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Early Intervention and Newborn Screening

Parallel Roads or Divergent Highways?

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Both early intervention (EI) programs for infants and toddlers with disabilities and newborn screening (NBS) programs to identify specific disorders shortly after birth rest on the assumption that the best way to prevent or lessen the impact of a disorder is to provide treatment as early as possible. Despite this shared vision, the two programs differ in substantial ways, including historical beginnings and subsequent evolution; program entry and eligibility; service models; evidence, efficacy, and outcomes; roles and responsibilities vis-à-vis families; and financing. Each program is well established and highly successful. But 4 crosscutting issues need to be addressed: (1) documenting meaningful outcomes for children and families; (2) deciding whether and how to provide highly effective yet very expensive treatments; (3) implementing surveillance activities to monitor progress and determine optimal timing for intervention; and (4) identifying appropriate intersections between EI and NBS. Solving these challenges will require national and state leadership, stakeholder engagement, collaboration between EI and NBS, and a significant investment of resources if the promise of maximizing benefit for children and families is to be realized.
Key words: *early intervention, newborn screening, program improvement*

EARLY INTERVENTION (EI) and newborn screening (NBS) rest on the same basic assumption, namely, that treatments or services for children with special health care needs or disabilities must begin as early as possible to maximize benefits for children and families. Both are long-standing, state-based programs with established infrastructures guided by national legislation, regulations, and advisory groups. One would think that two programs fulfilling essentially the same mission would have much in common and could be viewed as alternative yet parallel roads heading toward the same ultimate destination. This may be true. But EI and NBS differ in so many fundamental ways that they could also be construed as diverging roads with different destinations.

Having spent decades working in both programs, I believe that EI and NBS share a similar vision and the same general destination. But history and context have led each on very different, partially successful, but ultimately imperfect pathways. Each has a set of core objectives and activities that must be achieved but for which there are stark differences in both assumptions and approach. And both face imminent challenges that need be addressed in the next few years. Comparing and contrasting these assumptions, approaches, and challenges could identify ways in which two successful programs could learn from each other, and

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perhaps even work together, to the ultimate benefit of children, families, and society.

HISTORICAL PERSPECTIVES ON PROGRAM DEVELOPMENT

As well-established programs in the United States, EI and NBS have important but different legacies that shaped initial program development and subsequent program evolution.

Early intervention

Early intervention is a multidisciplinary enterprise, but its roots lie in special education. In 1975, Public Law No. 94-142 (Education for All Handicapped Children Act) was passed, landmark legislation that required free and appropriate public education for all children with disabilities. Although the legislation had language regarding services for preschool-aged children (3–5 years), an emphasis on the infant–toddler years was not enacted until 1986 after intense lobbying by researchers and disability advocacy groups. Part H of the newly named Individuals with Disabilities Education Act (IDEA) added an optional infant and toddler component, further strengthened in 1997 as Part C of IDEA, under the auspice of the Office of Special Education Programs, U.S. Department of Education.

The infant–toddler program is not mandatory, but all states and territories participate, which means they must provide a minimum set of program components to children with or at risk for disability ages birth through 2 years in order to receive federal funding. Although EI is a state-based program, federal requirements strongly affect the way state programs are structured and operate. For more than 40 years, the U.S. Office of Special Education Programs has supported a technical assistance center (e.g., Early Childhood Technical Assistance Center [ECTA]; https://ectacenter.org/) to help states build capacity to improve outcomes for children and families. States are required to submit annual reports to the U.S. Department of Education describing the number and characteristics of participating children, services provided, and progress toward child and family outcomes (U.S. Department of Education, 2020). States have the discretion to identify a lead agency for EI, and these agencies vary widely. The state education agency is the EI lead in about a fourth of states, with the remainder being housed in a variety of health and human service agencies.

Newborn screening

In contrast to EI's special education legacy, NBS evolved out of public health (American Academy of Pediatrics Newborn Screening Task Force, 2000; Association of Public Health Laboratories [APHL], 2013). In the 1960s, Robert Guthrie developed a method using dried blood spots to detect phenylketonuria (PKU), which causes permanent and severe intellectual disability if untreated. A dietary change can almost completely prevent disability, but it must be provided early in life to be effective. Phenylketonuria set the standard for future expectations of NBS—find disorders that can easily be identified and must be treated in the presymptomatic stage. A few states gradually began to offer screening for PKU through state laboratories. As success with PKU became obvious, advocates and researchers began to press for adding more disorders, leading to the evolution of multidisorder screening panels. More states began to offer screening and more disorders were added, primarily based on the availability of screening methods and new treatments, aided by selective state-level advocacy efforts.

No federal mandate exists for NBS, and historically state NBS programs have varied widely in the number of disorders screened. However, in 2006, the U.S. Department of Health and Human Services (DHHS) created a national advisory committee to establish a process for recommending disorders that should be included in screening (Kemper et al., 2014). Committee deliberations inform decisions about the DHHS Recommended Uniform Screening Panel (RUSP). States are not required to adopt the RUSP, but most

eventually do and the Committee's work has led to great (although not complete) harmonization of NBS program targets. Other federal agencies play important roles in NBS. The Health Resources and Services Administration supports aspects of technical assistance and national data collection efforts. The Centers for Disease Control and Prevention (CDC) provides technical support and competitive funding for state laboratories so that screening methods are accurate and consistent, and the National Institutes of Health supports pilot studies and other research on new treatments, technologies, and outcome measures. Ultimately, however, NBS is a state-based initiative, and all NBS programs operate under the auspice of the state's health department.

PROGRAM ENTRY AND ELIGIBILITY

Children enter EI and NBS in substantially different ways. Most children enter EI because they show signs of a delay or have an obvious risk factor. Newborn screening screens children before any symptoms are apparent.

Early intervention

Similar to special education programs for older children with disabilities, EI relies on categorical eligibility for services. Entry into EI requires that a child either exhibit a developmental delay or be at risk for one. IDEA requires that all states participating in Part C serve children with developmental delays (as documented by standardized developmental assessments) and children with "established conditions" likely to lead to a developmental delay. For example, a newborn with Down syndrome may not exhibit delays during the early months of life but almost certainly will experience delayed development and thus is immediately eligible for services. Eligibility is determined by a comprehensive multidisciplinary assessment. States have the option to serve "high-risk" children, those children with medical (e.g., low birth weight) or other risk factors that might lead to delay, though very few do. States have the authority to determine the criteria used to define developmental delay, what conditions are "established" disorders, whether to serve high-risk children, and the factors that constitute high risk. Currently, more than 400,000 infants and toddlers are served in EI programs across the nation (U.S. Department of Education, 2020). This figure represents a little over 3% of the national population, but there is considerable cross-state variability, ranging from a low of less than 1% of children to a high of more than 8%. More than half of the children served in EI are in the 2–3 years age range, as would be expected, given that eligibility is heavily dependent on demonstration of a developmental delay. Only about 12% of children are in the birth to 12-month age range.

Newborn screening

Newborn screening focuses on specific genetic or metabolic disorders and is offered by all U.S. states and territories. There are no eligibility requirements for NBS, a universally available program for all of the approximately 4,000,000 infants born in the United States each year. Most NBS is done without consent, and although many states allow parents to decline screening for religious reasons, nationally more than 97% of children are screened each year. Currently, there are 35 disorders on the RUSP, including metabolic (e.g., PKU), endocrine (e.g., primary congenital hyperthyroidism), hemoglobin (e.g., sickle cell disease), and other disorders (e.g., cystic fibrosis, hearing loss, spinal muscular atrophy). Another 26 "secondary conditions" are also recommended for screening, disorders that are automatically detected when screening for the 35 core conditions. Thus, NBS and EI differ fundamentally in their approach to identifying eligible children. In EI, screening and eligibility begin *after* a problem is suspected. In contrast, NBS is specifically designed to identify children *before* symptoms appear, on the assumption that presymptomatic treatments are more

efficacious than treatments initiated after clinical manifestation.

SERVICE MODELS

Early intervention

Eligibility determination is necessary for entry into EI, but EI is primarily a system of services. Some forms of screening and case finding can occur, but these are not usually population-based efforts, focusing instead on children with one or more risk factors. Numerous instruments have been developed for presymptomatic screening at the earliest possible age, but clear behavioral markers for neurodevelopmental disorders have proved elusive (Micai et al., 2020); thus, the focus is primarily on testing children with suspected delays. Eligible children are referred to the local service entity; a multidisciplinary team of professionals works with the family to develop an Individualized Family Service Plan (IFSP), which includes information about the child's strengths and needs, family resources, priorities and concerns, services to be provided, and outcomes expected. A service coordinator is assigned and is responsible for ensuring that specified services are provided.

Almost all EI services are home based (U.S. Department of Education, 2020), typically two to four visits per month. Services are individualized on the basis of child needs and family preferences and could include speech and language therapy, physical therapy, occupational therapy, or early childhood special education. Often one team member is designated as the primary service provider, who consults with other team members and integrates their input during the home visit. Certain rights and privileges are afforded to parents, and due process procedures are in place for families who believe they have not received adequate services, but only a small number of complaints are filed.

Newborn screening

In contrast to EI, NBS is first and foremost a program to identify children with disorders that are on the state's NBS panel. Most of the disorders screened are tested by state laboratories using a dried blood spot collected in every hospital. Spots are sent to a central laboratory facility and tested within a week to 10 days, especially for disorders that are time sensitive (urgency of early treatments). Two disorders—hearing loss and critical congenital heart disease (CCHD)—are tested in the hospital, because they do not rely on blood spot testing. Screening tests undergo strict validation procedures, and each state establishes cutoff scores for "screen-positive" cases. Generally, states try to establish cutoff scores to ensure that all infants who might have a disorder are identified (no false-negatives), but using these cutoffs often means that there are some or even many false-positive cases—infants with a positive screen who do not actually have the disorder. As a result, screening is never considered diagnostic. For some disorders, states conduct a "second-tier" screen that can eliminate some false-positives, but final resolution of the remaining cases must be done through a diagnostic confirmation process. State laboratories do not provide diagnostic confirmation. Cases are referred to designated diagnostic and treatment centers (typically associated with a university or specialty hospital), where parents bring their children for additional tests, genetic counseling, and treatment recommendations. Usually, these services are coordinated with the family's pediatrician or other primary care physician. Unfortunately, because most disorders included in NBS are rare, primary care physicians may not be aware of what to do. The American College of Medical Genetics (www.acmg.net) has developed Act sheets that provide guidance on short-term actions, family communications, diagnostic procedures, and general treatment guidelines. Nonetheless, some disorders require highly specialized treatments, such as stem cell transplants and gene therapy, that must be provided by specialty clinics.

Annually, more than 6,500 infants are diagnosed with a disorder as a result of NBS, not counting those with hearing loss or CCHD

(state and national data on these disorders are more difficult to obtain, because screening for them is hospital based) (APHL, 2020). Newborn screening is often described as a system, with many different agencies and professionals involved at different points in the process (APHL, 2013). But once children receive a diagnosis, services become widely distributed and highly variable depending on the disorder. State laboratories typically do not have the resources or mandate to track what happens with families and children. Newborn screening does not have legislation specifying family rights and privileges, and there is no due process or dispute resolution procedure for families who are not satisfied with NBS services.

EVIDENCE, EFFICACY, AND OUTCOMES

Early intervention

The 1970s saw a strong push for evidence of the effectiveness of EI for infants and toddlers with disabilities, as a precursor to federal legislation. Primarily, this evidence drew on studies demonstrating the benefits of early education for infants and toddlers from low-income environments, and follow-up studies have shown that benefits of high-quality programs persist into the adult years (Campbell et al., 2012) and can have a long-term economic payoff (Garca et al., 2020). But rigorous studies demonstrating that EI is effective for infants and toddlers with disabilities are difficult to conduct; the disorders represented in EI vary immensely, and a no-treatment comparison group is virtually impossible. Thus, the evidence for EI as a program continues to draw on developmental science, neuroscience, and cumulative data showing the importance of high-quality early childhood experiences (Black et al., 2017; National Research Council Institute of Medicine Committee on Integrating the Science of Early Childhood Development, 2000). There is considerable emphasis on "evidence-based" practices in EI, but increasingly, it is recognized that EI is a system of complex and interrelated services for children and families (Guralnick, 2019). Instead of a discrete treatment or practice, more emphasis has been placed on core components that should underlie EI (e.g., family-centered practices, responsive teaching, routines-based interventions) or curriculum approaches (Juffer, Bakermans-Kranenburg, & van Ijzendoorn, 2008; Rogers et al., 2014). Each component and most curriculum models have a strong theoretical and evidence base, and collectively it is clear that, done well, EI can make a real difference in children's development and family well-being (Crnic, Neece, McIntyre, Blacher, & Baker, 2017; Guralnick, 2011). Randomized trials of selected curriculum models have been published (e.g., Green et al., 2017), but these studies typically are conducted in a research context and are rarely implemented with strong fidelity or with sufficient magnitude in typical intervention settings. It is not known whether the current minimal level of EI services is sufficient to achieve the potential that EI could have if fully implemented.

In contrast to efficacy studies, which test the effects of specific treatments or models, outcome studies focus on whether a broad program of services has achieved agreed-upon goals. In 1986, Congress established four broad goals for EI: (1) enhance the development of infants and toddlers with disabilities; (2) reduce educational costs by minimizing the need for special education; (3) minimize the likelihood of institutionalization and maximize independent living; and (4) enhance the capacity of families to meet their child's needs. Documenting ultimate attainment of these goals would be an enormous and long-term undertaking, and there has been no substantial effort to do so. Instead, the U.S. Department of Education established more immediate (but quite broad) outcomes for children and families. Following an extensive national effort to identify important child and family outcomes (Bailey, Bruder, et al., 2006; Barton, Taylor, Spiker, & Hebbeler, 2016), the Department established a set of

reporting requirements. States must report annually on three child outcomes and three family outcomes, as well as selected indicators of program quality (e.g., timely receipt of services). States may choose the method by which they gather outcome data, but the data must be reported in a common format. For example, states are expected to report "of those children who entered the program below age expectations in each outcome, the percent who substantially increased their rate of growth by the time they turned 3 years of age or exited the program." In the most recent reporting year, the national data summarized by the ECTA (2019) report the following average percentages of children who have increased their rate of growth in the three child outcomes:

- Have positive social-emotional skills (57%);
- Acquire and use knowledge and skills (71%); and
- Use appropriate behaviors to meet needs (74%).

Comparable data report the following percentages of families who agree that EI services have helped them:

- Know their rights (90%);
- Effectively communicate their child's needs (91%); and
- Help their children develop and learn (92%).

Newborn screening

The efficacy of NBS has never been evaluated systematically as a broad enterprise, despite being lauded as one of the 10 great public health achievements of 2001–2010 (CDC, 2011). Almost all evidence for NBS efficacy is condition specific, often in the form of clinical trials testing the efficacy of diet, drugs, hematopoietic stem cell transplantation, or gene therapy in the treatment of specific disorders (De Vivo et al., 2019; Grosse, Lam, Wiggins, & Kemper, 2017; Kemper et al., 2017). The evidence review commissioned by the Secretary's Advisory Committee considers, among other factors (e.g., screening test accuracy), the treatment evidence in support of early identification, and the Committee makes a summary judgment about the level of certainty and benefit. Typically, the Committee recommends a disorder for the RUSP if there is "high certainty that screening for the targeted condition would lead to a significant net benefit" (Kemper et al., 2014). Almost always this evidence is based on short-term (1–3 years) outcomes, primarily because of the cost and other challenges inherent in collecting long-term data on rare disorders.

Beginning in 2012, the DHHS began funding the APHL to establish the Newborn Screening Technical Assistance and Evaluation Program (NewSTEPS) (Ojodu et al., 2018). NewSTEPS works with states to gather data on the national status of NBS and provides annual reports summarizing the number of babies screened by state and other state-level data (e.g., use of couriers, operating hours, short- and long-term follow-up) (APHL, 2013). A panel of expert stakeholders identified eight quality indicators that are tracked annually at a national level: (1) percentage of unacceptable dried blood spot specimens; (2) percentage of dried blood spot specimens with at least one missing data field; (3) percentage of eligible newborns not receiving a newborn screen; (4) percentage of infants who have no recorded final resolution or confirmed diagnosis; (5) timeliness of NBS activities; (6) percentage of infants with an out-of-range screening result requiring clinical diagnostic workup; (7) percentage of disorders with a confirmed diagnosis by an appropriate medical professional; and (8) percentage of missed cases by disorder (Ojodu et al., 2018). Some of these data have been relatively easy to document (e.g., timeliness, confirmed diagnoses, percentage of newborns not receiving a newborn screen), but others (e.g., percentage of missed cases) are more difficult to obtain. National reporting efforts for NBS almost entirely focus on system performance indicators and not on child or family outcomes.

FAMILIES

Early intervention

Although EI was initially designed primarily to benefit children, over the past 40 years, there has been increasing emphasis on the importance of families, shifting from a view of families as recipients of professional services to help children to a view of families as partners in a community-based, coordinated, family-centered service model (Brewer, McPherson, Magrab, & Hutchins, 1989; Brooks-Gunn, Berlin, & Fuligni, 2000). Families have rights and privileges, ought to be considered as partners in the planning process and ultimate decision-makers about the focus of services, and their outcomes as a result of EI should also be considered. This perspective has played out in different ways over recent decades, including the IFSP and a mandate that EI services should be provided in "natural environments," usually described as the home environment, where most EI services occur. A national effort was launched to reach consensus on family outcomes of EI (Bailey, Bruder, et al., 2006), resulting in the addition of family outcomes to national reporting systems (although these outcomes focus primarily on perceived helpfulness of services). Implementing family-centered practices in EI has proved to be a challenge, but virtually every publication about EI systems and approaches rests fundamentally on the assumption that family processes and child outcomes are inextricably linked and that any service approach that does not recognize these links will compromise effectiveness (Crnic et al., 2017; Guralnick, 2019).

Newborn screening

Although early publications in maternal and child health provided the foundational arguments for a family-centered approach to services (Brewer et al., 1989), for the most part these assumptions have not been embraced by the NBS community. Newborn screening is considered a public health emergency, so urgent that for the most part screening is done without consent. After some debate about whether family benefit should be a part of NBS, a 1994 report by the Institute of Medicine stated that genetic testing of infants should only be done to benefit the child and that "a person should not be used as a means for the benefit of others" (Andrews, Fullarton, Holtzman, & Motulsky, 1994). Subsequent reports reiterated this position (see Bailey, Beskow, Davis, & Skinner, 2006, for a summary), and although early guidelines for "scoring" conditions for NBS suitability included points for family and societal benefits (American College of Medical Genetics Newborn Screening Expert Group, 2006), those were soon removed from consideration. RUSP deliberations by the Advisory Committee on Heritable Disorders in Newborns and Children focus entirely on benefit to the child. But the debate continues, and expanded genetic testing will only exacerbate the issue, because broad-based genomic screening detects heritable disorders and carrier status. In a recent article, Ross and Clayton (2019), responding to an NBS sequencing study in which a child was found to carry a *BRCA2* mutation (elevated risk for breast cancer), state, "We reject family benefit as a moral reason to expand genomic sequencing of children beyond conditions that present in childhood" (p. 1). In addition to the moral concerns raised by this argument, there is certainly some fear in the NBS community that if family benefit were to become a primary litmus test for new disorders, it would open the door to far too many disorders than NBS programs could handle. Regardless, NBS provides a clear benefit to families, eliminating an otherwise long and frustrating search for a diagnosis and providing timely treatments of great benefit to their child.

FINANCING

Early intervention

The cost of EI is paid from a variety of sources. The federal government, through

Part C of IDEA, gives formula grants (based on state census) to each state to assist in implementing a statewide system of services. In FY 2019, the total grant pool was $470,000,000, or about $1,148 per child served. Although a substantial federal investment, these funds fall far short of the cost of EI. Most states supplement federal funding with state funds and have established a system of payments and billing that draws on multiple sources, including private insurance, the state's Children's Health Insurance Program, and Medicaid.

Newborn screening

The collection of dried blood spots and state laboratory screening are usually paid for by a combination of state funding and fee collection. Hospitals typically either bill insurance companies directly for NBS or that cost is included in the total labor and delivery charge. Considerable cross-state variability exists in fees charged by NBS programs. In 2019, fees ranged from $0 to $203 (APHL, 2020), not only because of the number of disorders screened, but also because of state policy on funding. Fees can cover the cost of screening and subsequent diagnosis and short-term follow-up, but treatment is not covered by NBS fees. Children with a diagnosed NBS condition are seen by either a primary physician or a specialty physician, who works with the family to determine the best course of treatment. The cost of these treatments is paid for by private insurance, Medicaid, and parent co-pays. Treatment costs can vary widely across disorders. Some disorders, such as PKU, require special medical food or food supplement, the cost of which may or may not be covered by insurance. Other disorders, such as severe combined immunodeficiency or spinal muscular atrophy, are treated through targeted therapeutics, stem cell transplants, or gene therapy, and those costs could extend to hundreds of thousands or even millions of dollars per child. The Affordable Care Act requires most health care plans to cover the costs of treatment for any disorder included on the RUSP. But in many states, this cost must be borne by Medicaid for a large percentage of eligible children.

REFLECTIONS AND RECOMMENDATIONS

Both EI and NBS are well-established programs designed to support children with special health care needs or disabilities. They share a simple but powerful underlying assumption: The best way to prevent or lessen the impact of a disorder or disability is to begin treatment as early as possible. But because they differ considerably in history, auspice, focus, financing, and approach, the two programs operate in virtually independent spheres. Newborn screening begins with a hospital heel prick of virtually every newborn, followed by rapid screening (usually within a week) in a state public health laboratory. Primary care physicians and specialty clinics follow up with "screen-positive" children to confirm the presence or absence of a disorder and begin disorder-specific medical treatments as soon as possible. In contrast, EI typically begins with the referral of a child with suspected developmental delays. An interdisciplinary team conducts a developmental evaluation to determine if the delay is sufficient to make a child eligible for EI services. Eligible children are referred to the local EI program, an IFSP is developed, and services begin, but unless a child has a condition that is obvious at birth, most children with disabilities will not enter EI until the second or third year of life.

Both EI and NBS face their own unique challenges related to program organization, quality assurance, and growth. But both programs must address at least four cross-cutting issues in the very near future: (a) documenting meaningful outcomes for children and families; (b) deciding whether and how to provide highly effective yet very expensive treatments; (c) implementing surveillance activities to monitor progress and identify treatment needs; and (d) identifying appropriate intersections between EI and NBS.

Documenting meaningful outcomes for children and families

Are EI and NBS achieving the outcomes for which they were intended? Annual reporting by states provides information on variables such as the number of children served, satisfaction with certain aspects of the program, and quality/performance indicators. But these data provide only a limited view of program success. For example, we can guess at the number of lives saved annually by NBS, but we cannot say anything about surviving children's health status, certainly not at the national level or over the long term. Lack of consensus on outcomes represents a significant barrier to a national assessment system. The magnitude of this problem was highlighted in a recent review of outcomes studied for just two of the disorders currently included in NBS, finding 97 different outcomes studied for PKU and 83 different outcomes for medium-chain acyl-CoA dehydrogenase deficiency (Pugliese et al., 2020). The American College of Medical Genetics oversees a Longitudinal Pediatric Data Resource (LPDR) funded by the National Institutes of Health, identifying condition-specific core elements that should be assessed and providing a mechanism by which researchers can enter data on a secure portal that can be accessed for research purposes. The LPDR fills a critical gap in data, but it does not generate national reports.

Early intervention has similar challenges. There are a few studies of specific curricula, methods, or models, but national data collection is limited to annual reporting on the three child outcomes described earlier and only at a superficial level. What does it really mean to say "As a result of EI, 57% of participating children have increased their rate of growth during the past year?" especially when each state has its own model for how to collect these data? Family outcomes are reported, but only the perceived helpfulness of EI, not actual outcomes.

Three initiatives are needed to determine the real benefits of EI and NBS. First, both EI and NBS need to engage multiple stakeholders (parents, policy makers, clinicians, researchers) to establish a meaningful set of measurable outcomes for children, going beyond morbidity and mortality to assess health status, functional abilities, associated disorders, and quality of life. Although it will be tempting to individualize data for each disorder, this effort should focus on more generalizable outcomes applicable across disorders and across programs. A comparable effort is needed for family outcomes. For example, we led an evidence-based process with substantial stakeholder input to recommend five outcomes for families participating in EI: understand their child's strengths, abilities, and special needs; know their rights and advocate effectively for their child; help their child develop and learn; have support systems; and gain access to desired services and activities in their community (Bailey, Bruder, et al., 2006). These outcomes should be reexamined for EI and can serve as a starting point to identify family outcomes for NBS. Much work remains to be done before family benefit becomes accepted as a core consideration in NBS, but focused efforts to identify potential family outcomes would be an important first step.

Second, both EI and NBS need to build on the resulting consensus outcome frameworks to develop and implement an annual report of outcomes for all children and families participating in these programs. Organizing and supporting such an initiative will require significant investment, national leadership, and state buy-in, but establishing a national reporting system is the only way to determine whether EI and NBS are providing demonstrable benefits and could lead to meaningful changes if important outcomes are not achieved at an acceptable level. Numerous working groups have bemoaned the lack of coordinated short- and long-term follow-up data systems at the state and national levels (e.g., Hinton et al., 2011). Pilot studies have shown the benefit of a public health-tracking system for NBS follow-up (Hinton et al., 2014), but much work remains to be done before such data are available.

Third, both EI and NBS need to identify policy questions for which systematic data are essential to inform decision making. These questions should form the basis for an ongoing series of nationally representative studies that (a) measure a broad range of child and family outcomes using robust and valid assessment procedures and (b) systematically examine factors associated with outcome attainment, as was the case with the National Early Intervention Longitudinal Study (NEILS) (Hebbler et al., 2007). Although NEILS yielded important information about services and outcomes (e.g., Bailey, Hebbeler, Scarborough, Spiker, & Mallik, 2004; Bailey, Hebbeler, et al., 2005; Bailey, Nelson, Hebbeler, & Spiker, 2007), the information is dated. No comparable study has been conducted for NBS. Such an endeavor will be expensive but needs to be rigorous and conducted in a way that takes into consideration the multiple factors likely to influence outcomes for children and families.

Deciding whether and how to provide highly effective yet costly treatments

Maximizing the attainment of desired outcomes for EI and NBS has been the focus of considerable research over the past 30 years. As new knowledge about effective treatments develops, programs are faced with the important but challenging question of whether, when, and how to provide treatments that substantially improve outcomes but at a far greater cost. And with escalating costs questions about equitable access for all families become more salient. For NBS, notable breakthroughs in medical research have led to a new generation of treatments that dramatically improve outcomes for children—hematopoietic stem cell transplantation, targeted therapeutics to replace missing proteins, and gene therapy to replace defective genes with normally functioning genes. These new treatments save lives and restore function in ways previously unimagined, but at great cost. For example, a gene therapy recently approved to treat spinal muscular atrophy comes with a $2 million price tag. Currently, much of this cost is covered by insurance or Medicaid, but literally hundreds of disorders could ultimately benefit from gene therapy.

Because the primary treatment modality in EI is the provision of specialized education or therapy, treatment advances have been less dramatic, and in practice, EI has changed little in the past 30 years. Limited program capacity and the extensive processes required to determine eligibility mean that the gap between initial concerns about development or behavior and onset of services can be months or even a year or more (Bailey et al., 2004; Conroy et al., 2018). Even then the services provided typically are limited to a 1-hr home visit two to four times per month. In contrast, an accumulated evidence base suggests that EI can be effective, but it must start early, be of sufficient intensity, and incorporate evidence-based treatments embedded within a developmental systems approach to be effective (Crnic et al., 2017; Guralnick, 2019), none of which characterizes most community-based EI systems (Bruder et al., 2020).

Both EI and NBS will need to determine the value of substantially increased investments in treatments relative to desired outcomes. Newborn screening has the backing of industry and government to rigorously test treatment safety and efficacy and a financing system that, at least for the time being, can pay for new therapies. But despite analyses showing that even very expensive treatments such as gene therapy or stem cell transplants can be cost-effective (Jalali et al., 2020), the spiraling cost of specialized (but highly effective) treatments presents a sustainability challenge to health care financing for NBS (Prosser, 2018).

Early intervention has no such support, and it is difficult to imagine what it would take to provide the level and type of services that will lead to significant and sustained improvement in outcomes for children with disabilities. Research will provide part of the answer, but serious advocacy efforts and intentional policy analyses will be required before EI

can achieve the full promise of benefit for children and families.

The role of surveillance in monitoring progress and determining when treatment should begin

Surveillance refers to a systematic effort to monitor children's developmental progress and health status, on the assumption that by doing so clinicians will know when treatments should begin. The need for surveillance in NBS is exemplified by disorders for which it is not clear whether or how symptoms will appear. For example, X-linked adrenoleukodystrophy requires an annual magnetic resonance scan of the brain to detect signs of disease onset and decide whether stem cell transplantation should be initiated. Many of the newer disorders included in NBS, such as spinal muscular atrophy, have a primary, severe version that is evident in the newborn period and requires immediate treatment, as well as less severe versions with milder impact or later onset for which the timing or necessity of treatment is less clear. Early intervention programs also face surveillance challenges—siblings of children with autism, premature infants, infants experiencing neonatal abstinence syndrome, children in high-risk environments, and children with genetic mutations that may or may not lead to developmental delay all could benefit from systematic surveillance.

Despite the potential benefits of surveillance, implementation remains a challenge. The agency or clinician who provides treatments or services is not necessarily the same as those responsible for surveillance activities, and linkages between the two are not always seamless. Surveillance can extend over a long period of time, and the findings that trigger the need for treatment may not be well defined. Surveillance can potentially serve as a surrogate for services, and parents might grow weary of surveillance activities over time and be less likely to participate upon repeated findings of "no problem." A systems analysis of surveillance models is needed for both EI and NBS. The analysis should cross multiple disorders, describe the contexts in which surveillance is most critical, identify the biggest challenges to effective surveillance, develop alternative surveillance models, and test their effectiveness.

IDENTIFYING APPROPRIATE INTERSECTIONS BETWEEN EI AND NBS

Early intervention currently plays no role in deliberations about net benefit for conditions nominated for NBS. This will come as a surprise to professionals in the EI community but certainly has been apparent to parents for many years. A partial explanation is that EI is not considered a "treatment" in a world that focuses primarily on medical interventions. And in reality, EI simply cannot produce the kind of data or demonstrate the magnitude of effect that is expected of medical treatments and certainly not for individual disorders.

But virtually none of the current treatments of conditions identified through NBS would be considered a cure. In fact, most children need a lifetime of surveillance (to monitor disease progression or changes in response to treatment), dietary adherence (as in the case of PKU), augmentation (as in the case of hearing impairment), or medical treatments (as in the case of annual intrathecal injections for children with spinal muscular atrophy). Many children identified by NBS would likely qualify for EI services (Grosse et al., 2017), sometimes even experiencing moderate to severe developmental delays (Wasserstein et al., 2016). Early intervention could play a critical role in enhancing the development of children identified through NBS, preventing secondary complications (through therapies provided by allied health professionals), and providing essential support for families (Bailey, Skinner, & Warren, 2005).

A national assessment is needed to determine the extent to which children with NBS disorders are referred for EI, how their services differ from those who are eligible through the usual developmental delay criteria, and whether and how they benefit from EI services. Such an assessment could

lead to substantial program improvements that would benefit both children and families. Importantly, likely eligibility for EI and the potential helpfulness of EI services ought to be built in as a formal part of RUSP deliberations to determine net benefit for decisions under consideration for NBS.

CONCLUSION

Both EI and NBS have a shared vision to improve outcomes for very young children with special needs. Notable achievements are evident in both programs, now well entrenched in health and education. But despite their many successes, EI and NBS programs face critical challenges that require substantial rethinking to ensure long-term success. Many children identified and treated through NBS would benefit from EI, but linkages between the two programs vary considerably across states. National leadership and interagency collaboration at both the national and state levels will be needed, as well as contributions from parents, advocates, industry, professional associations, and researchers. Although much can be accomplished within each program, it will be important to envision possible synergies that could emerge if EI and NBS could work together to ensure the best possible future for young children and their families.

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