

Adults with Down Syndrome: a Comprehensive Approach to Manage Complexity

Running title: Geriatric care for Adults with Down Syndrome

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1. adults with Down syndrome present with a high level of clinical and functional complexity
2. a 'geriatric approach' based on a comprehensive assessment and management could likely deliver the highest quality care in these subjects

Abstract

Background: Down syndrome (DS) is characterised by premature aging that affects selected organ systems, and persons with this condition can present patterns of comorbidities and deficits often observed in the older population without DS. However, information on the characteristics of adult persons with DS is limited.

Objective: Describe characteristics of adults with DS collected with a standardised, comprehensive assessment instrument.

Design: Cross sectional study

Setting and Subjects: 430 adults with DS (age range 18/75 years) from 3 countries (Italy, n=95; United States, n=175; Canada, n=160).

Methods: A standardised assessment instrument (interRAI Intellectual disability, or interRAI ID) was used to assess sample characteristics.

Results: Mean age ranged from 35.2 (SD 12.0) years in the US sample to 48.8 years (SD 9.0) in the Canadian sample. Most participants in the Italian and US sample were living in private homes, while more than half of those in the Canadian sample were institutionalised. Prevalences of geriatric conditions, including cognitive deficits, disability in the common activities of daily living, symptoms of withdrawal or anhedonia, aggressive behaviour, communication problems, falls and hearing problems were high in the study sample. Gastrointestinal symptoms, skin and dental problems and obesity were also frequently observed.

Conclusions: Adults with DS present with a high level of complexity, which may suggest the need for an approach based on a comprehensive assessment and management that can provide adequate care. Further research is needed to understand better the effectiveness of such an approach in the DS population.

Introduction

Life expectancy of persons with Down Syndrome (DS) has progressively increased in the last century, from 9 years in 1929 to almost 60 years in 2013 (Englund et al., 2013; Glasson et al., 2002; Yang et al., 2002). Such an exceptional increase has led some investigators to forecast that in a few decades persons with DS could live as long as the general population (Bittles and Glasson, 2004). Given these epidemiological changes, DS should be no longer considered a “paediatric” condition, but rather a condition that affects the whole life span. DS is considered a “segmental” progeroid syndrome, in which a premature aging affects selected organ systems, and persons with this condition might present patterns of comorbidities often observed in the older population (Carfi et al., 2017; Picciotti et al., 2017; Real de Asua et al., 2015; Schoufour et al., 2016; Vetrano et al., 2015). The clinical picture is often complicated by the presence of functional deficits, behavioural symptoms and nutritional and social problems (Carfi et al., 2014). This level of complexity requires the use of a comprehensive approach, in order to have a full understanding of the issues and needs of adults with DS (Carfi et al., 2015; Covelli et al., 2016). Such an approach is commonly adopted in geriatric medicine. The aim of the present study is understanding whether the use of a standardised comprehensive assessment tool on a sample of adults with DS from 3 countries (Italy, United States (US) and Canada) could provide useful information on their functional impairment and comorbidities.

Materials and Methods

Participants

Data on participants with DS were collected in three countries (Italy, US and Canada).

Italy

Participants were adults with DS, aged 18 or older, assessed at the Day Hospital (DH) of the Geriatric Department of Policlinico A. Gemelli, Università Cattolica del Sacro Cuore in Rome. No specific inclusion criterion was required to be admitted to the DH except for aged 18 or older. Participants did receive a clinical assessment, following a standardised protocol, including blood sample, electrocardiogram, ophthalmologist, dentist and ear-nose-throat specialist consultation, nutrition status and body composition measurement, echocardiography, and a comprehensive assessment by the use of the InterRAI Intellectual Disability (ID) instrument. Adults with DS were directed to the DH through DS associations and from family physicians. Data on 95 adults with DS that completed clinical assessment are presented in this manuscript.

US

The US data represents the full 2013 state-wide census of adults (i.e., 18+ years old) receiving intellectual/developmental disabilities services funded by the State of Arkansas Division of Developmental Disability Services, with assessments using the interRAI ID performed by trained professional assessors under contract to the state. These data include people who lived in private homes, staff homes, community residential settings, state Human Development Centers, homeless persons, and people classified as living in “other” arrangements. We report here on the 175 persons with DS in this population.

Canada

Data from Canada are based on studies of adults with intellectual or developmental disabilities living in the community and specialised institutions (Langlois and Martin, 2008). The community data comes from two separate studies, one of which focused exclusively on adults aged 50 years or more (the other study sample included adults younger than 50 years). All persons living in Ontario’s institutions were assessed, so the data represent the full population; note that these specialised institutions have since closed. Assessments were collected by front-line staff who had received extensive training related to the interRAI ID. A total of 160 adults with Down syndrome were identified in the data.

InterRAI ID

Adults with DS in the study were assessed using the interRAI ID instrument, which contains over 350 data elements including socio-demographic variables, numerous clinical items about physical and cognitive status, functioning, behaviours, as well as signs, symptoms, syndromes, and treatments being provided (Martin et al., 2007). Items are completed by an assessor based on history and basic signs and symptoms (e.g. face expressions, disruptive behaviours, pain frequency and intensity, etc.) collected directly from the subject or by an informant; a number of questions are asked directly to the subject concerning his/her preferences, outlook and wellbeing. Informants are selected among the closest relatives (parents or siblings) or long standing caregivers. All assessors followed a standardised training for administration of the interRAI ID. In US and Canada data, collection was performed by trained assessors in the field of intellectual and developmental disabilities, and by trained physicians in Italy.

Clusters of items are set up in algorithms and scales to deliver clinically relevant diagnostic triggers to inform subsequent clinical evaluation; such scales have proven internally consistent and valid among

adults with ID(Langlois and Martin, 2008; Martin et al., 2007). The cognitive performance scale (CPS) informs on current cognitive status(Morris et al., 1994). The scale scores were categorised into mild to moderate cognitive impairment (CPS scores 2 to 4) and severe cognitive impairment (CPS scores ≥ 5). The seven point Activities of Daily Living (ADL) Hierarchy Scale informs on functioning - i.e., independence in ADLs. Score ≥ 2 in the ADL Hierarchy Scale was used to identify participants requiring assistance in ADL(Morris et al., 1999). The Depression Rating Scale (DRS) was used to assess the presence of depressive symptoms with scores ≥ 3 indicative of depression(Burrows et al., 2000). A score from 1 to 4 on the Aggressive Behaviour Scale was used to define mild/moderate aggressive behaviour and scores ≥ 5 to define severe aggressive behaviour(Frederiksen et al., 1996). On the Social Withdrawal Scale, scores ≥ 1 identified symptoms of withdrawal or anhedonia(Rios and Perlman, 2017). Finally, mild/moderate communication problems were defined by Communication Scale scores from 2 to 5 and severe problems by scores from 6 to 8(Frederiksen et al., 1996).

Descriptive analyses are present in this manuscript. Data were analysed using SPSS (version 18.0) for the Italian data, and SAS (Version 9.3) for the US and Canada data.

Results

Mean age of persons with DS ranged from 35.2 (SD 12.0) years in the US sample to 48.8 years (SD 9.0) in the Canadian sample (Table 1). Most participants in the Italian and US sample were living in private homes, while more than half of those in the Canadian sample were institutionalised; these differences being related to different sample recruitment and not to different social care models. Geriatric conditions occurred at high rates in the study samples, including cognitive deficits, ADL disability, symptoms of withdrawal or anhedonia, aggressive behaviour, communication problems, falls and hearing problems, while rate of urinary and faecal incontinence varied across study sites, being more commonly observed in the US and Canadian samples. Gastrointestinal symptoms, and skin and dental problems were also frequently observed, while acute conditions such as psychiatric symptoms (which includes delusions, hallucinations or abnormal thought process), dizziness and pain occurring in the three days before the assessment were uncommon. Obesity (Body Mass Index, BMI ≥ 30 kg/m²) was also highly prevalent (BMI data not measured in Canada). Prevalence of physicians visits in the last 90 days varied across study sites ranging from 37.9% in Italy to 81.1% in the US.

Discussion

We present here data on a large sample of adults with DS which demonstrate that they present with a high level of comorbidity and complexity. Despite variation in different study sites related to the enrolment procedures adopted, the data clearly show that adults with DS are characterised by presence of functional and cognitive impairments, and common occurrence of mood disorders, oral and nutritional problems, and geriatric syndromes (including anhedonia, aggressive behaviour, falls, incontinence, communication and hearing problems) - and at fairly young mean ages. This is in line with previous studies showing higher prevalences of disease with trajectories of precocious onset (Glasson et al., 2014). However, no previous study has attempted to picture the prevalence of disease and functional impairment in this population both comprehensively, i.e. with a single multidimensional instrument, and transnationally.

The approach to deliver the highest quality care to this population is still being discussed, and as a result, persons with DS resulting in complex needs may not receive appropriate care (Glasson et al., 2014). Further, adults with DS and their families often face hard times initiating the transition from paediatric to adult-based services, and the responsibility for the care of adults with DS and other congenital disabilities is poorly defined (Jensen and Davis, 2013). In the current literature, there seem to be two different approaches. Some advocate for care by general practitioners (Jensen and Bulova, 2014), though yet there remains controversy whether general practitioners are sufficiently knowledgeable, experienced, or even available to administer the proper levels of care for adults with DS (Bittles and Glasson, 2004; Henderson et al., 2007; Jensen et al., 2013). The complexity of needs of adults with DS often means that there is an increase in workload for health professionals in the primary care setting. For this reason, an approach that favours care based on comprehensive assessment and close interaction between general practitioners and management team specialised in the area of DS have been proposed (Carfi et al., 2015, 2014). This approach has been adopted and tested in geriatric medicine and it was shown to be successful independently of patient age (Ellis et al., 2011).

Comprehensive, multidimensional assessment is key to full evaluation and understanding of complexity. It provides information on the various co-morbidities, syndromes, functional and cognitive deficits experienced by adults with DS, which are not all covered by the traditional medical assessment. Comprehensive assessment, therefore, allows a more specific and sensible care plan to be developed. As presented in this study, the adoption of a comprehensive assessment instrument specifically designed for persons with intellectual disabilities (interRAI ID), might lead to better identification of problems or

conditions associated with DS and improve diagnostic accuracy; it may also lead to initiation of needed services in a timely fashion.

Given the similarities between complex older adults in the general population and adults with DS, a 'geriatric approach' based on a comprehensive assessment and management likely represents the best intervention to optimise medical treatment, improve prognosis, restore, maintain and maximise functional autonomy, compensate for the loss of autonomy with an appropriate support, and, hopefully, improve quality of life in adults with DS. Further research is needed that examines the impact of such assessment on services and outcomes among adults with DS.

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Table 1. Sample characteristics

	Italy (n=95)	United States (n=175)	Canada (n=160)
Demographics			
Age, mean \pm SD (range)	38.1 \pm 13.3 (19/65)	35.2 \pm 12.0 (18/62)	48.8 \pm 9.0 (21/75)
Age > 40 y, n (%)	41 (43.2%)	58 (33.1%)	23 (14.4%)
Female, n (%)	47 (49.5%)	65 (37.1%)	62 (39.2%)
Usual residential status			
Private home	85 (89.4%)	146 (83.5%)	-
Group home	10 (10.6%)	12 (6.9%)	71 (44.4%)
Institution	-	17 (9.7%)	89 (55.6%)
Geriatric conditions			
Cognitive status ^a			
Mild/moderate impairment	60 (63.2%)	129 (73.7%)	67 (41.9%)
Severe impairment	14 (14.7%)	35 (20.0%)	74 (46.3%)
Assistance required in ADL ^b	27 (28.4%)	99 (56.6%)	101 (63.1%)
Depression ^c	22 (23.2%)	85 (48.6%)	18 (11.3%)
Symptoms of withdrawal/anhedonia ^d	23 (24.2%)	62 (35.4%)	45 (28.1%)
Aggressive behaviour ^e			
Mild/moderate	25 (26.3%)	81 (46.3%)	64 (40.0%)
Severe	1(1.1%)	16 (9.1%)	9 (5.6%)
Communication problems ^f			
Mild/moderate impairment	57 (60%)	98 (56.0%)	71 (44.4%)
Severe impairment	9 (9.5%)	63 (36.0%)	70 (43.8%)
At least one fall in the last 90 days	12 (12.7%)	30 (17.1%)	14 (8.8%)
Urinary incontinence	7 (7.4%)	49 (28.0%)	68 (42.5%)
Faecal incontinence	3 (3.2%)	49 (28.0%)	45 (28.1%)
Hearing problems ^g	28 (29.5%)	47 (26.9%)	34 (21.3%)
Symptoms & other conditions			
Pain ^h			
No/less than daily	82 (94.3)	155(88.6%)	148 (92.5%)
Daily but not severe	4 (4.6)	16(9.1%)	10 (6.3%)
Daily severe	1 (1.1)	4(2.3%)	1 (0.6%)
Dizziness in the last 3 days	5 (5.3%)	7 (4.0%)	3 (1.9%)
Gastrointestinal symptoms ⁱ in the last 3 days	38 (40%)	47 (26.9%)	22 (17.6%)
Psychiatric symptoms ^j in the last 3 days	5 (5.3%)	22 (12.6%)	7 (4.4%)
Skin problems ^k	28 (29.5%)	130 (74.3%)	77 (49.0%)
Nutrition			
Dental problems ^l	63 (66.3%)	40 (22.9%)	26 (16.4%)
Body mass Index (kg/m ²) ^m			
< 18.5	1 (1.1%)	3 (1.7%)	-
18-5-25	41(43.2%)	34 (19.4%)	-
25-30	27 (28.4%)	44 (25.1%)	-
30 or more	25 (26.3%)	94 (53.7%)	-
Visits and admissions (last 90 days)ⁿ			

Physician visits	36 (37.9%)	142 (81.1%)	107 (66.9%)
Inpatient acute hospital admission	5 (5.3%)	6 (3.4%)	2 (1.3%)
Emergency room visits	8 (8.4%)	10 (5.7%)	2 (1.3%)
Visits with mental health physician	3 (3.2%)	27 (15.4%)	14 (8.8%)

- a. Mild/moderate cognitive impairment is defined by Cognitive Performance Scale score 2–4, severe impairment by Cognitive Performance Scale score 5–6.
- b. Assistance required is defined by ADL hierarchical scale score 2 or higher.
- c. Depression Rating Scale score ≥ 3 .
- d. Social Withdrawal Scale score ≥ 1
- e. Mild/moderate aggressive behaviour is defined by Aggressive Behaviour Scale score 1–4, severe aggressive behaviour by Aggressive Behaviour Scale 5 or more.
- f. Mild/moderate communication problems are defined by Communication Scale score 2–5, severe problems by Communication Scale score 6–8.
- g. Defined as a difficulty to hear (with hearing appliance normally used) at least in some environments (e.g., when person speaks softly or is more than 2 metres away).
- h. Person complains or shows evidence of pain (including grimacing, teeth clenching, moaning, withdrawal when touched, or other nonverbal signs suggesting pain)
- i. Including acid reflux or constipation or diarrhoea or vomiting.
- j. Including delusion or hallucinations or abnormal thought process.
- k. Including bruises or rashes or itching or mottling or herpes zoster or intertrigo or eczema.
- l. Including presence of broken, fragmented, loose, or otherwise nonintact natural teeth or gum (soft tissue) inflammation or bleeding adjacent to natural teeth or tooth fragments.
- m. BMI data not collected in Canada
- n. Used the specified health service at least once in the last 90 days