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Health care in adults with Down syndrome: a longitudinal cohort study

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Abstract

Background Individuals with Down syndrome increasingly survive into adulthood, yet little is known about their healthcare patterns as adults. Our study sought to characterise patterns of health care among adults with Down syndrome based on whether they had fully transitioned to adult-oriented providers by their inception in this cohort.
Methods In this retrospective observational cohort study, healthcare utilisation and annualised patient charges were evaluated in patients with Down syndrome aged 18–45 years who received care in a single academic health centre from 2000 to 2008. Comparisons were made based on patients' provider mix (only adult-focused or 'mixed' child- and adult-focused providers).
Results The cohort included 205 patients with median index age = 28 years; 52% of these adult patients had incompletely transitioned to adult providers and received components of their care from child-focused providers. A higher proportion of these 'mixed' patients were seen exclusively by subspecialty providers (mixed = 81%, adult = 46%, $P < 0.001$), suggesting a need for higher intensity specialised services. Patients in the mixed provider group incurred higher annualised charges in analyses adjusted for age, mortality, total annualised encounters, and number of subspecialty disciplines accessed. These differences were most pronounced when stratified by whether patients were hospitalised during the study period (e.g. difference in adjusted means between mixed versus adult provider groups: $571 without hospitalisation, $19 061 with hospitalisation).
Conclusions In this unique longitudinal cohort of over 200 adults aged 18–45 years with Down syndrome, over half demonstrated incomplete transition to adult care. Persistent use of child-focused care, often with a subspecialty emphasis, has implications for healthcare charges. Future studies must identify reasons for distinct care patterns, examine their relationship with clinical outcomes, and evaluate which provider types deliver the highest quality care for adults with Down syndrome and a wide variety of comorbidities.

Keywords Down syndrome, healthcare transition, healthcare utilisation, health services research, intellectual disability

Background

Largely as a result of medical advances over the past several decades, nearly 90% of individuals with

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chronic and serious illnesses originating in childhood now survive to adulthood (American Academy of Pediatrics 1996). Experts estimate that as many as 900 000 individuals with special healthcare needs (SHCN) turn 18 years annually (Reiss & Gibson 2002; Okumura *et al.* 2007). Consequently, several professional medical organisations (American Academy of Pediatrics, American Academy of Family Physicians, American College of Physicians, and the Society for Adolescent Medicine), as well as Healthy People 2020, have chosen to highlight the importance of transitioning adolescents with SHCN to adult-focused providers over the past several decades (American Academy of Pediatrics *et al.* 2002; Rosen *et al.* 2003; United States Department of Health and Human Services 2010; Cooley & Sagerman 2011). Despite these recommendations, the timing and mechanisms for transition remain undefined in many institutions across the USA and often rely upon the preferences of providers and families. In fact, very little is known about current patterns of health care in adults with chronic conditions originating in childhood, regardless of their state within the transition process.

As physicians providing care to both adult and paediatric populations, we have observed many adults with SHCN retain child-focused providers while incompletely transitioning various components of their care to adult-oriented settings. Yet, this concept of incomplete transition and its implications on care provision and utilisation has not been well described previously. For this reason, we sought to characterise longitudinal patterns of both healthcare utilisation and charges among a cohort of adult survivors of childhood illness with a specific set of healthcare needs at different stages of transition.

Down syndrome is among the most commonly identified causes of developmental disability (Yang *et al.* 2002; Sherman *et al.* 2007). Occurring in 1:650–1000 live births (Wilson & Cooley 2006), patients with Down syndrome now live well into adulthood with an average life expectancy of nearly 60 years (Glasson *et al.* 2002). The healthcare needs of persons with Down syndrome overlap with many different facets of the SHCN experience given the diversity of comorbidities (e.g. cardiac, endocrine) and degree of intellectual disability possible with this chromosomal abnormality. As both paediatric and adult preventive healthcare recommendations specific to this population are well published, the diagnosis of Down syndrome should not be an inherent barrier to successful transition to adult-oriented care (Cooley & Graham 1991; American Academy of Pediatrics 2001; Baum *et al.* 2008; Chicoine & McGuire 2010; Bull & the Committee on Genetics 2011; Steingass *et al.* 2011). For these reasons, we believe that characterisation of longitudinal healthcare utilisation by adults with Down syndrome at different stages of transition offers an instructive assessment of care patterns that may be applicable to many different types of adults with chronic conditions originating in childhood.

Based on our experience as clinicians at an academic medical centre, we hypothesised that adults with Down syndrome who accessed care within paediatric (child-focused) settings would have higher complexity of illness and higher utilisation of health care than those who had fully transitioned to adult-oriented providers at their inception in the cohort (i.e. less complex patients would more easily have completed transition to adult-focused care). Additionally, we theorised that differences in total annualised charges would directly correlate with levels of healthcare utilisation.

Methods

Study cohort

Our cohort was identified from administrative data gathered from our University Hospital's Health System 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 in 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). The 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).

We then identified all encounters for the cohort during the study period and classified their settings (inpatient, outpatient, or emergency). Chart review was conducted to determine the number of subspecialty disciplines utilised by each patient, their sources of primary care, and whether they were housed in child- or adult-focused services. As one of the study objectives was to identify which adults with Down syndrome were receiving care from physicians trained in the care of adult patients, all providers with training in adult medicine (e.g. Internal Medicine, Family Medicine and combined Internal Medicine-Paediatrics) were categorised as adult-focused providers. Likewise, clinicians trained only in the care of paediatric patients were classified as child-focused providers. After incorporating the specialties of referring and primary care providers, electronic medical chart review and administrative claims were used to categorise individuals into one of four mutually exclusive provider groups based on their utilisation patterns: only child-focused providers (paediatric), only adult-focused providers (adult), both child- and adult-focused providers (mixed), and those for whom a referring primary care physician was unable to be identified (unknown PCP). Analysis was restricted to provider groups containing at least 20 patients, which led to the exclusion of the paediatric and unknown PCP provider groups for a final cohort of 205 patients (Fig. 1). It is important to note that no formal transition policy was in effect at our institution during the course of this study. Rather, decisions regarding the timing and mechanisms of transition were left to the discretion of individual providers and departments. Although there was a general rule that persons over 18 years of age should be hospitalised in adult settings, this could be modified by the physicians depending on the clinical circumstances of the patient.

Within this cohort, EMERSE was utilised to verify the presence of comorbidities commonly associated with Down syndrome (e.g. congenital heart disease, hypothyroidism, and atlanto-axial instability). 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). The Institutional Review Board at our University Hospital approved the study protocol.

Determination of additional variables

The length of time spent in the study cohort was calculated by the number of years each patient was aged 18–45 years within the study time frame (1 January 2000–30 June 2008). Where applicable, the time period was adjusted for date of death. Length of stay, the number of days spent in an intensive care unit (ICU), and the location of each hospitalisation (children's versus adult hospital, ICU versus ward service) were determined through manual chart review. Individual encounter charges were converted to 2010 US$ using the Bureau of Labor Statistics consumer price index (Bureau of Labor Statistics 2010). Annualised encounters and charges were calculated through division of their total respective sums by the number of years each patient was in the study. Although the specialties of referring physicians were incorporated into provider group assignments, only encounters within our health system ($n = 4601$) are included in this analysis because of the nature of our data source. As a result of inconsistent

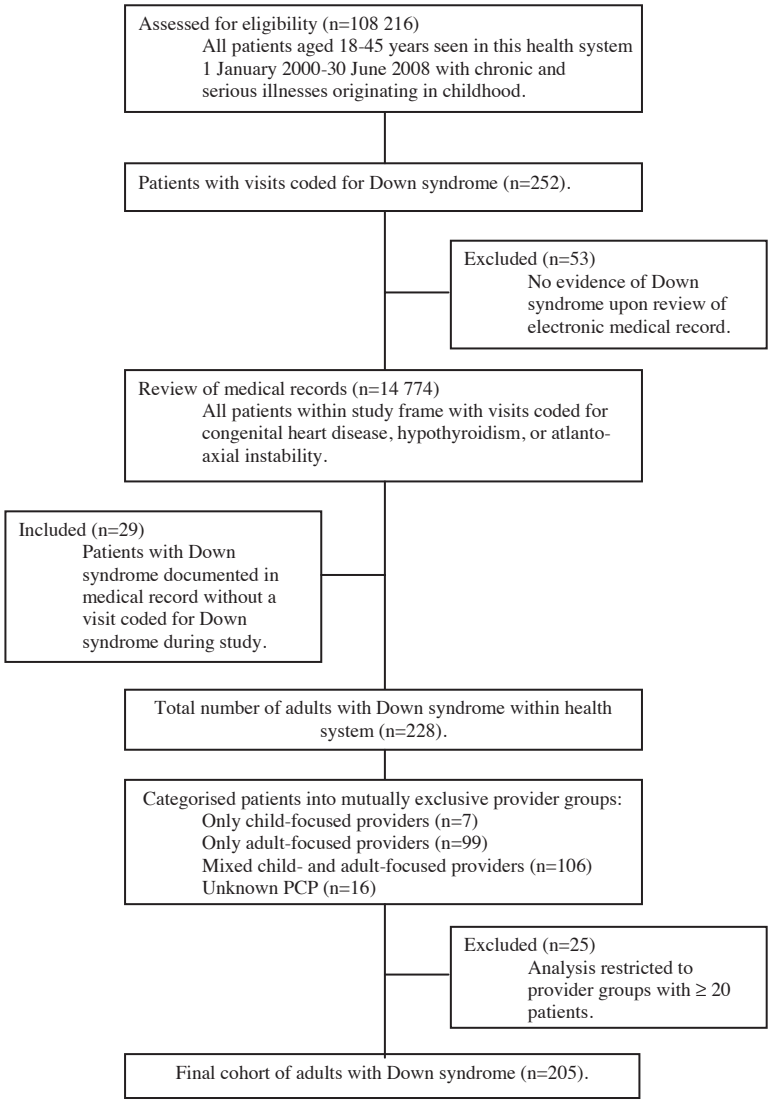

Figure 1 Study cohort identification.

documentation of medical insurance for patients in this dataset, insurance source was not included as a variable in this analysis.

Data analysis

Bivariate comparisons of utilisation patterns and patient attributes between provider groups were conducted using Kruskal–Wallis and chi-squared tests. To account for the skewed distribution of charge and encounter frequency data, both health system charges and encounter frequency data were log-transformed; ordinary least squares regression was then used to evaluate the association between provider groups and charges, adjusting for age at index encounter, hospitalisation status, mortality, log-transformed encounters, and the number of subspecialty disciplines accessed. Stata version 11 software (Stata Corp, College Station, TX, USA) was used for all analyses.

In additional analyses, we considered other explanatory variables for patterns of healthcare charges. These included the presence of specific comorbidities, the presence and severity of congenital heart disease, utilisation of specific subspecialty disciplines, referral status, and whether patients required any operative procedures during the study period. However, inclusion of these variables consistently led to worse fit of the regression model described above. Therefore, we present only the findings of our original model.

Results

Sample characteristics

We identified a total of 205 patients with Down syndrome aged 18–45 years who received care at our institution from 1 January 2000 to 30 June 2008, constituting 1113 patient-years of data. There were 13 deaths among the patients in this cohort during the study period (Tables 1,2). Comorbidities were common but not universal within this cohort: 52% of patients had hypothyroidism, 43% of patients had congenital heart disease, and 7% had atlanto-axial instability.

Encounters ($n = 4504$) were predominantly outpatient (94%). Thirty-six per cent of our cohort received primary care services through our health system, while 95% received specialty care services (Table 3). Over the 8.5-year study period, there were 173 total hospitalisations, reflecting inpatient stays for 40% of the cohort members (0.16 hospitalisations/patient-year) (Table 2). The medical specialties most frequently utilised by our cohort were paediatric cardiology (41%), adult neurology (21%), otolaryngology (19%), orthopaedic surgery (12%), ophthalmology (11%), and adult gastroenter-

Table 1 Patient characteristics by provider group

Characteristics (%)	Total	Adult care	Mixed care	*P*-value
n	205	99 (48%)	106 (52%)	–
Gender				
Female	90 (44%)	44 (44%)	46 (43%)	0.88
Self-reported race/ethnicity				
Caucasian	172 (84%)	80 (81%)	92 (87%)	0.24
African American	16 (8%)	9 (9%)	7 (7%)	0.51
Hispanic	3 (2%)	1 (1%)	2 (2%)	0.6
Asian	1 (1%)	1 (1%)	0	0.3
Other/unknown	13 (6%)	8 (8%)	5 (5%)	0.32
Median age (years) at index encounter (IQR)	28 (19–37)	35 (27–39)	20 (18–30)	<0.001
Presence of common Down syndrome comorbidities				
Congenital heart disease	89 (43%)	9 (9%)	80 (76%)	<0.001
Hypothyroidism	106 (52%)	50 (51%)	56 (53%)	0.74
Atlanto-axial instability	15 (7%)	7 (7%)	8 (8%)	0.9
Mortality during study years	13 (6%)	8 (8%)	5 (5%)	0.32
Median # years in study	6 (3.2–7.8)	6 (3.2–7.8)	5.8 (2.3–7.5)	0.12
Primary care provider specialty*				
Internal Medicine	72 (35%)	48 (49%)	24 (23%)	<0.001
Paediatrics	21 (10%)	0	21 (20%)	<0.001
Internal Medicine-Paediatrics	14 (7%)	6 (6%)	8 (8%)	0.67
Family Medicine	93 (45%)	41 (41%)	52 (49%)	0.27
General Practice	1 (1%)	1 (1%)	0	0.28
Non-Primary Care Specialty	4 (2%)	3 (3%)	1 (1%)	0.3

* Includes referring primary care providers, as well as those within the healthcare system analysed in this study.
IQR, interquartile range.

Table 2 Causes of death during study years*

Age (years)	Gender	Race/ ethnicity	Cause of death	Provider group
26	M	Caucasian	Hypoxaemia secondary to Eisenmenger syndrome, Electrolyte disturbances secondary to gastrointestinal illness	Mixed
31	F	Caucasian	Community-acquired pneumonia, hypoxaemic respiratory failure, Eisenmenger syndrome	Mixed
32	F	Caucasian	Unknown	Mixed
35	M	Unknown	Unknown	Mixed
36	M	Caucasian	Unknown	Adult
40	F	Caucasian	Unknown	Adult
42	M	Caucasian	Chondrosarcoma of pelvis	Mixed
44	F	Caucasian	Unknown	Adult
45	F	Caucasian	Post-operative cerebrovascular accident, pneumothorax	Adult
46	M	Caucasian	Unknown	Adult
46	F	Caucasian	Unknown	Adult
46	F	Caucasian	Pulmonary hypertension	Adult
47	F	Caucasian	Subdural haematoma secondary to fall	Adult

* Cases listed in order of ascending age.

Table 3 Outpatient utilisation patterns by location and provider group

Utilisation (%)	Total ($n = 205$)	Adult care ($n = 99$)	Mixed care ($n = 106$)	*P* value
Total encounters	4504	2134	2370	<0.001
Primary care				
Total patients	74 (36%)	54 (55%)	20 (20%)	<0.001
Subspecialty care				
Median # subspecialty disciplines accessed (IQR)	2 (1–3)	2 (1–3)	2 (1–3)	0.14
Total patients	195 (95%)	90 (91%)	105 (99%)	0.007
Outpatient care				
Median annualised visits	2.1	2.1	2.0	0.95
Total patients	201 (98%)	95 (96%)	106 (100%)	0.04
Total encounters	4220 (94%)	1977 (93%)	2243 (95%)	0.01
Emergency care				
Median annualised visits	0	0	0	0.1
Total patients	42 (21%)	25 (25%)	17 (16%)	0.1
Total encounters	111 (3%)	69 (3%)	42 (2%)	0.002

IQR, interquartile range.

ology (14%). There were no differences in the proportion of patients accessing the most common medical specialties with the exceptions of paediatric cardiology (adult = 0%, mixed = 78%, $P < 0.001$) and adult gastroenterology (adult = 20%, mixed = 9%, $P = 0.02$).

Provider group characteristics

The majority of adults with Down syndrome within our health system had incompletely transitioned to a full adult-care model and were seen by a combination of child- and adult-focused providers

Table 4 Congenital heart disease complexity by provider group

Complexity of congenital heart disease	Total ($n = 205$)	Adult care ($n = 99$)	Mixed care ($n = 106$)	*P* value
None	116 (57%)	90 (91%)	26 (25%)	<0.001
Mild	17 (8%)	3 (3%)	14 (13%)	<0.001
Moderate	40 (20%)	2 (2%)	38 (36%)	<0.001
Severe	32 (16%)	4 (4%)	28 (26%)	<0.001

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.

(mixed = 52%), compared with 48% of patients seeing entirely adult-focused providers (Table 1). Nearly all persons in our cohort received primary care from providers trained in adult medicine (e.g. Internal Medicine, Family Medicine, or combined Internal Medicine-Paediatrics). The median age at index encounter was significantly younger in the mixed provider group (20 years) than in patients seen only by adult-focused providers (35 years).

Although the vast majority of our cohort (95%) required evaluation by a subspecialist over the course of this 8.5-year study, a higher proportion of patients in the mixed provider group were seen only as referral patients in our health system (81%) compared with their peers in the adult provider group (46%). Both provider groups demonstrated similar proportions of hypothyroidism and atlanto-axial instability. However, only 9% of patients in the adult provider group had congenital heart disease compared with 76% among the mixed provider population (Table 3). No differences were observed in total numbers of subspecialty disciplines accessed between provider groups. However, we found that those patients with congenital heart disease in our cohort were more likely to have moderate or severe disease than mild disease (Table 4).

Differences in healthcare utilisation

Over this 8.5-year study, patients in both provider groups demonstrated similar overall patterns of healthcare utilisation (median annualised total visits: adult = 2.2, mixed = 2.3, $P = 0.74$). There were no differences in median annualised hospitalisations or emergency visits.

Only 12% of all hospital admissions during the study period required intensive care (total hospitalisations = 173, total ICU admissions = 20). There were no differences between provider groups in the number of days spent in an ICU or overall length of stay for each hospitalisation (Table 5).

Direct comparison of admissions to the adult hospital versus the children's hospital showed no differences in length of stay, the number of hospitalisations involving time in the ICU, the number of days in the ICU, total number of operations, or individual hospitalisation charges (data not shown).

Differences in total charges

As illustrated in Table 6, mean annualised total patient charges were significantly higher for patients who received care from a combination of child- and adult-focused providers (mixed) than for those seen entirely by adult-focused providers. This finding was present in unadjusted analyses and held after adjusting for age at index encounter, total annualised patient visits, mortality and the number of subspecialty disciplines accessed by each patient. These findings became even more pronounced when the model was also controlled for whether a patient required hospitalisation during the study period. For example: whereas the annualised difference in adjusted means between non-hospitalised mixed and adult provider groups was $571, the difference

Table 5 Inpatient utilisation patterns by location and provider group

Utilisation	Total (*n* = 205)	Adult care (*n* = 99)	Mixed care (*n* = 106)	*P* value
Inpatient care – all				
Median annualised visits	0	0	0	0.7
Total inpatients (% total)	81 (40%)	42 (42%)	39 (37%)	0.41
Total inpatient encounters	173	88	85	0.43
Median length of stay (days) (IQR)	4 (2–7)	4 (2–8)	4 (2–7)	0.3
Paediatric*				
Patients (% inpatients)	24 (30%)	0	24 (62%)	<0.001
Admissions (% admissions)	41 (24%)	0	41 (48%)	<0.001
Median length of stay (days) (IQR)	4 (1–7)	–	4 (1–7)	–
Adult*				
Patients (% inpatients)	64 (80%)	42 (100%)	22 (56%)	0.001
Admissions (% admissions)	132 (76%)	88 (100%)	44 (52%)	<0.001
Median length of stay (days) (IQR)	4 (2–7.5)	4 (2–8)	3 (2–6.5)	0.54
Inpatient care – ICU				
Patients (% inpatients)	19 (24%)	9 (21%)	10 (26%)	0.93
Admissions (% admissions)	20 (12%)	10 (11%)	10 (12%)	0.93
Median length of stay (days) (IQR)	8 (3.5–14)	8 (3–17)	5 (4–13)	0.7
Median # days in ICU (IQR)	4 (3–9)	6.5 (4–9)	3.5 (3–5)	0.15
Paediatric*				
Patients (% inpatients)	6 (7%)	0	6 (15%)	0.02
Admissions (% admissions)	6 (30%)	0	6 (60%)	0.003
Median length of stay (days) (IQR)	7 (5–13)	–	7 (5–13)	–
Median # days in ICU (IQR)	3.5 (3–4)	–	3.5 (3–4)	–
Adult*				
Patients (% inpatients)	13 (16%)	9 (21%)	4 (10%)	0.12
Admissions (% admissions)	14 (70%)	10 (100%)	4 (40%)	0.003
Median length of stay (days) (IQR)	8 (3–17)	8 (3–17)	3.5 (3–14.5)	0.57
Median # days in ICU (IQR)	4.5 (3–9)	6.5 (4–9)	4 (2–15.5)	0.57

* Manual chart review was used to differentiate between paediatric and adult inpatient encounters by provider group.
ICU, intensive care unit; IQR, interquartile range.

Table 6 Adjusted mean annualised total charges by provider group

	Adult care only	Mixed care*
Not hospitalised	$2 305	$2 876
Hospitalised*	$19 240	$38 301

* Comparisons between Provider Groups, as well as across Provider Groups within 'not hospitalised' and 'hospitalised' strata were significant, at $P < 0.001$.
Analysis controlled for age at index encounter, number of organ systems requiring care by a specialist, mortality, and log-transformed annualised total visits.

among patients requiring hospitalisation was $19 061. As noted previously in the Methods section, this trend persisted even when analysis was controlled for the presence and severity of congenital heart disease.

Comment

In this study of over 200 adults with Down syndrome who sought care at an integrated academic medical centre with distinct child- and adult-focused facilities, we found that approximately half of this cohort continued to receive components of their care from child-focused providers. Within this population, we observed higher proportions of patients being referred into our medical centre for

subspecialty care. This was especially noted within those seeking care for complex congenital heart disease.

The issue of incomplete transition to adult-oriented care for persons with SHCN is not unique to our health system. Review of the existing medical literature demonstrates that difficulties identifying competent adult-oriented providers can pose tremendous barriers to successful transitions (Lotstein *et al*. 2005; Knauth *et al*. 2006). Additionally, recently published data indicate that 50% of internists do not view themselves as prepared to provide primary care for young adults with SHCN (Okumura *et al*. 2010). Work done by Pace *et al*. (2011) supports this finding with nearly 25% of physicians surveyed feeling uncomfortable or neutral in providing medical care for persons with Down syndrome (Pace *et al*. 2011). However, dramatic increases in the survival of patients with SHCN necessitate ongoing efforts to improve the transition process to adult-focused providers.

Areas of general consensus within existing transition literature highlight the fact that transition is a process that needs to be both clinically and developmentally appropriate (Sawyer *et al*. 1997; Bryon & Madge 2001; Lotstein *et al*. 2005; White 2009). Experts in the field consistently emphasise that a primary goal of transition to adult-oriented providers is to optimise the lifelong potential of patients by addressing the fact that they are now adult survivors of childhood disease (American Academy of Pediatrics 2001; Bull & the Committee on Genetics 2011). However, researchers also note the difficulties providers face in tracking patterns of transition through which the process can be evaluated (Scal *et al*. 1999).

We had originally hypothesised that adults with Down syndrome receiving care in child-focused settings would have both higher complexity of illness and higher utilisation of health care than those who had fully transitioned to adult-focused providers by their inception in the cohort. We found this to be partially true. Despite similar patterns of healthcare utilisation, we observed marked differences between the provider groups. Patients receiving components of their care from child-focused providers (mixed) were found to have both higher proportions and increased complexity of congenital heart disease than their peers who had fully transitioned to adult providers by onset of the study. Additionally, lower proportions of patients in the mixed provider group received primary care at our institution, indicating a referral population that may be inherently more medically complex.

We also hypothesised that higher annualised charges would be associated with increased healthcare utilisation. We found no differences in annualised encounters between the adult and mixed provider groups. However, we did observe that patients who had incompletely transitioned to adult care (mixed) incurred higher annualised total charges than those in the adult provider group. This difference became more pronounced among those patients requiring hospitalisation during the study years.

These findings suggest that the reasons for which these adults were referred to child-focused providers in this tertiary care centre are closely linked to their need for medically intense care. Given that we are unable to access healthcare utilisation or charge data outside of our health system, we believe that the observations in this study more accurately represent domains in which high intensity care was needed by adults with Down syndrome, rather than their overall care patterns.

Previous work on transitions for patients with SHCN has focused mostly on patient and provider preferences, as well as on the availability of providers both knowledgeable and comfortable caring for this population (Hartman *et al*. 2000; Soanes & Timmons 2004; Steinbeck *et al*. 2008; Okumura *et al*. 2010; Pace *et al*. 2011). Our work for adults with Down syndrome suggests that complexity and severity of illness have significant implications for patterns of transition and healthcare charges, even within a patient population with the same central health issue in a single health system with uniform availability of specialists.

Limitations

This study has several limitations. First, our work reflects the experiences of patients with Down syndrome at a single tertiary care centre. The academic medical centre analysed here has distinct adult and children's hospitals that are fully integrated into one health system, both through a shared electronic medical record and through geographic proximity.

This set of circumstances may differ from other referral centres for patients with SHCN, in which children's hospitals are separated both geographically and administratively from adult-focused hospitals. Therefore, our health system may allow for more cross-disciplinary provider communication between child- and adult-focused care settings and potentially less impetus for full transition to adult-oriented care than is present in other health systems.

Second, the health system analysed in this study is a major referral centre for rare conditions linked to Down syndrome, such as congenital heart disease. This may have led to a more medically complex patient population at our institution than is typical for adults with Down syndrome. Although the overall proportion of patients with congenital heart disease within our cohort is similar to national estimates (American Academy of Pediatrics 2001; Wilson & Cooley 2006; Bull & the Committee on Genetics 2011), the available data do not provide enough detail to allow direct comparison of complexity of congenital heart disease.

Third, because of its status as a major referral centre, this health system also sees many patients strictly for subspecialty care. Therefore, we are unable to track global healthcare utilisation for all of the patients in this study. However, one strength of this study is that attribution of care settings relied on our investigators' knowledge of peer clinicians and their clinical settings, which would not be as feasible in broader multi-institutional efforts. We do not believe that failure to capture primary care encounters would have strongly biased our findings, as we expect that they were chiefly missing for the mixed provider group, which already has higher annualised charges within our cohort.

Fourth, identification of many additional comorbidities associated with Down syndrome was limited by the fact that the majority of this cohort was seen only by medical subspecialists in our institution. We suspect that comorbidities such as obstructive sleep apnoea, behavioural/psychiatric changes, hearing loss and vision abnormalities would not be well captured in the medical records of referral-only patients. We do feel that evaluation of a broader range of comorbidities associated with Down syndrome and their implications on service provision will be a useful step in future studies.

These limitations notwithstanding, we expect that the vast majority of adults with Down syndrome in the USA receive all or part of their ongoing medical care in tertiary care centres with subspecialty care resources. Therefore, our study cohort is likely similar to those of other referral centres. In other words, the challenges of transitioning populations of adults with Down syndrome and other chronic conditions of childhood are not unique to our health system.

Conclusion

Despite long-standing guidelines for transition to adult-oriented providers, little is known about healthcare patterns in adults with chronic illnesses originating in childhood. Our findings represent one of the first comparisons of healthcare utilisation and charges in any cohort of adults with a SHCN, and represent the first such study in a large cohort of adults with Down syndrome.

We found evidence that the reasons behind incomplete transition to adult-oriented providers among adults with Down syndrome appear to be related to the need for medically intense specialty care. Despite similar overall healthcare utilisation, these differing transition states appear to impact annualised charges. Although we cannot evaluate a specific 'transition point' through analysis of administrative and clinical data, it is remarkable that half of our cohort continued to see child-focused providers in some capacity, given their median age of the late 20s.

The prevalence of incomplete transition to adult-oriented providers observed in this study may serve as a reflection of several possibilities: incomplete adherence to existing recommendations for transition to full adult-oriented providers, continuing preferences of adult patients and their families for paediatric subspecialty care, or a lack of adult-oriented specialty providers with the necessary training to address the specific healthcare needs of this population. In light of such findings, it is important to consider whether existing transition guidelines should be adapted to openly accommodate the most clinically appropriate care, regardless of whether it is housed within an adult- or child-focused specialty. Whether transition to a full adult-care model is

clinically or psychosocially appropriate for these individuals remains to be seen. Further studies must evaluate the implications of observed care patterns on clinical outcomes.

References

American Academy of Pediatrics (1996) Transition of care provided for adolescents with special health care needs. American academy of pediatrics committee on children with disabilities and committee on adolescence. *Pediatrics* **98,** 1203–6.

American Academy of Pediatrics (2001) Health supervision for children with Down syndrome. *Pediatrics* **107,** 442–9.

American Academy of Pediatrics, American Academy of Family Physicians & American College of Physicians – American Society of Internal Medicine (2002) A consensus statement on health care transitions for young adults with special health care needs. *Pediatrics* **110,** 1304–6.

Baum R. A., Nash P. L., Foster J. E., Spader M., Ratliff-Schaub K. & Coury D. L. (2008) Primary care of children and adolescents with Down syndrome: an update. *Current Problems in Pediatric and Adolescent Health Care* **38,** 241–61.

Bryon M. & Madge S. (2001) Transition from paediatric to adult care: psychological principles. *Journal of the Royal Society of Medicine* **94** (Suppl. 40), 5–7.

Bull M. J. & the Committee on Genetics (2011) Health supervision for children with Down syndrome. *Pediatrics* **128,** 393–406.

Bureau of Labor Statistics (2010) *Inflation calculator*. Available at: http://data.bls.gov/cgi-bin/cpicalc/pl (retrieved 6 October 2010).

Chicoine B. & McGuire D. (2010) *Health issues for adults with Down syndrome*. Available at: http://www.advocatehealth.com/luth/documents/downsyndrome/healthissues.pdf (retrieved 19 November 2010].

Cooley W. C. & Graham J. M., Jr (1991) Down syndrome – an update and review for the primary pediatrician. *Clinical Pediatrics (Philadelphia)* **30,** 233–53.

Cooley W. C. & Sagerman P. J. (2011) Supporting the health care transition from adolescence to adulthood in the medical home. *Pediatrics* **128,** 182–200.

Glasson E. J., Sullivan S. G., Hussain R., Petterson B. A., Montgomery P. D. & Bittles A. H. (2002) The changing survival profile of people with Down's syndrome: implications for genetic counselling. *Clinical Genetics* **62,** 390–3.

Hanauer D. A. (2006) EMERSE: the electronic medical record search engine. *AMIA Annual Symposium Proceedings*, 941.

Hanauer D. A., Englesbe M. J., Cowan J. A., Jr & Campbell D. A. (2009) Informatics and the American college of surgeons national surgical quality improvement program: automated processes could replace manual record review. *Journal of the American College of Surgeons* **208,** 37–41.

Hartman A., Depoy E., Francis C. & Gilmer D. (2000) Adolescents with special health care needs in transition: three life histories. *Social Work in Health Care* **31,** 43–57.

Knauth A., Verstappen A., Reiss J. & Webb G. D. (2006) Transition and transfer from pediatric to adult care of the young adult with complex congenital heart disease. *Cardiology Clinics* **24,** 619–29.

Lotstein D. S., Mcpherson M., Strickland B. & Newacheck P. W. (2005) Transition planning for youth with special health care needs: results from the national survey of children with special health care needs. *Pediatrics* **115,** 1562–8.

Okumura M. J., Mcpheeters M. L. & Davis M. M. (2007) State and national estimates of insurance coverage and health care utilization for adolescents with chronic conditions from the national survey of children's health, 2003. *The Journal of Adolescent Health* **41,** 343–9.

Okumura M. J., Kerr E. A., Cabana M. D., Davis M. M., Demonner S. & Heisler M. (2010) Physician views on barriers to primary care for young adults with childhood-onset chronic disease. *Pediatrics* **125,** e748–754.

Pace J. E., Shin M. & Rasmussen S. A. (2011) Understanding physicians' attitudes toward people with Down syndrome. *American Journal of Medical Genetics. Part A* **155A,** 1258–63.

Reiss J. & Gibson R. (2002) Health care transition: destinations unknown. *Pediatrics* **110,** 1307–14.

Rosen D. S., Blum R. W., Britto M., Sawyer S. M. & Siegel D. M. (2003) Transition to adult health care for adolescents and young adults with chronic conditions: position paper of the society for adolescent medicine. *The Journal of Adolescent Health* **33,** 309–11.

Sawyer S. M., Blair S. & Bowes G. (1997) Chronic illness in adolescents: transfer or transition to adult services? *Journal of Paediatrics and Child Health* **33,** 88–90.

Scal P., Evans T., Blozis S., Okinow N. & Blum R. (1999) Trends in transition from pediatric to adult health care services for young adults with chronic conditions. *The Journal of Adolescent Health* **24,** 259–64.

Sherman S. L., Allen E. G., Bean L. H. & Freeman S. B. (2007) Epidemiology of Down syndrome. *Mental Retardation and Developmental Disabilities Research Reviews* **13,** 221–7.

Soanes C. & Timmons S. (2004) Improving transition: a qualitative study examining the attitudes of young people with chronic illness transferring to adult care. *Journal of Child Health Care* **8,** 102–12.

Steinbeck K. S., Brodie L. & Towns S. J. (2008) Transition in chronic illness: who is going where? *Journal of Paediatrics and Child Health* **44,** 478–82.

Steingass K. J., Chicoine B., Mcguire D. & Roizen N. J. (2011) Developmental disabilities grown up: Down syndrome. *Journal of Developmental and Behavioral Pediatrics* **32,** 548–58.

United States Department of Health and Human Services (2010) *Developing Healthy People 2020*. Topic Area: Disability and Health (DH-5). Available at: http://www.healthypeople.gov/2020/topicsobjectives2020/pdfs/Disability.pdf (initially retrieved 5 May 2010, confirmed 12 June 2012).

Warnes C. A., Liberthson R., Danielson G. K., Dore A., Harris L., Hoffman J. I. *et al.* (2001) Task force 1: the changing profile of congenital heart disease in adult life. *Journal of the American College of Cardiology* **37,** 1170–5.

White P. H. (2009) Destination known: planning the transition of youth with special health care needs to adult health care. *Adolescent Health Update* **21,** 1–8.

Wilson G. N. & Cooley W. C. (2006) *Preventative Health Care for Children with Genetic Conditions*. Cambridge University Press, Cambridge; New York.

Yang Q., Rasmussen S. A. & Friedman J. M. (2002) Mortality associated with Down's syndrome in the USA from 1983 to 1997: a population-based study. *Lancet* **359,** 1019–25.

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