

Brief Report**Adults with Down syndrome: a comprehensive approach to manage complexity****A. Carfi,¹  D. L. Vetrano,¹ D. Mascia,¹ E. Meloni,¹ E. R. Villani,¹ N. Acampora,¹ V. Brandi,¹ B. E. Fries,² L. Martin,³ R. Bernabei¹ & G. Onder¹**¹ *Centro Medicina dell'Invecchiamento, Department of Geriatrics, Università Cattolica del Sacro Cuore, Rome, Italy*² *Department of Geriatrics and Palliative Care, Department of Internal Medicine and Department of Health, Management and Policy, School of Public Health, University of Michigan, Ann Arbor, MI, USA*³ *Department of Health Sciences, Lakehead University, Thunder Bay, ON, Canada***Abstract**

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

The objective of the study is to describe characteristics of adults with DS collected with a standardised, comprehensive assessment instrument.

Methods Cross-sectional study.

Four hundred thirty adults with DS (age range 18/75 years) from three countries (Italy, $n = 95$; USA, $n = 175$; and Canada, $n = 160$).

A standardised assessment instrument (interRAI intellectual disability) was used to assess sample characteristics.

Results Mean age ranged from 35.2 (standard deviation 12.0) years in the US sample to 48.8 (standard deviation 9.0) years 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.

Keywords ageing, comprehensive assessment, Down syndrome

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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 (Glasson *et al.* 2002; Yang *et al.* 2002; Englund *et al.* 2013). 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 & Glasson 2004). Given these epidemiological changes, DS should be no longer considered a 'paediatric' condition but rather a condition that affects the whole lifespan. DS is considered a 'segmental' progeroid syndrome, in which a premature ageing affects selected organ systems, and persons with this condition might present patterns of co-morbidities often observed in the older population (Real de Asua *et al.* 2015; Vetrano *et al.* 2015; Schoufour *et al.* 2016; Carfi *et al.* 2017; Picciotti *et al.* 2017). 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 to understand whether the use of a standardised comprehensive assessment tool on a sample of adults with DS from three countries (Italy, USA and Canada) could provide useful information on their functional impairment and co-morbidities.

Materials and methods

Participants

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

Italy

Participants were adults with DS, aged 18 or older and 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.

USA

The US data represent the full 2013 statewide 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 centres, 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 & Martin 2008). The community data come 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 DS were identified in the data.

InterRAI intellectual disability

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, and 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) collected directly from the subject or by

an informant; a number of questions are asked directly to the subject concerning his or her preferences, outlook and well-being. 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 the 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 (Martin *et al.* 2007; Langlois & Martin 2008). 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 7-point Activities of Daily Living (ADL) Hierarchy Scale informs on functioning – that is, independence in ADL. Score ≥ 2 in the ADL Hierarchy Scale was used to identify participants requiring assistance in ADL (Morris *et al.* 1999). The Depression Rating Scale 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 & 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 (SPSS Inc. 2009, Chicago, IL, USA) (version 18.0) for the Italian data and SAS (SAS Institute Inc., Cary, NC, USA) (version 9.3) for the US and Canada data.

Results

Mean age of persons with DS ranged from 35.2 (standard deviation 12.0) years in the US sample to 48.8 (standard deviation 9.0) years 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 include delusions, hallucinations or abnormal thought process), dizziness and pain occurring in the 3 days before the assessment were uncommon. Obesity (body mass index ≥ 30 kg/m²) was also highly prevalent (body mass index 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 USA.

Discussion

We present here data on a large sample of adults with DS, which demonstrate that they present with a high level of co-morbidity 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 the 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, that is, 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).

Table 1 Sample characteristics

	Italy (n = 95)	USA (n = 175)	Canada (n = 160)
Demographics			
Age, mean ± SD (range)	38.1 ± 13.3 (19/65)	35.2 ± 12.0 (18/62)	48.8 ± 9.0 (21/75)
Age >40 years, 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, n (%)	85 (89.4)	146 (83.5)	—
Group home, n (%)	10 (10.6)	12 (6.9)	71 (44.4)
Institution, n (%)	—	17 (9.7)	89 (55.6)
Geriatric conditions			
Cognitive status[†]			
Mild/moderate impairment, n (%)	60 (63.2)	129 (73.7)	67 (41.9)
Severe impairment, n (%)	14 (14.7)	35 (20.0)	74 (46.3)
Assistance required in ADL [‡] , n (%)	27 (28.4)	99 (56.6)	101 (63.1)
Depression [§] , n (%)	22 (23.2)	85 (48.6)	18 (11.3)
Symptoms of withdrawal/anhedonia [¶] , n (%)	23 (24.2)	62 (35.4)	45 (28.1)
Aggressive behaviour^{††}			
Mild/moderate, n (%)	25 (26.3)	81 (46.3)	64 (40.0)
Severe, n (%)	1 (1.1)	16 (9.1)	9 (5.6)
Communication problems^{‡‡}			
Mild/moderate impairment, n (%)	57 (60)	98 (56.0)	71 (44.4)
Severe impairment, n (%)	9 (9.5)	63 (36.0)	70 (43.8)
At least one fall in the last 90 days, n (%)	12 (12.7)	30 (17.1)	14 (8.8)
Urinary incontinence, n (%)	7 (7.4)	49 (28.0)	68 (42.5)
Faecal incontinence, n (%)	3 (3.2)	49 (28.0)	45 (28.1)
Hearing problems ^{§§} , n (%)	28 (29.5)	47 (26.9)	34 (21.3)
Symptoms and other conditions			
Pain^{¶¶}, n (%)			
No/less than daily, n (%)	82 (94.3)	155 (88.6)	148 (92.5)
Daily but not severe, n (%)	4 (4.6)	16 (9.1)	10 (6.3)
Daily severe, n (%)	1 (1.1)	4 (2.3)	1 (0.6)
Dizziness in the last 3 days, n (%)	5 (5.3)	7 (4.0)	3 (1.9)
Gastrointestinal symptoms ^{†††} in the last 3 days, n (%)	38 (40)	47 (26.9)	22 (17.6)
Psychiatric symptoms ^{‡‡‡} in the last 3 days, n (%)	5 (5.3)	22 (12.6)	7 (4.4)
Skin problems ^{§§§} , n (%)	28 (29.5)	130 (74.3)	77 (49.0)
Nutrition			
Dental problems ^{¶¶¶} , n (%)	63 (66.3)	40 (22.9)	26 (16.4)
Body mass index^{††††}, n (%)			
<18.5 kg/m ²	1 (1.1)	3 (1.7)	—
18–25 kg/m ²	41 (43.2)	34 (19.4)	—
25–30 kg/m ²	27 (28.4)	44 (25.1)	—
30 or more	25 (26.3)	94 (53.7)	—
Visits and admissions (last 90 days)^{‡‡‡‡}			
Physician visit, n (%)	36 (37.9)	142 (81.1)	107 (66.9)
Inpatient acute hospital admission, n (%)	5 (5.3)	6 (3.4)	2 (1.3)
Emergency room visits, n (%)	8 (8.4)	10 (5.7)	2 (1.3)
Visits with mental health physician, n (%)	3 (3.2)	27 (15.4)	14 (8.8)

[†]Mild/moderate cognitive impairment is defined by Cognitive Performance Scale score 2–4 and severe impairment by Cognitive Performance Scale score 5–6.

[‡]Assistance required is defined by ADL Hierarchical Scale score 2 or higher.

[§]Depression Rating Scale score ≥3.

[¶]Social Withdrawal Scale score ≥1.

^{††}Mild/moderate aggressive behaviour is defined by Aggressive Behaviour Scale score 1–4 and severe aggressive behaviour by Aggressive Behaviour Scale 5 or more.

^{##}Mild/moderate communication problems are defined by Communication Scale score 2–5 and severe problems by Communication Scale score 6–8.

^{§§}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 m away).

^{¶¶}Person complains or shows evidence of pain (including grimacing, teeth clenching, moaning, withdrawal when touched or other non-verbal signs suggesting pain).

^{†††}Including acid reflux or constipation or diarrhoea or vomiting.

^{‡‡}Including delusion or hallucinations or abnormal thought process.

^{§§§}Including bruises or rashes or itching or mottling or herpes zoster or intertrigo or eczema.

^{¶¶¶}Including the presence of broken, fragmented, loose or otherwise nonintact natural teeth or gum (soft tissue) inflammation or bleeding adjacent to natural teeth or tooth fragments.

^{††††}Body mass index data not collected in Canada.

^{‡‡‡}Used the specified health service at least once in the last 90 days.

ADL, Activities of Daily Living; SD, standard deviation.

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 & Davis 2013). In the current literature, there seem to be two different approaches. Some advocate for care by general practitioners (Jensen & Bulova 2014), although 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 & 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 has been proposed (Carfi *et al.* 2014; Carfi *et al.* 2015). 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 comorbidities, syndromes and 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 IDs (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 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.

Conflict of Interest

The authors declare no conflicts of interest.

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