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Prevention and awareness of birth defects across the lifespan using examples from congenital heart defects and spina bifida

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1 | INTRODUCTION

The emergence of birth defects programs in the United States accelerated in the 1970s and 1980s due to recognition that the use of the drug thalidomide during pregnancy resulted in fetal abnormalities (McBride, 1961; Smithells, 1962) and concerns around environmental exposures, such as Agent Orange exposure during the Vietnam War (Erickson et al., 1984). These experiences shaped the mission of many birth defect programs to focus on the surveillance of fetuses/infants affected by birth defects to monitor prevalence, identify and respond to clusters, and explore the epidemiology of birth defects as early warning systems to identify potential teratogens. This work helped identify additional risk factors for birth defects, support primary prevention opportunities, such as folic acid fortification and supplementation for neural tube defect prevention, and enabled evaluations of the success of those efforts (Harris et al., 2017).

Improved early identification of birth defects through prenatal detection and critical congenital heart defect screening, as well as advances in clinical interventions and treatments, have improved survival of infants and children with birth defects over the last few decades (Ho, Quigley, Tatwavedi, Britto, & Kurinczuk, 2021; Wang, Hu, Druschel, & Kirby, 2011). Conditions once largely limited to infancy are now impacting the health

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and well-being of adults. For example, there are more people living with congenital heart defects and spina bifida in the United States than infants born with these conditions each year (Danielson, McKing, Devine, & Correa, 2009; Gilboa et al., 2016; Hoffman & Kaplan, 2002; Ouyang, Grosse, Armour, & Waitzman, 2007; Parker et al., 2010; Reller, Strickland, Riehle-Colarusso, Mahle, & Correa, 2008). The clinical and public health communities have begun to expand their mission and activities to include a focus not only on surveillance during pregnancy/infancy and primary prevention opportunities, but also on the needs of children, adolescents and adults and secondary prevention that can support the health and well-being of individuals living with birth defects across the lifespan.

An initial step to promote the importance of advancing awareness of birth defects across the lifespan was the formal renaming of Birth Defects Prevention Month, which occurs in January each year, to Birth Defects Awareness Month (Centers for Disease Control and Prevention [CDC], 2021a). Historically, this event has provided an opportunity to promote primary prevention messages, including the importance of folic acid consumption before and during pregnancy, the avoidance of alcohol, and maintaining a healthy weight. The change from “prevention” to “awareness” enhances messaging opportunities to highlight issues that impact children, adolescents, and adults living with birth defects. Examples include ensuring children are connected to early intervention services to promote optimal development, supporting adolescents who are transitioning from pediatric to adult clinical care, ensuring adults living with a birth defect are receiving appropriate specialty care, and addressing the mental health needs of individuals living with birth defects. This name change can signal the commitment of the birth defects community to ensuring that the clinical and nonclinical needs of the populations we serve are met. However, much work remains.

In this editorial, we explore public health surveillance and research activities, and clinical and nonclinical outcomes across the lifespan, using examples from congenital heart defects and spina bifida, which improve our understanding of the needs of individuals living with birth defects to promote secondary prevention opportunities.

2 | SURVEILLANCE AND RESEARCH

Most current efforts to conduct population-based surveillance and research of birth defects have focused on monitoring fetuses/infants, and lifespan surveillance has sometimes been considered a special initiative or exclusively a research effort. Despite the public health burden, little population-based information is available on children, adolescents and adults living with birth defects, including secondary disability developing later in life. Surveillance and research are needed on clinical outcomes and services, such as survival, health care access and utilization, and comorbidities, including mental health (Cassell, Grosse, & Kirby, 2014). Surveillance and research are also needed on developmental and nonclinical outcomes and needs, such as behavioral issues, educational needs and attainment, employment, and disability. Population-based surveillance of individuals with birth defects can provide a better understanding of the needed services and resources on a population level. This type of surveillance complements clinic-based research that may

have more information on biomedical markers, measures of disease severity, and treatment history, but lower generalizability to the larger population of individuals with birth defects.

There are multiple methods for conducting birth defects surveillance across the lifespan, each with its own strengths and limitations. Existing efforts have found efficiencies by linking clinical data sources, such as hospital discharge data, Medicaid claims data, and clinical and surgical data (Bennett, Mann, & Ouyang, 2018; Glidewell et al., 2018; Jill Glidewell et al., 2021). However, these linkages may suffer from lower positive predictive value of International Classification of Disease (ICD) codes to identify cases (Khan et al., 2018; Rodriguez et al., 2018) and limited information on severity of disease, past treatments or procedures, and demographic characteristics, including race/ethnicity. These linkages may require administrative efforts, such as data use agreements, navigating and interpreting laws and regulations on sharing personally identifiable information (PII), or creative methods to link and deduplicate cases across data systems, such as probabilistic linkage or hashing algorithms (Dusetzina et al., 2014), when PII is missing or cannot be shared across entities. Standardization of variables across data sets and across surveillance sites takes additional time and resources. All of these issues may reduce data timeliness.

The sensitivity of this type of surveillance methodology for identifying all individuals with birth defects living in a defined area depends largely on the percentage of individuals with the specific birth defect who access care and whose birth defect is documented at those health care encounters, as well as the investigator's comprehensive access to health care data sources in that area. During initial visits to adult congenital heart defect centers, over 40% of adults with heart defects reported a gap in cardiac care of over 3 years and 8% had gaps over 10 years (Gurvitz et al., 2013). Therefore, congenital heart defect surveillance relying on health care encounters with a congenital heart defect-related ICD code may underestimate the prevalence and limit generalizability of results. Utilizing large administrative databases has shed light on health care access and expenditures among individuals with spina bifida; however, limitations exist such as accounting for patients that switch insurance plans, in particular between a public and private payor (Ouyang et al., 2007).

Other surveillance efforts have focused on self-reported or proxy-reported outcomes that may not be found in clinical data, such as quality of life, employment, and educational attainment (Farr et al., 2020). Identification of individuals with birth defects for surveillance efforts becomes more difficult after early childhood when individuals begin dropping out of specialty care, moving outside of their place of birth, or changing their name. Those who remain in specialty care may disproportionately have more severe defects or poorer overall health and may not represent the larger population of people living with birth defects. Tracking individuals with birth defects, starting in early childhood and continuing through adulthood, will limit detection bias and survivorship bias, but takes time and resources.

U.S. population-based surveillance of adolescents and adults with congenital heart defects began in 2012, with CDC funding three organizations to conduct congenital heart defect surveillance among adolescents and adults in their respective catchment areas, linking and deduplicating cases across data sources. This initial project showed the feasibility of conducting population-based surveillance of heart defects in adolescents and adults,

although highlighted differences in prevalence of people with congenital heart defects documented in their health records across sites. Subsequent CDC-funded projects expanded surveillance across the lifespan efforts to five sites in 2015, and seven sites currently. These surveillance systems have also propagated other projects to examine barriers to transition to adult cardiac care, distance to care (Gaydos et al., 2020; Insaf et al., 2021; Schlichting, Insaf, Lui, Zaidi, & Van Zutphen, 2020; Sommerhalter et al., 2017), and validity of ICD codes (Rodriguez et al., 2018) for heart defects.

To examine nonmedical outcomes, in 2016, three birth defects surveillance sites with active case-finding methods, together with CDC and March of Dimes, initiated the Congenital Heart Survey To Recognize Outcomes, Needs, and well-beinG (CH STRONG, http://www.chstrong.org/), with the objective to identify individuals with heart defects who survive into adulthood and survey them on health care use and barriers to care, health concerns, social and educational outcomes, reproductive health, and quality of life (Farr et al., 2020). Among individuals with heart defects born in 1980–1997, current contact information was found for two-thirds. Among the 1,656 participants ages 19–38 years at survey recruitment, over 40% were male and 30% were non-white. Initial CH STRONG findings show increased cardiac and noncardiac comorbidities in young adults with heart defects (Oster et al., 2021), substantial rates of disability (Downing et al., 2021), as well as a low percentage of young adults with heart defects with advance care directives (Farr et al., 2021).

In addition to surveillance efforts, clinic-based research can help us understand more about the care and services received in different clinical settings and clinical outcomes for people with birth defects. An example of clinic-based research is the National Spina Bifida Patient Registry (NSBPR, https://www.cdc.gov/ncbddd/spinabifida/nsbprregistry.html), established by CDC in 2008 to provide a "framework for a systematic approach to improving the quality of care received at spina bifida clinics nationwide" (CDC, 2021b). In response to needs identified by the Spina Bifida Association's (SBA) 2005 survey of spina bifida clinics, the NSBPR was created to assess clinical care administered to people living with spina bifida and describe how care may affect outcomes. More than 20 articles have been published to date using NSBPR data; findings from NSBPR have been used to improve health outcomes and quality of life for people with spina bifida (Thibadeau, 2017). The registry has also informed the development of new research and treatment protocols. For example, based on findings from 2009 to 2012 NSBPR data showing that 26% of patients experienced skin breakdown (Kim et al., 2015), a working group convened by CDC, in collaboration with SBA, developed a Skin Breakdown Prevention Care Bundle. These preventative interventions have been implemented in both NSBPR clinics and other spina bifida clinics.

Another example of a clinic-based effort that can inform care and treatment of individuals with birth defects is the CDC-supported Urologic Management to Preserve Initial Renal Function Protocol for Young Children with Spina Bifida (https://www.cdc.gov/ncbddd/spinabifida/umpire.html) (CDC, 2021c). The development of this protocol aims to determine the best approach to monitoring and testing in the first 10 years of life that can help maintain bladder and kidney function in infants and children with spina bifida (Routh et al., 2016). Infants born with spina bifida can have healthy kidneys at birth; however, they are at risk

for developing kidney failure and urological complications that can lead to morbidity and mortality. This effort aims to mitigate, to the best extent possible, these sequalae of spina bifida.

3 | CLINICAL SERVICES AND OUTCOMES

Individuals with birth defects are living longer (Gilboa et al., 2016; Ho et al., 2021; Shin et al., 2012; Wang et al., 2011) and thus there are needs for appropriate clinical care for children, adolescents, and adults living with birth defects, to support transition of care from pediatric to adult providers, and to ensure sufficient numbers of appropriately trained specialty providers. In addition, there is limited information on aging among those with birth defects and what special considerations might be for these populations.

Knowledge of health care access and outcomes of people with heart defects has grown over the last 10 years. Children and adolescents with heart defects have higher morbidity and mortality than those without heart defects (Razzaghi, Oster, & Reefhuis, 2015), and, among individuals with heart defects, adverse outcomes disproportionately affect racial/ethnic minorities (Lopez, Morris, Sexson Tejtel, Espaillat, & Salemi, 2020). Children and adolescents with heart defects experience substantial rates of additional cardiac and noncardiac conditions, such as heart failure, respiratory/pulmonary conditions, and mental health conditions (Khanna et al., 2019; Lui et al., 2019). Over 1 in 10 have additional noncardiac birth defects and/or genetic syndromes, such as Down syndrome. Adults with heart defects are also more likely than those without heart defects to report other cardiac and noncardiac comorbidities, such as heart failure, stroke, depressive symptoms, and dementia, and adults with heart defects are twice as likely to have any comorbidity (Agarwal et al., 2019; Bagge et al., 2018; Oster et al., 2021). Pregnant people with heart defects, compared to those without, also have higher rates of adverse conditions, such as pulmonary hypertension and cardiac conduction disorders, and adverse outcomes, such as stillbirth and preterm birth (Downing et al., 2020; Raskind-Hood et al., 2020; Schlichting, Insaf, Zaidi, Lui, & Van Zutphen, 2019).

Many individuals with heart defects fall out of specialty cardiology care, and under 10% may receive care at centers specializing in adult congenital heart disease care (Gurvitz et al., 2013; Krasuski & Bashore, 2016). Children begin to drop out of cardiology care in childhood, with higher loss to follow up among racial and ethnic minorities (Jackson et al., 2019). Barriers to transitioning from pediatric to adult cardiology care include replacing the strong relationship with the pediatric provider, locating an adult provider, and accessing adult health insurance (Gaydos et al., 2020), and over half of parents of children with heart conditions report not discussing transition issues with their child's provider (Downing, Oster, & Farr, 2017). While the number of centers specializing in care for adults with congenital heart defects has grown considerably over the past decade, it has been estimated that there are still too few adult congenital cardiologists to care for this growing population (Krasuski & Bashore, 2016).

Similarly, we do not know as much about clinical care for adults living with spina bifida as we do for infants, children, and adolescents. Yet, there are more adults living with spina

bifida than infants, children, and adolescents with spina bifida (Ouyang et al., 2007; Parker et al., 2010). Limited U.S. studies are available, as there are few clinical settings focused on care for adults with spina bifida. However, a recent study (Dicianno & Wilson, 2010) indicates that a significant proportion of hospitalizations for adults with spina bifida may be preventable, such as those for urinary tract infections and pressure ulcers, highlighting the importance of secondary prevention in birth defect awareness activities. A recent cross-sectional survey of adults with spina bifida documented challenges to transition from pediatric centers of care to adult care, where organized systems of care and providers with specialized knowledge about caring for individuals with spina bifida are limited (Dicianno & Wilson, 2010). Individuals living with spina bifida may not receive the care and services they need in a system that lacks experience addressing the needs of the spina bifida population as they age. It is important that we learn more about caring for adults with spina bifida to improve care and quality of life for all ages.

As we learn more about caring for the spina bifida population, we must continue to translate these findings for individuals living with spina bifida, their families, and their providers. This can be done through information dissemination and education. One way to disseminate information is through health care guidelines for spina bifida. In the past, guidelines for caring for individuals with spina bifida primarily focused on pediatric populations and did not fully address the health care needs of adults with spina bifida. In 2016, the SBA led an effort to revise health care guidelines, including the inclusion of new guidelines specific to addressing care across the lifespan. This effort involved more than 100 international experts in care of people with disabilities and spina bifida and led to development of 24 “Guidelines for the Care of People with Spina Bifida,” available on the SBA website (SBA, 2021). Another way to ensure research findings and health care guidelines for spina bifida reach the health care provider community and impact quality of care is through provider training and education. CDC collaborated with the American Academy of Pediatrics (AAP) to implement a virtual quality improvement project with two cohorts of health care providers in 2020 and 2021 to improve transition coordination for individuals diagnosed with spina bifida, consistent with the 2018 Clinical Report on Health Care Transitions (White et al., 2018), Guidelines for the Care of People with Spina Bifida (SBA, 2021), and Got Transitions Six Core Elements (Got transition, 2014). Following these efforts, the CDC and AAP launched a Project Extension for Community Health Care Outcomes, in which providers take a series of trainings and shared learning sessions, aimed at improving the transition of people living with spina bifida from pediatric to adult care.

4 | DEVELOPMENTAL, SOCIAL, EDUCATIONAL, AND QUALITY OF LIFE OUTCOMES

Aside from strictly medical needs, as mentioned earlier, individuals living with birth defects also have unique social, developmental, and behavioral needs. There are challenges with connecting families of people with birth defects to social services, especially for those who may have fallen out of care or who may be uninsured. Having a comprehensive medical home may help identify and diagnose issues of concern early and connect children and adults to the services they need (Lantin-Hermoso et al., 2017).

Children with heart defects may experience developmental delays and cognitive limitations stemming from genetic or chromosomal anomalies, impaired fetal brain development in utero, as well as postnatal surgeries and long hospital stays (Marelli, Miller, Marino, Jefferson, & Newburger, 2016). Children and adolescents with heart defects may be more likely than their counterparts to have difficulty with learning and concentration, communication, self-care, and fine and gross motor skills (Farr, Downing, Riehle-Colarusso, & Abarbanell, 2018). They are also more likely to be diagnosed with autism spectrum disorder and intellectual disability (Razzaghi et al., 2015). Compared to children and adolescents without heart defects, those with heart defects are more likely to receive special education services; miss more days of school in a year, affecting school performance; and participate less in extracurricular activities, limiting social and physical activity (Farr et al., 2018; Riehle-Colarusso et al., 2015). Adults with heart defects experience limitations with cognitive functioning, including executive functioning, and lower employment (Marelli et al., 2016; Cohen & Earing, 2018). While some studies have found reduced quality of life among individuals with heart defects, others have not (Fteropoulli, Stygall, Cullen, Deanfield, & Newman, 2013).

Individuals with spina bifida may have reduced quality of life (Sawin, Brei, & Houtrow, 2020), but contributing factors are poorly understood. In addition to possible medical issues, both societal characteristics (e.g., environments that support participation in school, the workforce, and general societal activities, as well as access to health care and support services) and personal factors (e.g., cultural, health literacy, relationships, and social determinants of health) may impact a person's perception of quality of life. Some of the NSBPR spina bifida clinics have recently embarked on a feasibility study to measure quality of life using an overall quality of life assessment and validated health-related quality of life instruments (Sawin et al., 2020). Children with spina bifida can have difficulty in learning environments due to challenges with paying attention and restlessness. They can also have difficulty reaching milestones (Holbein et al., 2017) as they transition into adulthood (Lindsay, McPherson, & Maxwell, 2017).

5 | OPPORTUNITIES: MEETING THE NEEDS OF PEOPLE LIVING WITH BIRTH DEFECTS ACROSS THE LIFESPAN

There are several actions that the public health, clinical, and nonclinical communities can take to advance the health and well-being of individuals of all ages living with birth defects. First, these communities can improve partnerships with organizations that support individuals living with birth defects. This includes organizations whose missions expressly support generalized birth defects work, condition-specific organizations, and organizations that serve individuals living with birth defects but that might not be actively engaged in birth defects activities. One example of such collaboration is the Congenital Heart Public Health Consortium (CHPHC), which is led by the AAP and has a steering committee comprised of 11 national CHD-focused clinical and patient-family advocacy organizations and federal advisors. The CHPHC mission is to prevent heart defects and improve outcomes for affected children and adults. The CHPHC works toward this mission by providing leadership and a unified voice for public health priorities, expanding opportunities for surveillance and

public health research, and informing public policy priorities that benefit public health and affected persons. Other examples include collaborations between CDC and AAP to improve the transition from pediatric to adult care among patients with spina bifida, and between CDC and the SBA to understand more about factors in the clinical setting that may impact outcomes for spina bifida patients. CDC recently awarded funding to the Association of Maternal and Child Health Programs, which formally engaged the National Birth Defects Prevention Network (NBDPN), to strengthen the relationship between Title V Maternal/Child Health and Children and Youth with Special Health Care Needs programs and birth defect surveillance programs. This project aims to identify and promote best practices for improved collaboration and build workforce capacity to support children and families impacted by birth defects to receive early intervention services that support health and well-being during early childhood. Given the complexity of the conditions, the clinical care systems, and the nonclinical environment (e.g., educational and employment issues), partnerships between federal and state programs, academic institutions, clinical care providers, other service providers, and organizations that represent individuals living with birth defects will advance the science and implementation of findings to ultimately improve health and well-being. Enhancing partnerships provides the opportunity to advance awareness and develop new and enhance existing initiatives by expanding the perspectives contributing to actionable solutions.

Second, to facilitate improved partnerships and inspire action, communication and education mechanisms can be enhanced to share public health information and reach new audiences. The National Action Plan to Improve Health Literacy, released May 2010 by the U.S. Department of Health and Human Services, strives to develop and disseminate health and safety information that is accurate, accessible, and actionable (U.S. Department of Health and Human Services, Office of Disease Prevention and Health Promotion, 2010). Due to widespread availability and usability, digital communication initiatives hold great potential for health promotion. CDC's National Birth Defects Awareness Month (BDAM) digital toolkit is a prime example of using a digital platform to disseminate evidence-based health information to empower families and people living with birth defects with knowledge and resources (CDC, 2021a). CDC's BDAM toolkit includes sections dedicated to specific audiences across the lifespan, including messages for people preparing for pregnancy, those caring for infants with birth defects, educators and people who support optimal childhood development, adolescents transitioning from pediatric to adult care, and adults living with birth defects who may be preparing to have children of their own. CDC's BDAM materials are freely available and can be used by partners to promote awareness of critical issues across the lifespan. Clinical and public health conferences, webinars, newsletters, and other communication tools can also be used to advance knowledge and propagate solutions to challenges facing this community. As another example of ways to reach new audiences, in 2020, the CDC and AAP initiated a program to increase awareness among primary care and urgent care providers of the need for lifelong specialty cardiac care among their patients with heart defects, including how to discuss cardiology care with their patients and where to refer their patients who may have fallen out of cardiology care. The goals of this initiative are to increase retention and re-engagement of people with heart defects in specialized cardiac care. Lastly, national and state programs can revisit their mission

and vision statements and other communication materials to incorporate messaging around the impact of birth defects across the lifespan. How organizations communicate about their programs can demonstrate commitment to supporting this population, increase knowledge about birth defects across the lifespan, and identify opportunities for action.

On the education and training side, there is limited number of providers that feel prepared to care for individuals with birth defects in adulthood, highlighting a need to provide education and training for the current workforce specific to caring for adults living with birth defects. There is also a need to develop and implement training materials for the next generation of the workforce that will be providing care and services for populations of individuals with birth defects. Education at colleges and universities could be more inclusive of lifespan issues related to birth defects and training received in schools of nursing and medicine as well as residencies and fellowships could expand to include information about the care of individuals with birth defects as they age and throughout the life course.

In addition to the opportunities above that all organizations can engage in, there are specific actions that public health birth defects programs can take to advance lifespan efforts. Among public health organizations, one of the primary obstacles to conducting lifespan surveillance is the limited understanding of its benefits and challenges. The field of Maternal and Child Health accepts Life Course Theory to explain the need to care for individuals across the lifespan and for interventions at sensitive time points (Elder, Johnson, & Crosnoe, 2003). This theory can also provide the needed justification for conducting lifespan surveillance and research. Promoting a clear understanding of lifespan surveillance and its benefits among key decision makers can provide the foundation for specific actions needed to progress in this area. While adequate fiscal and human resources are common barrier to birth defects programs expanding their focus to support lifespan surveillance and research efforts, programs can take action to prepare for and/or actively engage in supporting the health and well-being of individuals living with birth defects across the lifespan. First, programs can review their existing regulatory, organizational, and data sharing policies to identify opportunities and barriers to addressing this critical issue. Some programs might be authorized to support lifespan surveillance directly through longitudinal tracking and/or data acquisition and linkage from partner organizations. Others might find that, though not authorized themselves to conduct lifespan surveillance, they can share data with partner organizations to support these activities. Second, programs can determine if changes in data sharing policies or methods are required, such as obtaining IRB approval, executing new data sharing agreements, or using novel data linkage methods, such as hashing, to link data sets without compromising data security policies. Of note, groups such as birth defects advisory councils and committees can help educate public health and legislative leaders about the importance of lifespan surveillance and policies that promote or hinder robust data sharing, such as was the case with the South Carolina Birth Defects Act, which allows comprehensive birth defects data acquisition across the lifespan (South Carolina Legislative Services Agency, 2004). Lastly, programs can ensure that data storage and maintenance procedures are well defined for historically collected data. The NBPDN is an international group of individuals dedicated to birth defects surveillance, providing opportunities for members with expertise in lifespan surveillance to share their knowledge with other members. Of the 51 birth defects programs providing directory information

in the 2019 NBDPN Annual Report, 37 reported case information for births in 2004 or before (Stallings et al., 2019). These individuals would be at least 18 years old in 2022, providing a potentially rich source of information to support lifespan surveillance and research efforts. The NBDPN membership plans to convene a Lifespan Surveillance Workgroup beginning in 2022. This group will be comprised of birth defects surveillance programs, health departments, and other researchers who will collaborate to identify and document strengths and opportunities of U.S.-based birth defects surveillance programs to conduct lifespan surveillance. Resulting products could include white papers, tip sheets, and networking opportunities to navigate common obstacles to conducting lifespan surveillance. In addition, collaborative publications using existing data could help promote the value of this kind of work.

In conclusion, much has been learned about the needs of individuals living with congenital heart defects and spina bifida as they age from childhood to adulthood. The work on these birth defects can serve as models for work on other birth defects, about which less information may be known. Improved knowledge of the health and well-being of people living with heart defects and spina bifida has highlighted opportunities for the public health, clinical care, and nonclinical care communities to work collaboratively to serve people living with birth defects and promote secondary prevention opportunities. Additional efforts to improve partnerships, enhance communication, and leverage existing birth defects registries to track longer term outcomes may lead to further improvements in the health and well-being of this growing population.

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DATA AVAILABILITY STATEMENT

Data sharing not applicable to this article as no datasets were generated or analysed during the current study.

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