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Dennis J. Baumgardner  
Brian Chicoine

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## Challenges of Pediatric Disease in Adulthood

Dennis J. Baumgardner, MD,<sup>1</sup> Brian Chicoine, MD<sup>2,3</sup>

<sup>1</sup>Department of Family Medicine, Aurora UW Medical Group, Aurora Health Care, Milwaukee, WI; <sup>2</sup>Adult Down Syndrome Center, Advocate Medical Group, Park Ridge, IL; <sup>3</sup>Family Medicine, Advocate Lutheran General Hospital, Park Ridge, IL

Elsewhere in this issue of the *Journal of Patient-Centered Research and Reviews*, Doyle and colleagues explore functional outcomes and lived experiences of adults with X-linked hypophosphatemia.<sup>1</sup> This entity, generally diagnosed in the pediatric age group, progresses in adulthood with increasing pain and disability. The study authors identified 3 primary themes common to their patient population: presence of chronic pain, fear of falling, and perception of lack of clinician credence given to patient concerns.<sup>1</sup> The traits of resistance, adaptability, and compensation among patients with X-linked hypophosphatemia also were noted.<sup>1</sup>

We have previously reported on the issue of transitions of care from the pediatric to the adult clinical world among patients with significant childhood-onset disease.<sup>2</sup> That focused review explored the current status of such health care transitions, the logistics and pitfalls of these transitions, and measures that practices and health care systems could take to ensure more effective care.<sup>2</sup> This commentary further discusses the medical pitfalls and psychosocial issues surrounding childhood-onset disease in adult survivors once transition has taken place.

There are many well-described childhood-onset diseases that remain problematic for adults and cause significant physical and psychological morbidity — type 1 diabetes mellitus,<sup>3</sup> sickle cell disease,<sup>4</sup> autism,<sup>5</sup> and pediatric age-onset liver disease,<sup>6</sup> to name a few.

In this editorial, 4 other known medical and surgical conditions have been chosen as examples to illustrate the issues often faced by adults with childhood-onset disease. In all cases, the transition from pediatric to adult care has the potential to produce (at least transiently) lower physical and psychological well-being than at pretransition, and issues may vary by gender and the specific medical condition.<sup>7</sup> Certainly, very individualized experiences of transition are to be anticipated.

### Down Syndrome

We will discuss and primarily focus on Down syndrome (DS) as a prototype condition in this regard. In the past, individuals with DS less frequently lived into adulthood, but that number has increased significantly since the early 1980s, when life expectancy was about 25 years, to about 60 years now.<sup>8-10</sup> People with Down syndrome often have difficulty finding health care providers who can or will provide services for adults. The reasons include: 1) concerns regarding inadequate insurance reimbursement (particularly Medicaid), which may be particularly problematic due to longer time often needed for appointments; and 2) clinician's lack of knowledge, as there is often insufficient training in caring for adults with DS in medical school and residency.<sup>cf.11</sup> An additional challenge for adults with DS is finding a provider who will communicate directly with the individual with DS and will include them (to the level of the patient's capability) in health care decision-making.

Particular medical issues facing adults with DS can be placed broadly (with some overlap) into 3 categories: DS-associated diseases that occur in childhood or adolescence and persist into adulthood; DS-associated diseases that tend to first manifest in adulthood;

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Correspondence: Dennis J. Baumgardner, MD, Aurora Sinai Medical Center, 1020 N. 12th St., #4180, Milwaukee, WI 53233 (dennis.baumgardner@aurora.org)

and common adult comorbidities that may arise independent of, or are partially associated with, DS (diabetes mellitus, dyslipidemia, etc).<sup>8,9,12</sup>

Congenital heart disease (CHD) is one example in the first category. Approximately half of patients with DS have CHD, most commonly endocardial cushion and ventricular septal defects.<sup>12,13</sup> Many of these defects are repaired prior to adulthood; however, patients with DS are at higher risk of acquired valvular and other heart disease, which may not manifest until adulthood.<sup>12</sup> There is a higher risk of embolic (presumed to be related to congenital heart disease) and hemorrhagic stroke in adults with DS.<sup>14</sup> Similarly, orthopedic problems, and the much less common congenital defects of the gastrointestinal tract, also may be discovered and repaired prior to adulthood; celiac disease has a higher prevalence in DS and may first become recognized in adulthood.<sup>8,9,12,13</sup>

Constipation, obesity, hearing loss, and diseases from narrowed otolaryngologic passage (ie, congestion, cerumenosis, obstructive sleep apnea) as well as immune system, hematologic, and ophthalmologic problems are examples of issues that typically carry across the life span.<sup>8,9,12,13,15,16</sup> Seizures in DS have a trimodal distribution, with the second peak occurring after puberty.<sup>9</sup> Thyroid disease (most typically hypothyroidism) and osteopenia/osteoporosis may first manifest in those with DS in childhood or adulthood.<sup>8,9,12</sup>

A common example in the second category of medical issues is development of dementia. One community survey revealed that dementia was associated with mortality in the majority (70%) of older adults with DS.<sup>17</sup> Virtually all patients with DS will manifest neuropathological changes consistent with Alzheimer's disease, and most will manifest clinical/symptomatic Alzheimer's in older age, typically well before the median age of onset in the general adult population.<sup>18,19</sup> The diagnosis of Alzheimer's dementia may be difficult in this population due to baseline cognitive and behavioral function or clinical ascertainment of baseline functioning. The ability of the primary care clinician to follow patients with DS over a long period of time is helpful in the latter regard.<sup>18</sup> Much investigative work remains to be done regarding useful tests for early diagnosis, prevention, and treatment.<sup>18,20</sup>

There are gaps in our knowledge about most co-occurring conditions in adults with DS. Many co-occurring conditions have a different incidence profile. This can lead to over- and underdiagnosis. In addition, we do not have clear direction on how we should alter screening and prevention recommendations. Atherosclerosis is very uncommon, for example, as are solid tumors.<sup>8,14</sup> Should we be screening for certain cancers (eg, colonoscopy, mammogram) or treating patients with statins to lower the risk of myocardial infarctions (that are uncommon)?<sup>8,21</sup> Other conditions are more common — what are screening implications for Alzheimer's, sleep apnea, thyroid disease and others? There are also gaps in our knowledge in how best to include people with DS in their own health promotion. The problem is not that people with DS cannot learn how to participate in promoting their health (most individuals can), the problem is that we have not figured out the best ways to include them in the process.

Particular pitfalls to avoid for clinicians taking care of adult patients with DS include missing physical, sexual, or psychological abuse, failure to provide routine adult preventive care, overevaluation or overtreatment of minor hematologic and other laboratory "abnormalities" that may be typical of DS, delayed diagnosis of emerging disorders such as sleep apnea, leukemia, and early onset Alzheimer's, and failure to address palliative and end-of-life care at the appropriate time.<sup>8,9,15,22</sup>

A significant problem faced by adults with DS is the lack of understanding by providers of the interaction between mental and physical health.<sup>23,24</sup> Particularly when the individual has limited verbal skills, the likelihood that the presentation of the physical problem will be wholly or partially behavioral in etiology or the result of a change in their mental health is significant. This lack of knowledge may lead to inadequate or inaccurate treatment, often overdiagnosis of mental health problems without appreciation of the contribution of physical health. Some psychiatric-like presentations are manifestations of acute regression, which may be triggered by a significant emotional stress.<sup>24,25</sup> Similarly, there is overdiagnosis of mental health problems due to the lack of understanding of common characteristics of people with DS. For example, a very high percentage of people with DS talk

to themselves (“self-talk”). This is often misinterpreted as psychoses and can lead to overtreatment.<sup>23,26</sup>

Social issues are significant, and the following examples only scratch the surface. For many children with DS who transition to adulthood and graduate from the school system, there are significant challenges in finding employment and meaningful activity.<sup>27</sup> Finding appropriate residential opportunities is also a significant challenge for many. These have significant implications for physical and mental health. In older adults, the earlier onset of some health conditions, particularly Alzheimer’s, presents additional challenges. This is often complicated by the primary caregiver being an aging parent(s). Both authors of this editorial have observed the unique situation for siblings in which both the parent and the sibling with DS develop Alzheimer’s at the same time.

### **Cystic Fibrosis**

In developed countries, the majority of patients with cystic fibrosis (CF) live into adulthood.<sup>28,29</sup> This has resulted in the need for infrastructure to ensure a workforce of respiratory care clinicians and support staff who are adequately trained in the management of CF in adults.<sup>28,29</sup> Primary care clinics also need to be attuned to the nuances of care for this population<sup>29</sup> and where to best access referral services, including lung transplant teams, if needed.

Medically, major life-threatening presentations in adult patients with CF include respiratory infections, pneumothorax, respiratory failure, and gross hemoptysis.<sup>28</sup> Nonthreatening but important issues include the potential for additional morbidity (eg, diabetes), substance abuse, liver disease, and other adult medical problems.<sup>28</sup> Some clinical paradigms are changing with regard to adult CF care; for example, a decline in the prevalence of traditional CF airway pathogens has been observed.<sup>30</sup>

Major psychosocial issues facing adults with CF include having normal family and friend relationships, employment barriers (including the need for additional time to complete assignments and illness-day accommodations), coping with complex medical needs and treatments, depression, anxiety, and uncertainty and distress regarding early death.<sup>28,31</sup> Other issues cross over between the psychosocial

and the medical realm, including concealment of disease, reproductive challenges, and adherence to treatment.<sup>28,31</sup> Screening for and attention to socially complex young adults with CF at the time of transition to adult care may result in fewer avoidable hospitalizations.<sup>32</sup>

Fortunately, in the case of CF, there are now many facilities for adult CF care in the United States, and transition from pediatric providers generally occurs without significant gaps in care.<sup>33</sup> That said, major complications and premature death remain prevalent. Attention must be paid to proper palliative and end-of-life care.<sup>28,34</sup>

### **Congenital Heart Disease**

Once associated with a grim prognosis, most patients with CHD undergo successful neonatal cardiac surgery and survive into adulthood.<sup>35,36</sup> One author made the point that these individuals “are not ‘cured’ and almost all require lifelong cardiac care.”<sup>35</sup> While limited adult life span may be a central issue for patients with CF, some adults with CHD also still deal with “threatened futures.”<sup>31</sup> Like all examples discussed in this editorial, a primary care medical home and coordination with specialists is paramount for CHD management, and issues of lifestyle, prevention, and psychosocial care remain important.<sup>2,31,35</sup>

Significant medical/surgical issues remain for many adults with CHD, including risk of reoperation, heart failure, need for heart transplantation, complications like atrial fibrillation, and premature death.<sup>36</sup> Increased risk of premature death is particularly associated with the following CHD patient groups: those having had cyanotic heart disease (especially those with Eisenmenger syndrome [development of pulmonary hypertension leading to shunt reversal and cyanosis]); those with repaired tetralogy of Fallot; those with Fontan palliation procedures for single ventricle; and those with subaortic right ventricles.<sup>36</sup>

Currently, a number of percutaneous and other less invasive techniques for cardiac repair in adults with CHD are increasingly being used.<sup>37</sup> While this has decreased the number of more invasive surgeries, and increased the quality of life for these patients, long-term outcomes need to be studied.<sup>37</sup>

## Hirschsprung Disease

Lastly, the rare congenital condition Hirschsprung disease is generally surgically repaired during the first year of life, and most patients are diagnosed and repaired prior to young adulthood.<sup>38</sup> Despite successful repair, significant issues remain for adults who had been treated for this condition as children. This includes constipation, fecal or urinary incontinence, repeat surgery, and issues of sexual function.<sup>38,39</sup> A study of 66 adults with repaired Hirschsprung disease in France observed reports of significant emotional distress and concerns regarding body image, but higher educational levels and similar marital and parental status, as those of the general population.<sup>38</sup> While sexual function is generally preserved in adults with Hirschsprung disease, sexual distress was a significant issue in adult women and associated with perceived self-competence as an adolescent.<sup>39</sup> Hirschsprung disease has been a significant challenge for patients with DS regarding limitations on their independence due to incontinence.<sup>40</sup>

## Improving Clinical Support

What can we, as clinicians and caregivers, do to help this population? First, we must appreciate the blessing of these individuals living well into adulthood and frequently thriving. Beyond that, we must ensure that there is an adequately trained workforce to care for these adult survivors of serious congenital disease. In some cases, traditional pediatric specialists have been extending their care of these specific populations within their practice well into adulthood.<sup>41</sup>

We must strive to provide appropriate, thoughtful, and informed care of common adult conditions that may have their onset in childhood. This includes promotion of self-management of health conditions, when appropriate.<sup>7</sup> In addition, we should be attuned to “normal abnormal” laboratory findings in such individuals, as those with DS, in addition to unique adult-onset illnesses such as early Alzheimer’s dementia. We must be prepared to provide or refer for appropriate palliative and end-of-life care when the need arises.

Equally important, we need to strive to ensure adequate psychosocial support for these individuals beyond that needed for typical adolescent-to-adult transition.<sup>7,38</sup> This includes social services and community-based programs and resources as needed, screening for abusive situations, and, particularly in the case of CF, psychological support regarding patients’ concerns

surrounding a limited life span.<sup>31</sup> We must inquire of the patient and/or caregivers regarding perceived stigma due to the patient’s intellectual or physical disabilities and provide appropriate support.<sup>42</sup> Advocacy for the patient regarding appropriate work and adult education accommodations is important as well.<sup>43</sup>

As always, we should be the caring, competent, and compassionate caregivers that this population deserves.

### Note Added in Proof

The authors are reflecting on the suffering likely endured by adults with Down syndrome and others with cognitive and physical disabilities during this COVID-19 pandemic. Their isolation may be particularly acute. Day programs, work programs, Special Olympics, and other socially enriching activities are suspended. Grocery store “adventures” may not be done safely. Technology-based social networking from home may be severely restricted due to inherent lack of ability to communicate using these media or older parents not having such technologies in their homes. Some individuals may not have the cognitive ability to understand technical/logistical aspects of social distancing and mask wearing. They may be scared and confused. For those being hospitalized for COVID-19, this may be the first frightening experience ever of not having a parent or personal caregiver at their side. These patients need our particular attention during these times.

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