

## Primary care for adults with Down syndrome: adherence to preventive healthcare recommendations

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### Abstract

**Background** Due to significant medical improvements, persons with Down syndrome now live well into adulthood. Consequently, primary care for adults with Down syndrome needs to incorporate routine care with screening for condition-specific comorbidities. This study seeks to evaluate the adherence of primary care physicians to age- and condition-specific preventive care in a cohort of adults with Down syndrome.

**Methods** In this retrospective observational cohort study, preventive screening was evaluated in patients with Down syndrome aged 18–45 years who received primary care in an academic medical centre from 2000 to 2008. Comparisons were made based on the field of patients' primary care providers (Family or Internal Medicine).

**Results** This cohort included 62 patients, median index age = 33 years. Forty per cent of patients received primary care by Family Physicians, with 60% seen by Internal Medicine practices. Patient demographics, comorbidities and overall screening patterns were similar between provider groups. Despite near universal screening for obesity and hypothyroidism, adherence to preventive care recommendations was otherwise inconsistent.

Screening was 'moderate' (50–80%) for cardiac anomalies, reproductive health, dentition, and the combined measure of behaviour, psychological, or memory abnormalities. Less than 50% of patients were evaluated for obstructive sleep apnea, atlanto-axial instability, hearing loss or vision loss.

**Conclusions** We observed inconsistent preventive care in adults with Down syndrome over this 8.5-year study. This is concerning, given that the adverse effects of many of these conditions can be ameliorated if discovered in a timely fashion. Further studies must evaluate the implications of screening practices and more timely identification of comorbidities on clinical outcomes.

**Keywords** adult, Down syndrome, preventive care, primary care

### Introduction

Down syndrome is among the most commonly identifiable causes of developmental delay, occurring in an estimated 14.5 per 10 000 live births (Parker *et al.* 2010). Largely due to improvements in early medical interventions, increasing numbers of persons with Down syndrome now survive well into adulthood (American Academy of Pediatrics 1996). Nearly 80% survive into adolescence (Hayes *et al.* 1997), with an average life expectancy of nearly 60 years (Glasson *et al.* 2002).

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Individuals with Down syndrome are at increased risk of developing multiple comorbidities which require regular care by both specialists and primary care physicians. Conditions such as intellectual disability (ID), congenital heart disease, haematologic abnormalities, gastrointestinal atresia, obstructive sleep apnea, and hearing and vision abnormalities are well described in children with Down syndrome (Cooley & Graham 1991; Baum *et al.* 2008; Bull & the Committee on Genetics 2011). However, the onset of many of these comorbidities is not isolated to early childhood. In addition to conditions that typically accompany the ageing process, adults with Down syndrome can also develop medical conditions directly related to their underlying genetic abnormality. This includes increased incidence of conditions commonly associated with Down syndrome (e.g. hypothyroidism, obesity, and obstructive sleep apnea) (Rubin & Crocker 1989; Cooley & Graham 1991; American Academy of Pediatrics 2001; Roizen 2002; New York Commissioner's Task Force 2009; Rimmer *et al.* 2011; Steingass *et al.* 2011), as well as the development of new comorbidities, such as cardiac valvular abnormalities, 'accelerated aging', and 'early senescence' (Goldhaber *et al.* 1986, 1987; Rubin & Crocker 1989; Geggel *et al.* 1993; Pueschel *et al.* 1995; Cohen 1999; Smith 2001; Bittles *et al.* 2007; New York Commissioner's Task Force 2009; Steingass *et al.* 2011).

Given the rising incidence and prevalence of medical comorbidities in adults with Down syndrome, there is a tremendous need for preventive screening in this population. The adverse effects of many of these conditions can be ameliorated if discovered and treated in a timely fashion. Several recommendations for preventive health care in adults with Down syndrome have been published in the medical literature with varying levels of consensus (Chicoine and McGuire 2010; Rubin & Crocker 1989; Pueschel *et al.* 1995; Cohen 1999; Saenz 1999; Van Allen *et al.* 1999; Tyler & Edman 2004; Wilson & Cooley 2006; Henderson *et al.* 2007; Virji-Babul *et al.* 2007; New York Commissioner's Task Force 2009; Bull & the Committee on Genetics 2011; Steingass *et al.* 2011). However, few studies exist that evaluate adherence to these guidelines (Ferguson *et al.* 2009; Maatta *et al.* 2011; McGrath *et al.* 2011). Within paediatric populations, persons with Down

syndrome are significantly less likely to have a medical home despite higher unmet medical needs (McGrath *et al.* 2011). These discrepancies in care are likely to increase with age (McCabe *et al.* 2011) and are especially concerning given that persons with the number and type of comorbid conditions as adults with Down syndrome have less capacity to deal with a new physiological 'hit' than many of their unaffected age-matched peers.

The purpose of this study is to evaluate how successful primary care physicians in an academic medical centre were in providing age-, gender-, and Down syndrome-specific preventive care over an 8-year period in a cohort of adults with Down syndrome. Based on our experiences as clinicians at this institution, we hypothesised that primary care services were lacking for adults with Down syndrome. We also hypothesised that there would not be appreciable differences in preventive screening patterns based on the field of primary care.

## Methods

### Study cohort

Our cohort was identified from administrative data gathered from our university hospital's Central Data Repository, which captures all outpatient, inpatient, and emergency care within our health system. Patients aged 18–45 years were included in the initial cohort if they were seen at our institution at any time between 1 January 2000 and 30 June 2008 with Down syndrome listed within any of the 15 diagnostic fields available for each encounter ( $n = 252$ ) [*International Classification of Diseases, Ninth Revision, Clinical Modification* (ICD-9-CM) code 758.0] (Bryon & Madge 2001). Diagnosis of Down syndrome was then confirmed through manual chart review utilising a search engine for free-text documents within the electronic medical record known as the Electronic Medical Record Search Engine (EMERSE) (Hanauer 2006; Hanauer *et al.* 2009). Search terms included the following: 'Down syndrome', 'Down's syndrome', 'Downs syndrome', 'Trisomy 21', 'Tri21' and 'Tri 21'. This resulted in 53 cases without Down syndrome being excluded from our cohort.

Within the Central Data Repository, we then identified all patients (meeting the aforementioned age and date criteria) who had visits coded for the commonly associated Down syndrome comorbidities of congenital heart disease, hypothyroidism, and atlanto-axial instability. Congenital heart disease was identified using ICD-9-CM codes 745–745.9, 746–746.9, 747–747.49. Patients with hypothyroidism were identified using ICD-9-CM codes 243, 244.3, 244.8, 244.9. Atlanto-axial instability was identified with ICD-9-CM code 847.0. EMERSE (Hanauer 2006; Hanauer *et al.* 2009) was utilised to identify patients with documented Down syndrome in the electronic medical record with one of the aforementioned comorbidities who had not been previously identified by ICD-9-CM code 758.0 ( $n = 29$ ). Only patients with documented Down syndrome, verified by the authors upon review of the clinical record, were included in the cohort ( $n = 228$ ) (Fig. 1). Through chart review, we then identified which patients in our cohort received primary care services within our institution ( $n = 73$ ). Patients were assigned to mutually exclusive provider groups based on the specialty of their primary care providers (e.g. Family Medicine, Internal Medicine, Pediatrics, or Internal Medicine-Pediatrics). Only provider groups with  $\geq 20$  patients were included in analysis, which led to the exclusion of the Pediatric and Internal Medicine-Pediatrics provider groups for a final cohort of 62 patients. The Institutional Review Board in our health system approved the study protocol.

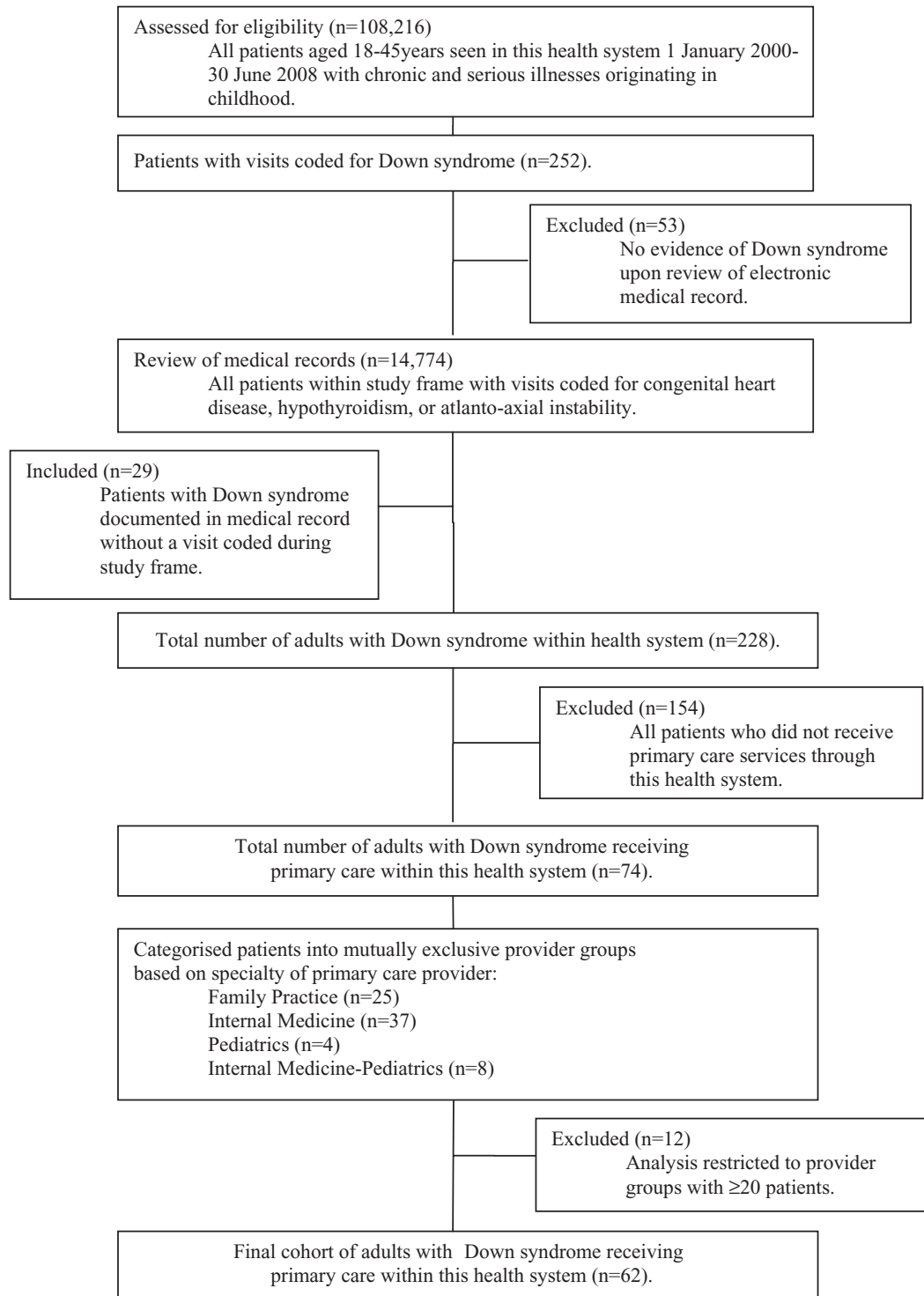
#### Assessment preventive healthcare domains

Through review of existing preventive healthcare guidelines for adults with Down syndrome within the medical literature, areas of broad consensus for preventive screening were determined to be age- and gender-appropriate health care in addition to regular screening for hypothyroidism, obesity, behavioural, psychiatric or intellectual changes, the development of cardiac valve, vision, dental, or hearing abnormalities, and musculoskeletal changes (Chicoine and McGuire 2010; Goldhaber *et al.* 1986, 1987; Pueschel & Scola 1987; Rubin & Crocker 1989; Cooley & Graham 1991; Stebbens *et al.* 1991; Yeates 1991; Carey 1992; Geggel *et al.* 1993; American Academy of Pediatrics 1994, 2001; Pueschel

*et al.* 1995; Verma *et al.* 1996; Cohen 1999; Saenz 1999; Van Allen *et al.* 1999; Feingold & Geggel 2001; Smith 2001; Van Riper & Cohen 2001; Roizen 2002; Bosch 2003; Tyler & Edman 2004; Murphy *et al.* 2005; Wilson & Cooley 2006; Henderson *et al.* 2007; Virji-Babul *et al.* 2007; Baum *et al.* 2008; Davidson 2008; Ferguson *et al.* 2009; New York Commissioner's Task Force 2009; Bull & the Committee on Genetics 2011; Rimmer *et al.* 2011; Steingass *et al.* 2011). Therefore, the following screening domains were chosen for evaluation within our study population: atlanto-axial instability, cardiac abnormalities, dental abnormalities, hearing abnormalities, hypothyroidism, obesity, obstructive sleep apnea, reproductive health care, vision, and a combined measure evaluating psychiatric disorders, behaviour difficulties, or memory regression. Criteria for each of these domains are defined in Table 1. EMERSE was utilised to identify whether patients had received preventive healthcare screening within these domains at any time prior to the end of the 8.5-year study. Manual chart review was used to identify any missing information and to settle any discrepancies identified within the findings from EMERSE. Data collection was performed by investigators KMJ and LCT. The investigators cross-checked 10% of each other's data abstraction and settled any discrepancies by group consensus through duplicated manual chart review.

#### Determination of additional variables

The length of time spent in the study cohort was calculated by the number of years each patient was within the ages of 18–45 years during the study time frame (1 January 2000–30 June 2008). Where applicable, the time period was adjusted for date of death. Annualised encounters were calculated through division of their total respective sums by the number of years each patient was in the study. Because of inconsistent documentation of medical insurance for patients in this dataset, insurance source was not included as a variable in this analysis. Complexity levels of congenital heart disease were determined through manual chart review, and were classified as mild, moderate or severe based on criteria published by the American College of Cardiology (Warnes *et al.* 2001).



**Figure 1** Study cohort identification.

**Table 1** Criteria for screening of preventive healthcare domains

Preventive healthcare domain	Definition of meeting diagnostic criteria for given medical condition	Definition of achieving screening criteria ( $\geq 1$ area within each domain at least once during study period)
Atlanto-axial instability	Documented atlanto-axial instability in the medical record or in radiographic results	Documented atlanto-axial instability Cervical Spine X-ray, CT, or MRI
Cardiac abnormality	Documented congenital heart disease or other cardiac abnormalities in the medical record or in echocardiographic results	Diagnosis of congenital heart disease or other cardiac abnormalities Formal cardiology screening (echocardiogram or EKG) Referral for cardiology evaluation
Dental care	Documented poor dentition in the medical record	Documented formal dental evaluation Referral for dental evaluation Discussion of dentition within the medical record
Hearing loss	Documented hearing abnormalities in medical record or results from formal audiology evaluation	Documented formal audiology assessment Referral for audiology assessment Discussion of hearing capacity within the medical record
Hypothyroidism	Documented hypothyroidism in the medical record	Documented hypothyroidism Screening thyroid stimulating hormone or free T4
Overweight/obesity	BMI $\geq 25$ Documented overweight/obesity in the medical record	Documented Body Mass Index Documented diagnosis of obesity/overweight Discussion of weight-related concerns within the medical record
Obstructive sleep apnea	Documented obstructive sleep apnea in the medical record or in results from formal sleep evaluation	Documented diagnosis of obstructive sleep apnea Documented formal sleep study Referral for sleep study Discussion of apnea risks within the medical record
Reproductive health care	Screening measure only, so no diagnosis formally tracked	Documented Pap smear/pelvic or testicular examination Discussion of sexual activity Prescription of contraception method Discussion of risk factors for sexually transmitted infections Screening and treatment for sexually transmitted infections
Psychiatric disorder, behaviour difficulties, or memory regression (combined measure)	Documented intellectual disability, behaviour problems, psychiatric condition, memory loss/regression, or dementia in the medical record	Documented intellectual disability, behaviour problems, psychiatric condition, memory loss/regression, or dementia Discussion of cognition or behaviour within the medical record
Vision abnormalities*	Documented vision abnormalities in the medical record	Documented ophthalmological evaluation Referral for ophthalmological evaluation Discussion of vision within the medical record

\* Ophthalmological care provided on handwritten notes that are not visible within the electronic medical record of this health system.

## Data analysis

Bivariate comparisons of utilisation patterns and patient attributes between provider groups were conducted using chi-squared, Kruskal–Wallis and *t*-tests. Stata version 11.1 software (Stata Corp, College Station, TX, USA) was used for all analyses.

## Results

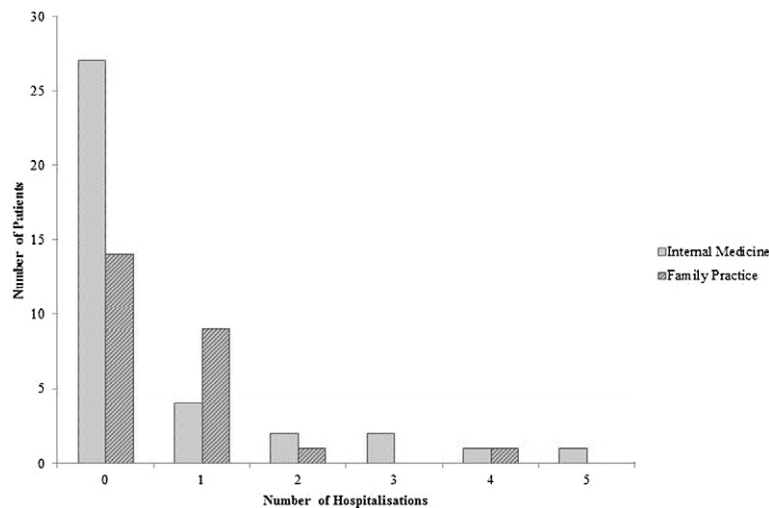
### Sample characteristics

We identified a total of 62 adults with Down syndrome, aged 18–45 years, who received primary care services at our institution at any time between 1 January 2000 and 30 June 2008. This constituted

**Table 2** Patient characteristics by provider group

Characteristics (%)	Total	Internal Medicine	Family Medicine	P value
<i>n</i>	62	37(60%)	25 (40%)	–
Gender				
Female	23 (37%)	11 (30%)	12 (48%)	0.14
Self-reported race/ethnicity				
Caucasian	53 (86%)	31 (84%)	22 (88%)	0.64
African American	5 (8%)	2 (5%)	3 (12%)	0.35
Hispanic	0	0	0	–
Asian	0	0	0	–
Other/unknown	4 (7%)	4 (11%)	0	0.09
Median age at index encounter (IQR)	33 years (23–39)	34 years (25–39)	28 years (22–37)	0.38
Mortality during study years	13 (6%)	8 (8%)	5 (5%)	0.32
Median number of specialty providers (IQR)	2 (1–3)	2 (1–4)	2 (1–3)	0.86
Number of patients ever hospitalised during study period	21 (34%)	10 (16%)	11 (18%)	0.17
Median annualised hospitalisations (IQR)	0 (0–0.13)	0 (0–0.13)	0 (0.014)	0.41
Median annualised total visits (IQR)	3.9 (2.1–6.2)	4.2 (2–6.8)	3.7 (2.8–5.9)	0.72

IQR, interquartile range.

**Figure 2** Hospitalisation patterns by provider group.

372 years of patient data. Members of this cohort were predominantly male (63%) with a median age at index encounter of 33 years (Table 2). Only 34% of our cohort were ever hospitalised during the study period; of those hospitalised, less than half had more than one inpatient stay (Fig. 2). There were five deaths during the study period, causes not available in the electronic medical record. Nearly all

persons in our cohort required some form of subspecialty care, with the median patient accessing two subspecialty providers. Comorbidities were common, the most frequent of which were overweight/obesity (71%), diagnosis of psychiatric disorders, behaviour difficulties or memory regression (52%), poor dentition (55%), and hypothyroidism (45%) (Table 3).

**Table 3** Presence of comorbidities by provider group

Diagnosis	Total (%), n = 62	Internal Medicine (%), n = 37	Family Medicine (%), n = 25	P value
Atlanto-axial instability	5 (8)	3 (8)	2 (8)	0.99
Cardiac abnormalities	10 (16)	7 (19)	3 (12)	0.47
Poor dentition	34 (55)	22 (59)	12 (48)	0.42
Hearing loss	17 (27)	9 (24)	8 (32)	0.46
Hypothyroidism	28 (45)	15 (41)	13 (52)	0.37
Overweight/obesity	44 (71)	26 (70)	18 (72)	0.71
Obstructive sleep apnea	15 (24)	11 (30)	4 (16)	0.14
(Combined measure) Psychiatric disorders, behaviour difficulties, or memory regression	32 (52)	14 (46)	15 (60)	0.19
Vision abnormalities	15 (24)	10 (27)	5 (20)	0.55

**Table 4** Congenital heart disease complexity by provider group

Complexity of congenital heart disease	Total (%), n = 62	Internal Medicine (%), n = 37	Family Medicine (%), n = 25	P value
None	52 (84)	30 (81)	22 (88)	0.47
Mild	2 (3)	1 (3)	1 (4)	0.78
Moderate	3 (5)	2 (5)	1 (4)	0.80
Severe	5 (8)	4 (11)	1 (4)	0.33

Sample diagnoses within each category (Warnes *et al.* 2001):

- Mild congenital heart disease: isolated patent foramen ovale, isolated small atrial septal defect, isolated small ventricular septal defect without associated lesions;
- Moderate congenital heart disease: anomalous pulmonary venous return, coarctation of the aorta, Ebstein's anomaly, Tetralogy of Fallot, atrioventricular canal defects;
- Severe congenital heart disease: all forms of cyanotic heart disease, double-outlet ventricle, Eisenmenger syndrome, pulmonary atresia, pulmonary vascular obstructive disease.

### Provider group characteristics

Forty per cent of patients in our cohort received primary care from Family Medicine practitioners, with 60% seen in Internal Medicine clinics. No differences in demographics were observed between provider groups, including age, gender, ethnicity or mortality (Table 3). The Internal Medicine and Family Medicine provider groups also displayed similar overall patterns of healthcare utilisation and hospitalisation. Direct comparison of comorbidities between provider groups shows similar prevalence of all conditions evaluated (Table 3), as well as the presence and severity of congenital heart disease (Table 4).

### Adherence to screening recommendations

Screening patterns for preventive healthcare domains in adults with Down syndrome were largely similar between the Internal Medicine and Family Medicine provider groups (Table 5). Both provider types achieved high levels of adherence in screening for obesity and hypothyroidism (>90% in both domains). Screening was 'moderate' (50–80% adherence) for cardiac anomalies, reproductive health care, poor dentition, and the combined measure of behavioural, psychological, or memory abnormalities. The only areas of discrepancy between provider groups were in reproductive health care (adherence = Family Medicine 72%,



**Table 5** Documented preventive screening domains by provider group

Diagnosis	Total (%), n = 62	Internal Medicine (%), n = 37	Family Medicine (%), n = 25	P value
Atlanto-axial instability	26 (42)	16 (43)	10 (40)	0.8
Cardiac abnormalities	42 (68)	30 (81)	12 (48)	0.006
Dental care	35 (56)	23 (62)	12 (48)	0.27
Hearing loss	29 (47)	18 (49)	11 (44)	0.72
Hypothyroidism	59 (95)	34 (92)	25 (100)	0.14
Overweight/obesity	61 (98)	36 (97)	25 (100)	0.41
Obstructive sleep apnea	22 (35)	13 (35)	9 (36)	0.94
Reproductive health care	34 (55)	16 (43)	18 (72)	0.03
(Combined measure)	32 (52)	17 (46)	15 (60)	0.28
Psychiatric disorders, behaviour difficulties, or memory regression				
Vision abnormalities	24 (39)	14 (38)	10 (40)	0.86

Internal Medicine 43%,  $P = 0.03$ ) and in screening for cardiac anomalies (adherence = Internal Medicine 81%, Family Medicine 48%,  $P = 0.006$ ). Both provider groups demonstrated low adherence (<50%) in screening for obstructive sleep apnea, atlanto-axial instability, hearing loss, and vision abnormalities.

## Discussion

As we approached this study, we hypothesised that primary care services at our institution would be insufficient for adults with Down syndrome and that no substantive differences would be observed between primary care provider types. Our findings support these hypotheses. Although few differences were observed in screening behaviours between providers in Internal Medicine and Family Medicine at our institution, it is important to note that overall adherence to existing preventive healthcare recommendations for adults with Down syndrome was inconsistent.

Our providers excelled at screening for obesity and hypothyroidism but demonstrated varying levels of adherence to the remaining preventive care domains. Given that screening needed to occur only once during this 8.5-year study period for patients to receive credit for any given domain, this is especially concerning. Nationally, screening patterns for similar domains in general adult populations dem-

onstrate annual adherence anywhere from 33% for depression screening (Tudiver *et al.* 2010) to 92% for cervical cancer screening (Hughes *et al.* 2010). In comparison, 52% of our cohort was screened for the combined measure of psychiatric disorders, behavioural disorders or memory regression, and only 55% of our cohort received reproductive health screening at least once over this 8.5-year study.

The purpose of primary care is to identify conditions for which prevention, early identification, and treatment can decrease an individual's morbidity and mortality (Robertson *et al.* 2011). For each of the preventive healthcare domains addressed in this study, early detection and intervention have the potential to make a substantive impact on the lives of persons with Down syndrome. For example, individuals with Down syndrome are at high risk for obesity because of multiple factors, including inactivity and hypothyroidism (Smith 2001; Henderson *et al.* 2007; Bull & the Committee on Genetics 2011; Steingass *et al.* 2011). However, they are also at high risk for obstructive sleep apnea independent of obesity, which itself is a known risk factor for obstructive sleep apnea. Interventions that decrease obesity, therefore, can have multiple positive downstream effects. Additionally, poor dentition has been identified as risk factor for increased morbidity (Garcia *et al.* 1998), while untreated hearing loss can manifest itself in decreased learning potential, social withdrawal or even clinical depression



(Meuwese-Jongejugd *et al.* 2006; Hild *et al.* 2008). Recently published work in the geriatric (non-Down syndrome) population indicates that changes in cognitive ability can function as an independent risk factor for mortality (Sachs *et al.* 2011). Given the accelerated ageing and early senescence experienced by individuals with Down syndrome (Rubin & Crocker 1989; Pueschel *et al.* 1995; Smith 2001; Bittles *et al.* 2007; New York Commissioner's Task Force 2009), we anticipate these findings would readily translate into our population that already has a baseline level of ID (Singer & Strauss 1997).

In addition to screening for Down syndrome-specific comorbidities, it is important to ensure both age- and gender-appropriate screening. This includes typical primary care domains such as reproductive health screening, diet, exercise, and primary prevention of general adult conditions, such as cardiovascular disease (Bittles *et al.* 2007). The invasive nature of some screening mechanisms must be weighed against each person's ability to undergo such procedures. Nonetheless, these conversations need to occur between providers, patients and their caregivers. Informed decisions need to be made regarding whether traditional screening methods can be utilised or whether alternative, less invasive procedures may be used to approximate care that might not otherwise have been provided (e.g. fingertip pelvic examination versus traditional speculum examination) (Kavoussi *et al.* 2009). Providers should also weigh the potential benefit of screening practices with their anticipated interventions should the procedure uncover new medical issues. In some situations, it is quite reasonable to forego invasive procedures if the outcome would not change a patient's decisions or a clinician's behaviour. However, given the rising life expectancy of persons with Down syndrome, it is inappropriate to ignore age- and gender-appropriate care simply because of the presence of ID.

When compared with national estimated prevalence rates of comorbidities in adults with Down syndrome (Rubin & Crocker 1989; Cooley & Graham 1991; Bell & Bhate 1992; Prasher 1995; Pueschel *et al.* 1995; American Academy of Pediatrics 2001; Melville *et al.* 2005; New York Commissioner's Task Force 2009; Steingass *et al.* 2011), the individuals observed in this cohort appear to be slightly less complex on average. Our medical

records indicate lower prevalence of several conditions in our cohort, with the exception of hypothyroidism, overweight/obesity and the combined measure of psychiatric disorders, behaviour difficulties, or memory regression (Fig. 3). We hypothesise that these numbers may be artificially underestimated, as prevalence estimates in our cohort are completely dependent upon what has been documented in the medical record and not prospective evaluation. We suspect the lower numbers of persons identified with obstructive sleep apnea reflect less screening performed at our institution than is done on a national level. Additionally, care by dentists and ophthalmologists is often documented in paper charts within our health system, which were not available for review by the investigators of this study.

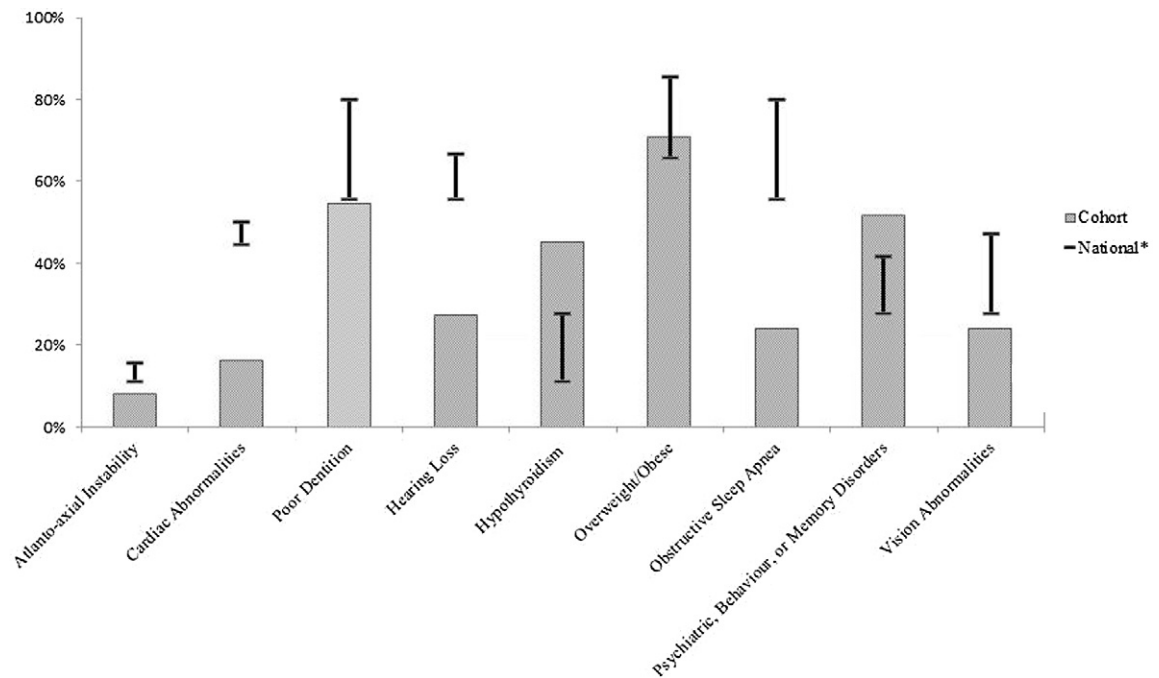
#### Limitations

This study has several limitations. First, our work reflects the experiences of patients with Down syndrome at a single tertiary care centre with multiple associated primary care clinics. This may not reflect trends in primary care practices that would be evident on a national level.

Second, the small population of patients in each of the primary care practices, especially in Internal Medicine-Pediatrics and Pediatrics, may prevent observation of true differences in care practices that would be evident within a larger cohort. However, despite the fact that the overall number of patients described in this study is not particularly large, this cohort of adults with Down syndrome is comparable in size to many previously described in the medical literature.

Third, documentation of receipt of screening domains was limited to what was recorded in the electronic medical records. This likely underestimated the domains of ophthalmology, dentistry, audiology and psychiatry, as these fields often store clinical charts outside of the electronic medical record typically utilised within this health system. Additionally, the scope of this study is to evaluate the presence or absence of screening behaviours and does not follow any subsequent interventions on the part of clinicians.

These limitations notwithstanding, we expect that the experience of caring for adults with Down syn-



**Figure 3** Prevalence of comorbidities in adults with Down syndrome. \*National prevalence estimates taken from: Rubin & Crocker 1989; Cooley & Graham 1991; Bell & Bhate 1992; Prasher 1995; American Academy of Pediatrics 2001; Melville *et al.* 2005; New York Commissioner's Task Force 2009; Steingass *et al.* 2011.

drome at our institution will serve as a reasonable approximation of primary care clinicians nationally. If anything, we would anticipate that the proximity to subspecialty care might artificially increase adherence rates to screening recommendations. Given the overall low adherence to preventive care recommendations observed in this study, this does not appear to be a concern.

## Conclusion

Despite well-published recommendations for preventive health care in adults with Down syndrome, little is known about adherence to these guidelines. Our findings represent one of the first comparisons of primary care practices in a cohort of adults with Down syndrome.

We observed that adherence to existing preventive care guidelines was inconsistent but did not result from the 'type' of primary care each patient received (Family Medicine vs. Internal Medicine).

Rather, the patterns of incomplete adherence to Down syndrome-specific guidelines appear to mirror the discomfort in providing care for adults with ID observed in adult-oriented clinicians previously documented in the medical literature (Okumura *et al.* 2010; Pace *et al.* 2011).

The reasons behind the inconsistent screening patterns observed in this study may serve as a reflection of several possibilities: poor documentation of issues discussed during a clinic visit, lack of awareness by the provider of existing recommendations, or more urgent medical concerns that supersede primary care discussions. In light of such findings, it is important to remember that improved screening has the potential to identify issues that can be readily treated and lead to decreased morbidity and mortality. This will allow patients and families to make more informed decisions about their care. To achieve improvements, preventive healthcare recommendations need to be widely disseminated and readily available to all primary care providers.

We hypothesise that the number and varying scope of existing preventive care recommendations for adults with Down syndrome has made it difficult for clinicians to implement many of these screening guidelines and adds confusion to already complex care management. We highlight the recommendations from the American Academy of Pediatrics (Bull & the Committee on Genetics 2011) and Steingass *et al.* (2011) as the most recently published high quality references for primary care physicians, and we encourage greater collaboration between stakeholders within the Down syndrome community to regularly disseminate recommendations incorporating expert opinion with evidence-based medicine. As a medical community, we need to place greater emphasis on improving the collection and storage of high-quality longitudinal data to better inform clinical guidelines. Further studies are needed to assess the implications of current screening recommendations and more timely identification of comorbidities on clinical outcomes.

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