & White, 1992) are gaining in popularity, the wide-ranging implications of advocacy have rarely been studied. A possible reason for this is that advocacy is difficult to operationalize. The current study extended the work of Balcazar, Keys, Bertram, and Rizzo (1996) by developing a scale for measuring advocacy in parents, the Parent Advocacy Scale (PAS). The PAS has five dimensions: the number of organizations, the focus of actions, membership in organizations, role in organizations, and the centrality of the advocacy role in the individual's life; it also has a total advocacy score. The PAS has good inter-rater reliability and internal consistency. Higher levels of advocacy on the PAS were found to be associated with higher education level and age of parents.

Parents of children with developmental disabilities face unique challenges in raising their child, and have been shown to encounter more stress than parents of non-disabled children (Beckman, 1991; Dyson, 1991). When faced with the challenge of raising a child with a developmental disability, families tend to develop strategies to cope with the stressors in their lives. One such strategy is that many parents become

advocates for their children. Munro (1991) defines effective advocacy as "a non-violent empowerment and support process, through which families with disabled relatives can constructively express dissatisfaction and contribute to creative solutions to problems existing in human service systems" (p. 1). It is generally acknowledged that advocacy is an important tool for parents to gain access to the resources necessary to improve their family's quality of life.

The concept of advocacy is closely related to the empowerment of the parent. Empowerment refers to "a process by which individuals gain mastery over their lives and a critical understanding of the environment" (Zimmerman, Israel, Shulz, & Checkoway, 1992, p. 708). With greater knowledge, confidence, and feelings of control, parents will be more equipped to participate actively in the development of a better life for their child. Empowerment and advocacy have become increasingly important because of the recent shift in family services away from professionally-centred practices, in which family deficits are emphasized and the staff are considered to be the experts. Increasingly, programs are becoming more family-centred, aiming "to bring information, knowledge, skills and access to resources to families so that they may gain greater power over their own lives" (Dempsey, 1996, p. 7). Dempsey describes this as a shift toward enabling practices, which emphasize individual empowerment and focus on competencies, responsibility, knowledge, and a sense of partnership between the professional and the individual or family. According to Munro (1991), learning systematic advocacy approaches will enhance parents' ability to participate in the delivery of services which, in turn, will benefit both the parent and the child. He writes that in this world of rapidly changing ideas and ideologies parents need to become "watchdogs" regarding treatment philosophies and the implications of conservative fiscal policies. As well, Munro asserts that, while the influence of professionals over government policies has decreased, families are gaining leverage in the practice of policymaking.

Recently, professionals have begun to encourage parents to develop effective advocacy skills, and advocacy training programs such as "Part-

ners in Policymaking" (Zirpoli et al., 1994) and "Tomorrow's Challenge" (Hixson, Stoff, & White, 1992) are gaining in popularity. Despite advocacy's admitted importance by both parents and professionals, the area has rarely been studied. One possible explanation for the lack of research in this highly relevant area is the absence of an appropriate measure of advocacy. Without a reliable measurement tool, it is not possible to rate the effectiveness of advocacy both within the families' daily routine and in their interaction with the services provided in the community. In order to enhance the available research on the role of advocacy in families of children with developmental disabilities, an appropriate measure of advocacy must be developed.

The greatest contribution to the conceptualization of advocacy has been made by Balcazar, Keys, Bertram, and Rizzo (1996). Based on their analysis of the Partners in Policymaking program, the authors propose a useful taxonomy of advocate development in order to determine the effect of advocacy training on individuals at the various levels of involvement in advocacy. The Partners in Policymaking program was designed to enhance the empowerment and involvement in policy making of individuals with developmental disabilities and their families. In the taxonomy created by Balcazar et al., individuals are placed in one of three categories depending on a number of factors. Members of the "Beginner" group belong to one organization, are passive members, receive few services, and are involved in less than three advocacy actions during the last nine months. The "Involved" members belong to at least one organization, are active members, obtain services as needed, and are involved in from three to ten advocacy actions to address personal needs during the last nine months. "Activist" members belong to several organizations at local and state levels, have a leadership role, obtain services for local group members, and are involved in over ten actions during the last nine months.

Balcazar et al. (1996) proposed stages of advocacy development are important for the categorizing and training of advocacy recruits. Results of their study demonstrated that previous experience and membership in advocacy organizations predicted the participants' advocacy

performance following the training sessions. For example, more experienced participants would benefit from training at the state and national levels because they had likely previously participated at the local levels. In contrast, less experienced participants would benefit from training at a personal or local level until they have obtained more knowledge and experience.

Balcazar et al. (1996) stages of advocacy development appear useful when utilized for interventions. They allow the participants to be categorized and properly trained, and at a general level, this approach could be quite effective. However, advocacy may be too complex to categorize advocates into three highly specific stages. For example, an individual may be performing at a Beginner level in regard to membership in organizations, and at an Activist level in terms of their actions in the past nine months. The three-stage model provides limited information about advocacy behaviour. A multidimensional construct that measures different aspects of advocacy separately would allow researchers to explore the concept of advocacy more thoroughly.

The taxonomy of advocacy development proposed by Balcazar et al. (1996) describes five dimensions along which participation in advocacy can be measured. The first dimension is the individual's membership in organizations. This can range from non-membership, to roles in organizations at the state level and beyond. The second dimension involves the individual's role in organizations, ranging from passive membership to active participation in leadership roles. The third dimension concerns the number of advocacy actions performed by individuals over a certain amount of time. The activity level of the individuals can range from no advocacy activities, to the maintenance of a level of activity indicative of a full-time career in the area. The fourth area is the focus of advocacy, ranging from an exclusive focus on the immediate family, to a wider focus on the needs of all individuals with developmental disabilities. The final dimension concerns the parents' feeling about the role of advocacy in their life, and their role within the community. While some individuals may feel that advocacy plays a minimal role in their feelings

about themselves and their role in their community, others may define themselves through the advocacy role.

In the present study we developed a multidimensional measure of advocacy, the Parent Advocacy Scale, based on the five dimensions of advocacy behaviour proposed by Balcazar at al. (1996). This scale is designed to measure advocacy in parents of children with developmental disabilities. Preliminary information is provided on the reliability of the PAS and the characteristics of advocates.

Method

Participants

Participants were recruited through organizations in Thunder Bay, Ontario, which serve the needs of individuals with developmental disabilities. A letter describing the study was given to service providers, who were asked to pass on the information to their clients. Both mothers and fathers of children with developmental disabilities were invited to participate, provided that they considered themselves to be actively involved in the care of their child. Participants included 25 mothers and 1 father of 26 children with developmental disabilities. The participants had a mean age of 40 (*SD* = 11.6; range = 23–66 years). Forty percent of the participants had a college or University degree, 38.5% had a high school diploma, one parent had not completed high school, and another declined to respond. The majority of the participants were married (69.2%). The others were either separated (11.5%), single (7.7%), divorced (7.7%) or widowed (3.8%). The children had a mean age of 13 (*SD* = 10.1) and were nearly evenly distributed between males (57.7%) and females (42.3%). The children had been diagnosed with Autism (34.6%), Down Syndrome (15.4%), a developmental disability due to other causes (23%) or developmental disability due to unknown causes (26.9%). The majority of the children continued to live at home with their parents (73.1%). This sample is generally representative of the population, although privacy constraints prevent a more detailed comparison.

Measures

Demographic Information. A family information checklist was used to obtain information regarding parents' age, gender, education, and marital status and the child's age, sex, type of disability and accommodations.

Parent Advocacy Scale. The Parent Advocacy Scale (PAS) was developed for this study and the questions are presented in the Appendix. This scale is used as a structured interview, and contains both closed and open-ended questions. Parents are first asked to describe the advocacy actions they have undertaken in the past nine months. The nine-month time frame was used by Balcazar et al. (1996), and was chosen for this study to be consistent with their work. Following from Balcazar et al., advocacy actions were categorized into five types: a) phone calls, b) office visits or meetings, c) letters and mass mailings, d) media reports, and e) other activities. Within each category, parents were asked how many actions they had undertaken, and approximately when those actions had taken place. They were asked whether their actions responded to personal or family issues, and whether their actions were undertaken for the needs of their own child, or on behalf of other individuals with developmental disabilities. A final question related to the outcome of the advocacy actions.

The second portion of the scale addresses parents' membership in organizations relating to individuals with developmental disabilities. Parents were asked to list and briefly describe the organizations or groups to which they belonged. They were also asked to describe their role in the organization, as well as their level of activity. Finally, parents were asked two open-ended questions about their feelings about advocacy. One question focused on the role of advocacy in parents' lives. The other concerned whether the advocacy activities they had described had altered the way they felt about themselves and their role in the community.

Procedure

Interviews were conducted in the parents' home by the first author. The

PAS was administered as part of a larger battery employed by the author for the purposes of her MA thesis. Parents were given Munro's (1991) definition of advocacy and were asked to consider how it related to their lives over the past nine months. The PAS took approximately 10 minutes to complete. The interviews were tape-recorded and later transcribed to ensure accuracy. All of the interviews were included in the analysis.

Parents' responses to the PAS were scored by two independent raters using the key presented in Table 1. Each dimension was rated on a scale ranging from 0 to 4. The scores on all five dimensions can be added together to form a single score ranging from 0 to 20.

Analysis

The frequency distributions for all measures were examined. Inter-rater reliability for the PAS scores provided by two raters was assessed using Pearson's correlation coefficients. Internal consistency of the PAS was measured using Chronbach's alpha. Finally correlations between PAS scores and demographic measures were computed.

Results

The frequency distributions for each of the 5 dimensions are presented in Table 2. Only one coding category, '3' for Activity Level in Organizations (being president or vice-president) was not assigned to any parents in this sample. For all 5 dimensions, some parents received scores at both extremes. The distribution of scores was normal for Actions and Role of Advocacy, however, Focus of Actions was skewed, with most parents focussing on their own child. Membership in Organizations also had a large modal value of '1'. The Total Advocacy score on the PAS was also normally distributed and had a mean =8.12, (SD = 5.26), with scores ranging from 0 to 20.

The inter-rater reliability coefficients for each of the four dimensions were found to be significant at the $p < .01$ level. The correlation between

Table 1
Scoring of Parent Advocacy Scale

Score	Actions	Focus of Actions	Membership in Organizations	Activity level in Organizations	Role of Advocacy
0	No advocacy Actions	Focus is only on own child	Not a member of any organizations	None or irregular attendance	Advocacy is irrelevant to person's life.
1	Fewer than five actions in nine months	Focus is mainly on own child, few actions on behalf of others	Membership in 1-3 support-based organizations	Regular attendance at meetings	Advocacy is an unwelcome necessity.
2	Actively makes efforts to access services through phone calls and meetings	Focuses both on own child and other individuals at the local level.	Membership in numerous advocacy-oriented, local organizations	Has an assigned role (secretary, treasurer, etc.) in 1-3 organizations	Advocacy is beneficial, but should be used cautiously.
3	Participates in a wide variety of advocacy activities	Works beyond the local level for own child, as well as other individuals	Membership in many local organizations, as well as some broader organizations.	Participates in role as president or vice-president of an organization.	Advocacy is very important and has had an effect on person's self-image.
4	Spends most of their time involved in advocacy. It may be a career.	Often works at the national or even international level on behalf of other individuals	Member of numerous local, provincial, and national organizations.	Extremely active, may have created their own grassroots organization.	Advocacy is central to self-image and has dramatically impacted view of self and role in the community.

Table 2
Summary of Scores on the PAS (n=26)

Score	Action	Focus of Actions	Membership in Organizations	Activity Level in Organizations	Centrality of Advocacy Role
0	3	13	4	8	3
1	6	5	15	6	6
2	9	3	3	8	7
3	5	2	3	0	5
4	3	3	1	4	5
Mean	1.96	1.11	1.31	1.46	2.11
S.D.	1.18	1.42	1.01	1.36	1.31

the two raters was $r = .95$ for Actions, $r = .89$ for the Focus of Actions, $r = .87$ for Membership in Organizations, $r = .90$ for Activity Level, $r = .82$ for Role of Advocacy, and $r = .95$ for the total score.

The internal consistency of the scale was acceptable ($a = .87$). The five dimensions showed moderate inter-correlations, ranging from $r = .37$ for Actions and Role of Advocacy, to $r = .76$ between Actions and Focus of Actions.The inter-item correlations are presented in Table 3.

To gain a preliminary understanding of the characteristics of individuals who are involved in different levels of advocacy, correlations between the PAS and demographic measures were examined. It was hypothesized that parents with a higher level of education would be more likely to participate more actively in advocacy related pursuits. Parents' level of education was found to be significantly correlated with parents' level of activity in organizations ($r = .61$, $p < .01$), the centrality of the advocacy role ($r = .80$, $p < .01$), and the overall score on the advocacy scale ($r = .58$, $p < .05$). A higher level of education was related to higher scores on all three dimensions. Parental age was also related to increased activity in organizations ($r = .52$, $p < .01$) and an increased total score on the advocacy scale ($r = .46$, $p < .05$).

Table 3
Inter-item Correlations for PAS Dimensions (n=26)

	Action	Focus of Actions	Membership in Organizations	Activity Level in Organizations	Centrality of Advocacy Role
Action	1				
Focus of Actions	0.7632	1			
Membership in Organizations	0.6462	0.586	1		
Activity Level in Organizations	0.4333	0.6105	0.5895	1	
Centrality of Advocacy Role	0.3655	0.6164	0.4265	0.7327	1

Discussion

The present study was an attempt to create a multidimensional scale to measure advocacy in parents of children with developmental disabilities, based on the work of Balcazar et al. (1996). The findings showed that it was possible to develop such a scale, the Parent Advocacy Scale (PAS) which has high internal consistency and good levels of inter-rater reliability. Preliminary analyses suggested that higher levels of advocacy are found in parents with more education and who are older. The study also provided information about the specific dimensions of advocacy that are related to education and age. The PAS dimensions indicated that education is related to the roles that parents play in organizations, and their feelings regarding the importance of the advocacy role in their lives. Parents with higher levels of education tend to have more active roles in organizations, and consider the role of advocate as more central in their feelings about themselves and their role in the community.

The PAS was found to have good reliability. The finding that older parents and parents with more education are more involved with advocacy provides some indication of the predictive validity of this instrument, and its potential for differentiating among the various aspects of advo-

cacy. Although the scale appears promising for studies that evaluate the effectiveness of advocacy, and the effects of advocacy on a family's functioning, further evidence of its validity is desirable. Concurrent validity could be obtained through comparisons with scales designed to measure empowerment (advocates should be more empowered than non-advocates), knowledge of rights and responsibilities (advocates should be more aware of their rights than non-advocates), and service use (advocates should access more services than non-advocates).

In this study, the PAS was administered as a structured interview in order to give parents the opportunity to speak freely regarding their experiences. Although the empowerment orientation of interview-based research was ideal for this study of advocacy, future research might also address the utility of the PAS as a self-administered questionnaire. This could be achieved through a simple reformatting of the questions in the Appendix.

The primary focus of the current study was to present a measure of advocacy following from the work of Balcazar et al. (1996). The reliability estimates observed in this study is promising, but must be confirmed by future research using larger samples. The work of Balcazar et al. (1996) has proven useful in categorizing parents for placement in advocacy training programs. The PAS is an extension of their work, which contains more detailed information about levels of advocacy and types of advocacy behaviours. One reason for wanting such information is that it would be useful in investigating issues such as the effects of advocacy on family stress and quality of life, and to identify factors that might mediate or moderate the stress reducing or stress augmenting effects of advocacy. The PAS shows promise for such investigations.

Appendix: Advocacy Questionnaire

In each of the following categories, describe the advocacy actions you have undertaken in the past 9 months:

Categories

A. Phone calls
B. Office Visits or Meetings
C. Letters and Mass Mailings
D. Media Reports
E. Other Activities

Actions undertaken in the past 9 months

- How many?
- Was this action in response to a personal or family issue?
- Was this action undertaken for the needs of your child, or on behalf of all individuals with developmental disabilities?
- What was the outcome of this action?

- Do you belong to any organizations or groups regarding children with developmental disabilities?
- What is your role in this group?
- How active are you in this group?
- How do you feel about the role of advocacy in your life?
- Have the advocacy actions you have described altered the way you think about yourself and your role in the community?
- Do you believe that you have the power to improve the quality of life of your child?

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Correspondence

Jennifer Nachshen
Department of Psychology
Queen's University
Kingston, ON
K7L 3N6

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A Tale of Two Sisters: Quality of Life Within Two Systems

Robert J. King
North Bay Psychiatric Hospital

Robert Barnett

Abstract

Recent publications by the Ontario Ministries of Health and Community and Social Services have the potential to significantly affect the quality of life of adults with developmental disabilities and mental health concerns. This article, in a case-report format, contrasts the life experiences of two sisters, each with developmental disabilities, who have had similar mental health concerns addressed from differing systemic perspectives. The promise for an enhanced impact on quality of life through inter-ministerial collaboration is highlighted.

Background Information: Evolving Service Delivery Systems in Ontario

This past decade in Ontario has witnessed the publication of multiple policy documents by the Ontario Ministries of Health (MOH) and Community & Social Services (MCSS), each with the potential to significantly affect the quality of life of adults with developmental disabilities and mental health concerns living in this province. The concept of *Quality of Life* itself has been identified as providing a fundamentally positive and growth-oriented principle that can be the basis for service delivery policy and practices (Schalock, 1997, p. 15). Core dimensions of the concept of quality of life, correlated examplars of these dimensions and objective measuring systems are now evolving, with an emphasis on ensuring interventions that seek to improve quality of life and recognize the degree to which they affect quality within the whole of the

person's life (Brown & Renwick, 1997, p. iv). From a clinician's perspective, Hollins (1997) has offered the opinion that a distinction must be maintained between the quality of services offered and the quality of the experience of that service by the individual (p. 35). Emphasizing the critical need to facilitate individual choices as a driving force in improving quality of life, Hollins (1997) pointedly reminds us that our attitudes and responses to people with developmental disabilities can have a profound effect upon the quality of life by enabling or disenabling their access to those supports that they need (p. 40).

Policy Documents, Guidelines, and Reforms

Putting People First: The Reform of Mental Health Services in Ontario, released in June 1993 by the MOH, proposed to reform Ontario's mental health system by placing individuals within mental health systems at the centre of the system, while balancing and coordinating services. The emphasis of this document on a shift from an institutional to community-based system was followed by the July 23, 1996 announcement by MCSS of a 4-year plan to move 978 individuals with developmental disabilities from institutional settings into the community. Moving people back into their communities will bring many of them closer to their families and friends. It will provide them with an opportunity of having a better quality of life, commented then MCSS Minister, Mr. David Tsubouchi (1996). To date, the MCSS deinstitutionalization process in Northeastern Ontario has involved many individuals with complex mental health needs, the intensity of which predictably increased in the context of critical periods of transition and readjustment in the lives of these individuals. The ministerial promise of a redirection of funds from institutions to local community agencies and services has yet to be adequately fulfilled, leaving the implementation of person-centered support plans difficult and at times, impossible to achieve.

Making Services Work for People sets out a new framework for services for children and people with developmental disabilities, requiring local systems to provide essential supports to those in greatest need, and to provide investment supports to reduce or eliminate the need for future

services (MCSS, 1997c). A systemic shift to a proactive, coordinated, streamlined system responding to individual needs appears to mirror the principles of mental health reform. A reinvestment strategy for this vision is articulated in the documents *Reinvestment Strategy for Adults with a Developmental Disability (*MCSS, 1997c) and *Reinvestment Strategy for Children and Youth (*MCSS, 1997d*)*. Individuals of focus for reinvestment in the former document are adults with developmental disabilities who need supports to help them live independently within a context of enhanced quality of life (MCSS, 1997c). Five types of supports and six system features are identified for increased reinvestment. These documents appear to be heavily reliant upon factors identified with an improved quality of life through research conducted at the Centre for Health Promotion (1994) at the University of Toronto, Ontario. No direct reference is made in these documents to addressing existing mental health needs to reduce future needs for long-term or intrusive supports. Rather, specific reference is made in these documents to strategies aimed at reducing marked behavioral problems in nonverbal individuals, to increase the quality of their lives and to reduce the burden born by a family supporting a relative with behavioral challenges in their home.

To address mutual issues of concern, the Ontario MOH and MCSS jointly released *Policy Guideline: The Provision of Services for Persons with a Dual Diagnosis*, a commitment to the development of a collaborative, cross-sectional approach to ensure individuals with developmental disabilities and mental health concerns have access to services and supports in either or in both the mental health and developmental sectors, as their needs require (MCSS, 1997b). This guideline is intended to support restructuring initiatives in both the MOH and MCSS, allowing interministerial coordination at a provincial, regional and district level. The potential positive impact of this guideline on the quality of life of individuals with developmental disabilities hinges upon the outcome of its implementation. This document fails at the present time to address the need for cross-sector services, appearing to perpetuate the delineation of specific roles of organizations and agencies within each Ministry, despite initially identifying this division as a barrier to serving

individuals with developmental disabilities and mental health concerns. To support the community integration of individuals currently living in MOH and MCSS institutions, the implementation of expressed principles in this document must address a number of agendas. These include the cost-saving agendas of Mental Health Reform and *Making Services Work for People*, the recommendations of the Hospital Restructuring Commission regarding closure of provincial psychiatric hospitals and the concerning selective focus of *Making Services Work for People*. The quality of many lives is dependent upon the development of innovative, interministerial initiatives at a primary, secondary and tertiary care level, in the context of continued efforts to optimally define and measure individual quality of life.

Case Report

Complex mental health needs of adults with developmental disabilities have, to date in Ontario, been addressed from diverse perspectives with respect to the particular Ministry (either the MOH or MCSS) supporting the individual. These case reports highlight the differences within the lives of two sisters with lifetime support from both systems, support only recently offered in a collaborative, interministerial manner.

Historically, the establishment in 1876 of an Asylum for Idiots in Ontario marked the first administrative distinction within social service policy in Canada between individuals with developmental disabilities and those with mental illnesses. Until 1974, however, all services to individuals with either developmental disabilities or mental health concerns were funded by the Ontario MOH and its bureaucratic predecessors. In 1974, the Developmental Services Act mandated that all institutional and community services to individuals with developmental disabilities were to be provided by the MCSS (Goldberg & Stavrakaki, 1992). In the subsequent two decades, the Ontario MOH and MCSS have struggled to collaborate, initially with little systemic success, to meet the needs of individuals with developmental disabilities and challenging behaviors. Many of these individuals are now retrospectively

being identified as having developmental disabilities and specific mental health concerns.

The Tale of Two Sisters

H & M are biological sisters, born in a small community in Northeastern Ontario, the first and third children in the sibline of six. H is now 61 years of age and single, M 56 years of age and widowed. Birth and early developmental histories of each are unknown. Information recorded by intake coordinators during their respective initial admissions to psychiatric facilities suggests that a maternal grandfather had been institutionalized and that a maternal aunt had a history of a bipolar disorder. Psychometrically, although not formally tested until age 47 (at a MCSS regional facility), intelligence testing suggested a moderate degree of disability for H. In contrast, testing at a provincial psychiatric hospital at age 27 demonstrated a mild degree of disability for M.

According to provincial psychiatric hospital records, M left home at age 16 to enter a convent but was not able to complete a course of training there and returned home until age 23. At this time she was first admitted to a regional psychiatric hospital (MOH), located several hundred kilometers from her home community, after threatening a sister with a gun. She was described by her attending psychiatrist as lacking in obedience, exhibiting childish behavior, was overactive and overtalkative and interfering. A diagnosis of Mental Deficiency Without Psychosis or Epilepsy, Moron Level (the accepted nomenclature at the time) was established. Thioridazine (Mellaril) a sedating antipsychotic was prescribed. She has subsequently continued to receive a variety of antipsychotic medications until 1996. Between the ages of 23 and 32, M was admitted to a regional psychiatric hospital (MOH) on 11 occasions, with little or no professional support in her home community between admissions. At age 34 in 1974, with the implementation of the Developmental Services Act, she was transferred to a regional MCSS facility for the mentally retarded and resided in this facility and a subsequent MCSS facility for 8 years until age 42. Highs characterized by self-talk, aimless wandering, trouble sleeping, irrational feelings and conversations

were documented in psychological and vocational service progress notes but were not reassessed psychiatrically in these settings.

Diagnoses during M's multiple psychiatric admissions included schizophrenia and mental retardation. Treatments with antipsychotic medication, environmental manipulation, habilitative changes and mood and behavioral monitoring, were tried. Community integration into a group home setting (under the jurisdiction of an Association for Community Living, a MCSS funded agency) at age 42 was followed by a marriage and independent living with her spouse for two years until his death when M was 48 years old. A re-entrance into the MOH system, with the diagnosis of a hypomanic phase of a bipolar disorder, appeared precipitated by stress associated with marital conflict, at age 47. A mood-stabilizing medication, Lithium Carbonate was first prescribed at this point.

Following the diagnosis of her Bipolar Disorder, M was admitted on 8 additional occasions, between age 48 and the present time, to a regional psychiatric facility (MOH) while living in a group home (MCSS) several hundred miles from this hospital. Community-based care of her bipolar disorder was provided by her family physician until age 48, with little communication or coordination of care occurring between her hospital-care providers and her family physician. At age 48 she was referred to an outreach tertiary care program (MOH) providing psychiatric assessments and treatment specifically to individuals with developmental disabilities and mental health concerns. The development of moderate renal failure and a lack of demonstrated efficacy in response to the continued prescription of Lithium Carbonate resulted in its discontinuation and substitution with an alternative mood stabilizing drug, Valproic Acid. The rapid cycling nature of her bipolar disorder (4 or more episodes of hypomania or depression in one year) was recognized at this time. The addition of Carbamazepine (Tegretol), a second mood stabilizing medication, was guided by the introduction of a formal objective monitoring system designed by a psychometrist working with a regional MCSS professional resource team and implemented by local Association for Community Living residential counsellors. Essential

hypertension, a head injury resulting in a delirium and persistent mild renal failure, further complicated her course. With continued assistance from a local psychiatrist, her family physician and extensive support from a local Association for Community Living, her mood has stabilized over the past 18 months.

H was admitted to a psychiatric hospital in Southern Ontario (MOH) at age 18, in the absence of an established regional mental health centre in Northeastern Ontario at the time, isolating her from community and family supports. She was described as angry, disobedient, experiencing tactile and auditory hallucinations in admitting records. She was given a diagnosis of a mental deficiency without psychosis or epilepsy, paralleling her sister's diagnosis and was prescribed Chlorpromazine, a sedating antipsychotic medication. Between the ages of 19 and 39 H was admitted to a regional psychiatric facility (MOH) on 5 subsequent occasions. A variety of antipsychotic medications were prescribed intermittently during this time frame. A diagnosis of severe mental retardation was maintained to age 39. With the passing of the Developmental Services Act, at this time, she was transferred to a mental retardation unit (MCSS) created adjacent to the MOH psychiatric hospital in which she had formally been admitted. At age 39 a diagnosis of a bipolar disorder was established by a family practitioner and Lithium Carbonate was prescribed. She was subsequently transferred to a separate regional MCSS institution where she resided from age 40 to age 57. Her course here was complicated by a head injury at age 41 with a resultant seizure disorder, which is managed medically with Dilantin, an anticonvulsant. She was ultimately transferred at age 57 to a group home affiliated with an Association for Community Living, differing from the association supporting her sister. She continues to reside in this setting. While residing in this setting at age 58, H was finally given a formal diagnosis of a rapid-cycling bipolar disorder. An objective charting mechanism was implemented and presented to the consulting psychiatrist to facilitate a diagnostic and pharmacological reassessment through the tertiary care service, which had provided care to her sister. It would require an additional year and a chance comment made by a caregiver, who had known

both sisters historically in different contexts, to establish their common genetic backgrounds. (Given M's marriage, M and H had different surnames at this time). At the time of H's tertiary care psychiatric assessment, concern was expressed by her caregivers regarding an increase in her cycling frequency, although a subsequent retrospective review indicated that the criteria for a rapid-cycling bipolar disorder had been met since at least age 41. Multiple pharmacological changes over the ensuing three years resulted in the current prescription of Risperidone, an atypical antipsychotic medication with mood stabilizing properties and two mood-stabilizing agents, Carbamazepine and Valproic Acid. Her course was further complicated by essential hypertension, fluctuations in the optimal control of lithium-induced hypothyroidism, intermittent exacerbations of her seizure disorder and Carbamazepine-induced hyponatremia (a low serum sodium level). Her caregivers believe that there has been a significant decrease in the intensity of episodes of hypomania and depression during this time frame but no change in her cycling frequency. Over the past three months a decline in her adaptive living skills has been noted in the absence of an identifiable metabolic or neurological cause.

Discussion

This case illustrates the complicated life courses of two biological sisters with differing degrees of developmental disabilities and severe forms of the same psychiatric disorder. Their life experiences differ considerably in the amount of time their needs have been addressed by the Ministries of Health and Community & Social Services respectively. They share, however, the common experience that until very recently the service provided by these two systems to address their habilitative and mental health needs has been offered in a segmented, noncoordinated manner.

Quality of Life Implications – Historical

It would appear reasonable to question to what extent the quality of M & H's lives were adversely affected by both a delay of a formal recognition

of their developmental disabilities and the diagnosis of their psychiatric disorders. M has been admitted to a provisional psychiatric hospital (MOH) on 18 occasions to date, intermittently throughout her life course, while living in her family home, in independent settings and in a group home setting. Significantly, during her 8-year placement in a MCSS regional institution, as her chronic-relapsing bipolar disorder evolved, she received no formal psychiatric care. In contrast H has been admitted to a provincial psychiatric hospital on 5 occasions while living in her family home. During the 18 years of support she received in a MCSS institution and the subsequent 2 years in which she has lived in a group home setting she has not been admitted to the psychiatric hospital, although the course of her bipolar disorder has paralleled that of her sister's disorder. The delay in the formal recognition of their bipolar disorders (M diagnosed at age 32 and H at age 39) is likely partially attributable to the concept of diagnostic overshadowing (Reiss, Levitan, & Szyszko, 1982). The failure on behalf of both MOH and MCSS caregivers to acknowledge their shared biological vulnerabilities to a bipolar disorder delayed the implementation of a psychotropic medication regime for H which had been demonstrated to be beneficial for M. In the early phases of the bipolar disorder of both M and H, the MOH failed to integrate community and hospital-based supports (likely the result of a combination of therapeutic nihilism engendered by a disbelief at the time that individuals with developmental disabilities could experience concurrent psychiatric disorders and a lack of community-based resources). Multiple admissions for both the sisters resulted in isolating them from both family and community supports. In the context of the Developmental Services Act, psychometric testing appeared to systemically deny access to psychiatric care for H and for a number of years, for M. It is possible that the course of H's illness, in particular, could have been favorably altered if she had greater access to psychiatric care during her years of MCSS institutionalization. Alternatively, her MCSS institutionalization may have spared her the adrenergic adverse effects she has more recently experienced (Carbamazepine-induced hyponatremia), and may have allowed an improved quality of life through the provision of an enhanced habilitative support network.

Quality of Life Implications in the Future

Both H and M are now community integrated, receiving coordinated, intensive care from interministerial teams formed on an ad hoc basis in recognition of a need to address their mental health concerns as one component in an attempt to improve the quality of their lives. Through decreasing the distress they have experienced in association with periods of hypomania and depression, maintaining their access to community-based leisure, spiritual and vocational opportunities and through allowing them to experience continuity in interpersonal relationships, perhaps for the first time in their lives, their quality of life appears to have improved. The quality of life of future generations of individuals with developmental disabilities, who are at significant risk of developing mental health concerns, is now critically dependent on an implementation of MCSS and MOH policy guidelines organized around quality of life issues. Concurrently extreme caution will be required to avoid diminishing the quality of life of individuals with developmental disabilities and mental health concerns currently living in either community-based settings or continuing to live in MOH and MCSS hospitals and institutions.

Changes in government policy in other jurisdictions, emphasizing a shift from institutional to community-based care, has not always systemically improved the quality of life of individuals on a person-centered basis. Legislative changes in Denmark and Sweden, premised on the principles of normalization and the right of individuals with developmental disabilities and mental health concerns to access generic supports, (while at the same time eliminating specialty care) have had controversial results (Day, 1992). The pervasiveness of support needs for mental health concerns in individuals with developmental disabilities following deinstitutionalization in a Canadian context has been clearly demonstrated (Fotheringham, Abdo, Ouelette-Kuntz, & Wolfgarth, 1993). Strauss and colleagues (Strauss, Shavelle, Baumeister, & Anderson, 1998) have recently documented concerning increases in mortality rates in individuals transferred to community settings from institutional settings in California.

Recommendations

- It is critical for both the MCSS and MOH to recognize that the development of *interministerial* teams composed of individuals with experience and expertise in addressing both mental health concerns and the primary needs of individuals with developmental disabilities, is crucial in optimizing the community-based quality of life of these individuals. Continued insistence on the maintenance of divided ministerial responsibilities and mandates will be detrimental to this goal.
- Given the demonstrated vulnerability of individuals with developmental disabilities to experience a high prevalence of chronic relapsing psychiatric disorders, a continuum of services ranging from community-based care to hospital-based intensive care is necessary to meet the needs of these individuals. A continuum of care lacking dedicated beds to ensure safety in the context of severe self-injurious or aggressive behaviour will fall short in attempts to meet these needs.
- Inherent in the recognition that individuals with developmental disabilities may experience fluctuating degrees of need in the context of relapsing, recurrent psychiatric disorders is the need for the development of a community-based habilitative support system with the capacity to provide varying degrees of support over lifetimes.
- The need for enhanced communication between community-based and facility-based services within and between both the MCSS and MOH, must be addressed. Individuals leaving MCSS or MOH institutions or hospitals with complex mental health needs require community support networks in advance of their discharge.
- A process allowing cross-sector reciprocating training initiatives must be endorsed by both ministries. A firm financial commitment to this endorsement is now required.
- It is incumbent upon individuals working within both systems to acknowledge that they are supporting the same individuals whose quality of life is entitled to be better served than history has allowed.

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Correspondence

Dr. Robert King
North Bay Psychiatric Hospital
Box 3010 Hwy 11 North
North Bay, ON
P1B 8L1
email: Robert.King@MOH.GOV.ON.CA

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Catalysts for Education in Developmental Disabilities: Interim Report

Bruce D. McCreary
Queen's University

The Donald Gordon Centre at Queen's University in Kingston was the site of a search symposium" about catalysing education in developmental disabilities, November 3–5, 2000. Sixty health care educators and other professionals with interests in the field across Canada, participated. Much of the symposium, in keeping with the "search" format, involved small group discussion on ways to remedy acknowledged shortcomings in teaching about developmental disabilities across Canada.

Bruce McCreary, Chairman of the Division of Developmental Disabilities in the Queen's Department of Psychiatry, opened the symposium with a perspective entitled "Where are we now?" He noted that developmental disabilities, recently reported to be responsible for 8.1% of health care spending in the Netherlands, receives far less attention than it deserves in Canadian Academic Health Science Centres (AHSC's). Noting that what was needed for improvement was reasonably clear, he suggested that how to achieve the necessary enhancements was the challenge for the symposium. He proposed a manifesto as a focus for deliberations during the symposium and for post- symposium advocacy by participants once they returned to their own provinces and AHSC'S:

> "Persons with developmental disabilities have complex and sometimes unique health problems. Where there is neglect of this issue in the training of 'human services' professionals, the result is an undue burden for the disabled individual, for his/her family or caregivers, and for the community where they live. Given that such neglect is unacceptable in Canada, where a life of dignity in the community is the

> goal for all persons with developmental disabilities, it is the commitment of this Academic Health Sciences Centre that relevant instruction will be provided to all professionals who study here."

Sheila Hollins, Professor of Psychiatry of Leaming Disability at St. George's Medical School, University of London, reviewed her UK experience in teaching medical students about developmental disabilities. She stressed the importance of "hands on" practicum teaching so that students might become comfortable in communicating with persons who have significant intellectual and language impairments. She assigns her students in pairs to meet disabled persons living in the community where they complete health checks. She observes that when the students' experiences are reported back to small discussion groups, there is clear evidence of awareness about the individual's special needs and what will be required to meet them in practice. Professor Hollins also outlined an introductory teaching session that is provided to all health care professionals including nurses, speech language pathologists, other therapists and medical students. With this basic orientation students can then proceed to clinical teaching relevant to their particular discipline such as that described for medical students.

Following Professor Hollins' lecture, small group discussion identified the important gaps in contemporary instruction across Canada:

- many students miss out entirely on instruction in developmental disabilities.
- when instruction is provided, it reflects the availability of faculty members who happen to have a special interest in the fleld.
- instruction about adult problems lags far behind that provided about child/family problems.
- there is little or no coordination of effort by instructors from various health care disciplines within individual centres.
- there is little infrastructure to foster coordination of effort and to facilitate supervision in practicum settings.

- government and agencies with obvious interests in ensuring the availability of health care professionals do little to create attractive "career paths".

Francine Knoops and Pamela Forsythe, representing the Canadian Psychiatric Association, related their experiences with linking to mental health advocacy groups to lobby for government support for improved services for individuals with mental health problems. Noting that provincial governments are responsible for health, social services and education, they encouraged approaches that would align professional and advocacy group interests before proceeding to lobby for financial support. Federal government interests are rather more specialized, particularly with respect to human resources development.

Further small group discussion was directed to the development of approaches that would address the gaps already identified. Faculty associated with an academic unit that has been operating for several years at Queen's outlined their experiences in associating service, research and organizational endeavours with the education of a variety of health care professionals. The key outcomes generated in small group discussions and plenary sessions included:

- An immediate need to engage all faculty with a special interest in developmental disabilities in a given AHSC in informal discussions that will promote the formation of an interdepartmental, interdisciplinary academic unit.
- A need to inform the AHSC administration of the desirability of a formal agreement that would ensure cooperation by a relevant provincial government ministry or ministries, one or more agencies that would serve as practicum sites for instruction, and the proposed academic unit.
- The creation of a network of interest in these matters across Canada by building on the relationships established during the symposium.

The Developmental Consulting Program at Queen's will serve as a focus

for further networking. Discussions have been initiated on the development of a professional association relevant to the field of developmental disabilities, the preparation of models of care and clinical guidelines of use across Canada, and the sharing of curricula/programs of instruction between the sixteen Canadian Academic Health Sciences Centres. The full conference proceedings and background chapters prepared prior to the symposium by Queen's faculty will be published in early 2001.

> "The future is not some place we are going to, but one we are creating. The paths are not to be found, but made and the activity of making them changes both the maker and the destination."
>
> – John Scharr